

background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25122] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1619 correlate with, and may be deduced from, the identity of the target genes which GAM1619 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25123] Nucleotide sequences of the GAM1619 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1619 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1619 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1619 are further described hereinbelow with reference to Table Table1.

[25124] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of

Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1619 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25125] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1620 (GAM1620) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25126] GAM1620 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1620 was detected is described hereinabove with reference to Figs. 2–8.

[25127] GAM1620 gene, herein designated GAM GENE, and GAM1620 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25128] GAM1620 gene, herein designated GAM GENE, encodes a GAM1620 precursor RNA, herein designated GAM PRECURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1620 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to

the nucleotide sequence of GAM1620 precursor RNA is designated SEQ ID:1593, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1593 is located at position 4851368 relative to contig NT_022517.13, on chromosome 3.

[25129] GAM1620 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1620 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25130] An enzyme complex designated DICER COMPLEX, dices the GAM1620 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1620 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other

necessary proteins. A probable (over 89%) nucleotide sequence of GAM1620 RNA is designated SEQ ID:3269, and is provided hereinbelow with reference to the sequence listing part.

- [25131] GAM1620 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1620 target RNA, herein designated GAM TARGET RNA. GAM1620 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25132] GAM1620 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1620 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1620 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target

binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1620 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1620 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25133] The complementary binding of GAM1620 RNA, herein designated GAM RNA, to target binding sites on GAM1620 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1620 target RNA, herein designated GAM TARGET RNA, into GAM1620 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25134] It is appreciated that GAM1620 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1620 target genes. The mRNA of each one of this plurality of GAM1620 target genes comprises one or more target binding sites, each having a nucleotide sequence

which is at least partly complementary to GAM1620 RNA, herein designated GAM RNA, and which when bound by GAM1620 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1620 target proteins.

[25135] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1620 gene, herein designated GAM GENE, on one or more GAM1620 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25136] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1620 correlate with, and may

be deduced from, the identity of the target genes which GAM1620 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25137] Nucleotide sequences of the GAM1620 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1620 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1620 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1620 are further described hereinbelow with reference to Table Table1.

[25138] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1620 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25139] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1621 (GAM1621) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25140] GAM1621 is a novel bioinformatically detected regulatory,

non protein coding, micro RNA (miRNA) gene. The method by which GAM1621 was detected is described hereinabove with reference to Figs. 2–8.

[25141] GAM1621 gene, herein designated GAM GENE, and GAM1621 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25142] GAM1621 gene, herein designated GAM GENE, encodes a GAM1621 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1621 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1621 precursor RNA is designated SEQ ID:1594, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1594 is located at position 11570630 relative to contig NT_008470.13, on chromosome 9.

[25143] GAM1621 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1621 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the

fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25144] An enzyme complex designated DICER COMPLEX, dices the GAM1621 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1621 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1621 RNA is designated SEQ ID:3270, and is provided hereinbelow with reference to the sequence listing part.

[25145] GAM1621 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1621 target RNA, herein designated GAM TARGET RNA. GAM1621 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, design-

nated 5UTR, PROTEIN CODING and 3UTR respectively.

[25146] GAM1621 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1621 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1621 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1621 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1621 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25147] The complementary binding of GAM1621 RNA, herein

designated GAM RNA, to target binding sites on GAM1621 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1621 target RNA, herein designated GAM TARGET RNA, into GAM1621 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25148] It is appreciated that GAM1621 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1621 target genes. The mRNA of each one of this plurality of GAM1621 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1621 RNA, herein designated GAM RNA, and which when bound by GAM1621 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1621 target proteins.

[25149] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1621 gene, herein designated GAM GENE, on one or more GAM1621 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA

genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25150] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1621 correlate with, and may be deduced from, the identity of the target genes which GAM1621 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25151] Nucleotide sequences of the GAM1621 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1621 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1621 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1621 are further described hereinbelow with reference to Table Table1.

[25152] Nucleotide sequences of target binding sites, such as

BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1621 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25153] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1622 (GAM1622) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25154] GAM1622 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1622 was detected is described hereinabove with reference to Figs. 2-8.

[25155] GAM1622 gene, herein designated GAM GENE, and GAM1622 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25156] GAM1622 gene, herein designated GAM GENE, encodes a GAM1622 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1622 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a pro-

tein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1622 precursor RNA is designated SEQ ID:1595, and is provided hereinbelow with reference to the sequence listing part.

[25157] GAM1622 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1622 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25158] An enzyme complex designated DICER COMPLEX, dices the GAM1622 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1622 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide se-

quence of GAM1622 RNA is designated SEQ ID:3271, and is provided hereinbelow with reference to the sequence listing part.

- [25159] GAM1622 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1622 target RNA, herein designated GAM TARGET RNA. GAM1622 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25160] GAM1622 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1622 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1622 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration

only, and is not meant to be limiting GAM1622 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1622 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25161] The complementary binding of GAM1622 RNA, herein designated GAM RNA, to target binding sites on GAM1622 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1622 target RNA, herein designated GAM TARGET RNA, into GAM1622 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25162] It is appreciated that GAM1622 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1622 target genes. The mRNA of each one of this plurality of GAM1622 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1622 RNA,

herein designated GAM RNA, and which when bound by GAM1622 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1622 target proteins.

[25163] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1622 gene, herein designated GAM GENE, on one or more GAM1622 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25164] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1622 correlate with, and may be deduced from, the identity of the target genes which

GAM1622 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25165] Nucleotide sequences of the GAM1622 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1622 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1622 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1622 are further described hereinbelow with reference to Table Table1.

[25166] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1622 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25167] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1623 (GAM1623) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25168] GAM1623 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method

by which GAM1623 was detected is described hereinabove with reference to Figs. 2–8.

[25169] GAM1623 gene, herein designated GAM GENE, and GAM1623 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25170] GAM1623 gene, herein designated GAM GENE, encodes a GAM1623 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1623 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1623 precursor RNA is designated SEQ ID:1596, and is provided hereinbelow with reference to the sequence listing part.

[25171] GAM1623 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1623 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of

the second half thereof.

- [25172] An enzyme complex designated DICER COMPLEX, dices the GAM1623 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1623 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1623 RNA is designated SEQ ID:3272, and is provided hereinbelow with reference to the sequence listing part.
- [25173] GAM1623 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1623 target RNA, herein designated GAM TARGET RNA. GAM1623 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25174] GAM1623 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in

untranslated regions of GAM1623 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1623 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1623 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1623 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25175] The complementary binding of GAM1623 RNA, herein designated GAM RNA, to target binding sites on GAM1623 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, in-

hibits translation of GAM1623 target RNA, herein designated GAM TARGET RNA, into GAM1623 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25176] It is appreciated that GAM1623 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1623 target genes. The mRNA of each one of this plurality of GAM1623 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1623 RNA, herein designated GAM RNA, and which when bound by GAM1623 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1623 target proteins.

[25177] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1623 gene, herein designated GAM GENE, on one or more GAM1623 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the

known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25178] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1623 correlate with, and may be deduced from, the identity of the target genes which GAM1623 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25179] Nucleotide sequences of the GAM1623 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1623 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1623 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1623 are further described hereinbelow with reference to Table Table1.

[25180] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to

GAM1623 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25181] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1624 (GAM1624) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25182] GAM1624 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1624 was detected is described hereinabove with reference to Figs. 2–8.

[25183] GAM1624 gene, herein designated GAM GENE, and GAM1624 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25184] GAM1624 gene, herein designated GAM GENE, encodes a GAM1624 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1624 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1624 precursor RNA is designated SEQ ID:1597, and is provided hereinbelow with

reference to the sequence listing part. Nucleotide sequence SEQ ID:1597 is located at position 2349674 relative to contig NT_021907.13, on chromosome 1.

[25185] GAM1624 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1624 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25186] An enzyme complex designated DICER COMPLEX, dices the GAM1624 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1624 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1624 RNA is designated SEQ ID:3273, and

is provided hereinbelow with reference to the sequence listing part.

- [25187] GAM1624 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1624 target RNA, herein designated GAM TARGET RNA. GAM1624 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25188] GAM1624 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1624 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1624 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1624 RNA,

herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1624 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25189] The complementary binding of GAM1624 RNA, herein designated GAM RNA, to target binding sites on GAM1624 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1624 target RNA, herein designated GAM TARGET RNA, into GAM1624 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25190] It is appreciated that GAM1624 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1624 target genes. The mRNA of each one of this plurality of GAM1624 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1624 RNA, herein designated GAM RNA, and which when bound by

GAM1624 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1624 target proteins.

[25191] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1624 gene, herein designated GAM GENE, on one or more GAM1624 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25192] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1624 correlate with, and may be deduced from, the identity of the target genes which GAM1624 binds and inhibits, and the function of these

target genes, as elaborated hereinbelow.

[25193] Nucleotide sequences of the GAM1624 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1624 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1624 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1624 are further described hereinbelow with reference to Table Table1.

[25194] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1624 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25195] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1625 (GAM1625) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25196] GAM1625 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1625 was detected is described hereinabove

with reference to Figs. 2–8.

[25197] GAM1625 gene, herein designated GAM GENE, and GAM1625 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25198] GAM1625 gene, herein designated GAM GENE, encodes a GAM1625 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1625 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1625 precursor RNA is designated SEQ ID:1598, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1598 is located at position 56616163 relative to contig NT_026437.9, on chromosome 14.

[25199] GAM1625 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1625 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial

inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25200] An enzyme complex designated DICER COMPLEX, dices the GAM1625 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1625 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1625 RNA is designated SEQ ID:3274, and is provided hereinbelow with reference to the sequence listing part.

[25201] GAM1625 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1625 target RNA, herein designated GAM TARGET RNA. GAM1625 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25202] GAM1625 RNA, herein designated GAM RNA, binds com-

plementarily to one or more target binding sites located in untranslated regions of GAM1625 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1625 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1625 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1625 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25203] The complementary binding of GAM1625 RNA, herein designated GAM RNA, to target binding sites on GAM1625 target RNA, herein designated GAM TARGET RNA, such as

BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1625 target RNA, herein designated GAM TARGET RNA, into GAM1625 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25204] It is appreciated that GAM1625 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1625 target genes. The mRNA of each one of this plurality of GAM1625 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1625 RNA, herein designated GAM RNA, and which when bound by GAM1625 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1625 target proteins.

[25205] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1625 gene, herein designated GAM GENE, on one or more GAM1625 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary

binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25206] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1625 correlate with, and may be deduced from, the identity of the target genes which GAM1625 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25207] Nucleotide sequences of the GAM1625 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1625 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1625 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1625 are further described hereinbelow with reference to Table Table1.

[25208] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the

complementarity of each of these target binding sites to GAM1625 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25209] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1626 (GAM1626) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25210] GAM1626 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1626 was detected is described hereinabove with reference to Figs. 2–8.

[25211] GAM1626 gene, herein designated GAM GENE, and GAM1626 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25212] GAM1626 gene, herein designated GAM GENE, encodes a GAM1626 precursor RNA, herein designated GAM PRECURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1626 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1626 precursor RNA is

designated SEQ ID:1599, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1599 is located at position 4540528 relative to contig NT_009775.11, on chromosome 12.

[25213] GAM1626 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1626 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25214] An enzyme complex designated DICER COMPLEX, dices the GAM1626 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1626 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide se-

quence of GAM1626 RNA is designated SEQ ID:3275, and is provided hereinbelow with reference to the sequence listing part.

- [25215] GAM1626 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1626 target RNA, herein designated GAM TARGET RNA. GAM1626 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25216] GAM1626 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1626 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1626 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration

only, and is not meant to be limiting GAM1626 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1626 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25217] The complementary binding of GAM1626 RNA, herein designated GAM RNA, to target binding sites on GAM1626 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1626 target RNA, herein designated GAM TARGET RNA, into GAM1626 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25218] It is appreciated that GAM1626 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1626 target genes. The mRNA of each one of this plurality of GAM1626 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1626 RNA,

herein designated GAM RNA, and which when bound by GAM1626 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1626 target proteins.

[25219] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1626 gene, herein designated GAM GENE, on one or more GAM1626 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25220] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1626 correlate with, and may be deduced from, the identity of the target genes which

GAM1626 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25221] Nucleotide sequences of the GAM1626 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1626 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1626 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1626 are further described hereinbelow with reference to Table Table1.

[25222] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1626 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25223] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1627 (GAM1627) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25224] GAM1627 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method

by which GAM1627 was detected is described hereinabove with reference to Figs. 2–8.

[25225] GAM1627 gene, herein designated GAM GENE, and GAM1627 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25226] GAM1627 gene, herein designated GAM GENE, encodes a GAM1627 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1627 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1627 precursor RNA is designated SEQ ID:1600, and is provided hereinbelow with reference to the sequence listing part.

[25227] GAM1627 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1627 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of

the second half thereof.

[25228] An enzyme complex designated DICER COMPLEX, dices the GAM1627 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1627 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1627 RNA is designated SEQ ID:3276, and is provided hereinbelow with reference to the sequence listing part.

[25229] GAM1627 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1627 target RNA, herein designated GAM TARGET RNA. GAM1627 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25230] GAM1627 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in

untranslated regions of GAM1627 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1627 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1627 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1627 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25231] The complementary binding of GAM1627 RNA, herein designated GAM RNA, to target binding sites on GAM1627 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, in-

hibits translation of GAM1627 target RNA, herein designated GAM TARGET RNA, into GAM1627 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25232] It is appreciated that GAM1627 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1627 target genes. The mRNA of each one of this plurality of GAM1627 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1627 RNA, herein designated GAM RNA, and which when bound by GAM1627 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1627 target proteins.

[25233] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1627 gene, herein designated GAM GENE, on one or more GAM1627 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the

known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25234] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1627 correlate with, and may be deduced from, the identity of the target genes which GAM1627 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25235] Nucleotide sequences of the GAM1627 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1627 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1627 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1627 are further described hereinbelow with reference to Table Table1.

[25236] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to

GAM1627 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25237] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1628 (GAM1628) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25238] GAM1628 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1628 was detected is described hereinabove with reference to Figs. 2–8.

[25239] GAM1628 gene, herein designated GAM GENE, and GAM1628 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25240] GAM1628 gene, herein designated GAM GENE, encodes a GAM1628 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1628 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1628 precursor RNA is designated SEQ ID:1601, and is provided hereinbelow with

reference to the sequence listing part. Nucleotide sequence SEQ ID:1601 is located at position 280476 relative to contig NT_007758.8, on chromosome 7.

[25241] GAM1628 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1628 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25242] An enzyme complex designated DICER COMPLEX, dices the GAM1628 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1628 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1628 RNA is designated SEQ ID:3277, and

is provided hereinbelow with reference to the sequence listing part.

- [25243] GAM1628 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1628 target RNA, herein designated GAM TARGET RNA. GAM1628 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25244] GAM1628 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1628 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1628 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1628 RNA,

herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1628 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25245] The complementary binding of GAM1628 RNA, herein designated GAM RNA, to target binding sites on GAM1628 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1628 target RNA, herein designated GAM TARGET RNA, into GAM1628 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25246] It is appreciated that GAM1628 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1628 target genes. The mRNA of each one of this plurality of GAM1628 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1628 RNA, herein designated GAM RNA, and which when bound by

GAM1628 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1628 target proteins.

[25247] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1628 gene, herein designated GAM GENE, on one or more GAM1628 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25248] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1628 correlate with, and may be deduced from, the identity of the target genes which GAM1628 binds and inhibits, and the function of these

target genes, as elaborated hereinbelow.

[25249] Nucleotide sequences of the GAM1628 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1628 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1628 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1628 are further described hereinbelow with reference to Table Table1.

[25250] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1628 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25251] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1629 (GAM1629) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25252] GAM1629 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1629 was detected is described hereinabove

with reference to Figs. 2–8.

[25253] GAM1629 gene, herein designated GAM GENE, and GAM1629 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25254] GAM1629 gene, herein designated GAM GENE, encodes a GAM1629 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1629 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1629 precursor RNA is designated SEQ ID:1602, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1602 is located at position 15567257 relative to contig NT_007592.11, on chromosome 6.

[25255] GAM1629 has 20 highly conserved, expressed mouse homologs (>90% sequence similarity), found in mouse EST's. Nucleotide sequences similar to the nucleotide sequences of which mouse homologs, and the respective mouse EST's in which these homologs were detected are:

[25256] SEQ ID:99575 detected in mouse EST Accession BG092976.1.

[25257] SEQ ID:99576 detected in mouse EST Accession

AA798467.1.

[25258] SEQ ID:99577 detected in mouse EST Accession
BG147005.1.

[25259] SEQ ID:99578 detected in mouse EST Accession
BE311050.1.

[25260] SEQ ID:99579 detected in mouse EST Accession
BE625094.1.

[25261] SEQ ID:99580 detected in mouse EST Accession
BG914583.1.

[25262] SEQ ID:99581 detected in mouse EST Accession
BF784199.1.

[25263] SEQ ID:99582 detected in mouse EST Accession
BG975946.1.

[25264] SEQ ID:99583 detected in mouse EST Accession
BI689042.1.

[25265] SEQ ID:99584 detected in mouse EST Accession
BI696541.1.

[25266] SEQ ID:99585 detected in mouse EST Accession
BF140373.1.

[25267] SEQ ID:99589 detected in mouse EST Accession
AI194277.1.

[25268] SEQ ID:99590 detected in mouse EST Accession
BG803992.1.

[25269] SEQ ID:99592 detected in mouse EST Accession
BF465841.1.

[25270] SEQ ID:99593 detected in mouse EST Accession
AA798123.1.

[25271] SEQ ID:99594 detected in mouse EST Accession
BG148023.1.

[25272] SEQ ID:99595 detected in mouse EST Accession
AI316639.1.

[25273] SEQ ID:99596 detected in mouse EST Accession
BI659175.1.

[25274] SEQ ID:99597 detected in mouse EST Accession
BB067326.2.

[25275] SEQ ID:99598 detected in mouse EST Accession
BI080618.1.

[25276] GAM1629 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1629 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of

the second half thereof.

[25277] An enzyme complex designated DICER COMPLEX, dices the GAM1629 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1629 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1629 RNA is designated SEQ ID:3278, and is provided hereinbelow with reference to the sequence listing part.

[25278] GAM1629 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1629 target RNA, herein designated GAM TARGET RNA. GAM1629 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25279] GAM1629 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in

untranslated regions of GAM1629 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1629 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1629 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1629 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25280] The complementary binding of GAM1629 RNA, herein designated GAM RNA, to target binding sites on GAM1629 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, in-

hibits translation of GAM1629 target RNA, herein designated GAM TARGET RNA, into GAM1629 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25281] It is appreciated that GAM1629 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1629 target genes. The mRNA of each one of this plurality of GAM1629 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1629 RNA, herein designated GAM RNA, and which when bound by GAM1629 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1629 target proteins.

[25282] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1629 gene, herein designated GAM GENE, on one or more GAM1629 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the

known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25283] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1629 correlate with, and may be deduced from, the identity of the target genes which GAM1629 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25284] Nucleotide sequences of the GAM1629 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1629 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1629 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1629 are further described hereinbelow with reference to Table Table1.

[25285] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to

GAM1629 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25286] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1630 (GAM1630) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25287] GAM1630 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1630 was detected is described hereinabove with reference to Figs. 2–8.

[25288] GAM1630 gene, herein designated GAM GENE, and GAM1630 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25289] GAM1630 gene, herein designated GAM GENE, encodes a GAM1630 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1630 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1630 precursor RNA is designated SEQ ID:1603, and is provided hereinbelow with

reference to the sequence listing part. Nucleotide sequence SEQ ID:1603 is located at position 9039266 relative to contig NT_030059.8, on chromosome 10.

[25290] GAM1630 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1630 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25291] An enzyme complex designated DICER COMPLEX, dices the GAM1630 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1630 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1630 RNA is designated SEQ ID:3279, and

is provided hereinbelow with reference to the sequence listing part.

- [25292] GAM1630 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1630 target RNA, herein designated GAM TARGET RNA. GAM1630 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25293] GAM1630 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1630 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1630 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1630 RNA,

herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1630 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25294] The complementary binding of GAM1630 RNA, herein designated GAM RNA, to target binding sites on GAM1630 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1630 target RNA, herein designated GAM TARGET RNA, into GAM1630 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25295] It is appreciated that GAM1630 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1630 target genes. The mRNA of each one of this plurality of GAM1630 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1630 RNA, herein designated GAM RNA, and which when bound by

GAM1630 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1630 target proteins.

[25296] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1630 gene, herein designated GAM GENE, on one or more GAM1630 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25297] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1630 correlate with, and may be deduced from, the identity of the target genes which GAM1630 binds and inhibits, and the function of these

target genes, as elaborated hereinbelow.

[25298] Nucleotide sequences of the GAM1630 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1630 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1630 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1630 are further described hereinbelow with reference to Table Table1.

[25299] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1630 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25300] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1631 (GAM1631) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25301] GAM1631 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1631 was detected is described hereinabove

with reference to Figs. 2–8.

[25302] GAM1631 gene, herein designated GAM GENE, and GAM1631 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25303] GAM1631 gene, herein designated GAM GENE, encodes a GAM1631 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1631 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1631 precursor RNA is designated SEQ ID:1604, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1604 is located at position 2907543 relative to contig NT_015805.11, on chromosome 2.

[25304] GAM1631 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1631 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial

inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25305] An enzyme complex designated DICER COMPLEX, dices the GAM1631 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1631 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1631 RNA is designated SEQ ID:3280, and is provided hereinbelow with reference to the sequence listing part.

[25306] GAM1631 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1631 target RNA, herein designated GAM TARGET RNA. GAM1631 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25307] GAM1631 RNA, herein designated GAM RNA, binds com-

plementarily to one or more target binding sites located in untranslated regions of GAM1631 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1631 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1631 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1631 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25308] The complementary binding of GAM1631 RNA, herein designated GAM RNA, to target binding sites on GAM1631 target RNA, herein designated GAM TARGET RNA, such as

BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1631 target RNA, herein designated GAM TARGET RNA, into GAM1631 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25309] It is appreciated that GAM1631 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1631 target genes. The mRNA of each one of this plurality of GAM1631 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1631 RNA, herein designated GAM RNA, and which when bound by GAM1631 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1631 target proteins.

[25310] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1631 gene, herein designated GAM GENE, on one or more GAM1631 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary

binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25311] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1631 correlate with, and may be deduced from, the identity of the target genes which GAM1631 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25312] Nucleotide sequences of the GAM1631 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1631 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1631 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1631 are further described hereinbelow with reference to Table Table1.

[25313] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the

complementarity of each of these target binding sites to GAM1631 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25314] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1632 (GAM1632) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25315] GAM1632 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1632 was detected is described hereinabove with reference to Figs. 2–8.

[25316] GAM1632 gene, herein designated GAM GENE, and GAM1632 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25317] GAM1632 gene, herein designated GAM GENE, encodes a GAM1632 precursor RNA, herein designated GAM PRECURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1632 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1632 precursor RNA is

designated SEQ ID:1605, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1605 is located at position 2383351 relative to contig NT_035212.2, on chromosome 12.

[25318] GAM1632 precursor RNA, herein designated GAM PRECURSOR RNA, folds onto itself, forming GAM1632 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25319] An enzyme complex designated DICER COMPLEX, dices the GAM1632 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1632 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide se-

quence of GAM1632 RNA is designated SEQ ID:3281, and is provided hereinbelow with reference to the sequence listing part.

- [25320] GAM1632 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1632 target RNA, herein designated GAM TARGET RNA. GAM1632 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25321] GAM1632 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1632 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1632 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration

only, and is not meant to be limiting GAM1632 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1632 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25322] The complementary binding of GAM1632 RNA, herein designated GAM RNA, to target binding sites on GAM1632 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1632 target RNA, herein designated GAM TARGET RNA, into GAM1632 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25323] It is appreciated that GAM1632 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1632 target genes. The mRNA of each one of this plurality of GAM1632 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1632 RNA,

herein designated GAM RNA, and which when bound by GAM1632 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1632 target proteins.

[25324] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1632 gene, herein designated GAM GENE, on one or more GAM1632 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25325] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1632 correlate with, and may be deduced from, the identity of the target genes which

GAM1632 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25326] Nucleotide sequences of the GAM1632 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1632 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1632 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1632 are further described hereinbelow with reference to Table Table1.

[25327] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1632 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25328] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1633 (GAM1633) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25329] GAM1633 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method

by which GAM1633 was detected is described hereinabove with reference to Figs. 2–8.

[25330] GAM1633 gene, herein designated GAM GENE, and GAM1633 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25331] GAM1633 gene, herein designated GAM GENE, encodes a GAM1633 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1633 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1633 precursor RNA is designated SEQ ID:1606, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1606 is located at position 3436496 relative to contig NT_023148.9, on chromosome 5.

[25332] GAM1633 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1633 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the

RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25333] An enzyme complex designated DICER COMPLEX, dices the GAM1633 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1633 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1633 RNA is designated SEQ ID:3282, and is provided hereinbelow with reference to the sequence listing part.

[25334] GAM1633 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1633 target RNA, herein designated GAM TARGET RNA. GAM1633 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25335] GAM1633 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1633 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1633 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1633 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1633 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25336] The complementary binding of GAM1633 RNA, herein designated GAM RNA, to target binding sites on GAM1633

target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1633 target RNA, herein designated GAM TARGET RNA, into GAM1633 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25337] It is appreciated that GAM1633 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1633 target genes. The mRNA of each one of this plurality of GAM1633 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1633 RNA, herein designated GAM RNA, and which when bound by GAM1633 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1633 target proteins.

[25338] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1633 gene, herein designated GAM GENE, on one or more GAM1633 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the

background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25339] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1633 correlate with, and may be deduced from, the identity of the target genes which GAM1633 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25340] Nucleotide sequences of the GAM1633 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1633 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1633 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1633 are further described hereinbelow with reference to Table Table1.

[25341] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of

Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1633 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25342] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1634 (GAM1634) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25343] GAM1634 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1634 was detected is described hereinabove with reference to Figs. 2–8.

[25344] GAM1634 gene, herein designated GAM GENE, and GAM1634 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25345] GAM1634 gene, herein designated GAM GENE, encodes a GAM1634 precursor RNA, herein designated GAM PRECURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1634 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to

the nucleotide sequence of GAM1634 precursor RNA is designated SEQ ID:1607, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1607 is located at position 2349674 relative to contig NT_021907.13, on chromosome 1.

[25346] GAM1634 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1634 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25347] An enzyme complex designated DICER COMPLEX, dices the GAM1634 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1634 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other

necessary proteins. A probable (over 89%) nucleotide sequence of GAM1634 RNA is designated SEQ ID:3283, and is provided hereinbelow with reference to the sequence listing part.

- [25348] GAM1634 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1634 target RNA, herein designated GAM TARGET RNA. GAM1634 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25349] GAM1634 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1634 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1634 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target

binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1634 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1634 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25350] The complementary binding of GAM1634 RNA, herein designated GAM RNA, to target binding sites on GAM1634 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1634 target RNA, herein designated GAM TARGET RNA, into GAM1634 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25351] It is appreciated that GAM1634 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1634 target genes. The mRNA of each one of this plurality of GAM1634 target genes comprises one or more target binding sites, each having a nucleotide sequence

which is at least partly complementary to GAM1634 RNA, herein designated GAM RNA, and which when bound by GAM1634 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1634 target proteins.

[25352] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1634 gene, herein designated GAM GENE, on one or more GAM1634 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25353] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1634 correlate with, and may

be deduced from, the identity of the target genes which GAM1634 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25354] Nucleotide sequences of the GAM1634 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1634 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1634 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1634 are further described hereinbelow with reference to Table Table1.

[25355] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1634 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25356] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1635 (GAM1635) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25357] GAM1635 is a novel bioinformatically detected regulatory,

non protein coding, micro RNA (miRNA) gene. The method by which GAM1635 was detected is described hereinabove with reference to Figs. 2–8.

[25358] GAM1635 gene, herein designated GAM GENE, and GAM1635 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25359] GAM1635 gene, herein designated GAM GENE, encodes a GAM1635 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1635 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1635 precursor RNA is designated SEQ ID:1608, and is provided hereinbelow with reference to the sequence listing part.

[25360] GAM1635 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1635 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial

inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25361] An enzyme complex designated DICER COMPLEX, dices the GAM1635 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1635 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1635 RNA is designated SEQ ID:3284, and is provided hereinbelow with reference to the sequence listing part.

[25362] GAM1635 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1635 target RNA, herein designated GAM TARGET RNA. GAM1635 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25363] GAM1635 RNA, herein designated GAM RNA, binds com-

plementarily to one or more target binding sites located in untranslated regions of GAM1635 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1635 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1635 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1635 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25364] The complementary binding of GAM1635 RNA, herein designated GAM RNA, to target binding sites on GAM1635 target RNA, herein designated GAM TARGET RNA, such as

BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1635 target RNA, herein designated GAM TARGET RNA, into GAM1635 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25365] It is appreciated that GAM1635 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1635 target genes. The mRNA of each one of this plurality of GAM1635 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1635 RNA, herein designated GAM RNA, and which when bound by GAM1635 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1635 target proteins.

[25366] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1635 gene, herein designated GAM GENE, on one or more GAM1635 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary

binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25367] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1635 correlate with, and may be deduced from, the identity of the target genes which GAM1635 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25368] Nucleotide sequences of the GAM1635 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1635 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1635 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1635 are further described hereinbelow with reference to Table Table1.

[25369] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the

complementarity of each of these target binding sites to GAM1635 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25370] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1636 (GAM1636) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25371] GAM1636 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1636 was detected is described hereinabove with reference to Figs. 2–8.

[25372] GAM1636 gene, herein designated GAM GENE, and GAM1636 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25373] GAM1636 gene, herein designated GAM GENE, encodes a GAM1636 precursor RNA, herein designated GAM PRECURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1636 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1636 precursor RNA is

designated SEQ ID:1609, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1609 is located at position 808824 relative to contig NT_025004.11, on chromosome 18.

[25374] GAM1636 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1636 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25375] An enzyme complex designated DICER COMPLEX, dices the GAM1636 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1636 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide se-

quence of GAM1636 RNA is designated SEQ ID:3285, and is provided hereinbelow with reference to the sequence listing part.

- [25376] GAM1636 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1636 target RNA, herein designated GAM TARGET RNA. GAM1636 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25377] GAM1636 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1636 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1636 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration

only, and is not meant to be limiting GAM1636 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1636 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25378] The complementary binding of GAM1636 RNA, herein designated GAM RNA, to target binding sites on GAM1636 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1636 target RNA, herein designated GAM TARGET RNA, into GAM1636 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25379] It is appreciated that GAM1636 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1636 target genes. The mRNA of each one of this plurality of GAM1636 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1636 RNA,

herein designated GAM RNA, and which when bound by GAM1636 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1636 target proteins.

[25380] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1636 gene, herein designated GAM GENE, on one or more GAM1636 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25381] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1636 correlate with, and may be deduced from, the identity of the target genes which

GAM1636 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25382] Nucleotide sequences of the GAM1636 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1636 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1636 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1636 are further described hereinbelow with reference to Table Table1.

[25383] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1636 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25384] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1637 (GAM1637) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25385] GAM1637 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method

by which GAM1637 was detected is described hereinabove with reference to Figs. 2–8.

[25386] GAM1637 gene, herein designated GAM GENE, and GAM1637 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25387] GAM1637 gene, herein designated GAM GENE, encodes a GAM1637 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1637 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1637 precursor RNA is designated SEQ ID:1610, and is provided hereinbelow with reference to the sequence listing part.

[25388] GAM1637 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1637 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of

the second half thereof.

- [25389] An enzyme complex designated DICER COMPLEX, dices the GAM1637 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1637 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1637 RNA is designated SEQ ID:3286, and is provided hereinbelow with reference to the sequence listing part.
- [25390] GAM1637 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1637 target RNA, herein designated GAM TARGET RNA. GAM1637 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25391] GAM1637 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in

untranslated regions of GAM1637 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1637 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1637 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1637 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25392] The complementary binding of GAM1637 RNA, herein designated GAM RNA, to target binding sites on GAM1637 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, in-

hibits translation of GAM1637 target RNA, herein designated GAM TARGET RNA, into GAM1637 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25393] It is appreciated that GAM1637 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1637 target genes. The mRNA of each one of this plurality of GAM1637 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1637 RNA, herein designated GAM RNA, and which when bound by GAM1637 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1637 target proteins.

[25394] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1637 gene, herein designated GAM GENE, on one or more GAM1637 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the

known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25395] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1637 correlate with, and may be deduced from, the identity of the target genes which GAM1637 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25396] Nucleotide sequences of the GAM1637 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1637 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1637 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1637 are further described hereinbelow with reference to Table Table1.

[25397] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to

GAM1637 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25398] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1638 (GAM1638) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25399] GAM1638 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1638 was detected is described hereinabove with reference to Figs. 2–8.

[25400] GAM1638 gene, herein designated GAM GENE, and GAM1638 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25401] GAM1638 gene, herein designated GAM GENE, encodes a GAM1638 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1638 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1638 precursor RNA is designated SEQ ID:1611, and is provided hereinbelow with

reference to the sequence listing part.

[25402] GAM1638 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1638 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25403] An enzyme complex designated DICER COMPLEX, dices the GAM1638 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1638 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 94%) nucleotide sequence of GAM1638 RNA is designated SEQ ID:3287, and is provided hereinbelow with reference to the sequence listing part.

[25404] GAM1638 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1638 target RNA, herein designated GAM TARGET RNA. GAM1638 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25405] GAM1638 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1638 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1638 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1638 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a

GAM1638 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25406] The complementary binding of GAM1638 RNA, herein designated GAM RNA, to target binding sites on GAM1638 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1638 target RNA, herein designated GAM TARGET RNA, into GAM1638 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25407] It is appreciated that GAM1638 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1638 target genes. The mRNA of each one of this plurality of GAM1638 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1638 RNA, herein designated GAM RNA, and which when bound by GAM1638 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1638

target proteins.

[25408] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1638 gene, herein designated GAM GENE, on one or more GAM1638 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25409] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1638 correlate with, and may be deduced from, the identity of the target genes which GAM1638 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25410] Nucleotide sequences of the GAM1638 precursor RNA,

herein designated GAM PRECURSOR RNA, and of the diced GAM1638 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1638 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1638 are further described hereinbelow with reference to Table Table1.

[25411] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1638 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25412] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1639 (GAM1639) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25413] GAM1639 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1639 was detected is described hereinabove with reference to Figs. 2-8.

[25414] GAM1639 gene, herein designated GAM GENE, and

GAM1639 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25415] GAM1639 gene, herein designated GAM GENE, encodes a GAM1639 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1639 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1639 precursor RNA is designated SEQ ID:1612, and is provided hereinbelow with reference to the sequence listing part.

[25416] GAM1639 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1639 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25417] An enzyme complex designated DICER COMPLEX, dices the GAM1639 folded precursor RNA, herein designated

GAM FOLDED PRECURSOR RNA, into GAM1639 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1639 RNA is designated SEQ ID:3288, and is provided hereinbelow with reference to the sequence listing part.

- [25418] GAM1639 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1639 target RNA, herein designated GAM TARGET RNA. GAM1639 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25419] GAM1639 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1639 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1639

RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1639 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1639 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25420] The complementary binding of GAM1639 RNA, herein designated GAM RNA, to target binding sites on GAM1639 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1639 target RNA, herein designated GAM TARGET RNA, into GAM1639 target protein, herein designated GAM TARGET PROTEIN. GAM target

protein is therefore outlined by a broken line.

[25421] It is appreciated that GAM1639 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1639 target genes. The mRNA of each one of this plurality of GAM1639 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1639 RNA, herein designated GAM RNA, and which when bound by GAM1639 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1639 target proteins.

[25422] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1639 gene, herein designated GAM GENE, on one or more GAM1639 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other

genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25423] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1639 correlate with, and may be deduced from, the identity of the target genes which GAM1639 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25424] Nucleotide sequences of the GAM1639 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1639 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1639 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1639 are further described hereinbelow with reference to Table Table1.

[25425] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1639 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25426] Fig. 8 further provides a conceptual description of another

novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1640 (GAM1640) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25427] GAM1640 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1640 was detected is described hereinabove with reference to Figs. 2–8.

[25428] GAM1640 gene, herein designated GAM GENE, and GAM1640 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25429] GAM1640 gene, herein designated GAM GENE, encodes a GAM1640 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1640 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1640 precursor RNA is designated SEQ ID:1613, and is provided hereinbelow with reference to the sequence listing part.

[25430] GAM1640 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1640 folded

precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25431] An enzyme complex designated DICER COMPLEX, dices the GAM1640 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1640 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1640 RNA is designated SEQ ID:3289, and is provided hereinbelow with reference to the sequence listing part.

[25432] GAM1640 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1640 target RNA, herein designated GAM TARGET

RNA. GAM1640 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25433] GAM1640 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1640 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1640 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1640 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1640 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an

example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25434] The complementary binding of GAM1640 RNA, herein designated GAM RNA, to target binding sites on GAM1640 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1640 target RNA, herein designated GAM TARGET RNA, into GAM1640 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25435] It is appreciated that GAM1640 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1640 target genes. The mRNA of each one of this plurality of GAM1640 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1640 RNA, herein designated GAM RNA, and which when bound by GAM1640 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1640 target proteins.

[25436] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with

specific reference to translational inhibition exerted by GAM1640 gene, herein designated GAM GENE, on one or more GAM1640 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25437] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1640 correlate with, and may be deduced from, the identity of the target genes which GAM1640 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25438] Nucleotide sequences of the GAM1640 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1640 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of

GAM1640 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1640 are further described hereinbelow with reference to Table Table1.

[25439] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1640 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25440] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1641 (GAM1641) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25441] GAM1641 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1641 was detected is described hereinabove with reference to Figs. 2-8.

[25442] GAM1641 gene, herein designated GAM GENE, and GAM1641 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25443] GAM1641 gene, herein designated GAM GENE, encodes a

GAM1641 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1641 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1641 precursor RNA is designated SEQ ID:1614, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1614 is located at position 2081201 relative to contig NT_011109.13, on chromosome 19.

[25444] GAM1641 has 4 highly conserved, expressed mouse homologs (>90% sequence similarity), found in mouse EST's. Nucleotide sequences similar to the nucleotide sequences of which mouse homologs, and the respective mouse EST's in which these homologs were detected are:

[25445] SEQ ID:99996 detected in mouse EST Accession AA606484.1.

[25446] SEQ ID:99997 detected in mouse EST Accession AA541834.1.

[25447] SEQ ID:99998 detected in mouse EST Accession AA097502.1.

[25448] SEQ ID:99999 detected in mouse EST Accession BI111547.1.

[25449] GAM1641 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1641 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25450] An enzyme complex designated DICER COMPLEX, dices the GAM1641 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1641 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1641 RNA is designated SEQ ID:3290, and is provided hereinbelow with reference to the sequence listing part.

[25451] GAM1641 target gene, herein designated GAM TARGET

GENE, encodes a corresponding messenger RNA, GAM1641 target RNA, herein designated GAM TARGET RNA. GAM1641 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25452] GAM1641 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1641 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1641 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1641 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1641 target RNA, herein designated GAM TARGET

RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25453] The complementary binding of GAM1641 RNA, herein designated GAM RNA, to target binding sites on GAM1641 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1641 target RNA, herein designated GAM TARGET RNA, into GAM1641 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25454] It is appreciated that GAM1641 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1641 target genes. The mRNA of each one of this plurality of GAM1641 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1641 RNA, herein designated GAM RNA, and which when bound by GAM1641 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1641 target proteins.

[25455] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1641 gene, herein designated GAM GENE, on one or more GAM1641 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25456] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1641 correlate with, and may be deduced from, the identity of the target genes which GAM1641 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25457] Nucleotide sequences of the GAM1641 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced

GAM1641 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1641 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1641 are further described hereinbelow with reference to Table Table1.

[25458] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1641 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25459] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1642 (GAM1642) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25460] GAM1642 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1642 was detected is described hereinabove with reference to Figs. 2-8.

[25461] GAM1642 gene, herein designated GAM GENE, and GAM1642 target gene, herein designated GAM TARGET

GENE, are human genes contained in the human genome.

[25462] GAM1642 gene, herein designated GAM GENE, encodes a GAM1642 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1642 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1642 precursor RNA is designated SEQ ID:1615, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1615 is located at position 5764678 relative to contig NT_008984.13, on chromosome 11.

[25463] GAM1642 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1642 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25464] An enzyme complex designated DICER COMPLEX, dices

the GAM1642 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1642 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1642 RNA is designated SEQ ID:3291, and is provided hereinbelow with reference to the sequence listing part.

[25465] GAM1642 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1642 target RNA, herein designated GAM TARGET RNA. GAM1642 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25466] GAM1642 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1642 target RNA, herein designated GAM TARGET RNA. This complementary binding is

due to the fact that the nucleotide sequence of GAM1642 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1642 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1642 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25467] The complementary binding of GAM1642 RNA, herein designated GAM RNA, to target binding sites on GAM1642 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1642 target RNA, herein designated GAM TARGET RNA, into GAM1642 target protein,

herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25468] It is appreciated that GAM1642 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1642 target genes. The mRNA of each one of this plurality of GAM1642 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1642 RNA, herein designated GAM RNA, and which when bound by GAM1642 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1642 target proteins.

[25469] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1642 gene, herein designated GAM GENE, on one or more GAM1642 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by

those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25470] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1642 correlate with, and may be deduced from, the identity of the target genes which GAM1642 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25471] Nucleotide sequences of the GAM1642 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1642 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1642 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1642 are further described hereinbelow with reference to Table Table1.

[25472] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1642 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25473] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1643 (GAM1643) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25474] GAM1643 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1643 was detected is described hereinabove with reference to Figs. 2–8.

[25475] GAM1643 gene, herein designated GAM GENE, and GAM1643 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25476] GAM1643 gene, herein designated GAM GENE, encodes a GAM1643 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1643 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1643 precursor RNA is designated SEQ ID:1616, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1616 is located at position 3434852 rela-

tive to contig NT_009714.13, on chromosome 12.

- [25477] GAM1643 has a highly conserved, expressed mouse homolog (>90% sequence similarity), the nucleotide sequence of which is similar to the nucleotide sequence of SEQ ID:99524, which mouse homolog is detected in mouse EST accession BG288438.1.
- [25478] GAM1643 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1643 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.
- [25479] An enzyme complex designated DICER COMPLEX, dices the GAM1643 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1643 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex

comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1643 RNA is designated SEQ ID:3292, and is provided hereinbelow with reference to the sequence listing part.

- [25480] GAM1643 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1643 target RNA, herein designated GAM TARGET RNA. GAM1643 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25481] GAM1643 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1643 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1643 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III re-

spectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1643 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1643 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25482] The complementary binding of GAM1643 RNA, herein designated GAM RNA, to target binding sites on GAM1643 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1643 target RNA, herein designated GAM TARGET RNA, into GAM1643 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25483] It is appreciated that GAM1643 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1643 target genes. The mRNA of each one of this plurality of GAM1643 target genes comprises one or more

target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1643 RNA, herein designated GAM RNA, and which when bound by GAM1643 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1643 target proteins.

[25484] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1643 gene, herein designated GAM GENE, on one or more GAM1643 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25485] It is yet further appreciated that specific functions, and

accordingly utilities, of GAM1643 correlate with, and may be deduced from, the identity of the target genes which GAM1643 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25486] Nucleotide sequences of the GAM1643 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1643 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1643 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1643 are further described hereinbelow with reference to Table Table1.

[25487] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1643 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25488] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1644 (GAM1644) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25489] GAM1644 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1644 was detected is described hereinabove with reference to Figs. 2–8.

[25490] GAM1644 gene, herein designated GAM GENE, and GAM1644 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25491] GAM1644 gene, herein designated GAM GENE, encodes a GAM1644 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1644 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1644 precursor RNA is designated SEQ ID:1617, and is provided hereinbelow with reference to the sequence listing part.

[25492] GAM1644 has 20 highly conserved, expressed mouse homologs (>90% sequence similarity), found in mouse EST's. Nucleotide sequences similar to the nucleotide sequences of which mouse homologs, and the respective mouse EST's in which these homologs were detected are:

[25493] SEQ ID:98160 detected in mouse EST Accession AA623023.1.

[25494] SEQ ID:98161 detected in mouse EST Accession
AA561578.1.

[25495] SEQ ID:98162 detected in mouse EST Accession
BF453421.1.

[25496] SEQ ID:98163 detected in mouse EST Accession
BE687835.1.

[25497] SEQ ID:98164 detected in mouse EST Accession
AA177678.1.

[25498] SEQ ID:98165 detected in mouse EST Accession
BE448444.1.

[25499] SEQ ID:98166 detected in mouse EST Accession
AA499506.1.

[25500] SEQ ID:98167 detected in mouse EST Accession
AI551780.1.

[25501] SEQ ID:98168 detected in mouse EST Accession
BG228324.1.

- [25502] SEQ ID:98169 detected in mouse EST Accession
BB620404.1.
- [25503] SEQ ID:98170 detected in mouse EST Accession
AA111127.1.
- [25504] SEQ ID:98171 detected in mouse EST Accession
AA929520.1.
- [25505] SEQ ID:98172 detected in mouse EST Accession
BE634222.1.
- [25506] SEQ ID:98173 detected in mouse EST Accession
BE692018.1.
- [25507] SEQ ID:98174 detected in mouse EST Accession
BI647812.1.
- [25508] SEQ ID:98175 detected in mouse EST Accession
BF722531.1.
- [25509] SEQ ID:98176 detected in mouse EST Accession
BG071829.1.
- [25510] SEQ ID:98177 detected in mouse EST Accession
AI118571.1.
- [25511] SEQ ID:98178 detected in mouse EST Accession
BI689430.1.
- [25512] SEQ ID:98179 detected in mouse EST Accession
BI248537.1.
- [25513] GAM1644 precursor RNA, herein designated GAM PRE-

CURSOR RNA, folds onto itself, forming GAM1644 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25514] An enzyme complex designated DICER COMPLEX, dices the GAM1644 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1644 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1644 RNA is designated SEQ ID:3293, and is provided hereinbelow with reference to the sequence listing part.

[25515] GAM1644 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA,

GAM1644 target RNA, herein designated GAM TARGET RNA. GAM1644 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25516] GAM1644 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1644 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1644 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1644 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1644 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts tar-

get binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25517] The complementary binding of GAM1644 RNA, herein designated GAM RNA, to target binding sites on GAM1644 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1644 target RNA, herein designated GAM TARGET RNA, into GAM1644 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25518] It is appreciated that GAM1644 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1644 target genes. The mRNA of each one of this plurality of GAM1644 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1644 RNA, herein designated GAM RNA, and which when bound by GAM1644 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1644 target proteins.

[25519] It is further appreciated by one skilled in the art that the

mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1644 gene, herein designated GAM GENE, on one or more GAM1644 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25520] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1644 correlate with, and may be deduced from, the identity of the target genes which GAM1644 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25521] Nucleotide sequences of the GAM1644 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1644 RNA, herein designated GAM RNA, and a

schematic representation of the secondary folding of GAM1644 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1644 are further described hereinbelow with reference to Table Table1.

[25522] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1644 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25523] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1645 (GAM1645) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25524] GAM1645 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1645 was detected is described hereinabove with reference to Figs. 2-8.

[25525] GAM1645 gene, herein designated GAM GENE, and GAM1645 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25526] GAM1645 gene, herein designated GAM GENE, encodes a GAM1645 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1645 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1645 precursor RNA is designated SEQ ID:1618, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1618 is located at position 1698742 relative to contig NT_028309.8, on chromosome 11.

[25527] GAM1645 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1645 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25528] An enzyme complex designated DICER COMPLEX, dices the GAM1645 folded precursor RNA, herein designated

GAM FOLDED PRECURSOR RNA, into GAM1645 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1645 RNA is designated SEQ ID:3294, and is provided hereinbelow with reference to the sequence listing part.

- [25529] GAM1645 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1645 target RNA, herein designated GAM TARGET RNA. GAM1645 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25530] GAM1645 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1645 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1645

RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1645 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1645 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25531] The complementary binding of GAM1645 RNA, herein designated GAM RNA, to target binding sites on GAM1645 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1645 target RNA, herein designated GAM TARGET RNA, into GAM1645 target protein, herein designated GAM TARGET PROTEIN. GAM target

protein is therefore outlined by a broken line.

[25532] It is appreciated that GAM1645 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1645 target genes. The mRNA of each one of this plurality of GAM1645 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1645 RNA, herein designated GAM RNA, and which when bound by GAM1645 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1645 target proteins.

[25533] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1645 gene, herein designated GAM GENE, on one or more GAM1645 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other

genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25534] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1645 correlate with, and may be deduced from, the identity of the target genes which GAM1645 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25535] Nucleotide sequences of the GAM1645 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1645 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1645 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1645 are further described hereinbelow with reference to Table Table1.

[25536] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1645 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25537] Fig. 8 further provides a conceptual description of another

novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1646 (GAM1646) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25538] GAM1646 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1646 was detected is described hereinabove with reference to Figs. 2–8.

[25539] GAM1646 gene, herein designated GAM GENE, and GAM1646 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25540] GAM1646 gene, herein designated GAM GENE, encodes a GAM1646 precursor RNA, herein designated GAM PRECURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1646 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1646 precursor RNA is designated SEQ ID:1619, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1619 is located at position 6294511 relative to contig NT_024981.11, on chromosome 18.

[25541] GAM1646 has 15 highly conserved, expressed mouse homologs (>90% sequence similarity), found in mouse EST's. Nucleotide sequences similar to the nucleotide sequences of which mouse homologs, and the respective mouse EST's in which these homologs were detected are:

[25542] SEQ ID:99993 detected in mouse EST Accession BF719273.1.

[25543] SEQ ID:99994 detected in mouse EST Accession BF020008.1.

[25544] SEQ ID:99995 detected in mouse EST Accession BB615397.1.

[25545] SEQ ID:98180 detected in mouse EST Accession AW743974.1.

[25546] SEQ ID:98181 detected in mouse EST Accession BE947979.1.

[25547] SEQ ID:98182 detected in mouse EST Accession BF463146.1.

[25548] SEQ ID:98183 detected in mouse EST Accession BF718970.1.

[25549] SEQ ID:98184 detected in mouse EST Accession AW456243.1.

[25550] SEQ ID:98185 detected in mouse EST Accession AW494497.1.

[25551] SEQ ID:98186 detected in mouse EST Accession
AW987984.1.

[25552] SEQ ID:98187 detected in mouse EST Accession
BB624634.1.

[25553] SEQ ID:98188 detected in mouse EST Accession
AW987905.1.

[25554] SEQ ID:98189 detected in mouse EST Accession
BI111596.1.

[25555] SEQ ID:98190 detected in mouse EST Accession
BE915985.1.

[25556] SEQ ID:98191 detected in mouse EST Accession
BF472656.1.

[25557] GAM1646 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1646 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25558] An enzyme complex designated DICER COMPLEX, dices

the GAM1646 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1646 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1646 RNA is designated SEQ ID:3295, and is provided hereinbelow with reference to the sequence listing part.

[25559] GAM1646 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1646 target RNA, herein designated GAM TARGET RNA. GAM1646 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25560] GAM1646 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1646 target RNA, herein designated GAM TARGET RNA. This complementary binding is

due to the fact that the nucleotide sequence of GAM1646 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1646 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1646 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25561] The complementary binding of GAM1646 RNA, herein designated GAM RNA, to target binding sites on GAM1646 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1646 target RNA, herein designated GAM TARGET RNA, into GAM1646 target protein,

herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25562] It is appreciated that GAM1646 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1646 target genes. The mRNA of each one of this plurality of GAM1646 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1646 RNA, herein designated GAM RNA, and which when bound by GAM1646 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1646 target proteins.

[25563] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1646 gene, herein designated GAM GENE, on one or more GAM1646 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by

those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25564] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1646 correlate with, and may be deduced from, the identity of the target genes which GAM1646 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25565] Nucleotide sequences of the GAM1646 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1646 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1646 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1646 are further described hereinbelow with reference to Table Table1.

[25566] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1646 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25567] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1647 (GAM1647) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25568] GAM1647 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1647 was detected is described hereinabove with reference to Figs. 2–8.

[25569] GAM1647 gene, herein designated GAM GENE, and GAM1647 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25570] GAM1647 gene, herein designated GAM GENE, encodes a GAM1647 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1647 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1647 precursor RNA is designated SEQ ID:1620, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1620 is located at position 2349674 rela-

tive to contig NT_021907.13, on chromosome 1.

[25571] GAM1647 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1647 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25572] An enzyme complex designated DICER COMPLEX, dices the GAM1647 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1647 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1647 RNA is designated SEQ ID:3296, and is provided hereinbelow with reference to the sequence listing part.

[25573] GAM1647 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1647 target RNA, herein designated GAM TARGET RNA. GAM1647 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25574] GAM1647 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1647 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1647 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1647 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a

GAM1647 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25575] The complementary binding of GAM1647 RNA, herein designated GAM RNA, to target binding sites on GAM1647 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1647 target RNA, herein designated GAM TARGET RNA, into GAM1647 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25576] It is appreciated that GAM1647 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1647 target genes. The mRNA of each one of this plurality of GAM1647 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1647 RNA, herein designated GAM RNA, and which when bound by GAM1647 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1647

target proteins.

[25577] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1647 gene, herein designated GAM GENE, on one or more GAM1647 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25578] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1647 correlate with, and may be deduced from, the identity of the target genes which GAM1647 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25579] Nucleotide sequences of the GAM1647 precursor RNA,

herein designated GAM PRECURSOR RNA, and of the diced GAM1647 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1647 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1647 are further described hereinbelow with reference to Table Table1.

[25580] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1647 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25581] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1648 (GAM1648) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25582] GAM1648 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1648 was detected is described hereinabove with reference to Figs. 2-8.

[25583] GAM1648 gene, herein designated GAM GENE, and

GAM1648 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25584] GAM1648 gene, herein designated GAM GENE, encodes a GAM1648 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1648 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1648 precursor RNA is designated SEQ ID:1621, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1621 is located at position 755864 relative to contig NT_005416.8, on chromosome 2.

[25585] GAM1648 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1648 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25586] An enzyme complex designated DICER COMPLEX, dices the GAM1648 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1648 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1648 RNA is designated SEQ ID:3297, and is provided hereinbelow with reference to the sequence listing part.

[25587] GAM1648 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1648 target RNA, herein designated GAM TARGET RNA. GAM1648 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25588] GAM1648 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1648 target RNA, herein des-

ignated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1648 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1648 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1648 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25589] The complementary binding of GAM1648 RNA, herein designated GAM RNA, to target binding sites on GAM1648 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1648 target RNA, herein design-

nated GAM TARGET RNA, into GAM1648 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25590] It is appreciated that GAM1648 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1648 target genes. The mRNA of each one of this plurality of GAM1648 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1648 RNA, herein designated GAM RNA, and which when bound by GAM1648 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1648 target proteins.

[25591] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1648 gene, herein designated GAM GENE, on one or more GAM1648 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other

recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25592] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1648 correlate with, and may be deduced from, the identity of the target genes which GAM1648 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25593] Nucleotide sequences of the GAM1648 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1648 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1648 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1648 are further described hereinbelow with reference to Table Table1.

[25594] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1648 RNA, herein designated GAM RNA, are de-

scribed hereinbelow with reference to Table Table2.

[25595] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1649 (GAM1649) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25596] GAM1649 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1649 was detected is described hereinabove with reference to Figs. 2–8.

[25597] GAM1649 gene, herein designated GAM GENE, and GAM1649 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25598] GAM1649 gene, herein designated GAM GENE, encodes a GAM1649 precursor RNA, herein designated GAM PRECURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1649 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1649 precursor RNA is designated SEQ ID:1622, and is provided hereinbelow with reference to the sequence listing part.

[25599] GAM1649 has 13 highly conserved, expressed mouse homologs (>90% sequence similarity), found in mouse EST's. Nucleotide sequences similar to the nucleotide sequences of which mouse homologs, and the respective mouse EST's in which these homologs were detected are:

[25600] SEQ ID:98145 detected in mouse EST Accession BI409473.1.

[25601] SEQ ID:98146 detected in mouse EST Accession W36780.1.

[25602] SEQ ID:98147 detected in mouse EST Accession BE652434.1.

[25603] SEQ ID:98148 detected in mouse EST Accession BG861731.1.

[25604] SEQ ID:98149 detected in mouse EST Accession BE311296.1.

[25605] SEQ ID:98151 detected in mouse EST Accession AA245034.1.

[25606] SEQ ID:98152 detected in mouse EST Accession W42007.1.

[25607] SEQ ID:98153 detected in mouse EST Accession BE952277.1.

[25608] SEQ ID:98154 detected in mouse EST Accession BG083593.1.

[25609] SEQ ID:98155 detected in mouse EST Accession
AA756210.1.

[25610] SEQ ID:98156 detected in mouse EST Accession
AA624596.1.

[25611] SEQ ID:98157 detected in mouse EST Accession
BI414655.1.

[25612] SEQ ID:98682 detected in mouse EST Accession
AA636285.1.

[25613] GAM1649 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1649 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25614] An enzyme complex designated DICER COMPLEX, dices the GAM1649 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1649 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a

hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1649 RNA is designated SEQ ID:3298, and is provided hereinbelow with reference to the sequence listing part.

- [25615] GAM1649 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1649 target RNA, herein designated GAM TARGET RNA. GAM1649 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25616] GAM1649 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1649 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1649 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustra-

tion, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1649 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1649 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25617] The complementary binding of GAM1649 RNA, herein designated GAM RNA, to target binding sites on GAM1649 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1649 target RNA, herein designated GAM TARGET RNA, into GAM1649 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25618] It is appreciated that GAM1649 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of

GAM1649 target genes. The mRNA of each one of this plurality of GAM1649 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1649 RNA, herein designated GAM RNA, and which when bound by GAM1649 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1649 target proteins.

[25619] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1649 gene, herein designated GAM GENE, on one or more GAM1649 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a

tiny RNA world, Science 294,779 (2001)).

[25620] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1649 correlate with, and may be deduced from, the identity of the target genes which GAM1649 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25621] Nucleotide sequences of the GAM1649 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1649 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1649 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1649 are further described hereinbelow with reference to Table Table1.

[25622] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1649 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25623] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1650 (GAM1650) gene, which modulates expression of

respective target genes thereof, the function and utility of which target genes is known in the art.

[25624] GAM1650 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1650 was detected is described hereinabove with reference to Figs. 2–8.

[25625] GAM1650 gene, herein designated GAM GENE, and GAM1650 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25626] GAM1650 gene, herein designated GAM GENE, encodes a GAM1650 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1650 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1650 precursor RNA is designated SEQ ID:1623, and is provided hereinbelow with reference to the sequence listing part.

[25627] GAM1650 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1650 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typi-

cal of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25628] An enzyme complex designated DICER COMPLEX, dices the GAM1650 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1650 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1650 RNA is designated SEQ ID:3299, and is provided hereinbelow with reference to the sequence listing part.

[25629] GAM1650 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1650 target RNA, herein designated GAM TARGET RNA. GAM1650 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a pro-

tein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25630] GAM1650 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1650 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1650 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1650 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1650 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25631] The complementary binding of GAM1650 RNA, herein designated GAM RNA, to target binding sites on GAM1650 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1650 target RNA, herein designated GAM TARGET RNA, into GAM1650 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25632] It is appreciated that GAM1650 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1650 target genes. The mRNA of each one of this plurality of GAM1650 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1650 RNA, herein designated GAM RNA, and which when bound by GAM1650 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1650 target proteins.

[25633] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1650 gene, herein designated GAM GENE, on one or more GAM1650 target gene, herein designated GAM TAR-

GET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25634] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1650 correlate with, and may be deduced from, the identity of the target genes which GAM1650 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25635] Nucleotide sequences of the GAM1650 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1650 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1650 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1650 are further described hereinbelow with reference to Table Table1.

[25636] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1650 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25637] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1651 (GAM1651) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25638] GAM1651 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1651 was detected is described hereinabove with reference to Figs. 2-8.

[25639] GAM1651 gene, herein designated GAM GENE, and GAM1651 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25640] GAM1651 gene, herein designated GAM GENE, encodes a GAM1651 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1651 precursor RNA, herein

designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1651 precursor RNA is designated SEQ ID:1624, and is provided hereinbelow with reference to the sequence listing part.

[25641] GAM1651 precursor RNA, herein designated GAM PRECURSOR RNA, folds onto itself, forming GAM1651 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25642] An enzyme complex designated DICER COMPLEX, dices the GAM1651 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1651 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other

necessary proteins. A probable (over 89%) nucleotide sequence of GAM1651 RNA is designated SEQ ID:3300, and is provided hereinbelow with reference to the sequence listing part.

- [25643] GAM1651 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1651 target RNA, herein designated GAM TARGET RNA. GAM1651 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25644] GAM1651 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1651 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1651 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target

binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1651 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1651 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25645] The complementary binding of GAM1651 RNA, herein designated GAM RNA, to target binding sites on GAM1651 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1651 target RNA, herein designated GAM TARGET RNA, into GAM1651 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25646] It is appreciated that GAM1651 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1651 target genes. The mRNA of each one of this plurality of GAM1651 target genes comprises one or more target binding sites, each having a nucleotide sequence

which is at least partly complementary to GAM1651 RNA, herein designated GAM RNA, and which when bound by GAM1651 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1651 target proteins.

[25647] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1651 gene, herein designated GAM GENE, on one or more GAM1651 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25648] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1651 correlate with, and may

be deduced from, the identity of the target genes which GAM1651 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25649] Nucleotide sequences of the GAM1651 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1651 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1651 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1651 are further described hereinbelow with reference to Table Table1.

[25650] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1651 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25651] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1652 (GAM1652) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25652] GAM1652 is a novel bioinformatically detected regulatory,

non protein coding, micro RNA (miRNA) gene. The method by which GAM1652 was detected is described hereinabove with reference to Figs. 2–8.

[25653] GAM1652 gene, herein designated GAM GENE, and GAM1652 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25654] GAM1652 gene, herein designated GAM GENE, encodes a GAM1652 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1652 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1652 precursor RNA is designated SEQ ID:1625, and is provided hereinbelow with reference to the sequence listing part.

[25655] GAM1652 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1652 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial

inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25656] An enzyme complex designated DICER COMPLEX, dices the GAM1652 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1652 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1652 RNA is designated SEQ ID:3301, and is provided hereinbelow with reference to the sequence listing part.

[25657] GAM1652 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1652 target RNA, herein designated GAM TARGET RNA. GAM1652 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25658] GAM1652 RNA, herein designated GAM RNA, binds com-

plementarily to one or more target binding sites located in untranslated regions of GAM1652 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1652 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1652 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1652 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25659] The complementary binding of GAM1652 RNA, herein designated GAM RNA, to target binding sites on GAM1652 target RNA, herein designated GAM TARGET RNA, such as

BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1652 target RNA, herein designated GAM TARGET RNA, into GAM1652 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25660] It is appreciated that GAM1652 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1652 target genes. The mRNA of each one of this plurality of GAM1652 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1652 RNA, herein designated GAM RNA, and which when bound by GAM1652 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1652 target proteins.

[25661] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1652 gene, herein designated GAM GENE, on one or more GAM1652 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary

binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25662] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1652 correlate with, and may be deduced from, the identity of the target genes which GAM1652 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25663] Nucleotide sequences of the GAM1652 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1652 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1652 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1652 are further described hereinbelow with reference to Table Table1.

[25664] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the

complementarity of each of these target binding sites to GAM1652 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25665] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1653 (GAM1653) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25666] GAM1653 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1653 was detected is described hereinabove with reference to Figs. 2–8.

[25667] GAM1653 gene, herein designated GAM GENE, and GAM1653 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25668] GAM1653 gene, herein designated GAM GENE, encodes a GAM1653 precursor RNA, herein designated GAM PRECURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1653 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1653 precursor RNA is

designated SEQ ID:1626, and is provided hereinbelow with reference to the sequence listing part.

[25669] GAM1653 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1653 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25670] An enzyme complex designated DICER COMPLEX, dices the GAM1653 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1653 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 94%) nucleotide sequence of GAM1653 RNA is designated SEQ ID:3302, and is provided hereinbelow with reference to the sequence

listing part.

- [25671] GAM1653 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1653 target RNA, herein designated GAM TARGET RNA. GAM1653 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25672] GAM1653 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1653 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1653 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1653 RNA, herein designated GAM RNA, may have a different number

of target binding sites in untranslated regions of a GAM1653 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25673] The complementary binding of GAM1653 RNA, herein designated GAM RNA, to target binding sites on GAM1653 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1653 target RNA, herein designated GAM TARGET RNA, into GAM1653 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25674] It is appreciated that GAM1653 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1653 target genes. The mRNA of each one of this plurality of GAM1653 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1653 RNA, herein designated GAM RNA, and which when bound by GAM1653 RNA, herein designated GAM RNA, causes inhi-

bition of translation of respective one or more GAM1653 target proteins.

[25675] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1653 gene, herein designated GAM GENE, on one or more GAM1653 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25676] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1653 correlate with, and may be deduced from, the identity of the target genes which GAM1653 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25677] Nucleotide sequences of the GAM1653 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1653 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1653 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1653 are further described hereinbelow with reference to Table Table1.

[25678] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1653 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25679] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1654 (GAM1654) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25680] GAM1654 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1654 was detected is described hereinabove with reference to Figs. 2-8.

[25681] GAM1654 gene, herein designated GAM GENE, and GAM1654 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25682] GAM1654 gene, herein designated GAM GENE, encodes a GAM1654 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1654 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1654 precursor RNA is designated SEQ ID:1627, and is provided hereinbelow with reference to the sequence listing part.

[25683] GAM1654 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1654 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25684] An enzyme complex designated DICER COMPLEX, dices

the GAM1654 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1654 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1654 RNA is designated SEQ ID:3303, and is provided hereinbelow with reference to the sequence listing part.

[25685] GAM1654 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1654 target RNA, herein designated GAM TARGET RNA. GAM1654 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25686] GAM1654 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1654 target RNA, herein designated GAM TARGET RNA. This complementary binding is

due to the fact that the nucleotide sequence of GAM1654 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1654 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1654 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25687] The complementary binding of GAM1654 RNA, herein designated GAM RNA, to target binding sites on GAM1654 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1654 target RNA, herein designated GAM TARGET RNA, into GAM1654 target protein,

herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25688] It is appreciated that GAM1654 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1654 target genes. The mRNA of each one of this plurality of GAM1654 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1654 RNA, herein designated GAM RNA, and which when bound by GAM1654 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1654 target proteins.

[25689] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1654 gene, herein designated GAM GENE, on one or more GAM1654 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by

those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25690] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1654 correlate with, and may be deduced from, the identity of the target genes which GAM1654 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25691] Nucleotide sequences of the GAM1654 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1654 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1654 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1654 are further described hereinbelow with reference to Table Table1.

[25692] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1654 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25693] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1655 (GAM1655) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25694] GAM1655 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1655 was detected is described hereinabove with reference to Figs. 2–8.

[25695] GAM1655 gene, herein designated GAM GENE, and GAM1655 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25696] GAM1655 gene, herein designated GAM GENE, encodes a GAM1655 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1655 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1655 precursor RNA is designated SEQ ID:1628, and is provided hereinbelow with reference to the sequence listing part.

[25697] GAM1655 precursor RNA, herein designated GAM PRE-

CURSOR RNA, folds onto itself, forming GAM1655 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25698] An enzyme complex designated DICER COMPLEX, dices the GAM1655 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1655 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1655 RNA is designated SEQ ID:3304, and is provided hereinbelow with reference to the sequence listing part.

[25699] GAM1655 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA,

GAM1655 target RNA, herein designated GAM TARGET RNA. GAM1655 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25700] GAM1655 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1655 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1655 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1655 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1655 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts tar-

get binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25701] The complementary binding of GAM1655 RNA, herein designated GAM RNA, to target binding sites on GAM1655 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1655 target RNA, herein designated GAM TARGET RNA, into GAM1655 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25702] It is appreciated that GAM1655 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1655 target genes. The mRNA of each one of this plurality of GAM1655 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1655 RNA, herein designated GAM RNA, and which when bound by GAM1655 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1655 target proteins.

[25703] It is further appreciated by one skilled in the art that the

mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1655 gene, herein designated GAM GENE, on one or more GAM1655 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25704] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1655 correlate with, and may be deduced from, the identity of the target genes which GAM1655 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25705] Nucleotide sequences of the GAM1655 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1655 RNA, herein designated GAM RNA, and a

schematic representation of the secondary folding of GAM1655 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1655 are further described hereinbelow with reference to Table Table1.

[25706] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1655 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25707] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1656 (GAM1656) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25708] GAM1656 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1656 was detected is described hereinabove with reference to Figs. 2-8.

[25709] GAM1656 gene, herein designated GAM GENE, and GAM1656 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25710] GAM1656 gene, herein designated GAM GENE, encodes a GAM1656 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1656 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1656 precursor RNA is designated SEQ ID:1629, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1629 is located at position 4851350 relative to contig NT_022517.13, on chromosome 3.

[25711] GAM1656 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1656 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25712] An enzyme complex designated DICER COMPLEX, dices the GAM1656 folded precursor RNA, herein designated

GAM FOLDED PRECURSOR RNA, into GAM1656 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1656 RNA is designated SEQ ID:3305, and is provided hereinbelow with reference to the sequence listing part.

- [25713] GAM1656 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1656 target RNA, herein designated GAM TARGET RNA. GAM1656 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25714] GAM1656 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1656 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1656

RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1656 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1656 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25715] The complementary binding of GAM1656 RNA, herein designated GAM RNA, to target binding sites on GAM1656 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1656 target RNA, herein designated GAM TARGET RNA, into GAM1656 target protein, herein designated GAM TARGET PROTEIN. GAM target

protein is therefore outlined by a broken line.

[25716] It is appreciated that GAM1656 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1656 target genes. The mRNA of each one of this plurality of GAM1656 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1656 RNA, herein designated GAM RNA, and which when bound by GAM1656 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1656 target proteins.

[25717] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1656 gene, herein designated GAM GENE, on one or more GAM1656 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other

genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25718] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1656 correlate with, and may be deduced from, the identity of the target genes which GAM1656 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25719] Nucleotide sequences of the GAM1656 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1656 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1656 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1656 are further described hereinbelow with reference to Table Table1.

[25720] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1656 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25721] Fig. 8 further provides a conceptual description of another

novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1657 (GAM1657) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25722] GAM1657 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1657 was detected is described hereinabove with reference to Figs. 2–8.

[25723] GAM1657 gene, herein designated GAM GENE, and GAM1657 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25724] GAM1657 gene, herein designated GAM GENE, encodes a GAM1657 precursor RNA, herein designated GAM PRECURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1657 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1657 precursor RNA is designated SEQ ID:1630, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1630 is located at position 33703762 relative to contig NT_007592.11, on chromosome 6.

[25725] GAM1657 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1657 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25726] An enzyme complex designated DICER COMPLEX, dices the GAM1657 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1657 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1657 RNA is designated SEQ ID:3306, and is provided hereinbelow with reference to the sequence listing part.

[25727] GAM1657 target gene, herein designated GAM TARGET

GENE, encodes a corresponding messenger RNA, GAM1657 target RNA, herein designated GAM TARGET RNA. GAM1657 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25728] GAM1657 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1657 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1657 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1657 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1657 target RNA, herein designated GAM TARGET

RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25729] The complementary binding of GAM1657 RNA, herein designated GAM RNA, to target binding sites on GAM1657 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1657 target RNA, herein designated GAM TARGET RNA, into GAM1657 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25730] It is appreciated that GAM1657 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1657 target genes. The mRNA of each one of this plurality of GAM1657 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1657 RNA, herein designated GAM RNA, and which when bound by GAM1657 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1657 target proteins.

[25731] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1657 gene, herein designated GAM GENE, on one or more GAM1657 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25732] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1657 correlate with, and may be deduced from, the identity of the target genes which GAM1657 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25733] Nucleotide sequences of the GAM1657 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced

GAM1657 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1657 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1657 are further described hereinbelow with reference to Table Table1.

[25734] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1657 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25735] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1658 (GAM1658) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25736] GAM1658 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1658 was detected is described hereinabove with reference to Figs. 2-8.

[25737] GAM1658 gene, herein designated GAM GENE, and GAM1658 target gene, herein designated GAM TARGET

GENE, are human genes contained in the human genome.

[25738] GAM1658 gene, herein designated GAM GENE, encodes a GAM1658 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1658 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1658 precursor RNA is designated SEQ ID:1631, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1631 is located at position 180066 relative to contig NT_010859.10, on chromosome 18.

[25739] GAM1658 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1658 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25740] An enzyme complex designated DICER COMPLEX, dices

the GAM1658 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1658 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1658 RNA is designated SEQ ID:3307, and is provided hereinbelow with reference to the sequence listing part.

[25741] GAM1658 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1658 target RNA, herein designated GAM TARGET RNA. GAM1658 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25742] GAM1658 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1658 target RNA, herein designated GAM TARGET RNA. This complementary binding is

due to the fact that the nucleotide sequence of GAM1658 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1658 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1658 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25743] The complementary binding of GAM1658 RNA, herein designated GAM RNA, to target binding sites on GAM1658 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1658 target RNA, herein designated GAM TARGET RNA, into GAM1658 target protein,

herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25744] It is appreciated that GAM1658 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1658 target genes. The mRNA of each one of this plurality of GAM1658 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1658 RNA, herein designated GAM RNA, and which when bound by GAM1658 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1658 target proteins.

[25745] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1658 gene, herein designated GAM GENE, on one or more GAM1658 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by

those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25746] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1658 correlate with, and may be deduced from, the identity of the target genes which GAM1658 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25747] Nucleotide sequences of the GAM1658 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1658 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1658 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1658 are further described hereinbelow with reference to Table Table1.

[25748] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1658 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25749] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1659 (GAM1659) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25750] GAM1659 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1659 was detected is described hereinabove with reference to Figs. 2–8.

[25751] GAM1659 gene, herein designated GAM GENE, and GAM1659 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25752] GAM1659 gene, herein designated GAM GENE, encodes a GAM1659 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1659 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1659 precursor RNA is designated SEQ ID:1632, and is provided hereinbelow with reference to the sequence listing part.

[25753] GAM1659 precursor RNA, herein designated GAM PRE-

CURSOR RNA, folds onto itself, forming GAM1659 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25754] An enzyme complex designated DICER COMPLEX, dices the GAM1659 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1659 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1659 RNA is designated SEQ ID:3308, and is provided hereinbelow with reference to the sequence listing part.

[25755] GAM1659 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA,

GAM1659 target RNA, herein designated GAM TARGET RNA. GAM1659 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25756] GAM1659 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1659 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1659 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1659 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1659 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts tar-

get binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25757] The complementary binding of GAM1659 RNA, herein designated GAM RNA, to target binding sites on GAM1659 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1659 target RNA, herein designated GAM TARGET RNA, into GAM1659 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25758] It is appreciated that GAM1659 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1659 target genes. The mRNA of each one of this plurality of GAM1659 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1659 RNA, herein designated GAM RNA, and which when bound by GAM1659 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1659 target proteins.

[25759] It is further appreciated by one skilled in the art that the

mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1659 gene, herein designated GAM GENE, on one or more GAM1659 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25760] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1659 correlate with, and may be deduced from, the identity of the target genes which GAM1659 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25761] Nucleotide sequences of the GAM1659 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1659 RNA, herein designated GAM RNA, and a

schematic representation of the secondary folding of GAM1659 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1659 are further described hereinbelow with reference to Table Table1.

[25762] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1659 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25763] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1660 (GAM1660) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25764] GAM1660 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1660 was detected is described hereinabove with reference to Figs. 2-8.

[25765] GAM1660 gene, herein designated GAM GENE, and GAM1660 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25766] GAM1660 gene, herein designated GAM GENE, encodes a GAM1660 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1660 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1660 precursor RNA is designated SEQ ID:1633, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1633 is located at position 28903785 relative to contig NT_007592.11, on chromosome 6.

[25767] GAM1660 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1660 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25768] An enzyme complex designated DICER COMPLEX, dices the GAM1660 folded precursor RNA, herein designated

GAM FOLDED PRECURSOR RNA, into GAM1660 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 94%) nucleotide sequence of GAM1660 RNA is designated SEQ ID:3309, and is provided hereinbelow with reference to the sequence listing part.

- [25769] GAM1660 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1660 target RNA, herein designated GAM TARGET RNA. GAM1660 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25770] GAM1660 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1660 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1660

RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1660 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1660 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25771] The complementary binding of GAM1660 RNA, herein designated GAM RNA, to target binding sites on GAM1660 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1660 target RNA, herein designated GAM TARGET RNA, into GAM1660 target protein, herein designated GAM TARGET PROTEIN. GAM target

protein is therefore outlined by a broken line.

[25772] It is appreciated that GAM1660 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1660 target genes. The mRNA of each one of this plurality of GAM1660 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1660 RNA, herein designated GAM RNA, and which when bound by GAM1660 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1660 target proteins.

[25773] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1660 gene, herein designated GAM GENE, on one or more GAM1660 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other

genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25774] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1660 correlate with, and may be deduced from, the identity of the target genes which GAM1660 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25775] Nucleotide sequences of the GAM1660 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1660 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1660 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1660 are further described hereinbelow with reference to Table Table1.

[25776] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1660 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25777] Fig. 8 further provides a conceptual description of another

novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1661 (GAM1661) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25778] GAM1661 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1661 was detected is described hereinabove with reference to Figs. 2–8.

[25779] GAM1661 gene, herein designated GAM GENE, and GAM1661 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25780] GAM1661 gene, herein designated GAM GENE, encodes a GAM1661 precursor RNA, herein designated GAM PRECURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1661 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1661 precursor RNA is designated SEQ ID:1634, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1634 is located at position 22191257 relative to contig NT_007299.11, on chromosome 6.

[25781] GAM1661 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1661 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25782] An enzyme complex designated DICER COMPLEX, dices the GAM1661 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1661 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1661 RNA is designated SEQ ID:3310, and is provided hereinbelow with reference to the sequence listing part.

[25783] GAM1661 target gene, herein designated GAM TARGET

GENE, encodes a corresponding messenger RNA, GAM1661 target RNA, herein designated GAM TARGET RNA. GAM1661 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25784] GAM1661 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1661 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1661 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1661 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1661 target RNA, herein designated GAM TARGET

RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25785] The complementary binding of GAM1661 RNA, herein designated GAM RNA, to target binding sites on GAM1661 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1661 target RNA, herein designated GAM TARGET RNA, into GAM1661 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25786] It is appreciated that GAM1661 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1661 target genes. The mRNA of each one of this plurality of GAM1661 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1661 RNA, herein designated GAM RNA, and which when bound by GAM1661 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1661 target proteins.

[25787] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1661 gene, herein designated GAM GENE, on one or more GAM1661 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25788] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1661 correlate with, and may be deduced from, the identity of the target genes which GAM1661 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25789] Nucleotide sequences of the GAM1661 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced

GAM1661 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1661 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1661 are further described hereinbelow with reference to Table Table1.

[25790] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1661 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25791] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1662 (GAM1662) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25792] GAM1662 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1662 was detected is described hereinabove with reference to Figs. 2-8.

[25793] GAM1662 gene, herein designated GAM GENE, and GAM1662 target gene, herein designated GAM TARGET

GENE, are human genes contained in the human genome.

[25794] GAM1662 gene, herein designated GAM GENE, encodes a GAM1662 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1662 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1662 precursor RNA is designated SEQ ID:1635, and is provided hereinbelow with reference to the sequence listing part.

[25795] GAM1662 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1662 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25796] An enzyme complex designated DICER COMPLEX, dices the GAM1662 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1662 RNA,

herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1662 RNA is designated SEQ ID:3311, and is provided hereinbelow with reference to the sequence listing part.

- [25797] GAM1662 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1662 target RNA, herein designated GAM TARGET RNA. GAM1662 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25798] GAM1662 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1662 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1662 RNA, herein designated GAM RNA, is an accurate or a par-

tial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1662 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1662 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25799] The complementary binding of GAM1662 RNA, herein designated GAM RNA, to target binding sites on GAM1662 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1662 target RNA, herein designated GAM TARGET RNA, into GAM1662 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25800] It is appreciated that GAM1662 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1662 target genes. The mRNA of each one of this plurality of GAM1662 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1662 RNA, herein designated GAM RNA, and which when bound by GAM1662 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1662 target proteins.

[25801] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1662 gene, herein designated GAM GENE, on one or more GAM1662 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific com-

plementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25802] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1662 correlate with, and may be deduced from, the identity of the target genes which GAM1662 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25803] Nucleotide sequences of the GAM1662 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1662 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1662 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1662 are further described hereinbelow with reference to Table Table1.

[25804] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1662 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25805] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present in-

vention, referred to here as Genomic Address Messenger 1663 (GAM1663) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25806] GAM1663 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1663 was detected is described hereinabove with reference to Figs. 2–8.

[25807] GAM1663 gene, herein designated GAM GENE, and GAM1663 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25808] GAM1663 gene, herein designated GAM GENE, encodes a GAM1663 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1663 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1663 precursor RNA is designated SEQ ID:1636, and is provided hereinbelow with reference to the sequence listing part.

[25809] GAM1663 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1663 folded precursor RNA, herein designated GAM FOLDED PRECUR-

SOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25810] An enzyme complex designated DICER COMPLEX, dices the GAM1663 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1663 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1663 RNA is designated SEQ ID:3312, and is provided hereinbelow with reference to the sequence listing part.

[25811] GAM1663 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1663 target RNA, herein designated GAM TARGET RNA. GAM1663 target RNA, herein designated GAM TAR-

GET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25812] GAM1663 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1663 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1663 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1663 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1663 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in

the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25813] The complementary binding of GAM1663 RNA, herein designated GAM RNA, to target binding sites on GAM1663 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1663 target RNA, herein designated GAM TARGET RNA, into GAM1663 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25814] It is appreciated that GAM1663 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1663 target genes. The mRNA of each one of this plurality of GAM1663 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1663 RNA, herein designated GAM RNA, and which when bound by GAM1663 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1663 target proteins.

[25815] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by

GAM1663 gene, herein designated GAM GENE, on one or more GAM1663 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25816] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1663 correlate with, and may be deduced from, the identity of the target genes which GAM1663 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25817] Nucleotide sequences of the GAM1663 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1663 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1663 folded precursor RNA, herein designated GAM

FOLDED PRECURSOR RNA, of GAM1663 are further described hereinbelow with reference to Table Table1.

[25818] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1663 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25819] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1664 (GAM1664) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25820] GAM1664 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1664 was detected is described hereinabove with reference to Figs. 2-8.

[25821] GAM1664 gene, herein designated GAM GENE, and GAM1664 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25822] GAM1664 gene, herein designated GAM GENE, encodes a GAM1664 precursor RNA, herein designated GAM PRE-

CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1664 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1664 precursor RNA is designated SEQ ID:1637, and is provided hereinbelow with reference to the sequence listing part.

[25823] GAM1664 has a highly conserved, expressed mouse homolog (>90% sequence similarity), the nucleotide sequence of which is similar to the nucleotide sequence of SEQ ID:97948, which mouse homolog is detected in mouse EST accession AW475179.1.

[25824] GAM1664 precursor RNA, herein designated GAM PRECURSOR RNA, folds onto itself, forming GAM1664 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25825] An enzyme complex designated DICER COMPLEX, dices

the GAM1664 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1664 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1664 RNA is designated SEQ ID:3313, and is provided hereinbelow with reference to the sequence listing part.

[25826] GAM1664 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1664 target RNA, herein designated GAM TARGET RNA. GAM1664 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25827] GAM1664 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1664 target RNA, herein designated GAM TARGET RNA. This complementary binding is

due to the fact that the nucleotide sequence of GAM1664 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1664 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1664 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25828] The complementary binding of GAM1664 RNA, herein designated GAM RNA, to target binding sites on GAM1664 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1664 target RNA, herein designated GAM TARGET RNA, into GAM1664 target protein,

herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25829] It is appreciated that GAM1664 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1664 target genes. The mRNA of each one of this plurality of GAM1664 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1664 RNA, herein designated GAM RNA, and which when bound by GAM1664 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1664 target proteins.

[25830] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1664 gene, herein designated GAM GENE, on one or more GAM1664 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by

those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25831] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1664 correlate with, and may be deduced from, the identity of the target genes which GAM1664 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25832] Nucleotide sequences of the GAM1664 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1664 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1664 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1664 are further described hereinbelow with reference to Table Table1.

[25833] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1664 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25834] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1665 (GAM1665) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25835] GAM1665 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1665 was detected is described hereinabove with reference to Figs. 2–8.

[25836] GAM1665 gene, herein designated GAM GENE, and GAM1665 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25837] GAM1665 gene, herein designated GAM GENE, encodes a GAM1665 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1665 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1665 precursor RNA is designated SEQ ID:1638, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1638 is located at position 2349674 rela-

tive to contig NT_021907.13, on chromosome 1.

[25838] GAM1665 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1665 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25839] An enzyme complex designated DICER COMPLEX, dices the GAM1665 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1665 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1665 RNA is designated SEQ ID:3314, and is provided hereinbelow with reference to the sequence listing part.

[25840] GAM1665 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1665 target RNA, herein designated GAM TARGET RNA. GAM1665 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25841] GAM1665 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1665 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1665 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1665 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a

GAM1665 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25842] The complementary binding of GAM1665 RNA, herein designated GAM RNA, to target binding sites on GAM1665 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1665 target RNA, herein designated GAM TARGET RNA, into GAM1665 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25843] It is appreciated that GAM1665 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1665 target genes. The mRNA of each one of this plurality of GAM1665 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1665 RNA, herein designated GAM RNA, and which when bound by GAM1665 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1665

target proteins.

[25844] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1665 gene, herein designated GAM GENE, on one or more GAM1665 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25845] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1665 correlate with, and may be deduced from, the identity of the target genes which GAM1665 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25846] Nucleotide sequences of the GAM1665 precursor RNA,

herein designated GAM PRECURSOR RNA, and of the diced GAM1665 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1665 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1665 are further described hereinbelow with reference to Table Table1.

[25847] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1665 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25848] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1666 (GAM1666) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25849] GAM1666 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1666 was detected is described hereinabove with reference to Figs. 2-8.

[25850] GAM1666 gene, herein designated GAM GENE, and

GAM1666 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25851] GAM1666 gene, herein designated GAM GENE, encodes a GAM1666 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1666 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1666 precursor RNA is designated SEQ ID:1639, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1639 is located at position 3434852 relative to contig NT_009714.13, on chromosome 12.

[25852] GAM1666 has a highly conserved, expressed mouse homolog (>90% sequence similarity), the nucleotide sequence of which is similar to the nucleotide sequence of SEQ ID:97973, which mouse homolog is detected in mouse EST accession BG288438.1.

[25853] GAM1666 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1666 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typi-

cal of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25854] An enzyme complex designated DICER COMPLEX, dices the GAM1666 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1666 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1666 RNA is designated SEQ ID:3315, and is provided hereinbelow with reference to the sequence listing part.

[25855] GAM1666 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1666 target RNA, herein designated GAM TARGET RNA. GAM1666 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a pro-

tein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25856] GAM1666 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1666 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1666 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1666 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1666 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25857] The complementary binding of GAM1666 RNA, herein designated GAM RNA, to target binding sites on GAM1666 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1666 target RNA, herein designated GAM TARGET RNA, into GAM1666 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25858] It is appreciated that GAM1666 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1666 target genes. The mRNA of each one of this plurality of GAM1666 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1666 RNA, herein designated GAM RNA, and which when bound by GAM1666 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1666 target proteins.

[25859] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1666 gene, herein designated GAM GENE, on one or more GAM1666 target gene, herein designated GAM TAR-

GET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25860] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1666 correlate with, and may be deduced from, the identity of the target genes which GAM1666 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25861] Nucleotide sequences of the GAM1666 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1666 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1666 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1666 are further described hereinbelow with reference to Table Table1.

[25862] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1666 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25863] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1667 (GAM1667) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25864] GAM1667 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1667 was detected is described hereinabove with reference to Figs. 2-8.

[25865] GAM1667 gene, herein designated GAM GENE, and GAM1667 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25866] GAM1667 gene, herein designated GAM GENE, encodes a GAM1667 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1667 precursor RNA, herein

designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1667 precursor RNA is designated SEQ ID:1640, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1640 is located at position 1259555 relative to contig NT_025965.9, on chromosome X.

[25867] GAM1667 has 7 highly conserved, expressed mouse homologs (>90% sequence similarity), found in mouse EST's. Nucleotide sequences similar to the nucleotide sequences of which mouse homologs, and the respective mouse EST's in which these homologs were detected are:

[25868] SEQ ID:99543 detected in mouse EST Accession AI098543.1.

[25869] SEQ ID:98077 detected in mouse EST Accession BG807367.1.

[25870] SEQ ID:98078 detected in mouse EST Accession BG915676.1.

[25871] SEQ ID:98079 detected in mouse EST Accession AI115901.1.

[25872] SEQ ID:98080 detected in mouse EST Accession BE371710.1.

[25873] SEQ ID:98083 detected in mouse EST Accession

BI788766.1.

[25874] SEQ ID:98084 detected in mouse EST Accession

BI789775.1.

[25875] GAM1667 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1667 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25876] An enzyme complex designated DICER COMPLEX, dices the GAM1667 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1667 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1667 RNA is designated SEQ ID:3316, and

is provided hereinbelow with reference to the sequence listing part.

- [25877] GAM1667 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1667 target RNA, herein designated GAM TARGET RNA. GAM1667 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25878] GAM1667 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1667 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1667 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1667 RNA,

herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1667 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25879] The complementary binding of GAM1667 RNA, herein designated GAM RNA, to target binding sites on GAM1667 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1667 target RNA, herein designated GAM TARGET RNA, into GAM1667 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25880] It is appreciated that GAM1667 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1667 target genes. The mRNA of each one of this plurality of GAM1667 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1667 RNA, herein designated GAM RNA, and which when bound by

GAM1667 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1667 target proteins.

[25881] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1667 gene, herein designated GAM GENE, on one or more GAM1667 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25882] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1667 correlate with, and may be deduced from, the identity of the target genes which GAM1667 binds and inhibits, and the function of these

target genes, as elaborated hereinbelow.

[25883] Nucleotide sequences of the GAM1667 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1667 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1667 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1667 are further described hereinbelow with reference to Table Table1.

[25884] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1667 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25885] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1668 (GAM1668) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25886] GAM1668 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1668 was detected is described hereinabove

with reference to Figs. 2–8.

[25887] GAM1668 gene, herein designated GAM GENE, and GAM1668 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25888] GAM1668 gene, herein designated GAM GENE, encodes a GAM1668 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1668 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1668 precursor RNA is designated SEQ ID:1641, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1641 is located at position 1235091 relative to contig NT_009526.11, on chromosome 12.

[25889] GAM1668 has 3 highly conserved, expressed mouse homologs (>90% sequence similarity), found in mouse EST's. Nucleotide sequences similar to the nucleotide sequences of which mouse homologs, and the respective mouse EST's in which these homologs were detected are:

[25890] SEQ ID:100020 detected in mouse EST Accession AI326210.1.

[25891] SEQ ID:100021 detected in mouse EST Accession

BI965652.1.

[25892] SEQ ID:98851 detected in mouse EST Accession

BI965972.1.

[25893] GAM1668 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1668 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25894] An enzyme complex designated DICER COMPLEX, dices the GAM1668 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1668 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1668 RNA is designated SEQ ID:3317, and

is provided hereinbelow with reference to the sequence listing part.

- [25895] GAM1668 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1668 target RNA, herein designated GAM TARGET RNA. GAM1668 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25896] GAM1668 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1668 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1668 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1668 RNA,

herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1668 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25897] The complementary binding of GAM1668 RNA, herein designated GAM RNA, to target binding sites on GAM1668 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1668 target RNA, herein designated GAM TARGET RNA, into GAM1668 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25898] It is appreciated that GAM1668 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1668 target genes. The mRNA of each one of this plurality of GAM1668 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1668 RNA, herein designated GAM RNA, and which when bound by

GAM1668 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1668 target proteins.

[25899] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1668 gene, herein designated GAM GENE, on one or more GAM1668 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25900] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1668 correlate with, and may be deduced from, the identity of the target genes which GAM1668 binds and inhibits, and the function of these

target genes, as elaborated hereinbelow.

[25901] Nucleotide sequences of the GAM1668 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1668 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1668 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1668 are further described hereinbelow with reference to Table Table1.

[25902] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1668 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25903] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1669 (GAM1669) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25904] GAM1669 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1669 was detected is described hereinabove

with reference to Figs. 2–8.

[25905] GAM1669 gene, herein designated GAM GENE, and GAM1669 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25906] GAM1669 gene, herein designated GAM GENE, encodes a GAM1669 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1669 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1669 precursor RNA is designated SEQ ID:1642, and is provided hereinbelow with reference to the sequence listing part.

[25907] GAM1669 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1669 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

- [25908] An enzyme complex designated DICER COMPLEX, dices the GAM1669 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1669 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1669 RNA is designated SEQ ID:3318, and is provided hereinbelow with reference to the sequence listing part.
- [25909] GAM1669 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1669 target RNA, herein designated GAM TARGET RNA. GAM1669 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25910] GAM1669 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1669 target RNA, herein des-

ignated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1669 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1669 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1669 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25911] The complementary binding of GAM1669 RNA, herein designated GAM RNA, to target binding sites on GAM1669 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1669 target RNA, herein desig-

nated GAM TARGET RNA, into GAM1669 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25912] It is appreciated that GAM1669 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1669 target genes. The mRNA of each one of this plurality of GAM1669 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1669 RNA, herein designated GAM RNA, and which when bound by GAM1669 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1669 target proteins.

[25913] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1669 gene, herein designated GAM GENE, on one or more GAM1669 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other

recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25914] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1669 correlate with, and may be deduced from, the identity of the target genes which GAM1669 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25915] Nucleotide sequences of the GAM1669 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1669 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1669 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1669 are further described hereinbelow with reference to Table Table1.

[25916] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1669 RNA, herein designated GAM RNA, are de-

scribed hereinbelow with reference to Table Table2.

[25917] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1670 (GAM1670) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25918] GAM1670 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1670 was detected is described hereinabove with reference to Figs. 2–8.

[25919] GAM1670 gene, herein designated GAM GENE, and GAM1670 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25920] GAM1670 gene, herein designated GAM GENE, encodes a GAM1670 precursor RNA, herein designated GAM PRECURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1670 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1670 precursor RNA is designated SEQ ID:1643, and is provided hereinbelow with reference to the sequence listing part. Nucleotide se–

quence SEQ ID:1643 is located at position 5677198 relative to contig NT_028309.8, on chromosome 11.

[25921] GAM1670 has 2 highly conserved, expressed mouse homologs (>90% sequence similarity), found in mouse EST's. Nucleotide sequences similar to the nucleotide sequences of which mouse homologs, and the respective mouse EST's in which these homologs were detected are:

[25922] SEQ ID:99535 detected in mouse EST Accession BI688569.1.

[25923] SEQ ID:99541 detected in mouse EST Accession BF021563.1.

[25924] GAM1670 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1670 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25925] An enzyme complex designated DICER COMPLEX, dices the GAM1670 folded precursor RNA, herein designated

GAM FOLDED PRECURSOR RNA, into GAM1670 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1670 RNA is designated SEQ ID:3319, and is provided hereinbelow with reference to the sequence listing part.

- [25926] GAM1670 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1670 target RNA, herein designated GAM TARGET RNA. GAM1670 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25927] GAM1670 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1670 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1670

RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1670 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1670 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25928] The complementary binding of GAM1670 RNA, herein designated GAM RNA, to target binding sites on GAM1670 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1670 target RNA, herein designated GAM TARGET RNA, into GAM1670 target protein, herein designated GAM TARGET PROTEIN. GAM target

protein is therefore outlined by a broken line.

[25929] It is appreciated that GAM1670 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1670 target genes. The mRNA of each one of this plurality of GAM1670 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1670 RNA, herein designated GAM RNA, and which when bound by GAM1670 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1670 target proteins.

[25930] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1670 gene, herein designated GAM GENE, on one or more GAM1670 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other

genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25931] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1670 correlate with, and may be deduced from, the identity of the target genes which GAM1670 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25932] Nucleotide sequences of the GAM1670 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1670 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1670 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1670 are further described hereinbelow with reference to Table Table1.

[25933] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1670 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25934] Fig. 8 further provides a conceptual description of another

novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1671 (GAM1671) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25935] GAM1671 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1671 was detected is described hereinabove with reference to Figs. 2–8.

[25936] GAM1671 gene, herein designated GAM GENE, and GAM1671 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25937] GAM1671 gene, herein designated GAM GENE, encodes a GAM1671 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1671 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1671 precursor RNA is designated SEQ ID:1644, and is provided hereinbelow with reference to the sequence listing part.

[25938] GAM1671 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1671 folded

precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25939] An enzyme complex designated DICER COMPLEX, dices the GAM1671 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1671 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1671 RNA is designated SEQ ID:3320, and is provided hereinbelow with reference to the sequence listing part.

[25940] GAM1671 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1671 target RNA, herein designated GAM TARGET

RNA. GAM1671 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25941] GAM1671 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1671 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1671 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1671 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1671 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an

example only these target binding sites may be located in the 3'UTR region, the 5'UTR region, or in both 3'UTR and 5'UTR regions.

[25942] The complementary binding of GAM1671 RNA, herein designated GAM RNA, to target binding sites on GAM1671 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1671 target RNA, herein designated GAM TARGET RNA, into GAM1671 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25943] It is appreciated that GAM1671 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1671 target genes. The mRNA of each one of this plurality of GAM1671 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1671 RNA, herein designated GAM RNA, and which when bound by GAM1671 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1671 target proteins.

[25944] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with

specific reference to translational inhibition exerted by GAM1671 gene, herein designated GAM GENE, on one or more GAM1671 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25945] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1671 correlate with, and may be deduced from, the identity of the target genes which GAM1671 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25946] Nucleotide sequences of the GAM1671 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1671 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of

GAM1671 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1671 are further described hereinbelow with reference to Table Table1.

[25947] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1671 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25948] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1672 (GAM1672) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25949] GAM1672 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1672 was detected is described hereinabove with reference to Figs. 2-8.

[25950] GAM1672 gene, herein designated GAM GENE, and GAM1672 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25951] GAM1672 gene, herein designated GAM GENE, encodes a

GAM1672 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1672 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1672 precursor RNA is designated SEQ ID:1645, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1645 is located at position 65229 relative to contig NT_024901.11, on chromosome 17.

[25952] GAM1672 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1672 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25953] An enzyme complex designated DICER COMPLEX, dices the GAM1672 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1672 RNA,

herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1672 RNA is designated SEQ ID:3321, and is provided hereinbelow with reference to the sequence listing part.

- [25954] GAM1672 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1672 target RNA, herein designated GAM TARGET RNA. GAM1672 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [25955] GAM1672 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1672 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1672 RNA, herein designated GAM RNA, is an accurate or a par-

tial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1672 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1672 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25956] The complementary binding of GAM1672 RNA, herein designated GAM RNA, to target binding sites on GAM1672 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1672 target RNA, herein designated GAM TARGET RNA, into GAM1672 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25957] It is appreciated that GAM1672 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1672 target genes. The mRNA of each one of this plurality of GAM1672 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1672 RNA, herein designated GAM RNA, and which when bound by GAM1672 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1672 target proteins.

[25958] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1672 gene, herein designated GAM GENE, on one or more GAM1672 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific com-

plementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25959] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1672 correlate with, and may be deduced from, the identity of the target genes which GAM1672 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25960] Nucleotide sequences of the GAM1672 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1672 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1672 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1672 are further described hereinbelow with reference to Table Table1.

[25961] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1672 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25962] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present in-

vention, referred to here as Genomic Address Messenger 1673 (GAM1673) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25963] GAM1673 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1673 was detected is described hereinabove with reference to Figs. 2–8.

[25964] GAM1673 gene, herein designated GAM GENE, and GAM1673 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25965] GAM1673 gene, herein designated GAM GENE, encodes a GAM1673 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1673 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1673 precursor RNA is designated SEQ ID:1646, and is provided hereinbelow with reference to the sequence listing part.

[25966] GAM1673 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1673 folded precursor RNA, herein designated GAM FOLDED PRECUR-

SOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25967] An enzyme complex designated DICER COMPLEX, dices the GAM1673 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1673 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1673 RNA is designated SEQ ID:3322, and is provided hereinbelow with reference to the sequence listing part.

[25968] GAM1673 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1673 target RNA, herein designated GAM TARGET RNA. GAM1673 target RNA, herein designated GAM TAR-

GET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25969] GAM1673 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1673 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1673 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1673 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1673 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in

the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[25970] The complementary binding of GAM1673 RNA, herein designated GAM RNA, to target binding sites on GAM1673 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1673 target RNA, herein designated GAM TARGET RNA, into GAM1673 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[25971] It is appreciated that GAM1673 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1673 target genes. The mRNA of each one of this plurality of GAM1673 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1673 RNA, herein designated GAM RNA, and which when bound by GAM1673 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1673 target proteins.

[25972] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by

GAM1673 gene, herein designated GAM GENE, on one or more GAM1673 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[25973] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1673 correlate with, and may be deduced from, the identity of the target genes which GAM1673 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[25974] Nucleotide sequences of the GAM1673 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1673 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1673 folded precursor RNA, herein designated GAM

FOLDED PRECURSOR RNA, of GAM1673 are further described hereinbelow with reference to Table Table1.

[25975] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1673 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[25976] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1674 (GAM1674) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[25977] GAM1674 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1674 was detected is described hereinabove with reference to Figs. 2-8.

[25978] GAM1674 gene, herein designated GAM GENE, and GAM1674 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[25979] GAM1674 gene, herein designated GAM GENE, encodes a GAM1674 precursor RNA, herein designated GAM PRE-

CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1674 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1674 precursor RNA is designated SEQ ID:1647, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1647 is located at position 365849 relative to contig NT_010799.11, on chromosome 17.

[25980] GAM1674 has 15 highly conserved, expressed mouse homologs (>90% sequence similarity), found in mouse EST's. Nucleotide sequences similar to the nucleotide sequences of which mouse homologs, and the respective mouse EST's in which these homologs were detected are:

[25981] SEQ ID:100065 detected in mouse EST Accession BG808025.1.

[25982] SEQ ID:100066 detected in mouse EST Accession BG808010.1.

[25983] SEQ ID:100067 detected in mouse EST Accession BG807850.1.

[25984] SEQ ID:100068 detected in mouse EST Accession BG805970.1.

[25985] SEQ ID:100127 detected in mouse EST Accession

AA671455.1.

[25986] SEQ ID:100128 detected in mouse EST Accession
AW824753.1.

[25987] SEQ ID:100129 detected in mouse EST Accession
BE943987.1.

[25988] SEQ ID:100130 detected in mouse EST Accession
BE691413.1.

[25989] SEQ ID:100131 detected in mouse EST Accession
BE691447.1.

[25990] SEQ ID:100132 detected in mouse EST Accession
AA144533.1.

[25991] SEQ ID:100133 detected in mouse EST Accession
BG145596.1.

[25992] SEQ ID:100134 detected in mouse EST Accession
BE692802.1.

[25993] SEQ ID:100135 detected in mouse EST Accession
AW208601.1.

[25994] SEQ ID:100136 detected in mouse EST Accession
BB610842.1.

[25995] SEQ ID:100137 detected in mouse EST Accession
BB609859.1.

[25996] GAM1674 precursor RNA, herein designated GAM PRE-
CURSOR RNA, folds onto itself, forming GAM1674 folded

precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[25997] An enzyme complex designated DICER COMPLEX, dices the GAM1674 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1674 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1674 RNA is designated SEQ ID:3323, and is provided hereinbelow with reference to the sequence listing part.

[25998] GAM1674 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1674 target RNA, herein designated GAM TARGET

RNA. GAM1674 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[25999] GAM1674 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1674 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1674 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1674 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1674 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an

example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26000] The complementary binding of GAM1674 RNA, herein designated GAM RNA, to target binding sites on GAM1674 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1674 target RNA, herein designated GAM TARGET RNA, into GAM1674 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26001] It is appreciated that GAM1674 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1674 target genes. The mRNA of each one of this plurality of GAM1674 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1674 RNA, herein designated GAM RNA, and which when bound by GAM1674 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1674 target proteins.

[26002] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with

specific reference to translational inhibition exerted by GAM1674 gene, herein designated GAM GENE, on one or more GAM1674 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26003] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1674 correlate with, and may be deduced from, the identity of the target genes which GAM1674 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26004] Nucleotide sequences of the GAM1674 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1674 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of

GAM1674 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1674 are further described hereinbelow with reference to Table Table1.

[26005] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1674 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26006] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1675 (GAM1675) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26007] GAM1675 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1675 was detected is described hereinabove with reference to Figs. 2-8.

[26008] GAM1675 gene, herein designated GAM GENE, and GAM1675 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26009] GAM1675 gene, herein designated GAM GENE, encodes a

GAM1675 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1675 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1675 precursor RNA is designated SEQ ID:1648, and is provided hereinbelow with reference to the sequence listing part.

[26010] GAM1675 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1675 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26011] An enzyme complex designated DICER COMPLEX, dices the GAM1675 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1675 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a

hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1675 RNA is designated SEQ ID:3324, and is provided hereinbelow with reference to the sequence listing part.

- [26012] GAM1675 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1675 target RNA, herein designated GAM TARGET RNA. GAM1675 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [26013] GAM1675 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1675 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1675 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustra-

tion, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1675 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1675 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26014] The complementary binding of GAM1675 RNA, herein designated GAM RNA, to target binding sites on GAM1675 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1675 target RNA, herein designated GAM TARGET RNA, into GAM1675 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26015] It is appreciated that GAM1675 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of

GAM1675 target genes. The mRNA of each one of this plurality of GAM1675 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1675 RNA, herein designated GAM RNA, and which when bound by GAM1675 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1675 target proteins.

[26016] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1675 gene, herein designated GAM GENE, on one or more GAM1675 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a

tiny RNA world, Science 294,779 (2001)).

[26017] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1675 correlate with, and may be deduced from, the identity of the target genes which GAM1675 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26018] Nucleotide sequences of the GAM1675 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1675 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1675 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1675 are further described hereinbelow with reference to Table Table1.

[26019] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1675 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26020] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1676 (GAM1676) gene, which modulates expression of

respective target genes thereof, the function and utility of which target genes is known in the art.

[26021] GAM1676 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1676 was detected is described hereinabove with reference to Figs. 2–8.

[26022] GAM1676 gene, herein designated GAM GENE, and GAM1676 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26023] GAM1676 gene, herein designated GAM GENE, encodes a GAM1676 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1676 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1676 precursor RNA is designated SEQ ID:1649, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1649 is located at position 28311228 relative to contig NT_007819.11, on chromosome 7.

[26024] GAM1676 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1676 folded precursor RNA, herein designated GAM FOLDED PRECUR-

SOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26025] An enzyme complex designated DICER COMPLEX, dices the GAM1676 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1676 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1676 RNA is designated SEQ ID:3325, and is provided hereinbelow with reference to the sequence listing part.

[26026] GAM1676 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1676 target RNA, herein designated GAM TARGET RNA. GAM1676 target RNA, herein designated GAM TAR-

GET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[26027] GAM1676 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1676 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1676 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1676 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1676 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in

the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26028] The complementary binding of GAM1676 RNA, herein designated GAM RNA, to target binding sites on GAM1676 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1676 target RNA, herein designated GAM TARGET RNA, into GAM1676 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26029] It is appreciated that GAM1676 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1676 target genes. The mRNA of each one of this plurality of GAM1676 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1676 RNA, herein designated GAM RNA, and which when bound by GAM1676 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1676 target proteins.

[26030] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by

GAM1676 gene, herein designated GAM GENE, on one or more GAM1676 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26031] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1676 correlate with, and may be deduced from, the identity of the target genes which GAM1676 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26032] Nucleotide sequences of the GAM1676 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1676 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1676 folded precursor RNA, herein designated GAM

FOLDED PRECURSOR RNA, of GAM1676 are further described hereinbelow with reference to Table Table1.

[26033] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1676 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26034] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1677 (GAM1677) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26035] GAM1677 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1677 was detected is described hereinabove with reference to Figs. 2-8.

[26036] GAM1677 gene, herein designated GAM GENE, and GAM1677 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26037] GAM1677 gene, herein designated GAM GENE, encodes a GAM1677 precursor RNA, herein designated GAM PRE-

CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1677 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1677 precursor RNA is designated SEQ ID:1650, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1650 is located at position 2183835 relative to contig NT_034383.2, on chromosome 1.

[26038] GAM1677 precursor RNA, herein designated GAM PRECURSOR RNA, folds onto itself, forming GAM1677 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26039] An enzyme complex designated DICER COMPLEX, dices the GAM1677 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1677 RNA, herein designated GAM RNA, a single stranded ~22 nt

long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1677 RNA is designated SEQ ID:3326, and is provided hereinbelow with reference to the sequence listing part.

[26040] GAM1677 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1677 target RNA, herein designated GAM TARGET RNA. GAM1677 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[26041] GAM1677 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1677 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1677 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide se-

quence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1677 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1677 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26042] The complementary binding of GAM1677 RNA, herein designated GAM RNA, to target binding sites on GAM1677 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1677 target RNA, herein designated GAM TARGET RNA, into GAM1677 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26043] It is appreciated that GAM1677 target gene, herein desig-

nated GAM TARGET GENE, in fact represents a plurality of GAM1677 target genes. The mRNA of each one of this plurality of GAM1677 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1677 RNA, herein designated GAM RNA, and which when bound by GAM1677 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1677 target proteins.

[26044] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1677 gene, herein designated GAM GENE, on one or more GAM1677 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have

not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26045] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1677 correlate with, and may be deduced from, the identity of the target genes which GAM1677 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26046] Nucleotide sequences of the GAM1677 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1677 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1677 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1677 are further described hereinbelow with reference to Table Table1.

[26047] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1677 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26048] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger

1678 (GAM1678) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26049] GAM1678 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1678 was detected is described hereinabove with reference to Figs. 2–8.

[26050] GAM1678 gene, herein designated GAM GENE, and GAM1678 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26051] GAM1678 gene, herein designated GAM GENE, encodes a GAM1678 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1678 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1678 precursor RNA is designated SEQ ID:1651, and is provided hereinbelow with reference to the sequence listing part.

[26052] GAM1678 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1678 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure.

As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26053] An enzyme complex designated DICER COMPLEX, dices the GAM1678 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1678 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1678 RNA is designated SEQ ID:3327, and is provided hereinbelow with reference to the sequence listing part.

[26054] GAM1678 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1678 target RNA, herein designated GAM TARGET RNA. GAM1678 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA

of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[26055] GAM1678 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1678 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1678 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1678 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1678 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and

5UTR regions.

[26056] The complementary binding of GAM1678 RNA, herein designated GAM RNA, to target binding sites on GAM1678 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1678 target RNA, herein designated GAM TARGET RNA, into GAM1678 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26057] It is appreciated that GAM1678 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1678 target genes. The mRNA of each one of this plurality of GAM1678 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1678 RNA, herein designated GAM RNA, and which when bound by GAM1678 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1678 target proteins.

[26058] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1678 gene, herein designated GAM GENE, on one or

more GAM1678 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26059] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1678 correlate with, and may be deduced from, the identity of the target genes which GAM1678 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26060] Nucleotide sequences of the GAM1678 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1678 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1678 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1678 are further de-

scribed hereinbelow with reference to Table Table1.

[26061] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1678 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26062] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1679 (GAM1679) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26063] GAM1679 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1679 was detected is described hereinabove with reference to Figs. 2-8.

[26064] GAM1679 gene, herein designated GAM GENE, and GAM1679 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26065] GAM1679 gene, herein designated GAM GENE, encodes a GAM1679 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike

most ordinary genes, GAM1679 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1679 precursor RNA is designated SEQ ID:1652, and is provided hereinbelow with reference to the sequence listing part.

[26066] GAM1679 precursor RNA, herein designated GAM PRECURSOR RNA, folds onto itself, forming GAM1679 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26067] An enzyme complex designated DICER COMPLEX, dices the GAM1679 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1679 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex

comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1679 RNA is designated SEQ ID:3328, and is provided hereinbelow with reference to the sequence listing part.

- [26068] GAM1679 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1679 target RNA, herein designated GAM TARGET RNA. GAM1679 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [26069] GAM1679 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1679 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1679 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III re-

spectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1679 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1679 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26070] The complementary binding of GAM1679 RNA, herein designated GAM RNA, to target binding sites on GAM1679 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1679 target RNA, herein designated GAM TARGET RNA, into GAM1679 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26071] It is appreciated that GAM1679 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1679 target genes. The mRNA of each one of this plurality of GAM1679 target genes comprises one or more

target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1679 RNA, herein designated GAM RNA, and which when bound by GAM1679 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1679 target proteins.

[26072] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1679 gene, herein designated GAM GENE, on one or more GAM1679 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26073] It is yet further appreciated that specific functions, and

accordingly utilities, of GAM1679 correlate with, and may be deduced from, the identity of the target genes which GAM1679 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26074] Nucleotide sequences of the GAM1679 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1679 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1679 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1679 are further described hereinbelow with reference to Table Table1.

[26075] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1679 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26076] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1680 (GAM1680) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26077] GAM1680 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1680 was detected is described hereinabove with reference to Figs. 2–8.

[26078] GAM1680 gene, herein designated GAM GENE, and GAM1680 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26079] GAM1680 gene, herein designated GAM GENE, encodes a GAM1680 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1680 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1680 precursor RNA is designated SEQ ID:1653, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1653 is located at position 4851359 relative to contig NT_022517.13, on chromosome 3.

[26080] GAM1680 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1680 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typi-

cal of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26081] An enzyme complex designated DICER COMPLEX, dices the GAM1680 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1680 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1680 RNA is designated SEQ ID:3329, and is provided hereinbelow with reference to the sequence listing part.

[26082] GAM1680 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1680 target RNA, herein designated GAM TARGET RNA. GAM1680 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a pro-

tein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[26083] GAM1680 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1680 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1680 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1680 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1680 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26084] The complementary binding of GAM1680 RNA, herein designated GAM RNA, to target binding sites on GAM1680 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1680 target RNA, herein designated GAM TARGET RNA, into GAM1680 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26085] It is appreciated that GAM1680 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1680 target genes. The mRNA of each one of this plurality of GAM1680 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1680 RNA, herein designated GAM RNA, and which when bound by GAM1680 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1680 target proteins.

[26086] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1680 gene, herein designated GAM GENE, on one or more GAM1680 target gene, herein designated GAM TAR-

GET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26087] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1680 correlate with, and may be deduced from, the identity of the target genes which GAM1680 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26088] Nucleotide sequences of the GAM1680 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1680 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1680 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1680 are further described hereinbelow with reference to Table Table1.

[26089] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1680 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26090] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1681 (GAM1681) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26091] GAM1681 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1681 was detected is described hereinabove with reference to Figs. 2-8.

[26092] GAM1681 gene, herein designated GAM GENE, and GAM1681 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26093] GAM1681 gene, herein designated GAM GENE, encodes a GAM1681 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1681 precursor RNA, herein

designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1681 precursor RNA is designated SEQ ID:1654, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1654 is located at position 4851359 relative to contig NT_022517.13, on chromosome 3.

[26094] GAM1681 precursor RNA, herein designated GAM PRECURSOR RNA, folds onto itself, forming GAM1681 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26095] An enzyme complex designated DICER COMPLEX, dices the GAM1681 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1681 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short

~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1681 RNA is designated SEQ ID:3330, and is provided hereinbelow with reference to the sequence listing part.

- [26096] GAM1681 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1681 target RNA, herein designated GAM TARGET RNA. GAM1681 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [26097] GAM1681 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1681 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1681 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated

BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1681 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1681 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26098] The complementary binding of GAM1681 RNA, herein designated GAM RNA, to target binding sites on GAM1681 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1681 target RNA, herein designated GAM TARGET RNA, into GAM1681 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26099] It is appreciated that GAM1681 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1681 target genes. The mRNA of each one of this

plurality of GAM1681 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1681 RNA, herein designated GAM RNA, and which when bound by GAM1681 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1681 target proteins.

[26100] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1681 gene, herein designated GAM GENE, on one or more GAM1681 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

- [26101] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1681 correlate with, and may be deduced from, the identity of the target genes which GAM1681 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.
- [26102] Nucleotide sequences of the GAM1681 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1681 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1681 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1681 are further described hereinbelow with reference to Table Table1.
- [26103] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1681 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.
- [26104] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1682 (GAM1682) gene, which modulates expression of respective target genes thereof, the function and utility of

which target genes is known in the art.

[26105] GAM1682 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1682 was detected is described hereinabove with reference to Figs. 2–8.

[26106] GAM1682 gene, herein designated GAM GENE, and GAM1682 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26107] GAM1682 gene, herein designated GAM GENE, encodes a GAM1682 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1682 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1682 precursor RNA is designated SEQ ID:1655, and is provided hereinbelow with reference to the sequence listing part.

[26108] GAM1682 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1682 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the

fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26109] An enzyme complex designated DICER COMPLEX, dices the GAM1682 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1682 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1682 RNA is designated SEQ ID:3331, and is provided hereinbelow with reference to the sequence listing part.

[26110] GAM1682 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1682 target RNA, herein designated GAM TARGET RNA. GAM1682 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, design-

nated 5UTR, PROTEIN CODING and 3UTR respectively.

[26111] GAM1682 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1682 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1682 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1682 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1682 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26112] The complementary binding of GAM1682 RNA, herein

designated GAM RNA, to target binding sites on GAM1682 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1682 target RNA, herein designated GAM TARGET RNA, into GAM1682 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26113] It is appreciated that GAM1682 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1682 target genes. The mRNA of each one of this plurality of GAM1682 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1682 RNA, herein designated GAM RNA, and which when bound by GAM1682 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1682 target proteins.

[26114] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1682 gene, herein designated GAM GENE, on one or more GAM1682 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA

genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26115] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1682 correlate with, and may be deduced from, the identity of the target genes which GAM1682 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26116] Nucleotide sequences of the GAM1682 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1682 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1682 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1682 are further described hereinbelow with reference to Table Table1.

[26117] Nucleotide sequences of target binding sites, such as

BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1682 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26118] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1683 (GAM1683) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26119] GAM1683 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1683 was detected is described hereinabove with reference to Figs. 2-8.

[26120] GAM1683 gene, herein designated GAM GENE, and GAM1683 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26121] GAM1683 gene, herein designated GAM GENE, encodes a GAM1683 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1683 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a pro-

tein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1683 precursor RNA is designated SEQ ID:1656, and is provided hereinbelow with reference to the sequence listing part.

[26122] GAM1683 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1683 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26123] An enzyme complex designated DICER COMPLEX, dices the GAM1683 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1683 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide se-

quence of GAM1683 RNA is designated SEQ ID:3332, and is provided hereinbelow with reference to the sequence listing part.

- [26124] GAM1683 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1683 target RNA, herein designated GAM TARGET RNA. GAM1683 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [26125] GAM1683 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1683 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1683 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration

only, and is not meant to be limiting GAM1683 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1683 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26126] The complementary binding of GAM1683 RNA, herein designated GAM RNA, to target binding sites on GAM1683 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1683 target RNA, herein designated GAM TARGET RNA, into GAM1683 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26127] It is appreciated that GAM1683 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1683 target genes. The mRNA of each one of this plurality of GAM1683 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1683 RNA,

herein designated GAM RNA, and which when bound by GAM1683 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1683 target proteins.

[26128] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1683 gene, herein designated GAM GENE, on one or more GAM1683 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26129] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1683 correlate with, and may be deduced from, the identity of the target genes which

GAM1683 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26130] Nucleotide sequences of the GAM1683 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1683 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1683 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1683 are further described hereinbelow with reference to Table Table1.

[26131] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1683 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26132] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1684 (GAM1684) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26133] GAM1684 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method

by which GAM1684 was detected is described hereinabove with reference to Figs. 2–8.

[26134] GAM1684 gene, herein designated GAM GENE, and GAM1684 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26135] GAM1684 gene, herein designated GAM GENE, encodes a GAM1684 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1684 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1684 precursor RNA is designated SEQ ID:1657, and is provided hereinbelow with reference to the sequence listing part.

[26136] GAM1684 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1684 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of

the second half thereof.

- [26137] An enzyme complex designated DICER COMPLEX, dices the GAM1684 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1684 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1684 RNA is designated SEQ ID:3333, and is provided hereinbelow with reference to the sequence listing part.
- [26138] GAM1684 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1684 target RNA, herein designated GAM TARGET RNA. GAM1684 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [26139] GAM1684 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in

untranslated regions of GAM1684 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1684 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1684 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1684 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26140] The complementary binding of GAM1684 RNA, herein designated GAM RNA, to target binding sites on GAM1684 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, in-

hibits translation of GAM1684 target RNA, herein designated GAM TARGET RNA, into GAM1684 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26141] It is appreciated that GAM1684 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1684 target genes. The mRNA of each one of this plurality of GAM1684 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1684 RNA, herein designated GAM RNA, and which when bound by GAM1684 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1684 target proteins.

[26142] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1684 gene, herein designated GAM GENE, on one or more GAM1684 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the

known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26143] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1684 correlate with, and may be deduced from, the identity of the target genes which GAM1684 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26144] Nucleotide sequences of the GAM1684 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1684 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1684 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1684 are further described hereinbelow with reference to Table Table1.

[26145] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to

GAM1684 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26146] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1685 (GAM1685) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26147] GAM1685 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1685 was detected is described hereinabove with reference to Figs. 2–8.

[26148] GAM1685 gene, herein designated GAM GENE, and GAM1685 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26149] GAM1685 gene, herein designated GAM GENE, encodes a GAM1685 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1685 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1685 precursor RNA is designated SEQ ID:1658, and is provided hereinbelow with

reference to the sequence listing part.

[26150] GAM1685 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1685 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26151] An enzyme complex designated DICER COMPLEX, dices the GAM1685 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1685 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1685 RNA is designated SEQ ID:3334, and is provided hereinbelow with reference to the sequence listing part.

[26152] GAM1685 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1685 target RNA, herein designated GAM TARGET RNA. GAM1685 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[26153] GAM1685 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1685 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1685 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1685 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a

GAM1685 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26154] The complementary binding of GAM1685 RNA, herein designated GAM RNA, to target binding sites on GAM1685 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1685 target RNA, herein designated GAM TARGET RNA, into GAM1685 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26155] It is appreciated that GAM1685 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1685 target genes. The mRNA of each one of this plurality of GAM1685 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1685 RNA, herein designated GAM RNA, and which when bound by GAM1685 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1685

target proteins.

[26156] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1685 gene, herein designated GAM GENE, on one or more GAM1685 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26157] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1685 correlate with, and may be deduced from, the identity of the target genes which GAM1685 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26158] Nucleotide sequences of the GAM1685 precursor RNA,

herein designated GAM PRECURSOR RNA, and of the diced GAM1685 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1685 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1685 are further described hereinbelow with reference to Table Table1.

[26159] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1685 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26160] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1686 (GAM1686) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26161] GAM1686 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1686 was detected is described hereinabove with reference to Figs. 2-8.

[26162] GAM1686 gene, herein designated GAM GENE, and

GAM1686 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26163] GAM1686 gene, herein designated GAM GENE, encodes a GAM1686 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1686 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1686 precursor RNA is designated SEQ ID:1659, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1659 is located at position 387404 relative to contig NT_022508.11, on chromosome 3.

[26164] GAM1686 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1686 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

- [26165] An enzyme complex designated DICER COMPLEX, dices the GAM1686 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1686 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1686 RNA is designated SEQ ID:3335, and is provided hereinbelow with reference to the sequence listing part.
- [26166] GAM1686 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1686 target RNA, herein designated GAM TARGET RNA. GAM1686 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [26167] GAM1686 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1686 target RNA, herein des-

ignated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1686 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1686 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1686 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26168] The complementary binding of GAM1686 RNA, herein designated GAM RNA, to target binding sites on GAM1686 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1686 target RNA, herein design-

nated GAM TARGET RNA, into GAM1686 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26169] It is appreciated that GAM1686 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1686 target genes. The mRNA of each one of this plurality of GAM1686 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1686 RNA, herein designated GAM RNA, and which when bound by GAM1686 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1686 target proteins.

[26170] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1686 gene, herein designated GAM GENE, on one or more GAM1686 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other

recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26171] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1686 correlate with, and may be deduced from, the identity of the target genes which GAM1686 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26172] Nucleotide sequences of the GAM1686 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1686 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1686 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1686 are further described hereinbelow with reference to Table Table1.

[26173] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1686 RNA, herein designated GAM RNA, are de-

scribed hereinbelow with reference to Table Table2.

[26174] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1687 (GAM1687) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26175] GAM1687 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1687 was detected is described hereinabove with reference to Figs. 2–8.

[26176] GAM1687 gene, herein designated GAM GENE, and GAM1687 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26177] GAM1687 gene, herein designated GAM GENE, encodes a GAM1687 precursor RNA, herein designated GAM PRECURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1687 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1687 precursor RNA is designated SEQ ID:1660, and is provided hereinbelow with reference to the sequence listing part. Nucleotide se–

quence SEQ ID:1660 is located at position 387404 relative to contig NT_022508.11, on chromosome 3.

[26178] GAM1687 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1687 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26179] An enzyme complex designated DICER COMPLEX, dices the GAM1687 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1687 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1687 RNA is designated SEQ ID:3336, and is provided hereinbelow with reference to the sequence

listing part.

- [26180] GAM1687 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1687 target RNA, herein designated GAM TARGET RNA. GAM1687 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [26181] GAM1687 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1687 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1687 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1687 RNA, herein designated GAM RNA, may have a different number

of target binding sites in untranslated regions of a GAM1687 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26182] The complementary binding of GAM1687 RNA, herein designated GAM RNA, to target binding sites on GAM1687 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1687 target RNA, herein designated GAM TARGET RNA, into GAM1687 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26183] It is appreciated that GAM1687 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1687 target genes. The mRNA of each one of this plurality of GAM1687 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1687 RNA, herein designated GAM RNA, and which when bound by GAM1687 RNA, herein designated GAM RNA, causes inhi-

bition of translation of respective one or more GAM1687 target proteins.

[26184] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1687 gene, herein designated GAM GENE, on one or more GAM1687 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26185] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1687 correlate with, and may be deduced from, the identity of the target genes which GAM1687 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26186] Nucleotide sequences of the GAM1687 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1687 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1687 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1687 are further described hereinbelow with reference to Table Table1.

[26187] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1687 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26188] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1688 (GAM1688) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26189] GAM1688 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1688 was detected is described hereinabove with reference to Figs. 2-8.

[26190] GAM1688 gene, herein designated GAM GENE, and GAM1688 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26191] GAM1688 gene, herein designated GAM GENE, encodes a GAM1688 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1688 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1688 precursor RNA is designated SEQ ID:1661, and is provided hereinbelow with reference to the sequence listing part.

[26192] GAM1688 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1688 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26193] An enzyme complex designated DICER COMPLEX, dices

the GAM1688 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1688 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1688 RNA is designated SEQ ID:3337, and is provided hereinbelow with reference to the sequence listing part.

- [26194] GAM1688 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1688 target RNA, herein designated GAM TARGET RNA. GAM1688 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [26195] GAM1688 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1688 target RNA, herein designated GAM TARGET RNA. This complementary binding is

due to the fact that the nucleotide sequence of GAM1688 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1688 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1688 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26196] The complementary binding of GAM1688 RNA, herein designated GAM RNA, to target binding sites on GAM1688 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1688 target RNA, herein designated GAM TARGET RNA, into GAM1688 target protein,

herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26197] It is appreciated that GAM1688 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1688 target genes. The mRNA of each one of this plurality of GAM1688 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1688 RNA, herein designated GAM RNA, and which when bound by GAM1688 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1688 target proteins.

[26198] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1688 gene, herein designated GAM GENE, on one or more GAM1688 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by

those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26199] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1688 correlate with, and may be deduced from, the identity of the target genes which GAM1688 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26200] Nucleotide sequences of the GAM1688 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1688 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1688 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1688 are further described hereinbelow with reference to Table Table1.

[26201] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1688 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26202] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1689 (GAM1689) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26203] GAM1689 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1689 was detected is described hereinabove with reference to Figs. 2–8.

[26204] GAM1689 gene, herein designated GAM GENE, and GAM1689 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26205] GAM1689 gene, herein designated GAM GENE, encodes a GAM1689 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1689 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1689 precursor RNA is designated SEQ ID:1662, and is provided hereinbelow with reference to the sequence listing part.

[26206] GAM1689 precursor RNA, herein designated GAM PRE-

CURSOR RNA, folds onto itself, forming GAM1689 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26207] An enzyme complex designated DICER COMPLEX, dices the GAM1689 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1689 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1689 RNA is designated SEQ ID:3338, and is provided hereinbelow with reference to the sequence listing part.

[26208] GAM1689 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA,

GAM1689 target RNA, herein designated GAM TARGET RNA. GAM1689 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[26209] GAM1689 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1689 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1689 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1689 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1689 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts tar-

get binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26210] The complementary binding of GAM1689 RNA, herein designated GAM RNA, to target binding sites on GAM1689 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1689 target RNA, herein designated GAM TARGET RNA, into GAM1689 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26211] It is appreciated that GAM1689 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1689 target genes. The mRNA of each one of this plurality of GAM1689 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1689 RNA, herein designated GAM RNA, and which when bound by GAM1689 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1689 target proteins.

[26212] It is further appreciated by one skilled in the art that the

mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1689 gene, herein designated GAM GENE, on one or more GAM1689 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26213] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1689 correlate with, and may be deduced from, the identity of the target genes which GAM1689 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26214] Nucleotide sequences of the GAM1689 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1689 RNA, herein designated GAM RNA, and a

schematic representation of the secondary folding of GAM1689 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1689 are further described hereinbelow with reference to Table Table1.

[26215] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1689 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26216] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1690 (GAM1690) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26217] GAM1690 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1690 was detected is described hereinabove with reference to Figs. 2-8.

[26218] GAM1690 gene, herein designated GAM GENE, and GAM1690 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26219] GAM1690 gene, herein designated GAM GENE, encodes a GAM1690 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1690 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1690 precursor RNA is designated SEQ ID:1663, and is provided hereinbelow with reference to the sequence listing part.

[26220] GAM1690 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1690 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26221] An enzyme complex designated DICER COMPLEX, dices the GAM1690 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1690 RNA, herein designated GAM RNA, a single stranded ~22 nt

long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1690 RNA is designated SEQ ID:3339, and is provided hereinbelow with reference to the sequence listing part.

[26222] GAM1690 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1690 target RNA, herein designated GAM TARGET RNA. GAM1690 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[26223] GAM1690 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1690 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1690 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide se-

quence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1690 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1690 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26224] The complementary binding of GAM1690 RNA, herein designated GAM RNA, to target binding sites on GAM1690 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1690 target RNA, herein designated GAM TARGET RNA, into GAM1690 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26225] It is appreciated that GAM1690 target gene, herein desig-

nated GAM TARGET GENE, in fact represents a plurality of GAM1690 target genes. The mRNA of each one of this plurality of GAM1690 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1690 RNA, herein designated GAM RNA, and which when bound by GAM1690 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1690 target proteins.

[26226] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1690 gene, herein designated GAM GENE, on one or more GAM1690 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have

not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26227] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1690 correlate with, and may be deduced from, the identity of the target genes which GAM1690 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26228] Nucleotide sequences of the GAM1690 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1690 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1690 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1690 are further described hereinbelow with reference to Table Table1.

[26229] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1690 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26230] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger

1691 (GAM1691) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26231] GAM1691 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1691 was detected is described hereinabove with reference to Figs. 2–8.

[26232] GAM1691 gene, herein designated GAM GENE, and GAM1691 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26233] GAM1691 gene, herein designated GAM GENE, encodes a GAM1691 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1691 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1691 precursor RNA is designated SEQ ID:1664, and is provided hereinbelow with reference to the sequence listing part.

[26234] GAM1691 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1691 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure.

As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26235] An enzyme complex designated DICER COMPLEX, dices the GAM1691 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1691 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1691 RNA is designated SEQ ID:3340, and is provided hereinbelow with reference to the sequence listing part.

[26236] GAM1691 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1691 target RNA, herein designated GAM TARGET RNA. GAM1691 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA

of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[26237] GAM1691 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1691 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1691 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1691 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1691 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and

5UTR regions.

[26238] The complementary binding of GAM1691 RNA, herein designated GAM RNA, to target binding sites on GAM1691 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1691 target RNA, herein designated GAM TARGET RNA, into GAM1691 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26239] It is appreciated that GAM1691 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1691 target genes. The mRNA of each one of this plurality of GAM1691 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1691 RNA, herein designated GAM RNA, and which when bound by GAM1691 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1691 target proteins.

[26240] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1691 gene, herein designated GAM GENE, on one or

more GAM1691 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26241] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1691 correlate with, and may be deduced from, the identity of the target genes which GAM1691 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26242] Nucleotide sequences of the GAM1691 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1691 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1691 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1691 are further de-

scribed hereinbelow with reference to Table Table1.

[26243] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1691 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26244] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1692 (GAM1692) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26245] GAM1692 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1692 was detected is described hereinabove with reference to Figs. 2-8.

[26246] GAM1692 gene, herein designated GAM GENE, and GAM1692 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26247] GAM1692 gene, herein designated GAM GENE, encodes a GAM1692 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike

most ordinary genes, GAM1692 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1692 precursor RNA is designated SEQ ID:1665, and is provided hereinbelow with reference to the sequence listing part.

[26248] GAM1692 has 7 highly conserved, expressed mouse homologs (>90% sequence similarity), found in mouse EST's. Nucleotide sequences similar to the nucleotide sequences of which mouse homologs, and the respective mouse EST's in which these homologs were detected are:

[26249] SEQ ID:100000 detected in mouse EST Accession BF152625.1.

[26250] SEQ ID:100001 detected in mouse EST Accession AI614809.1.

[26251] SEQ ID:100002 detected in mouse EST Accession AA065653.1.

[26252] SEQ ID:100003 detected in mouse EST Accession AI153186.1.

[26253] SEQ ID:100004 detected in mouse EST Accession AW744693.1.

[26254] SEQ ID:100005 detected in mouse EST Accession BF101508.1.

[26255] SEQ ID:100006 detected in mouse EST Accession BG261697.1.

[26256] GAM1692 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1692 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26257] An enzyme complex designated DICER COMPLEX, dices the GAM1692 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1692 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1692 RNA is designated SEQ ID:3341, and is provided hereinbelow with reference to the sequence

listing part.

- [26258] GAM1692 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1692 target RNA, herein designated GAM TARGET RNA. GAM1692 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [26259] GAM1692 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1692 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1692 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1692 RNA, herein designated GAM RNA, may have a different number

of target binding sites in untranslated regions of a GAM1692 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26260] The complementary binding of GAM1692 RNA, herein designated GAM RNA, to target binding sites on GAM1692 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1692 target RNA, herein designated GAM TARGET RNA, into GAM1692 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26261] It is appreciated that GAM1692 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1692 target genes. The mRNA of each one of this plurality of GAM1692 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1692 RNA, herein designated GAM RNA, and which when bound by GAM1692 RNA, herein designated GAM RNA, causes inhi-

bition of translation of respective one or more GAM1692 target proteins.

[26262] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1692 gene, herein designated GAM GENE, on one or more GAM1692 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26263] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1692 correlate with, and may be deduced from, the identity of the target genes which GAM1692 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26264] Nucleotide sequences of the GAM1692 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1692 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1692 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1692 are further described hereinbelow with reference to Table Table1.

[26265] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1692 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26266] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1693 (GAM1693) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26267] GAM1693 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1693 was detected is described hereinabove with reference to Figs. 2-8.

[26268] GAM1693 gene, herein designated GAM GENE, and GAM1693 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26269] GAM1693 gene, herein designated GAM GENE, encodes a GAM1693 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1693 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1693 precursor RNA is designated SEQ ID:1666, and is provided hereinbelow with reference to the sequence listing part.

[26270] GAM1693 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1693 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26271] An enzyme complex designated DICER COMPLEX, dices

the GAM1693 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1693 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1693 RNA is designated SEQ ID:3342, and is provided hereinbelow with reference to the sequence listing part.

[26272] GAM1693 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1693 target RNA, herein designated GAM TARGET RNA. GAM1693 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[26273] GAM1693 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1693 target RNA, herein designated GAM TARGET RNA. This complementary binding is

due to the fact that the nucleotide sequence of GAM1693 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1693 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1693 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26274] The complementary binding of GAM1693 RNA, herein designated GAM RNA, to target binding sites on GAM1693 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1693 target RNA, herein designated GAM TARGET RNA, into GAM1693 target protein,

herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26275] It is appreciated that GAM1693 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1693 target genes. The mRNA of each one of this plurality of GAM1693 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1693 RNA, herein designated GAM RNA, and which when bound by GAM1693 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1693 target proteins.

[26276] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1693 gene, herein designated GAM GENE, on one or more GAM1693 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by

those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26277] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1693 correlate with, and may be deduced from, the identity of the target genes which GAM1693 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26278] Nucleotide sequences of the GAM1693 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1693 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1693 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1693 are further described hereinbelow with reference to Table Table1.

[26279] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1693 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26280] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1694 (GAM1694) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26281] GAM1694 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1694 was detected is described hereinabove with reference to Figs. 2–8.

[26282] GAM1694 gene, herein designated GAM GENE, and GAM1694 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26283] GAM1694 gene, herein designated GAM GENE, encodes a GAM1694 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1694 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1694 precursor RNA is designated SEQ ID:1667, and is provided hereinbelow with reference to the sequence listing part.

[26284] GAM1694 precursor RNA, herein designated GAM PRE-

CURSOR RNA, folds onto itself, forming GAM1694 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26285] An enzyme complex designated DICER COMPLEX, dices the GAM1694 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1694 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1694 RNA is designated SEQ ID:3343, and is provided hereinbelow with reference to the sequence listing part.

[26286] GAM1694 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA,

GAM1694 target RNA, herein designated GAM TARGET RNA. GAM1694 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[26287] GAM1694 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1694 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1694 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1694 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1694 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts tar-

get binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26288] The complementary binding of GAM1694 RNA, herein designated GAM RNA, to target binding sites on GAM1694 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1694 target RNA, herein designated GAM TARGET RNA, into GAM1694 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26289] It is appreciated that GAM1694 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1694 target genes. The mRNA of each one of this plurality of GAM1694 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1694 RNA, herein designated GAM RNA, and which when bound by GAM1694 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1694 target proteins.

[26290] It is further appreciated by one skilled in the art that the

mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1694 gene, herein designated GAM GENE, on one or more GAM1694 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26291] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1694 correlate with, and may be deduced from, the identity of the target genes which GAM1694 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26292] Nucleotide sequences of the GAM1694 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1694 RNA, herein designated GAM RNA, and a

schematic representation of the secondary folding of GAM1694 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1694 are further described hereinbelow with reference to Table Table1.

[26293] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1694 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26294] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1695 (GAM1695) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26295] GAM1695 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1695 was detected is described hereinabove with reference to Figs. 2-8.

[26296] GAM1695 gene, herein designated GAM GENE, and GAM1695 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26297] GAM1695 gene, herein designated GAM GENE, encodes a GAM1695 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1695 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1695 precursor RNA is designated SEQ ID:1668, and is provided hereinbelow with reference to the sequence listing part.

[26298] GAM1695 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1695 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26299] An enzyme complex designated DICER COMPLEX, dices the GAM1695 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1695 RNA, herein designated GAM RNA, a single stranded ~22 nt

long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1695 RNA is designated SEQ ID:3344, and is provided hereinbelow with reference to the sequence listing part.

[26300] GAM1695 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1695 target RNA, herein designated GAM TARGET RNA. GAM1695 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[26301] GAM1695 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1695 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1695 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide se-

quence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1695 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1695 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26302] The complementary binding of GAM1695 RNA, herein designated GAM RNA, to target binding sites on GAM1695 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1695 target RNA, herein designated GAM TARGET RNA, into GAM1695 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26303] It is appreciated that GAM1695 target gene, herein desig-

nated GAM TARGET GENE, in fact represents a plurality of GAM1695 target genes. The mRNA of each one of this plurality of GAM1695 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1695 RNA, herein designated GAM RNA, and which when bound by GAM1695 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1695 target proteins.

[26304] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1695 gene, herein designated GAM GENE, on one or more GAM1695 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have

not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26305] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1695 correlate with, and may be deduced from, the identity of the target genes which GAM1695 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26306] Nucleotide sequences of the GAM1695 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1695 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1695 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1695 are further described hereinbelow with reference to Table Table1.

[26307] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1695 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26308] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger

1696 (GAM1696) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26309] GAM1696 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1696 was detected is described hereinabove with reference to Figs. 2–8.

[26310] GAM1696 gene, herein designated GAM GENE, and GAM1696 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26311] GAM1696 gene, herein designated GAM GENE, encodes a GAM1696 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1696 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1696 precursor RNA is designated SEQ ID:1669, and is provided hereinbelow with reference to the sequence listing part.

[26312] GAM1696 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1696 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure.

As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26313] An enzyme complex designated DICER COMPLEX, dices the GAM1696 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1696 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1696 RNA is designated SEQ ID:3345, and is provided hereinbelow with reference to the sequence listing part.

[26314] GAM1696 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1696 target RNA, herein designated GAM TARGET RNA. GAM1696 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA

of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.

[26315] GAM1696 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1696 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1696 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1696 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1696 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and

5UTR regions.

[26316] The complementary binding of GAM1696 RNA, herein designated GAM RNA, to target binding sites on GAM1696 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1696 target RNA, herein designated GAM TARGET RNA, into GAM1696 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26317] It is appreciated that GAM1696 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1696 target genes. The mRNA of each one of this plurality of GAM1696 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1696 RNA, herein designated GAM RNA, and which when bound by GAM1696 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1696 target proteins.

[26318] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1696 gene, herein designated GAM GENE, on one or

more GAM1696 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26319] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1696 correlate with, and may be deduced from, the identity of the target genes which GAM1696 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26320] Nucleotide sequences of the GAM1696 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1696 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1696 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1696 are further de-

scribed hereinbelow with reference to Table Table1.

[26321] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1696 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26322] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1697 (GAM1697) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26323] GAM1697 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1697 was detected is described hereinabove with reference to Figs. 2-8.

[26324] GAM1697 gene, herein designated GAM GENE, and GAM1697 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26325] GAM1697 gene, herein designated GAM GENE, encodes a GAM1697 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike

most ordinary genes, GAM1697 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1697 precursor RNA is designated SEQ ID:1670, and is provided hereinbelow with reference to the sequence listing part.

[26326] GAM1697 precursor RNA, herein designated GAM PRECURSOR RNA, folds onto itself, forming GAM1697 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26327] An enzyme complex designated DICER COMPLEX, dices the GAM1697 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1697 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex

comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1697 RNA is designated SEQ ID:3346, and is provided hereinbelow with reference to the sequence listing part.

- [26328] GAM1697 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1697 target RNA, herein designated GAM TARGET RNA. GAM1697 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [26329] GAM1697 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1697 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1697 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III re-

spectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1697 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1697 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26330] The complementary binding of GAM1697 RNA, herein designated GAM RNA, to target binding sites on GAM1697 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1697 target RNA, herein designated GAM TARGET RNA, into GAM1697 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26331] It is appreciated that GAM1697 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1697 target genes. The mRNA of each one of this plurality of GAM1697 target genes comprises one or more

target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1697 RNA, herein designated GAM RNA, and which when bound by GAM1697 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1697 target proteins.

[26332] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1697 gene, herein designated GAM GENE, on one or more GAM1697 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26333] It is yet further appreciated that specific functions, and

accordingly utilities, of GAM1697 correlate with, and may be deduced from, the identity of the target genes which GAM1697 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26334] Nucleotide sequences of the GAM1697 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1697 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1697 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1697 are further described hereinbelow with reference to Table Table1.

[26335] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1697 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26336] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1698 (GAM1698) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26337] GAM1698 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1698 was detected is described hereinabove with reference to Figs. 2–8.

[26338] GAM1698 gene, herein designated GAM GENE, and GAM1698 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26339] GAM1698 gene, herein designated GAM GENE, encodes a GAM1698 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1698 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1698 precursor RNA is designated SEQ ID:1671, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1671 is located at position 1235063 relative to contig NT_009526.11, on chromosome 12.

[26340] GAM1698 has 3 highly conserved, expressed mouse homologs (>90% sequence similarity), found in mouse EST's. Nucleotide sequences similar to the nucleotide sequences of which mouse homologs, and the respective mouse EST's in which these homologs were detected are:

[26341] SEQ ID:100017 detected in mouse EST Accession
BI965972.1.

[26342] SEQ ID:100018 detected in mouse EST Accession
AI326210.1.

[26343] SEQ ID:100019 detected in mouse EST Accession
BI965652.1.

[26344] GAM1698 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1698 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26345] An enzyme complex designated DICER COMPLEX, dices the GAM1698 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1698 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short ~22nt RNA segment is catalyzed by an enzyme complex

comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1698 RNA is designated SEQ ID:3347, and is provided hereinbelow with reference to the sequence listing part.

- [26346] GAM1698 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1698 target RNA, herein designated GAM TARGET RNA. GAM1698 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [26347] GAM1698 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1698 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1698 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated BINDING SITE I, BINDING SITE II and BINDING SITE III re-

spectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1698 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1698 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26348] The complementary binding of GAM1698 RNA, herein designated GAM RNA, to target binding sites on GAM1698 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1698 target RNA, herein designated GAM TARGET RNA, into GAM1698 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26349] It is appreciated that GAM1698 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1698 target genes. The mRNA of each one of this plurality of GAM1698 target genes comprises one or more

target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1698 RNA, herein designated GAM RNA, and which when bound by GAM1698 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1698 target proteins.

[26350] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1698 gene, herein designated GAM GENE, on one or more GAM1698 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26351] It is yet further appreciated that specific functions, and

accordingly utilities, of GAM1698 correlate with, and may be deduced from, the identity of the target genes which GAM1698 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26352] Nucleotide sequences of the GAM1698 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1698 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1698 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1698 are further described hereinbelow with reference to Table Table1.

[26353] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1698 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26354] Fig. 8 further provides a conceptual description of another novel bioinformatically detected gene of the present invention, referred to here as Genomic Address Messenger 1699 (GAM1699) gene, which modulates expression of respective target genes thereof, the function and utility of which target genes is known in the art.

[26355] GAM1699 is a novel bioinformatically detected regulatory, non protein coding, micro RNA (miRNA) gene. The method by which GAM1699 was detected is described hereinabove with reference to Figs. 2–8.

[26356] GAM1699 gene, herein designated GAM GENE, and GAM1699 target gene, herein designated GAM TARGET GENE, are human genes contained in the human genome.

[26357] GAM1699 gene, herein designated GAM GENE, encodes a GAM1699 precursor RNA, herein designated GAM PRE-CURSOR RNA. Similar to other miRNA genes, and unlike most ordinary genes, GAM1699 precursor RNA, herein designated GAM PRECURSOR RNA, does not encode a protein. A nucleotide sequence identical or highly similar to the nucleotide sequence of GAM1699 precursor RNA is designated SEQ ID:1672, and is provided hereinbelow with reference to the sequence listing part. Nucleotide sequence SEQ ID:1672 is located at position 365814 relative to contig NT_010799.11, on chromosome 17.

[26358] GAM1699 has 15 highly conserved, expressed mouse homologs (>90% sequence similarity), found in mouse EST's. Nucleotide sequences similar to the nucleotide sequences of which mouse homologs, and the respective mouse EST's in which these homologs were detected are:

[26359] SEQ ID:97958 detected in mouse EST Accession
BE953126.1.

[26360] SEQ ID:97959 detected in mouse EST Accession
AA144533.1.

[26361] SEQ ID:97960 detected in mouse EST Accession
AA671455.1.

[26362] SEQ ID:97961 detected in mouse EST Accession
AW824753.1.

[26363] SEQ ID:97962 detected in mouse EST Accession
BE943987.1.

[26364] SEQ ID:97963 detected in mouse EST Accession
BE691447.1.

[26365] SEQ ID:97964 detected in mouse EST Accession
BG808025.1.

[26366] SEQ ID:97965 detected in mouse EST Accession
BG808010.1.

[26367] SEQ ID:97966 detected in mouse EST Accession
BG807850.1.

[26368] SEQ ID:97967 detected in mouse EST Accession
BG805970.1.

[26369] SEQ ID:97968 detected in mouse EST Accession
BG145596.1.

[26370] SEQ ID:97969 detected in mouse EST Accession

AW208601.1.

[26371] SEQ ID:97970 detected in mouse EST Accession
BB609859.1.

[26372] SEQ ID:97971 detected in mouse EST Accession
BB610842.1.

[26373] SEQ ID:97972 detected in mouse EST Accession
BE691413.1.

[26374] GAM1699 precursor RNA, herein designated GAM PRE-CURSOR RNA, folds onto itself, forming GAM1699 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, which has a two-dimensional hairpin structure. As is well known in the art, this hairpin structure, is typical of RNA encoded by miRNA genes, and is due to the fact that the nucleotide sequence of the first half of the RNA encoded by a miRNA gene is an accurate or partial inversed-reversed sequence of the nucleotide sequence of the second half thereof.

[26375] An enzyme complex designated DICER COMPLEX, dices the GAM1699 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, into GAM1699 RNA, herein designated GAM RNA, a single stranded ~22 nt long RNA segment. As is known in the art, dicing of a hairpin structured RNA precursor product into a short

~22nt RNA segment is catalyzed by an enzyme complex comprising an enzyme called Dicer together with other necessary proteins. A probable (over 89%) nucleotide sequence of GAM1699 RNA is designated SEQ ID:3348, and is provided hereinbelow with reference to the sequence listing part.

- [26376] GAM1699 target gene, herein designated GAM TARGET GENE, encodes a corresponding messenger RNA, GAM1699 target RNA, herein designated GAM TARGET RNA. GAM1699 target RNA, herein designated GAM TARGET RNA, comprises three regions, as is typical of mRNA of a protein coding gene: a 5 untranslated region, a protein coding region and a 3 untranslated region, designated 5UTR, PROTEIN CODING and 3UTR respectively.
- [26377] GAM1699 RNA, herein designated GAM RNA, binds complementarily to one or more target binding sites located in untranslated regions of GAM1699 target RNA, herein designated GAM TARGET RNA. This complementary binding is due to the fact that the nucleotide sequence of GAM1699 RNA, herein designated GAM RNA, is an accurate or a partial inversed-reversed sequence of the nucleotide sequence of each of the target binding sites. As an illustration, Fig. 8 shows 3 such target binding sites, designated

BINDING SITE I, BINDING SITE II and BINDING SITE III respectively. It is appreciated that the number of target binding sites shown in Fig. 8 is meant as an illustration only, and is not meant to be limiting GAM1699 RNA, herein designated GAM RNA, may have a different number of target binding sites in untranslated regions of a GAM1699 target RNA, herein designated GAM TARGET RNA. It is further appreciated that while Fig. 8 depicts target binding sites in the 3UTR region, this is meant as an example only these target binding sites may be located in the 3UTR region, the 5UTR region, or in both 3UTR and 5UTR regions.

[26378] The complementary binding of GAM1699 RNA, herein designated GAM RNA, to target binding sites on GAM1699 target RNA, herein designated GAM TARGET RNA, such as BINDING SITE I, BINDING SITE II and BINDING SITE III, inhibits translation of GAM1699 target RNA, herein designated GAM TARGET RNA, into GAM1699 target protein, herein designated GAM TARGET PROTEIN. GAM target protein is therefore outlined by a broken line.

[26379] It is appreciated that GAM1699 target gene, herein designated GAM TARGET GENE, in fact represents a plurality of GAM1699 target genes. The mRNA of each one of this

plurality of GAM1699 target genes comprises one or more target binding sites, each having a nucleotide sequence which is at least partly complementary to GAM1699 RNA, herein designated GAM RNA, and which when bound by GAM1699 RNA, herein designated GAM RNA, causes inhibition of translation of respective one or more GAM1699 target proteins.

[26380] It is further appreciated by one skilled in the art that the mode of translational inhibition illustrated by Fig. 8 with specific reference to translational inhibition exerted by GAM1699 gene, herein designated GAM GENE, on one or more GAM1699 target gene, herein designated GAM TARGET GENE, is in fact common to other known miRNA genes. As mentioned hereinabove with reference to the background section, although a specific complementary binding site has been demonstrated only for some of the known miRNA genes (primarily Lin-4 and Let-7), all other recently discovered miRNA genes are also believed by those skilled in the art to modulate expression of other genes by complementary binding, although specific complementary binding sites of these other miRNA genes have not yet been found (Ruvkun G., Perspective: Glimpses of a tiny RNA world, Science 294,779 (2001)).

[26381] It is yet further appreciated that specific functions, and accordingly utilities, of GAM1699 correlate with, and may be deduced from, the identity of the target genes which GAM1699 binds and inhibits, and the function of these target genes, as elaborated hereinbelow.

[26382] Nucleotide sequences of the GAM1699 precursor RNA, herein designated GAM PRECURSOR RNA, and of the diced GAM1699 RNA, herein designated GAM RNA, and a schematic representation of the secondary folding of GAM1699 folded precursor RNA, herein designated GAM FOLDED PRECURSOR RNA, of GAM1699 are further described hereinbelow with reference to Table Table1.

[26383] Nucleotide sequences of target binding sites, such as BINDING SITE-I, BINDING SITE-II and BINDING SITE-III of Fig. 8, found on, and schematic representation of the complementarity of each of these target binding sites to GAM1699 RNA, herein designated GAM RNA, are described hereinbelow with reference to Table Table2.

[26384] Fig. 16 further provides a conceptual description of novel bioinformatically detected regulatory gene, referred to here as Genomic Record 1700(GR1700) gene, which encodes an operon-like cluster of novel micro RNA-like genes, each of which in turn modulates expression of at

least one target gene, the function and utility of which at least one target gene is known in the art.

[26385] GR1700 gene, herein designated GR GENE, is a novel bioinformatically detected regulatory, non protein coding, RNA gene. The method by which GR1700 gene was detected is described hereinabove with reference to Figs. 6–15.

[26386] GR1700 gene encodes GR1700 precursor RNA, herein designated GR PRECURSOR RNA, an RNA molecule, typically several hundred nucleotides long.

[26387] GR1700 precursor RNA folds spatially, forming GR1700 folded precursor RNA, herein designated GR FOLDED PRECURSOR RNA. It is appreciated that GR1700 folded precursor RNA, herein designated GR FOLDED PRECURSOR RNA, comprises a plurality of what is known in the art as hairpin structures. These hairpin structures are due to the fact that the nucleotide sequence of GR1700 precursor RNA comprises a plurality of segments, the first half of each such segment having a nucleotide sequence which is at least a partial inversed–reversed sequence of the second half thereof, as is well known in the art.

[26388] GR1700 folded precursor RNA, herein designated GR FOLDED PRECURSOR RNA, is naturally processed by cellu–

lar enzymatic activity into at least 2 separate GAM precursor RNAs, GAM135 precursor RNA and GAM136 precursor RNA, herein schematically represented by GAM1 PRECURSOR and GAM2 PRECURSOR respectively, each of which GAM precursor RNAs being a hairpin shaped RNA segment, corresponding to GAM PRECURSOR RNA of Fig. 8.

[26389] The above mentioned GAM precursor RNAs are diced by DICER COMPLEX of Fig. 8, yielding respective short RNA segments of about 22 nucleotides in length, GAM135 RNA and GAM136 RNA respectively, herein schematically represented by GAM1 RNA and GAM2 RNA respectively, each of which GAM RNAs corresponding to GAM RNA of Fig. 8.

[26390] GAM135 RNA, herein schematically represented by GAM1 binds complementarily to a target binding site located in an untranslated region of GAM135 target RNA, herein schematically represented by GAM1 TARGET RNA, which target binding site corresponds to a target binding site such as BINDING SITE I, BINDING SITE II or BINDING SITE III of Fig. 8, thereby inhibiting translation of GAM135 target RNA, herein schematically represented by GAM1 TARGET RNA into GAM135 target protein, herein schematically represented by GAM1 TARGET PROTEIN, both of Fig. 8.

[26391] GAM136 RNA, herein schematically represented by GAM2

binds complementarily to a target binding site located in an untranslated region of GAM136 target RNA, herein schematically represented by GAM2 TARGET RNA, which target binding site corresponds to a target binding site such as BINDING SITE I, BINDING SITE II or BINDING SITE III of Fig. 8, thereby inhibiting translation of GAM136 target RNA, herein schematically represented by GAM2 TARGET RNA into GAM136 target protein, herein schematically represented by GAM2 TARGET PROTEIN, both of Fig. 8.

[26392] It is appreciated that specific functions, and accordingly utilities, of GR1700 gene, herein designated GR GENE, correlate with, and may be deduced from, the identity of the target genes, which are inhibited by GAM RNAs comprised in the operon-like cluster of GR1700 gene:

GAM135 target protein and GAM136 target protein, herein schematically represented by GAM1 TARGET PROTEIN and GAM TARGET PROTEIN respectively. The function of these target genes is elaborated hereinabove with reference to GAM135 and GAM136

[26393] Fig. 16 further provides a conceptual description of novel bioinformatically detected regulatory gene, referred to here as Genomic Record 1701 (GR1701) gene, which encodes an operon-like cluster of novel micro RNA-like

genes, each of which in turn modulates expression of at least one target gene, the function and utility of which at least one target gene is known in the art.

[26394] GR1701 gene, herein designated GR GENE, is a novel bioinformatically detected regulatory, non protein coding, RNA gene. The method by which GR1701 gene was detected is described hereinabove with reference to Figs. 6–15.

[26395] GR1701 gene encodes GR1701 precursor RNA, herein designated GR PRECURSOR RNA, an RNA molecule, typically several hundred nucleotides long.

[26396] GR1701 precursor RNA folds spatially, forming GR1701 folded precursor RNA, herein designated GR FOLDED PRECURSOR RNA. It is appreciated that GR1701 folded precursor RNA, herein designated GR FOLDED PRECURSOR RNA, comprises a plurality of what is known in the art as hairpin structures. These hairpin structures are due to the fact that the nucleotide sequence of GR1701 precursor RNA comprises a plurality of segments, the first half of each such segment having a nucleotide sequence which is at least a partial inversed–reversed sequence of the second half thereof, as is well known in the art.

[26397] GR1701 folded precursor RNA, herein designated GR

FOLDED PRECURSOR RNA, is naturally processed by cellular enzymatic activity into at least 2 separate GAM precursor RNAs, GAM250 precursor RNA and GAM251 precursor RNA, herein schematically represented by GAM1 PRECURSOR and GAM2 PRECURSOR respectively, each of which GAM precursor RNAs being a hairpin shaped RNA segment, corresponding to GAM PRECURSOR RNA of Fig. 8.

[26398] The above mentioned GAM precursor RNAs are diced by DICER COMPLEX of Fig. 8, yielding respective short RNA segments of about 22 nucleotides in length, GAM250 RNA and GAM251 RNA respectively, herein schematically represented by GAM1 RNA and GAM2 RNA respectively, each of which GAM RNAs corresponding to GAM RNA of Fig. 8.

[26399] GAM250 RNA, herein schematically represented by GAM1 binds complementarily to a target binding site located in an untranslated region of GAM250 target RNA, herein schematically represented by GAM1 TARGET RNA, which target binding site corresponds to a target binding site such as BINDING SITE I, BINDING SITE II or BINDING SITE III of Fig. 8, thereby inhibiting translation of GAM250 target RNA, herein schematically represented by GAM1 TARGET RNA into GAM250 target protein, herein schematically represented by GAM1 TARGET PROTEIN, both of Fig. 8.

[26400] GAM251 RNA, herein schematically represented by GAM2 binds complementarily to a target binding site located in an untranslated region of GAM251 target RNA, herein schematically represented by GAM2 TARGET RNA, which target binding site corresponds to a target binding site such as BINDING SITE I, BINDING SITE II or BINDING SITE III of Fig. 8, thereby inhibiting translation of GAM251 target RNA, herein schematically represented by GAM2 TARGET RNA into GAM251 target protein, herein schematically represented by GAM2 TARGET PROTEIN, both of Fig. 8.

[26401] It is appreciated that specific functions, and accordingly utilities, of GR1701 gene, herein designated GR GENE, correlate with, and may be deduced from, the identity of the target genes, which are inhibited by GAM RNAs comprised in the operon-like cluster of GR1701 gene: GAM250 target protein and GAM251 target protein, herein schematically represented by GAM1 TARGET PROTEIN and GAM TARGET PROTEIN respectively. The function of these target genes is elaborated hereinabove with reference to GAM250 and GAM251

[26402] Fig. 16 further provides a conceptual description of novel bioinformatically detected regulatory gene, referred to here as Genomic Record 1702(GR1702) gene, which en-

codes an operon-like cluster of novel micro RNA-like genes, each of which in turn modulates expression of at least one target gene, the function and utility of which at least one target gene is known in the art.

[26403] GR1702 gene, herein designated GR GENE, is a novel bioinformatically detected regulatory, non protein coding, RNA gene. The method by which GR1702 gene was detected is described hereinabove with reference to Figs. 6-15.

[26404] GR1702 gene encodes GR1702 precursor RNA, herein designated GR PRECURSOR RNA, an RNA molecule, typically several hundred nucleotides long.

[26405] GR1702 precursor RNA folds spatially, forming GR1702 folded precursor RNA, herein designated GR FOLDED PRECURSOR RNA. It is appreciated that GR1702 folded precursor RNA, herein designated GR FOLDED PRECURSOR RNA, comprises a plurality of what is known in the art as hairpin structures. These hairpin structures are due to the fact that the nucleotide sequence of GR1702 precursor RNA comprises a plurality of segments, the first half of each such segment having a nucleotide sequence which is at least a partial inversed-reversed sequence of the second half thereof, as is well known in the art.

[26406] GR1702 folded precursor RNA, herein designated GR FOLDED PRECURSOR RNA, is naturally processed by cellular enzymatic activity into at least 2 separate GAM precursor RNAs, GAM749 precursor RNA and GAM750 precursor RNA, herein schematically represented by GAM1 PRECURSOR and GAM2 PRECURSOR respectively, each of which GAM precursor RNAs being a hairpin shaped RNA segment, corresponding to GAM PRECURSOR RNA of Fig. 8.

[26407] The above mentioned GAM precursor RNAs are diced by DICER COMPLEX of Fig. 8, yielding respective short RNA segments of about 22 nucleotides in length, GAM749 RNA and GAM750 RNA respectively, herein schematically represented by GAM1 RNA and GAM2 RNA respectively, each of which GAM RNAs corresponding to GAM RNA of Fig. 8.

[26408] GAM749 RNA, herein schematically represented by GAM1 binds complementarily to a target binding site located in an untranslated region of GAM749 target RNA, herein schematically represented by GAM1 TARGET RNA, which target binding site corresponds to a target binding site such as BINDING SITE I, BINDING SITE II or BINDING SITE III of Fig. 8, thereby inhibiting translation of GAM749 target RNA, herein schematically represented by GAM1 TARGET RNA into GAM749 target protein, herein schematically

represented by GAM1 TARGET PROTEIN, both of Fig. 8.

[26409] GAM750 RNA, herein schematically represented by GAM2 binds complementarily to a target binding site located in an untranslated region of GAM750 target RNA, herein schematically represented by GAM2 TARGET RNA, which target binding site corresponds to a target binding site such as BINDING SITE I, BINDING SITE II or BINDING SITE III of Fig. 8, thereby inhibiting translation of GAM750 target RNA into GAM750 target protein, herein schematically represented by GAM2 TARGET PROTEIN, both of Fig. 8.

[26410] It is appreciated that specific functions, and accordingly utilities, of GR1702 gene, herein designated GR GENE, correlate with, and may be deduced from, the identity of the target genes, which are inhibited by GAM RNAs comprised in the operon-like cluster of GR1702 gene:

GAM749 target protein and GAM750 target protein, herein schematically represented by GAM1 TARGET PROTEIN and GAM TARGET PROTEIN respectively. The function of these target genes is elaborated hereinabove with reference to GAM749 and GAM750

[26411] Fig. 16 further provides a conceptual description of novel bioinformatically detected regulatory gene, referred to

here as Genomic Record 1703(GR1703) gene, which encodes an operon-like cluster of novel micro RNA-like genes, each of which in turn modulates expression of at least one target gene, the function and utility of which at least one target gene is known in the art.

[26412] GR1703 gene, herein designated GR GENE, is a novel bioinformatically detected regulatory, non protein coding, RNA gene. The method by which GR1703 gene was detected is described hereinabove with reference to Figs. 6–15.

[26413] GR1703 gene encodes GR1703 precursor RNA, herein designated GR PRECURSOR RNA, an RNA molecule, typically several hundred nucleotides long.

[26414] GR1703 precursor RNA folds spatially, forming GR1703 folded precursor RNA, herein designated GR FOLDED PRECURSOR RNA. It is appreciated that GR1703 folded precursor RNA, herein designated GR FOLDED PRECURSOR RNA, comprises a plurality of what is known in the art as hairpin structures. These hairpin structures are due to the fact that the nucleotide sequence of GR1703 precursor RNA comprises a plurality of segments, the first half of each such segment having a nucleotide sequence which is at least a partial inversed-reversed sequence of the sec-

ond half thereof, as is well known in the art.

[26415] GR1703 folded precursor RNA, herein designated GR FOLDED PRECURSOR RNA, is naturally processed by cellular enzymatic activity into at least 2 separate GAM precursor RNAs, GAM854 precursor RNA and GAM855 precursor RNA, herein schematically represented by GAM1 PRECURSOR and GAM2 PRECURSOR respectively, each of which GAM precursor RNAs being a hairpin shaped RNA segment, corresponding to GAM PRECURSOR RNA of Fig. 8.

[26416] The above mentioned GAM precursor RNAs are diced by DICER COMPLEX of Fig. 8, yielding respective short RNA segments of about 22 nucleotides in length, GAM854 RNA and GAM855 RNA respectively, herein schematically represented by GAM1 RNA and GAM2 RNA respectively, each of which GAM RNAs corresponding to GAM RNA of Fig. 8.

[26417] GAM854 RNA, herein schematically represented by GAM1 binds complementarily to a target binding site located in an untranslated region of GAM854 target RNA, herein schematically represented by GAM1 TARGET RNA, which target binding site corresponds to a target binding site such as BINDING SITE I, BINDING SITE II or BINDING SITE III of Fig. 8, thereby inhibiting translation of GAM854 target RNA, herein schematically represented by GAM1 TARGET

RNA into GAM854 target protein, herein schematically represented by GAM1 TARGET PROTEIN, both of Fig. 8.

[26418] GAM855 RNA, herein schematically represented by GAM2 binds complementarily to a target binding site located in an untranslated region of GAM855 target RNA, herein schematically represented by GAM2 TARGET RNA, which target binding site corresponds to a target binding site such as BINDING SITE I, BINDING SITE II or BINDING SITE III of Fig. 8, thereby inhibiting translation of GAM855 target RNA into GAM855 target protein, herein schematically represented by GAM2 TARGET PROTEIN, both of Fig. 8.

[26419] It is appreciated that specific functions, and accordingly utilities, of GR1703 gene, herein designated GR GENE, correlate with, and may be deduced from, the identity of the target genes, which are inhibited by GAM RNAs comprised in the operon-like cluster of GR1703 gene: GAM854 target protein and GAM855 target protein, herein schematically represented by GAM1 TARGET PROTEIN and GAM TARGET PROTEIN respectively. The function of these target genes is elaborated hereinabove with reference to GAM854 and GAM855

[26420] Fig. 16 further provides a conceptual description of novel

bioinformatically detected regulatory gene, referred to here as Genomic Record 1704(GR1704) gene, which encodes an operon-like cluster of novel micro RNA-like genes, each of which in turn modulates expression of at least one target gene, the function and utility of which at least one target gene is known in the art.

[26421] GR1704 gene, herein designated GR GENE, is a novel bioinformatically detected regulatory, non protein coding, RNA gene. The method by which GR1704 gene was detected is described hereinabove with reference to Figs. 6-15.

[26422] GR1704 gene encodes GR1704 precursor RNA, herein designated GR PRECURSOR RNA, an RNA molecule, typically several hundred nucleotides long.

[26423] GR1704 precursor RNA folds spatially, forming GR1704 folded precursor RNA, herein designated GR FOLDED PRECURSOR RNA. It is appreciated that GR1704 folded precursor RNA, herein designated GR FOLDED PRECURSOR RNA, comprises a plurality of what is known in the art as hairpin structures. These hairpin structures are due to the fact that the nucleotide sequence of GR1704 precursor RNA comprises a plurality of segments, the first half of each such segment having a nucleotide sequence which is

at least a partial inversed-reversed sequence of the second half thereof, as is well known in the art.

[26424] GR1704 folded precursor RNA, herein designated GR FOLDED PRECURSOR RNA, is naturally processed by cellular enzymatic activity into at least 2 separate GAM precursor RNAs, GAM1442 precursor RNA and GAM1443 precursor RNA, herein schematically represented by GAM1 PRECURSOR and GAM2 PRECURSOR respectively, each of which GAM precursor RNAs being a hairpin shaped RNA segment, corresponding to GAM PRECURSOR RNA of Fig. 8.

[26425] The above mentioned GAM precursor RNAs are diced by DICER COMPLEX of Fig. 8, yielding respective short RNA segments of about 22 nucleotides in length, GAM1442 RNA and GAM1443 RNA respectively, herein schematically represented by GAM1 RNA and GAM2 RNA respectively, each of which GAM RNAs corresponding to GAM RNA of Fig. 8.

[26426] GAM1442 RNA, herein schematically represented by GAM1 binds complementarily to a target binding site located in an untranslated region of GAM1442 target RNA, herein schematically represented by GAM1 TARGET RNA, which target binding site corresponds to a target binding site

such as BINDING SITE I, BINDING SITE II or BINDING SITE III of Fig. 8, thereby inhibiting translation of GAM1442 target RNA, herein schematically represented by GAM1 TARGET RNA into GAM1442 target protein, herein schematically represented by GAM1 TARGET PROTEIN, both of Fig. 8.

[26427] GAM1443 RNA, herein schematically represented by GAM2 binds complementarily to a target binding site located in an untranslated region of GAM1443 target RNA, herein schematically represented by GAM2 TARGET RNA, which target binding site corresponds to a target binding site such as BINDING SITE I, BINDING SITE II or BINDING SITE III of Fig. 8, thereby inhibiting translation of GAM1443 target RNA, herein schematically represented by GAM2 TARGET RNA into GAM1443 target protein, herein schematically represented by GAM2 TARGET PROTEIN, both of Fig. 8.

[26428] It is appreciated that specific functions, and accordingly utilities, of GR1704 gene, herein designated GR GENE, correlate with, and may be deduced from, the identity of the target genes, which are inhibited by GAM RNAs comprised in the operon-like cluster of GR1704 gene: GAM1442 target protein and GAM1443 target protein, herein schematically represented by GAM1 TARGET PROTEIN and GAM TARGET PROTEIN respectively. The function

of these target genes is elaborated hereinabove with reference to GAM1442 and GAM1443

[26429] Fig. 16 further provides a conceptual description of novel bioinformatically detected regulatory gene, referred to here as Genomic Record 1705 (GR1705) gene, which encodes an operon-like cluster of novel micro RNA-like genes, each of which in turn modulates expression of at least one target gene, the function and utility of which at least one target gene is known in the art.

[26430] GR1705 gene, herein designated GR GENE, is a novel bioinformatically detected regulatory, non protein coding, RNA gene. The method by which GR1705 gene was detected is described hereinabove with reference to Figs. 6–15.

[26431] GR1705 gene encodes GR1705 precursor RNA, herein designated GR PRECURSOR RNA, an RNA molecule, typically several hundred nucleotides long.

[26432] GR1705 precursor RNA folds spatially, forming GR1705 folded precursor RNA, herein designated GR FOLDED PRECURSOR RNA. It is appreciated that GR1705 folded precursor RNA, herein designated GR FOLDED PRECURSOR RNA, comprises a plurality of what is known in the art as hairpin structures. These hairpin structures are due to the

fact that the nucleotide sequence of GR1705 precursor RNA comprises a plurality of segments, the first half of each such segment having a nucleotide sequence which is at least a partial inversed-reversed sequence of the second half thereof, as is well known in the art.

[26433] GR1705 folded precursor RNA, herein designated GR FOLDED PRECURSOR RNA, is naturally processed by cellular enzymatic activity into at least 2 separate GAM precursor RNAs, GAM1566 precursor RNA and GAM1567 precursor RNA, herein schematically represented by GAM1 PRECURSOR and GAM2 PRECURSOR respectively, each of which GAM precursor RNAs being a hairpin shaped RNA segment, corresponding to GAM PRECURSOR RNA of Fig. 8.

[26434] The above mentioned GAM precursor RNAs are diced by DICER COMPLEX of Fig. 8, yielding respective short RNA segments of about 22 nucleotides in length, GAM1566 RNA and GAM1567 RNA respectively, herein schematically represented by GAM1 RNA and GAM2 RNA respectively, each of which GAM RNAs corresponding to GAM RNA of Fig. 8.

[26435] GAM1566 RNA, herein schematically represented by GAM1 binds complementarily to a target binding site located in

an untranslated region of GAM1566 target RNA, herein schematically represented by GAM1 TARGET RNA, which target binding site corresponds to a target binding site such as BINDING SITE I, BINDING SITE II or BINDING SITE III of Fig. 8, thereby inhibiting translation of GAM1566 target RNA, herein schematically represented by GAM1 TARGET RNA into GAM1566 target protein, herein schematically represented by GAM1 TARGET PROTEIN, both of Fig. 8.

[26436] GAM1567 RNA, herein schematically represented by GAM2 binds complementarily to a target binding site located in an untranslated region of GAM1567 target RNA, herein schematically represented by GAM2 TARGET RNA, which target binding site corresponds to a target binding site such as BINDING SITE I, BINDING SITE II or BINDING SITE III of Fig. 8, thereby inhibiting translation of GAM1567 target RNA, herein schematically represented by GAM2 TARGET RNA into GAM1567 target protein, herein schematically represented by GAM2 TARGET PROTEIN, both of Fig. 8.

[26437] It is appreciated that specific functions, and accordingly utilities, of GR1705 gene, herein designated GR GENE, correlate with, and may be deduced from, the identity of the target genes, which are inhibited by GAM RNAs comprised in the operon-like cluster of GR1705 gene:

GAM1566 target protein and GAM1567 target protein, herein schematically represented by GAM1 TARGET PROTEIN and GAM TARGET PROTEIN respectively. The function of these target genes is elaborated hereinabove with reference to GAM1566 and GAM1567

[26438] Fig. 16 further provides a conceptual description of novel bioinformatically detected regulatory gene, referred to here as Genomic Record 1706 (GR1706) gene, which encodes an operon-like cluster of novel micro RNA-like genes, each of which in turn modulates expression of at least one target gene, the function and utility of which at least one target gene is known in the art.

[26439] GR1706 gene, herein designated GR GENE, is a novel bioinformatically detected regulatory, non protein coding, RNA gene. The method by which GR1706 gene was detected is described hereinabove with reference to Figs. 6–15.

[26440] GR1706 gene encodes GR1706 precursor RNA, herein designated GR PRECURSOR RNA, an RNA molecule, typically several hundred nucleotides long.

[26441] GR1706 precursor RNA folds spatially, forming GR1706 folded precursor RNA, herein designated GR FOLDED PRECURSOR RNA. It is appreciated that GR1706 folded pre-

cursor RNA, herein designated GR FOLDED PRECURSOR RNA, comprises a plurality of what is known in the art as hairpin structures. These hairpin structures are due to the fact that the nucleotide sequence of GR1706 precursor RNA comprises a plurality of segments, the first half of each such segment having a nucleotide sequence which is at least a partial inversed-reversed sequence of the second half thereof, as is well known in the art.

[26442] GR1706 folded precursor RNA, herein designated GR FOLDED PRECURSOR RNA, is naturally processed by cellular enzymatic activity into at least 2 separate GAM precursor RNAs, GAM1673 precursor RNA and GAM1674 precursor RNA, herein schematically represented by GAM1 PRECURSOR and GAM2 PRECURSOR respectively, each of which GAM precursor RNAs being a hairpin shaped RNA segment, corresponding to GAM PRECURSOR RNA of Fig. 8.

[26443] The above mentioned GAM precursor RNAs are diced by DICER COMPLEX of Fig. 8, yielding respective short RNA segments of about 22 nucleotides in length, GAM1673 RNA and GAM1674 RNA respectively, herein schematically represented by GAM1 RNA and GAM2 RNA respectively, each of which GAM RNAs corresponding to GAM RNA of

Fig. 8.

[26444] GAM1673 RNA, herein schematically represented by GAM1 binds complementarily to a target binding site located in an untranslated region of GAM1673 target RNA, herein schematically represented by GAM1 TARGET RNA, which target binding site corresponds to a target binding site such as BINDING SITE I, BINDING SITE II or BINDING SITE III of Fig. 8, thereby inhibiting translation of GAM1673 target RNA into GAM1673 target protein, herein schematically represented by GAM1 TARGET PROTEIN, both of Fig. 8.

[26445] GAM1674 RNA, herein schematically represented by GAM2 binds complementarily to a target binding site located in an untranslated region of GAM1674 target RNA, herein schematically represented by GAM2 TARGET RNA, which target binding site corresponds to a target binding site such as BINDING SITE I, BINDING SITE II or BINDING SITE III of Fig. 8, thereby inhibiting translation of GAM1674 target RNA into GAM1674 target protein, herein schematically represented by GAM2 TARGET PROTEIN, both of Fig. 8.

[26446] It is appreciated that specific functions, and accordingly utilities, of GR1706 gene, herein designated GR GENE,

correlate with, and may be deduced from, the identity of the target genes, which are inhibited by GAM RNAs comprised in the operon-like cluster of GR1706 gene:

GAM1673 target protein and GAM1674 target protein, herein schematically represented by GAM1 TARGET PROTEIN and GAM TARGET PROTEIN respectively. The function of these target genes is elaborated hereinabove with reference to GAM1673 and GAM1674

[26447] BIBLIOGRAPHY

[26448] It is appreciated by persons skilled in the art that the present invention is not limited by what has been particularly shown and described hereinabove. Rather the scope of the present invention includes both combinations and subcombinations of the various features described hereinabove as well as variations and modifications which would occur to persons skilled in the art upon reading the specifications and which are not in the prior art.

[26449] 1.Gorin, M. B.; Tilghman, S. M.: Structure of the alpha-fetoprotein gene in the mouse. Proc. Nat. Acad. Sci. 77: 1351-1355, 1980.

[26450] 2.Zuker, C. S.: On the evolution of eyes: would you like it simple or compound? Science 265: 742-743, 1994.

[26451] 3.Davies, A. F.; Mirza, G.; Flinter, F.; Ragoussis, J.: An in-

terstitial deletion of 6p24–p25 proximal to the FKHL7 locus and including AP-2- α that affects anterior eye chamber development. *J. Med. Genet.* 36:708–710, 1999.

- [26452] 4. Sen Gupta, B.; Friedberg, F.; Detera-Wadleigh, S. D.: Molecular analysis of human and rat calmodulin complementary DNA clones: evidence for additional active genes in these species. *J. Biol. Chem.* 262:16663–16670, 1987.
- [26453] 5. Jeffrey, P. D.; Russo, A. A.; Polyak, K.; Gibbs, E.; Hurwitz, J.; Massague, J.; Pavletich, N. P.: Mechanism of CDK activation revealed by the structure of a cyclinA–CDK2 complex. *Nature* 376: 313–320, 1995.
- [26454] 6. Fishman, G. I.; Moreno, A. P.; Spray, D. C.; Levin, L. A.: Functional analysis of human cardiac gap junction channel mutants. *Proc. Nat. Acad. Sci.* 88: 3525–3529, 1991.
- [26455] 7. Kida, S.; Josselyn, S. A.; Pena de Ortiz, S.; Kogan, J. H.; Chevere, I.; Masushige, S.; Silva, A. J.: CREB required for the stability of new and reactivated fear memories. *Nature Neurosci.* 5: 348–355, 2002.
- [26456] 8. Yamauchi, M.; Yamauchi, N.; Meuth, M.: Molecular cloning of the human CTP synthetase gene by functional complementation with purified human metaphase chromosomes. *EMBO J.* 9: 2095–2099, 1990.

- [26457] 9.Gray, P. W.; Glaister, D.; Seeburg, P. H.; Guidotti, A.; Costa,E.: Cloning and expression of cDNA for human diazepam binding inhibitor,a natural ligand of an allosteric regulatory site of the gamma-aminobutyricacid type A receptor. *Proc. Nat. Acad. Sci.* 83: 7547–7551, 1986.
- [26458] 10.Hassett, C.; Aicher, L.; Sidhu, J. S.; Omiecinski, C. J.: Humanmicrosomal epoxide hydrolase: genetic polymorphism and functionalexpression in vitro of amino acid variants. *Hum. Molec. Genet.* 3:421–428, 1994.
- [26459] 11.Vestergaard, P.; Hermann, A.P.; Orskov, H.; Mosekilde, L.; TheDanish Osteoporosis Prevention Study: Effect of sex hormone replacementon the insulin-like growth factor system and bone mineral: a cross-sectionaland longitudinal study in 595 perimenopausal women participating inthe Danish osteoporosis prevention study. *J. Clin. Endocr. Metab.* 84:2286–2290, 1999.
- [26460] 12.Sander, M.; Chavoshan, B.; Harris, S. A.; Iannaccone, S. T.; Stull,J. T.; Thomas, G. D.; Victor, R. G.: Functional muscle ischemia inneuronal nitric oxide synthase-deficient skeletal muscle of childrenwith Duchenne muscular dystrophy. *Proc. Nat. Acad. Sci.* 97: 13818–13823,2000.
- [26461] 13.Eiberg, H.; Mohr, J.; Schmiegelow, K.; Nielsen, L. S.; Williamson,R.: Linkage relationships of paraoxonase (PON)

with other markers: indication of PON–cystic fibrosis syndrome. Clin. Genet. 28: 265–271, 1985.

- [26462] 14. Berger, J.; Garattini, E.; Hua, J.–C.; Udenfriend, S.: Cloning and sequencing of human intestinal alkaline phosphatase cDNA. Proc. Nat. Acad. Sci. 84: 695–698, 1987.
- [26463] 15. Simard, J.; Berube, D.; Sandberg, M.; Grzeschik, K.–H.; Gagne, R.; Hansson, V.; Jahnsen, T.: Assignment of the gene encoding the catalytic subunit C–beta of cAMP–dependent protein kinase to the p36 band on chromosome 1. Hum. Genet. 88: 653–657, 1992.
- [26464] 16. Engstrom, Y.; Francke, U.: Assignment of the structural gene for subunit M1 of human ribonucleotide reductase to the short arm of chromosome 11. Exp. Cell Res. 158: 477–483, 1985.
- [26465] 17. Ramamoorthy, S.; Bauman, A. L.; Moore, K. R.; Han, H.; Yang–Feng, T.; Chang, A. S.; Ganapathy, V.; Blakely, R. D.: Antidepressant– and cocaine–sensitive human serotonin transporter: molecular cloning, expression, and chromosomal localization. Proc. Nat. Acad. Sci. 90: 2542–2546, 1993.
- [26466] 18. Grimmond, S.; Van Hateren, N.; Siggers, P.; Arkell, R.; Larder, R.; Soares, M. B.; de Fatima Bonaldo, M.; Smith, L.; Tymowska–Lalanne, Z.; Wells, C.; Greenfield, A.: Sexually

dimorphic expression of proteasenexin-1 and vanin-1 in the developing mouse gonad prior to overt differentiationsuggests a role in mammalian sexual development. Hum. Molec. Genet. 9:1553-1560, 2000.

[26467] 19.ADHR Consortium: Autosomal dominant hypophosphataemic rickets associated with mutations in FGF23. Nature Genet. 26: 345-348,2000.

[26468] 20.Conlon, M. G.; Tomasini, B. R.; Schultz, R. L.; Mosher, D. F.:Plasma vitronectin polymorphism in normal subjects and patients withdisseminated intravascular coagulation. Blood 72: 185-190, 1988.

[26469] 21.Fink, T. M.; Jenne, D. E.; Lichter, P.: The human vitronectin(complement S-protein) gene maps to the centromeric region of 17q.Hum. Genet. 88: 569-572, 1992.

[26470] 22.Jenne, D.; Stanley, K. K.: Molecular cloning of S-protein, a linkbetween complement, coagulation and cell-substrate adhesion. EMBOJ. 4: 3153-3157, 1985.

[26471] 23.Kubota, K.; Katayama, S.; Matsuda, M.; Hayashi, M.: Three typesof vitronectin in human blood. Cell Struct. Funct. 13: 123-128,1988.

[26472] 24.Preissner, K. T.; Heimburger, N.; Anders, E.; Muller-Berghaus,G.: Physicochemical, immunological and functional comparison of humanS-protein and vitronectin: evi-

dence for the identity of both plasmaproteins. *Biochem. Biophys. Res. Commun.* 134: 951–956, 1986.

[26473] 25.Sun, W. H.; Mosher, D. F.: Polymorphism of vitronectin. (Letter) *Blood* 73:353–354, 1989.

[26474] 26.Gao, Y.; Ferguson, D. O.; Xie, W.; Manis, J. P.; Sekiguchi, J.; Frank, K. M.; Chaudhuri, J.; Horner, J.; De-Pinho, R. A.; Alt, F. W.: Interplay of p53 and DNA-repair protein XRCC4 in tumorigenesis, genomic stability and development. *Nature* 404: 897–900, 2000.

[26475] 27.Hirao, A.; Kong, Y.-Y.; Matsuoka, S.; Wakeham, A.; Ruland, J.; Yoshida, H.; Liu, D.; Elledge, S. J.; Mak, T. W.: DNA damage-induced activation of p53 by the checkpoint kinase Chk2. *Science* 287: 1824–1827, 2000.

[26476] 28.Kaname, T.; Miyauchi, T.; Kuwano, A.; Matsuda, Y.; Muramatsu, T.; Kajii, T.: Mapping basigin (BSG), a member of the immunoglobulin superfamily, to 19p13.3. *Cytogenet. Cell Genet.* 64: 195–197, 1993.

[26477] 29.Kanekura, T.; Miyauchi, T.; Tashiro, M.; Muramatsu, T.: Basigin, a new member of the immunoglobulin superfamily: genes in different mammalian species, glycosylation changes in the molecule from adult organs and possible variation in the N-terminal sequences. *Cell Struct. Funct.* 16: 23–30, 1991.

- [26478] 30. Kuno, N.; Kadomatsu, K.; Fan, Q.-W.; Hagihara, M.; Senda, T.; Mizutani, S.; Muramatsu, T.: Female sterility in mice lacking the basigin gene, which encodes a trans-membrane glycoprotein belonging to the immunoglobulin superfamily. *FEBS Lett.* 425: 191–194, 1998.
- [26479] 31. Miyauchi, T.; Kanekura, T.; Yamaoka, A.; Ozawa, M.; Miyazawa, S.; Muramatsu, T.: Basigin, a new, broadly distributed member of the immunoglobulin superfamily, has strong homology with both the immunoglobulin V domain and the beta-chain of major histocompatibility complex class II antigen. *J. Biochem.* 107: 316–323, 1990.
- [26480] 32. Miyauchi, T.; Masuzawa, Y.; Muramatsu, T.: The basigin group of the immunoglobulin superfamily: complete conservation of a segment in and around transmembrane domains of human and mouse basigin and chicken HT7 antigen. *J. Biochem.* 110: 770–774, 1991.
- [26481] 33. Naruhashi, K.; Kadomatsu, K.; Igakura, T.; Fan, Q.-W.; Kuno, N.; Muramatsu, H.; Miyauchi, T.; Hasegawa, T.; Itoh, A.; Muramatsu, T.; Nabeshima, T.: Abnormalities of sensory and memory functions in mice lacking Bsg gene. *Biochem. Biophys. Res. Commun.* 236: 733–737, 1997.
- [26482] 34. Klobutcher, L. A.; Nichols, E. A.; Kucherlapati, R. S.; Ruddle, F. H.: Assignment of the gene for human adeno-

sine kinase to chromosome10 using a somatic cell hybrid clone panel. Cytogenet. Cell Genet. 16:171–174, 1976.

- [26483] 35.McNally, T.; Helfrich, R. J.; Cowart, M.; Dorwin, S. A.; Meuth,J. L.; Idler, K. B.; Klute, K. A.; Simmer, R. L.; Kowaluk, E. A.;Halbert, D. N.: Cloning and expression of the adenosine kinase gene from rat and human tissues. Biochem. Biophys. Res. Commun. 231:645–650, 1997.
- [26484] 36.Snyder, F. F.; Lin, C. C.; Rudd, N. L.; Shearer, J. E.; Heikkila,E. M.; Hoo, J. J.: A de novo case of trisomy 10p: gene dosage studies of hexokinase, inorganic pyrophosphatase and adenosine kinase. Hum.Genet. 67: 187–189, 1984.
- [26485] 37.Spychala, J.; Datta, N. S.; Takabayashi, K.; Datta, M.; Fox, I.;Gribbin, T.; Mitchell, B.: Cloning of human adenosine kinase cDNA:sequenced similarity to microbial ribokinases and fructokinases, Proc.Nat. Acad. Sci. 93: 1232–1237, 1996.
- [26486] 38.Patten, J. L.; Smallwood, P. M.; Eil, C.; Johns, D. R.; Valle,D.; Steel, G.; Levine, M. A.: An initiator codon mutation in the gene encoding the alpha subunit of Gs in pseudohypoparathyroidism type IA (PHP IA). (Abstract) Am. J. Hum. Genet. 45 (suppl.): A212 only, 1989.
- [26487] 39.Yu, S.; Yu, D.; Lee, E.; Eckhaus, M.; Lee, R.; Corria, Z.;

Accili,D.; Westphal, H.; Weinstein, L. S.: Variable and tissue-specific hormone resistance in heterotrimeric Gs protein alpha-subunit (Gs-alpha) knockout mice is due to tissue-specific imprinting of the Gs-alpha gene. Proc. Nat. Acad. Sci. 95: 8715–8720, 1998.

[26488] 40. Shozu, M.; Akasofu, K.; Harada, T.; Kubota, Y.: A new cause of female pseudohermaphroditism: placental aromatase deficiency. J. Clin. Endocr. Metab. 72: 560–566, 1991.

[26489] 41. Gorn, A. H.; Rudolph, S. M.; Flannery, M. R.; Morton, C. C.; Weremowicz, S.; Wang, J.-T.; Krane, S. M.; Goldring, S. R.: Expression of two human skeletal calcitonin receptor isoforms cloned from a giant cell tumor of bone. J. Clin. Invest. 95: 2680–2691, 1995.

[26490] 42. Dudhia, J.; Bayliss, M. T.; Hardingham, T. E.: Human link protein gene: structure and transcription pattern in chondrocytes. Biochem. J. 303: 329–333, 1994.

[26491] 43. Xie, Y.; Heng, H. H. Q.: FISH mapping of centromere protein C (CENPC) on human chromosome 4q13–q21. Cytogenet. Cell Genet. 74: 192–193, 1996.

[26492] 44. Dalla Venezia, N.; Gilsanz, F.; Alloisio, N.; Ducluzeau, M.-T.; Benz, E. J., Jr.; Delaunay, J.: Homozygous 4.1(–) hereditary elliptocytosis associated with a point mutation

in the downstream initiation codon of protein 4.1 gene. *J. Clin. Invest.* 90: 1713–1717, 1992.

- [26493] 45. Elliott, K. J.; Ellis, S. B.; Berckhan, K. J.; Urrutia, A.; Chavez–Noriega, L. E.; Johnson, E. C.; Velicelebi, G.; Harpold, M. M.: Comparative structure of human neuronal $\alpha(2)$ – $\alpha(7)$ and $\beta(2)$ – $\beta(4)$ nicotinic acetylcholine receptor subunits and functional expression of the $\alpha(2)$, $\alpha(3)$, $\alpha(4)$, $\alpha(7)$, $\beta(2)$, and $\beta(4)$ subunits. *J. Molec. Neurosci.* 7: 217–228, 1996.
- [26494] 46. Budarf, M. L.; Korenberg, J. R.; Simon, M.; Emanuel, B. S.: Regional assignment of the guanine nucleotide binding protein, GNAZ, to chromosome 22 (22q11.1–q11.2). (Abstract) *Cytogenet. Cell Genet.* 58: 2046–2047, 1991.
- [26495] 47. Jayawardena–Wolf, J.; Benlagha, K.; Chiu, Y.–H.; Mehr, R.; Bendelac, A.: CD1d endosomal trafficking is independently regulated by an intrinsic CD1d–encoded tyrosine motif and by the invariant chain. *Immunity* 15: 897–908, 2001.
- [26496] 48. Yang, W.–S.; Nevin, D. N.; Peng, R.; Brunzell, J. D.; Deeb, S. S.: A mutation in the promoter of the lipoprotein lipase (LPL) gene in a patient with familial combined hyperlipidemia and low LPL activity. *Proc. Nat. Acad. Sci.* 92: 4462–4466, 1995.

- [26497] 49.Lloyd, A.; Modi, W.; Sprenger, H.; Cevario, S.; Oppenheim, J.;Kelvin, D.: Assignment of genes for interleukin-8 receptors (IL8R)A and B to human chromosome band 2q35. *Cytogenet. Cell Genet.* 63:238–240, 1993.
- [26498] 50.Murgia, C.; Blaikie, P.; Kim, N.; Dans, M.; Petrie, H. T.; Giancotti,F. G.: Cell cycle and adhesion defects in mice carrying a targeteddeletion of the integrin beta-4 cytoplasmic domain. *EMBO J.* 17:3940–3951, 1998.
- [26499] 51.Roberts, A. N.; Leighton, B.; Todd, J. A.; Cockburn, D.; Schofield,P. N.; Sutton, R.; Holt, S.; Boyd, Y.; Day, A. J.; Foot, E. A.; Willis,A. C.; Reid, K. B. M.; Cooper, G. J. S.: Molecular and functionalcharacterization of amylin, a peptide associated with type 2 diabetesmellitus. *Proc. Nat. Acad. Sci.* 86: 9662–9666, 1989.
- [26500] 52.Bouhouche, A.; Benomar, A.; Birouk, N.; Mularoni, A.; Meggouh,F.; Tassin, J.; Grid, D.; Vandenberghe, A.; Yahyaoui, M.; Chkili,T.; Brice, A.; LeGuern, E.: A locus for an axonal form of autosomalrecessive Charcot-Marie-Tooth disease maps to chromosome 1q21.2–q21.3. *Am.J. Hum. Genet.* 65: 722–727, 1999.
- [26501] 53.Curran, M. E.; Splawski, I.; Timothy, K. W.; Vincent, G. M.; Green,E. D.; Keating, M. T.: A molecular basis for cardiac arrhythmia:HERG mutations cause long QT syndrome.

Cell 80: 795–803, 1995.

[26502] 54. Postel, E. H.; Mango, S. E.; Flint, S. J.: A nuclease-hypersensitive element of the human c-myc promoter interacts with a transcription initiation factor. *Molec. Cell. Biol.* 9: 5123–5133, 1989.

[26503] 55. Harada, T.; Harada, C.; Nakayama, N.; Okuyama, S.; Yoshida, K.; Kohsaka, S.; Matsuda, H.; Wada, K.: Modification of glial-neuronal cell interactions prevents photoreceptor apoptosis during light-induced retinal degeneration. *Neuron* 26: 533–541, 2000.

[26504] 56. Barnett, T.; Goebel, S. J.; Nothdurft, M. A.; Elting, J. J.: Carcinoembryonic antigen family: characterization of cDNAs coding for NCA and CEA and suggestion of nonrandom sequence variation in their conserved loop-domains. *Genomics* 3:59–66, 1988.

[26505] 57. Fakharzadeh, S.; Trusko, S. P.; George, D. L.: Tumorigenic potential associated with enhanced expression of a gene that is amplified in a mouse tumor cell line. *EMBO J.* 10: 1565–1569, 1991.

[26506] 58. Bonaiti-Pellie, C.; Briard-Guillemot, M. L.; Feingold, J.; Frezal, J.: Associated congenital malformations in retinoblastoma. *Clin. Genet.* 7: 37–39, 1975.

[26507] 59. Radford, D. M.; Nakai, H.; Pegg, A. E.; Shows, T. B.:

Mapping genes for rate-limiting enzymes in polyamine biosynthesis. (Abstract) Am.J. Hum. Genet. 41: A35 only, 1987.

- [26508] 60. Ruppert, J. M.; Kinzler, K. W.; Wong, A. J.; Bigner, S. H.; Kao, F.-T.; Law, M. L.; Seunavaz, H. N.; O'Brien, S. J.; Vogelstein, B.: The GLI-Kruppel family of human genes. Molec. Cell. Biol. 8: 3104-3113, 1988.
- [26509] 61. Grachtchouk, M.; Mo, R.; Yu, S.; Zhang, X.; Sasaki, H.; Hui, C.; Dlugosz, A. A.: Basal cell carcinomas in mice over-expressing Gli2 in skin. (Letter) Nature Genet. 24: 216-217, 2000.
- [26510] 62. Matsumoto, N.; Fujimoto, M.; Kato, R.; Niikawa, N.: Assignment of the human GLI2 gene to 2q14 by fluorescence in situ hybridization. Genomics 36:220-221, 1996.
- [26511] 63. Ishikawa, T.; Kibe, T.; Wada, Y.: Deletion of small nuclear ribonucleoprotein polypeptide N (SNRPN) in Prader-Willi syndrome detected by fluorescence in situ hybridization: two sibs with the typical phenotype without a cytogenetic deletion in chromosome 15q. Am. J. Med. Genet. 62:350-352, 1996.
- [26512] 64. Gubina, E.; Ruiz-Hidalgo, M. J.; Baladron, V.; Laborda, J.: Assignment of DLK1 to human chromosome band 14q32 by in situ hybridization. Cytogenet. Cell Genet. 84:

206–207, 1999.

- [26513] 65. Helman, L. J.; Thiele, C. J.; Linehan, W. M.; Nelkin, B. D.; Baylin, S. B.; Israel, M. A.: Molecular markers of neuroendocrine development and evidence of environmental regulation. *Proc. Nat. Acad. Sci.* 84:2336–2339, 1987.
- [26514] 66. Jensen, C. H.; Krogh, T. N.; Hojrup, P.; Clausen, P. P.; Skjodt, K.; Larsson, L.-I.; Enghild, J. J.; Teisner, B.: Protein structure of fetal antigen 1 (FA1): a novel circulating human epidermal-growth-factor-like protein expressed in neuroendocrine tumors and its relation to the gene products of *dlk* and *pG2*. *Europ. J. Biochem.* 225: 83–92, 1994.
- [26515] 67. Schulz, P.; Stucka, R.; Feldmann, H.; Combriato, G.; Klobeck, H.-G.; Fittler, F.: Sequence of a cDNA clone encompassing the complete mature human prostate specific antigen (PSA) and an unspliced leader sequence. *Nucleic Acids Res.* 16: 6226 only, 1988.
- [26516] 68. Sutherland, G. R.; Baker, E.; Hyland, V. J.; Callen, D. F.; Close, J. A.; Tregear, G. W.; Evans, B. A.; Richards, R. I.: Human prostate-specific antigen (PSA) is a member of the glandular kallikrein gene family at 19q13. *Cytogenet. Cell Genet.* 48: 205–207, 1988.
- [26517] 69. Bookstein, R.; Lee, E. Y.-H. P.; To, H.; Young, L.-J.; Sery, T. W.; Hayes, R. C.; Friedmann, T.; Lee, W.-H.: Human

retinoblastoma susceptibility gene: genomic organization and analysis of heterozygous intragenic deletion mutants. Proc. Nat. Acad. Sci. 85: 2210–2214, 1988.

- [26518] 70. Brantley, M. A.; Worley, L.; Harbour, J. W.: Altered expression of Rb and p53 in uveal melanomas following plaque radiotherapy. Am. J. Ophthalmol. 133: 242–248, 2002.
- [26519] 71. Bremner, R.; Du, D. C.; Connolly-Wilson, M. J.; Bridge, P.; Ahmad, K. F.; Mostachfi, H.; Rushlow, D.; Dunn, J. M.; Gallie, B. L.: Deletion of RB exons 24 and 25 causes low-penetrance retinoblastoma. Am. J. Hum. Genet. 61: 556–570, 1997.
- [26520] 72. Briard-Guillemot, M. L.; Bonaiti-Pellie, C.; Feingold, J.; Frezal, J.: Etude genetique du retinoblastome. Human-genetik 24: 271–284, 1974.

- [26521] 73. Brownstein, S.; de Chadarevian, J.-P.; Little, J. M.: Tri-lateral retinoblastoma: report of two cases. *Arch. Ophthalmol.* 102: 257-262, 1984.
- [26522] 74. Buchkovich, K.; Duffy, L. A.; Harlow, E.: The retinoblastoma protein is phosphorylated during specific phases of the cell cycle. *Cell* 58:1097-1105, 1989.
- [26523] 75. Scarpatti, E. M.; Sadler, J. E.; O'Connell, P.; Nakamura, Y.; Leppert, M.; Ballard, L.; Lathrop, G. M.; Lalouel, J.-M.; White, R.: Identification and mapping of RFLPs for human tissue factor (HTF) to chromosome 1p. *Nucleic Acids Res.* 15: 9098 only, 1987.
- [26524] 76. Scarpatti, E. M.; Wen, D.; Broze, G. J., Jr.; Miletich, J. P.; Flandermeyer, R. R.; Siegel, N. R.; Sadler, J. E.: Human tissue factor: cDNA sequence and chromosome localization of the gene. *Biochemistry* 26: 5234-5238, 1987.
- [26525] 77. Spicer, E. K.; Horton, R.; Bloem, L.; Bach, R.; Williams, K. R.; Guha, A.; Kraus, J.; Lin, T.-C.; Nemerson, Y.; Konigsberg, W. H.: Isolation of cDNA clones coding for human tissue factor: primary structure of the protein and cDNA. *Proc. Nat. Acad. Sci.* 84: 5148-5152, 1987.
- [26526] 78. Toomey, J. R.; Kratzer, K. E.; Lasky, N. M.; Broze, G. J., Jr.: Effect of tissue factor deficiency on mouse and tumor development. *Proc. Nat. Acad. Sci.* 94: 6922-6926, 1997.

- [26527] 79.Lohr, G. W.; Waller, H. D.: Zur Biochemie einiger angeborenerhaemolytischer Anaemien. *Folia Haemat.* 8: 377–397, 1963.
- [26528] 80.Chu, F.–F.: The human glutathione peroxidase genes GPX2, GPX3, and GPX4 map to chromosomes 14, 5, and 19, respectively. *Cytogenet.Cell Genet.* 66: 96–98, 1994.
- [26529] 81.Seldin, M. F.: Personal Communication. Durham, N. C. 3/13/1989.
- [26530] 82.Weis, J. H.; Morton, C. C.; Bruns, G. A. P.; Weis, J. J.; Klickstein, L. B.; Wong, W. W.; Fearon, D. T.: A complement receptor locus: genes encoding C3b/C4b receptor and C3d/Epstein–Barr virus receptor map to 1q32. *J. Immun.* 138: 312–315, 1987.
- [26531] 83.Carson, S. D.; Henry, W. M.; Haley, L.; Byers, M.; Shows, T.: The gene for tissue factor (coagulation factor III) is localized on human chromosome 1pter–1p21. (Abstract) *Cytogenet. Cell Genet.* 40:600 only, 1985.
- [26532] 84.Carson, S. D.; Henry, W. M.; Shows, T. B.: Tissue factor gene localized to human chromosome 1 (1pter–1p21). *Science* 229: 991–993, 1985.
- [26533] 85.Kao, F.–T.; Hartz, J.; Horton, R.; Nemerson, Y.; Carson, S. D.: Regional assignment of human tissue factor gene (F3) to chromosome 1p21–p22. *Somat. Cell Molec. Genet.*

14: 407–410, 1988.

- [26534] 86.Mackman, N.; Fowler, B. J.; Edgington, T. S.; Morrissey, J. H.: Functional analysis of the human tissue factor promoter and induction by serum. *Proc. Nat. Acad. Sci.* 87: 2254–2258, 1990.
- [26535] 87.Wroe, S. F.; Kelsey, G.; Skinner, J. A.; Bodle, D.; Ball, S. T.; Beechey, C. V.; Peters, J.; Williamson, C. M.: An imprinted transcript, antisense to *Nesp*, adds complexity to the cluster of imprinted genes at the mouse *Gnas* locus. *Proc. Nat. Acad. Sci.* 97: 3342–3346, 2000.
- [26536] 88.Wrong, O.: Tegernsee giant. (Letter) *Lancet* 339: 194 only, 1992.
- [26537] 89.Yang, I.; Park, S.; Ryu, M.; Woo, J.; Kim, S.; Kim, J.; Kim, Y.; Choi, Y.: Characteristics of gsp-positive growth hormone-secreting pituitary tumors in Korean acromegalic patients. *Europ. J. Endocr.* 134:720–726, 1996.
- [26538] 90.Yu, D.; Yu, S.; Schuster, V.; Kruse, K.; Clericuzio, C. L.; Weinstein, L. S.: Identification of two novel deletion mutations within the *Gs-alpha* gene (*GNAS1*) in Albright hereditary osteodystrophy. *J. Clin. Endocr. Metab.* 84: 3254–3259, 1999.
- [26539] 91.Lerman, M. I.; Minna, J. D.: The 630-kb lung cancer homozygous deletion region on human chromosome

3p21.3: identification and evaluation of the resident candidate tumor suppressor genes. *Cancer Res.* 60:6116–6133, 2000.

[26540] 92. Hobbs, H. H.; Leidersdorf, E.; Leffert, C. C.; Cryer, D. R.; Brown, M. S.; Goldstein, J. L.: Evidence for a dominant gene that suppresses hypercholesterolemia in a family with defective low density lipoprotein receptors. *J. Clin. Invest.* 84: 656–664, 1989.

[26541] 93. Nejsum, L. N.; Kwon, T.-H.; Jensen, U. B.; Fumagalli, O.; Frokiaer, J.; Krane, C. M.; Menon, A. G.; King, L. S.; Agre, P. C.; Nielsen, S.: Functional requirement of aquaporin-5 in plasma membranes of sweat glands. *Proc. Nat. Acad. Sci.* 99: 511–516, 2002.

[26542] 94. Auffray, C.; Korman, A. J.; Roux-Dosseto, M.; Bono, R.; Strominger, J. L.: cDNA clone for the heavy chain of the human B cell alloantigen DC1: strong sequence homology to the HLA-DR heavy chain. *Proc. Nat. Acad. Sci.* 79: 6337–6341, 1982.

[26543] 95. Benacerraf, B.: Role of MHC gene products in immune response. (Nobel Lecture). *Science* 212: 1229–1238, 1981.

[26544] 96. Bono, M. R.; Strominger, J. L.: Direct evidence of homology between DC-1 antigen and murine I-A molecules. *Nature* 299: 836–840, 1982.

- [26545] 97. Bradley, D. S.; Nabozny, G. H.; Cheng, S.; Zhou, P.; Griffiths, M. M.; Luthra, H. S.; David, C. S.: HLA-DQB1 polymorphism determines incidence, onset, and severity of collagen-induced arthritis in transgenic mice: implications in human rheumatoid arthritis. *J. Clin. Invest.* 100:2227-2234, 1997.
- [26546] 98. Briata, P.; Radka, S. F.; Sartoris, S.; Lee, J. S.: Alternative splicing of HLA-DQB transcripts and secretion of HLA-DQ beta-chain proteins: allelic polymorphism in splicing and polyadenylation (sic) sites. *Proc. Nat. Acad. Sci.* 86: 1003-1007, 1989.
- [26547] 99. Cohen, D.; Cohen, O.; Marcadet, A.; Massart, C.; Lathrop, M.; Deschamps, I.; Hors, J.; Schuller, E.; Dausset, J.: Class II HLA-DC beta-chain DNA restriction fragments differentiate among HLA-DR2 individuals in insulin-dependent diabetes and multiple sclerosis. *Proc. Nat. Acad. Sci.* 81: 1774-1778, 1984.
- [26548] 100. Corte, G.; Calabi, F.; Damiani, G.; Bargellesi, A.; Tosi, R.; Sorrentino, R.: Human Ia molecules carrying DC1 determinants differ in both alpha- and beta-subunits from Ia molecules carrying DR determinants. *Nature* 292:357-360, 1981.
- [26549] 101. Cucca, F.; Lampis, R.; Congia, M.; Angius, E.; Nutland,

S.; Bain, S. C.; Barnett, A. H.; Todd, J. A.: A correlation between the relative predisposition of MHC class II alleles to type 1 diabetes and the structure of their proteins. *Hum. Molec. Genet.* 10: 2025–2037, 2001.

[26550] 102. Duquesnoy, R. J.; Marrari, M.; Annen, K.: Identification of an HLA–DR associated system of B cell alloantigens.

Transplant. Proc. 11: 1757–1760, 1979.

[26551] 103. Gyllensten, U. B.; Erlich, H. A.: Generation of single-stranded DNA by the polymerase chain reaction and its application to direct sequencing of the HLA–DQA locus. *Proc. Nat. Acad. Sci.* 85: 7652–7656, 1988.

[26552] 104. Helmuth, R.; Fildes, N.; Blake, E.; Luce, M. C.; Chimera, J.; Madej, R.; Gorodezky, C.; Stoneking, M.; Schmill, N.; Klitz, W.; Higuchi, R.; Erlich, H. A.: HLA–DQ–alpha allele and genotype frequencies in various human populations, determined by using enzymatic amplification and oligonucleotide probes. *Am. J. Hum. Genet.* 47: 515–523, 1990.

[26553] 105. Hsu, S. H.; Chan, M. M.; Bias, W. B.: Genetic control of major histocompatibility complex–linked immune responses to synthetic polypeptides in man. *Proc. Nat. Acad. Sci.* 78: 440–444, 1981.

[26554] 106. Kwok, W. W.; Lotshaw, C.; Milner, E. C. B.; Knitter–

Jack, N.;Nepom, G. T.: Mutational analysis of the HLA-DQ3.2 insulin-dependentdiabetes mellitus susceptibility gene. *Proc. Nat. Acad. Sci.* 86:1027–1030, 1989.

- [26555] 107.Lambert, N. C.; Evans, P. C.; Hashizumi, T. L.; Maloney, S.; Gooley,T.; Furst, D. E.; Nelson, J. L.: Cutting edge: persistent fetal microchimerismin T lymphocytes is associated with HLA-DQA1*0501: implications inautoimmunity. *J. Immun.* 164: 5545–5548, 2000.
- [26556] 108.Levine, F.; Mach, B.; Long, E.; Erlich, H.; Pious, D.: Mappingin the HLA-D region with deletion variants and cloned genes. (Abstract) *Cytogenet.Cell Genet.* 37: 523, 1984.
- [26557] 109.Meyer, C. G.; Gallin, M.; Erttmann, K. D.; Brattig, N.; Schnittger,L.; Gelhaus, A.; Tannich, E.; Begovich, A. B.; Erlich, H. A.; Horstmann,R. D.: HLA-D alleles associated with generalized disease, localizeddisease, and putative immunity in *Onchocerca volvulus* infection. *Proc.Nat. Acad. Sci.* 91: 7515–7519, 1994.
- [26558] 110.Moriuchi, J.; Moriuchi, T.; Silver, J.: Nucleotide sequence ofan HLA-DQ alpha chain derived from a DRw9 cell line: genetic and evolutionaryimplications. *Proc. Nat. Acad. Sci.* 82: 3420–3424, 1985.
- [26559] 111.Nabozny, G. H.; Baisch, J. M.; Cheng, S.; Cosgrove, D.;

Griffiths, M. M.; Luthra, H. S.; David, C. S.: HLA-DQ8 transgenic mice are highly susceptible to collagen-induced arthritis: a novel model for human polyarthritis. *J. Exp. Med.* 183: 27–37, 1996.

[26560] 112. Nadler, L. M.; Stashenko, P.; Hardy, R.; Tomaselli, K. J.; Yunis, E. J.; Schlossman, S. F.; Pesando, J. M.: Monoclonal antibody identifies a new Ia-like (p29,34) polymorphic system linked to the HLA-D/DR region. *Nature* 290:591–593, 1981.

[26561] 113. Okada, K.; Boss, J. M.; Prentice, H.; Spies, T.; Mengler, R.; Auffray, C.; Lillie, J.; Grossberger, D.; Strominger, J. L.: Gene organization of DC and DX subregions of the human major histocompatibility complex. *Proc. Nat. Acad. Sci.* 82: 3410–3414, 1985.

[26562] 114. Schenning, L.; Larhammar, D.; Bill, P.; Wiman, K.; Jonsson, A.-K.; Rask, L.; Peterson, P. A.: Both alpha and beta chains of HLA-DC class II histocompatibility antigens display extensive polymorphism in their amino-terminal domains. *EMBO J.* 3: 447–452, 1984.

[26563] 115. Shackelford, D. A.; Kaufman, J. F.; Korman, A. J.; Strominger, J. L.: HLA-DR antigens: structure, separation of subpopulations, gene cloning and function. *Immun. Rev.* 66: 129–183, 1982.

- [26564] 116. Erneux, C.; Roeckel, N.; Takazawa, K.; Mailleux, P.; Vassart, G.; Mattei, M. G.: Localization of the genes for human inositol 1,4,5-trisphosphate 3-kinase A (ITPKA) and B (ITPKB) to chromosome regions 15q14-q21 and 1q41-q43, respectively, by in situ hybridization. *Genomics* 14:546-547, 1992.
- [26565] 117. Takazawa, K.; Perret, J.; Dumont, J. E.; Erneux, C.: Human brain inositol 1,4,5-trisphosphate 3-kinase cDNA sequence. *Nucleic Acids Res.* 18: 7141 only, 1990.
- [26566] 118. Takazawa, K.; Perret, J.; Dumont, J. E.; Erneux, C.: Molecular cloning and expression of a human brain inositol 1,4,5-trisphosphate 3-kinase. *Biochem. Biophys. Res. Commun.* 174: 529-535, 1991.
- [26567] 119. Takazawa, K.; Perret, J.; Dumont, J. E.; Erneux, C.: Molecular cloning and expression of a new putative inositol 1,4,5-trisphosphate 3-kinase isoenzyme. *Biochem. J.* 278: 883-886, 1991.
- [26568] 120. 't Hart, L. M.; Stolk, R. P.; Dekker, J. M.; Nijpels, G.; Grobbee, D. E.; Heine, R. J.; Maassen, J. A.: Prevalence of variants in candidate genes for type 2 diabetes mellitus in the Netherlands: the Rotterdam study and the Hoorn study. *J. Clin. Endocr. Metab.* 84: 1002-1006, 1999.
- [26569] 121. Abe, H.; Yamada, N.; Kamata, K.; Kuwaki, T.; Shimada,

M.; Osuga, J.; Shionoiri, F.; Yahagi, N.; Kadowaki, T.; Tamemoto, H.; Ishibashi, S.; Yazaki, Y.; Makuuchi, M.: Hypertension, hypertriglyceridemia, and impaired endothelium-dependent vascular relaxation in mice lacking insulin receptor substrate-1. *J. Clin. Invest.* 101: 1784–1788, 1998.

[26570] 122. Almind, K.; Bjorbaek, C.; Vestergaard, H.; Hansen, T.; Echwald, S.; Pedersen, O.: Amino acid polymorphisms of insulin receptor substrate-1 in non-insulin-dependent diabetes mellitus. *Lancet* 342: 828–832, 1993.

[26571] 123. Almind, K.; Inoue, G.; Pedersen, O.; Kahn, C. R.: A common amino acid polymorphism in insulin receptor substrate-1 causes impaired insulin signaling: evidence from transfection studies. *J. Clin. Invest.* 97: 2569–2575, 1996.

[26572] 124. Araki, E.; Sun, X.-J.; Haag, B. L., III; Chuang, L.-M.; Zhang, Y.; Yang-Feng, T. L.; White, M. F.; Kahn, C. R.: Human skeletal muscle insulin receptor substrate-1: characterization of the cDNA, gene, and chromosomal localization. *Diabetes* 42: 1041–1054, 1993.

[26573] 125. Baroni, M. G.; D'Andrea, M. P.; Montali, A.; Pannitteri, G.; Barilla, F.; Campagna, F.; Mazzei, E.; Lovari, S.; Seccareccia, F.; Campa, P. P.; Ricci, G.; Pozzilli, P.; Urbinati, G.;

Arca, M.: A common mutation of the insulin receptor substrate-1 gene is a risk factor for coronary artery disease. *Arterioscler. Thromb. Vasc. Biol.* 19:2975–2980, 1999.

- [26574] 126. Bohni, R.; Riesgo-Escovar, J.; Oldham, S.; Brogiolo, W.; Stocker, H.; Andruss, B. F.; Beckingham, K.; Hafen, E.: Autonomous control of cell and organ size by CHICO, a *Drosophila* homolog of vertebrate IRS1–4. *Cell* 97: 865–875, 1999.
- [26575] 127. Fletcher, W. H.; Britz-Cunningham, S. H.; Zuppan, C. W.: Connexin43 mutations in sporadic and familial defects of laterality. (Letter) *New Eng. J. Med.* 333: 941–942, 1995.
- [26576] 128. Gebbia, M.; Towbin, J. A.; Casey, B.: Failure to detect connexin43 mutations in 38 cases of sporadic and familial heterotaxy. *Circulation* 94:1909–1912, 1996.
- [26577] 129. Gebbia, M.; Towbin, J. A.; Casey, B.: Connexin43 gene mutations and heterotaxy. Response. (Letter) *Circulation* 97: 118 only, 1998.
- [26578] 130. Guerrero, P. A.; Schuessler, R. B.; Davis, L. M.; Beyer, E. C.; Johnson, C. M.; Yamada, K. A.; Saffitz, J. E.: Slow ventricular conduction in mice heterozygous for a connexin43 null mutation. *J. Clin. Invest.* 99:1991–1998, 1997.
- [26579] 131. Li, J.-Y.; Hou, X.-E.; Dahlstrom, A.: GAP-43 and its relation to autonomic and sensory neurons in sciatic nerve

and gastrocnemius muscle in the rat. *J. Auton. Nerv. Syst.* 50: 299–309, 1995.

- [26580] 132. Liao, Y.; Day, K. H.; Damon, D. N.; Duling, B. R.: Endothelial cell-specific knockout of connexin 43 causes hypotension and bradycardia in mice. *Proc. Nat. Acad. Sci.* 98: 9989–9994, 2001.
- [26581] 133. Liu, X. Z.; Xia, X. J.; Adams, J.; Chen, Z. Y.; Welch, K. O.; Tekin, M.; Ouyang, X. M.; Kristiansen, A.; Pandya, A.; Balkany, T.; Arnos, K. S.; Nance, W. E.: Mutations in GJA1 (connexin 43) are associated with non-syndromic autosomal recessive deafness. *Hum. Molec. Genet.* 10: 2945–2951, 2001.
- [26582] 134. Reaume, A. G.; de Sousa, P. A.; Kulkarni, S.; Langille, B. L.; Zhu, D.; Davies, T. C.; Juneja, S. C.; Kidder, G. M.; Rossant, J.: Cardiac malformation in neonatal mice lacking connexin43. *Science* 267: 1831–1834, 1995.
- [26583] 135. Splitt, M. P.; Burn, J.; Goodship, J.: Connexin43 mutations in sporadic and familial defects of laterality. (Letter) *New Eng. J. Med.* 333: 941, 1995.
- [26584] 136. Splitt, M. P.; Tsai, M. Y.; Burn, J.; Goodship, J. A.: Absence of mutations in the regulatory domain of the gap junction protein connexin 43 in patients with viscerotaxial heterotaxy. *Heart* 77: 369–370, 1997.

- [26585] 137.Toth, T.; Hajdu, J.; Marton, T.; Nagy, B.; Papp, Z.: Connexin43gene mutations and heterotaxy. (Letter) Circulation 97: 117–118,1998.
- [26586] 138.Ya, J.; Erdstieck–Ernste, E. B. H. W.; de Boer, P. A. J.; vanKempen, M. J. A.; Jongsma, H.; Gros, D.; Moorman, A. F. M.; Lamers,W. H.: Heart defects in connexin43–deficient mice. Circ. Res. 82:360–366, 1998.
- [26587] 139.Anderson, R. A.; Koch, S.; Camerini–Otero, R. D.: Cardiovascularfindings in congenital contractural arachn–odactyly: report of an affectedkindred. Am. J. Med. Genet. 18: 265–271, 1984.
- [26588] 140.Babcock, D.; Gasner, C.; Francke, U.; Maslen, C.: A single mutationthat results in an asp–to–his substitution and partial exon skippingin a family with congenital con–tractual arachnodactyly. Hum. Genet. 103:22–28, 1998.
- [26589] 141.Mantamadiotis, T.; Lemberger, T.; Bleckmann, S. C.; Kern, H.;Kretz, O.; Villalba, A. M.; Tronche, F.; Kellendonk, C.; Gau, D.;Kapfhammer, J.; Otto, C.; Schmid, W.; Schutz, G.: Disruption of CREBfunction in brain leads to neurode–generation. Nature Genet. 31:47–54, 2002.
- [26590] 142.Montminy, M. R.; Bilezikjian, L. M.: Binding of a nu–clear proteinto the cyclic–AMP response element of the somatostatin gene. Nature 328:175–178, 1987.

- [26591] 143. Montminy, M. R.; Sevarino, K. A.; Wagner, J. A.; Mandel, G.; Goodman, R. H.: Identification of a cyclic-AMP-responsive element within the rat somatostatin gene. *Proc. Nat. Acad. Sci.* 83: 6682–6686, 1986.
- [26592] 144. Nguyen, L. Q.; Kopp, P.; Martinson, F.; Stanfield, K.; Roth, S. I.; Jameson, J. L.: A dominant negative CREB (cAMP response element-binding protein) isoform inhibits thyrocyte growth, thyroid-specific gene expression, differentiation, and function. *Molec. Endocr.* 14: 1448–1461, 2000.
- [26593] 145. Parker, D.; Ferreri, K.; Nakajima, T.; LaMorte, V. J.; Evans, R.; Koerber, S. C.; Hoeger, C.; Montminy, M. R.: Phosphorylation of CREB at ser-133 induces complex formation with CREB-binding protein via a direct mechanism. *Molec. Cell. Biol.* 16: 694–703, 1996.
- [26594] 146. Radhakrishnan, I.; Perez-Alvarado, G. C.; Parker, D.; Dyson, H. J.; Montminy, M. R.; Wright, P. E.: Solution structure of the KIX domain of CBP bound to the transactivation domain of CREB: a model for activator:coactivator interactions. *Cell* 91: 741–752, 1997.
- [26595] 147. Solomou, E. E.; Juang, Y.-T.; Gourley, M. F.; Kammer, G. M.; Tsokos, G. C.: Molecular basis of deficient IL-2 production in T cells from patients with systemic lupus erythematosus. *J. Immun.* 166: 4216–4222, 2001.

- [26596] 148. Taylor, A. K.; Klisak, I.; Mohandas, T.; Sparkes, R. S.; Li, C.; Gaynor, R.; Lusk, A. J.: Assignment of the human gene for CREB1 to chromosome 2q32.3–q34. *Genomics* 7: 416–421, 1990.
- [26597] 149. Blendy, J. A.; Kaestner, K. H.; Weinbauer, G. F.; Nieschlag, E.; Schutz, G.: Severe impairment of spermatogenesis in mice lacking the CREM gene. *Nature* 380: 162–165, 1996.
- [26598] 150. Foulkes, N. S.; Borrelli, E.; Sassone-Corsi, P.: CREM gene: use of alternative DNA-binding domains generates multiple antagonists of cAMP-induced transcription. *Cell* 64: 739–749, 1991.
- [26599] 151. Masquillier, D.; Foulkes, N. S.; Mattei, M.-G.; Sassone-Corsi, P.: Human CREM gene: evolutionary conservation, chromosomal localization, and inducibility of the transcript. *Cell Growth Differ.* 4: 931–937, 1993.
- [26600] 152. Nantel, F.; Monaco, L.; Foulkes, N. S.; Masquillier, D.; LeMeur, M.; Henriksen, K.; Dierich, A.; Parvinen, M.; Sassone-Corsi, P.: Spermiogenesis deficiency and germ-cell apoptosis in CREM-mutant mice. *Nature* 380: 159–162, 1996.
- [26601] 153. Meyerson, M.; Enders, G. H.; Wu, C.-L.; Su, L.-K.; Gorka, C.; Nelson, C.; Harlow, E.; Tsai, L.-H.: A family of

human cdc2-related protein kinases. EMBO J. 11: 2909–2917, 1992.

- [26602] 154. Ye, X.; Zhu, C.; Harper, J. W.: A premature-termination mutation in the *Mus musculus* cyclin-dependent kinase 3 gene. Proc. Nat. Acad. Sci. 98: 1682–1686, 2001.
- [26603] 155. Demetrick, D. J.; Zhang, H.; Beach, D. H.: Chromosomal mapping of human CDK2, CDK4, and CDK5 cell cycle kinase genes. Cytogenet. Cell Genet. 66: 72–74, 1994.
- [26604] 156. Harbour, J. W.; Luo, R. X.; Dei Santi, A.; Postigo, A. A.; Dean, D. C.: Cdk phosphorylation triggers sequential intramolecular interactions that progressively block Rb functions as cells move through G1. Cell 98: 859–869, 1999.
- [26605] 157. Bullrich, F.; MacLachlan, T. K.; Sang, N.; Druck, T.; Veronese, M. L.; Allen, S. L.; Chiorazzi, N.; Koff, A.; Heubner, K.; Croce, C. M.; Giordano, A.: Chromosomal mapping of members of the cdc2 family of protein kinases, cdk3, cdk6, PISLRE, and PITALRE, and a cdk inhibitor, p27-Kip1, to regions involved in human cancer. Cancer Res. 55: 1199–1205, 1995.
- [26606] 158. Bhandari, V.; Bateman, A.: Structure and chromosomal location of the human granulin gene. Biochem. Biophys. Res. Commun. 188: 57–63, 1992.
- [26607] 159. Bhandari, V.; Palfree, R. G. E.; Bateman, A.: Isolation

and sequence of the granulin precursor cDNA from human bone marrow reveals tandem cysteine-rich granulin domains. *Proc. Nat. Acad. Sci.* 89: 1715–1719, 1992.

[26608] 160. He, Z.; Bateman, A.: Progranulin gene expression regulates epithelial cell growth and promotes tumor growth in vivo. *Cancer Res.* 59: 3222–3229, 1999.

[26609] 161. Liao, L. M.; Lallone, R. L.; Seitz, R. S.; Buznikov, A.; Gregg, J. P.; Kornblum, H. I.; Nelson, S. F.; Bronstein, J. M.: Identification of a human glioma-associated growth factor gene, granulin, using differential immuno-absorption. *Cancer Res.* 60: 1353–1360, 2000.

[26610] 162. Thornton, M. A.; Poncz, M.; Korostishevsky, M.; Yakobson, E.; Usher, S.; Seligsohn, U.; Peretz, H.: The human platelet alpha-IIb gene is not closely linked to its integrin partner beta-3. *Blood* 94: 2039–2047, 1999.

[26611] 163. Birnbaum, R. A.; O'Marcaigh, A.; Wardak, Z.; Zhang, Y.-Y.; Dranoff, G.; Jacks, T.; Clapp, D. W.; Shannon, K. M.: Nf1 and Gmcsf interact in myeloid leukemogenesis. *Molec. Cell* 5: 189–195, 2000.

[26612] 164. Cantrell, M. A.; Anderson, D.; Cerretti, D. P.; Price, V.; McKereghan, K.; Tushinski, R. J.; Mochizuki, D. Y.; Larsen, A.; Grabstein, K.; Gillis, S.; Cosman, D.: Cloning, sequence, and expression of a human granulocyte/macrophage

colony-stimulating factor. Proc. Nat. Acad.Sci. 82:
6250-6254, 1985.

[26613] 165.Frolova, E. I.; Dolganov, G. M.; Mazo, I. A.; Smirnov, D. V.; Copeland,P.; Stewart, C.; O'Brien, S. J.; Dean, M.: Linkage mapping of thehuman CSF2 and IL3 genes. Proc. Nat. Acad. Sci. 88: 4821-4824, 1991.

[26614] 166.Grabstein, K. H.; Urdal, D. L.; Tushinski, R. J.; Mochizuki, D.Y.; Price, V. L.; Cantrell, M. A.; Gillis, S.; Conlon, P. J.: Inductionof macrophage tumoricidal activity by granulocyte-macrophage colony-stimulatingfactor. Science 232: 506-508, 1986.

[26615] 167.Huebner, K.; Isobe, M.; Croce, C. M.; Golde, D. W.; Kaufman, S.E.; Gasson, J. C.: The human gene encoding GM-CSF is at 5q21-q32,the chromosome region deleted in the 5q- anomaly. Science 230: 1282-1285,1985.

[26616] 168.Le Beau, M. M.; Westbrook, C. A.; Diaz, M. O.; Larson, R. A.; Rowley,J. D.; Gasson, J. C.; Golde, D. W.; Sherr, C. J.: Evidence for theinvolvement of GM-CSF and FMS in the deletion (5q) in myeloid disorders. Science 231:984-987, 1986.

[26617] 169.LeVine, A. M.; Reed, J. A.; Kurak, K. E.; Cianciolo, E.; Whitsett,J. A.: GM-CSF-deficient mice are susceptible to pulmonary group Bstreptococcal infection. J. Clin. Invest.

103: 563–569, 1999.

- [26618] 170. Metcalf, D.: The molecular biology and functions of the granulocyte–macrophage colony–stimulating factors. *Blood* 67: 257–267, 1986.
- [26619] 171. Pettenati, M. J.; Le Beau, M. M.; Lemons, R. S.; Shima, E. A.; Kawasaki, E. S.; Larson, R. A.; Sherr, C. J.; Diaz, M. O.; Rowley, J. D.: Assignment of CSF–1 to 5q33.1: evidence for clustering of genes regulating hematopoiesis and for their involvement in the deletion of the long arm of chromosome 5 in myeloid disorders. *Proc. Nat. Acad. Sci.* 84: 2970–2974, 1987.
- [26620] 172. Sieff, C. A.; Emerson, S. G.; Donahue, R. E.; Nathan, D. G.; Wang, E. A.; Wong, G. G.; Clark, S. C.: Human recombinant granulocyte–macrophage colony–stimulating factor: a multilineage hematopoietin. *Science* 230: 1171–1173, 1985.
- [26621] 173. Thangavelu, M.; Neuman, W. L.; Espinosa, R., III; Nakamura, Y.; Westbrook, C. A.; Le Beau, M. M.: A physical and genetic linkage map of the distal long arm of human chromosome 5. *Cytogenet. Cell Genet.* 59: 27–30, 1992.
- [26622] 174. Yang, Y.–C.; Kovacic, S.; Kriz, R.; Wolf, S.; Clark, S. C.; Wellems, T. E.; Nienhuis, A.; Epstein, N.: The human genes for GM–CSF and IL3 are closely linked in tandem on chro–

mosome 5. Blood 71: 958–961,1988.

[26623] 175.Modi, W. S.; Chen, Z.–Q.: Localization of the human CXC chemokinesubfamily on the long arm of chromosome 4 using radiation hybrids. Genomics 47:136–139, 1998.

[26624] 176.Proost, P.; Wuyts, A.; Conings, R.; Lenaerts, J.–P.; Billaud, A.;Opdenakker, G.; Van Damme, J.: Human and bovine granulocyte chemotacticprotein–2: complete amino acid sequence and functional characterizationas chemokines. Biochemistry 32: 10170–10177, 1993.

[26625] 177.Wong, G. G.; Witek, J. S.; Temple, P. A.; Wilkens, K. M.; Leary,A. C.; Luxenberg, D. P.; Jones, S. S.; Brown, E. L.; Kay, R. M.; Orr,E. C.; Shoemaker, C.; Golde, D. W.; Kaufman, R. J.; Hewick, R. M.;Wang, E. A.; Clark, S. C.: Human GM–CSF: molecular cloning of thecomplementary DNA and purification of the natural and recombinantproteins. Science 228: 810–815, 1985.

[26626] 178.Rovai, L. E.; Herschman, H. R.; Smith, J. B.: Cloning and characterizationof the human granulocyte chemotactic protein–2 gene. J. Immun. 158:5257–5266, 1997.

[26627] 179.Wuyts, A.; van Osselaer, N.; Haelens, A.; Samson, I.; Herdewijn,P.; Ben–Baruch, A.; Oppenheim, J. J.; Proost, P.; van Damme, J.:Characterization of synthetic human granulocyte chemotactic protein2: usage of chemokine recep–

tors CXCR1 and CXCR2 and in vivo inflammatory properties. *Biochemistry* 36: 2716–2723, 1997.

- [26628] 180. Kanda, N.; Fukushige, S.; Murotsu, T.; Yoshida, M. C.; Tsuchiya, M.; Asano, S.; Kaziro, Y.; Nagata, S.: Human gene coding for granulocyte–colony stimulating factor assigned to the q21–q22 region of chromosome 17. *Somat. Cell Molec. Genet.* 13: 679–684, 1987.
- [26629] 181. Abraham, J. A.; Mergia, A.; Whang, J. L.; Tumolo, A.; Friedman, J.; Hjerild, K. A.; Gospodarowicz, D.; Fiddes, J. C.: Nucleotide sequence of a bovine clone encoding the angiogenic protein, basic fibroblast growth factor. *Science* 233: 545–548, 1986.
- [26630] 182. Abraham, J. A.; Whang, J. L.; Tumolo, A.; Mergia, A.; Friedman, J.; Gospodarowicz, D.; Fiddes, J. C.: Human basic fibroblast growth factor: nucleotide sequence and genomic organization. *EMBO J.* 5:2523–2528, 1986.
- [26631] 183. Doniach, T.: Basic FGF as an inducer of anteroposterior neural pattern. *Cell* 83: 1067–1070, 1995.
- [26632] 184. Dono, R.; Texido, G.; Dussel, R.; Ehmke, H.; Zeller, R.: Impaired cerebral cortex development and blood pressure regulation in FGF2–deficient mice. *EMBO J.* 17: 4213–4225, 1998.
- [26633] 185. Fukushima, Y.; Byers, M. G.; Fiddes, J. C.; Shows, T.

B.: The human basic fibroblast growth factor gene (FGFB) is assigned to chromosome 4q25. *Cytogenet. Cell Genet.* 54: 159–160, 1990.

[26634] 186. Gritti, A.; Parati, E. A.; Cova, L.; Frolichsthal, P.; Galli, R.; Wanke, E.; Faravelli, L.; Morassutti, D. J.; Roisen, F.; Nickel, D. D.; Vescovi, A. L.: Multipotential stem cells from the adult mouse brain proliferate and self-renew in response to basic fibroblast growth factor. *J. Neurosci.* 16: 1091–1100, 1996.

[26635] 187. Kawaguchi, H.; Nakamura, K.; Tabata, Y.; Ikada, Y.; Aoyama, I.; Anzai, J.; Nakamura, T.; Hiyama, Y.; Tamura, M.: Acceleration of fracture healing in nonhuman primates by fibroblast growth factor-2. *J. Clin. Endocr. Metab.* 86: 875–880, 2001.

[26636] 188. Kurokawa, T.; Sasada, R.; Iwane, M.; Igarashi, K.: Cloning and expression of cDNA encoding human basic fibroblast growth factor. *FEBS Lett.* 213: 189–194, 1987.

[26637] 189. Lafage-Pochitaloff, M.; Galland, F.; Simonetti, J.; Prats, H.; Mattei, M.-G.; Birnbaum, D.: The human basic fibroblast growth factor gene is located on the long arm of chromosome 4 at bands q26–q27. *Oncogene Res.* 5: 241–244, 1990.

[26638] 190. Mattei, M.-G.; Pebusque, M.-J.; Birnbaum, D.: Chro-

mosomal localization of mouse Fgf2 and Fgf5 genes.

Mammalian Genome 2: 135–137, 1992.

- [26639] 191. Montero, A.; Okada, Y.; Tomita, M.; Ito, M.; Tsurukami, H.; Nakamura, T.; Doetschman, T.; Coffin, J. D.; Hurley, M. M.: Disruption of the fibroblast growth factor-2 gene results in decreased bone mass and bone formation. J. Clin. Invest. 105: 1085–1093, 2000.
- [26640] 192. Ortega, S.; Ittmann, M.; Tsang, S. H.; Ehrlich, M.; Basilico, C.: Neuronal defects and delayed wound healing in mice lacking fibroblast growth factor 2. Proc. Nat. Acad. Sci. 95: 5672–5677, 1998.
- [26641] 193. Plotnikov, A. N.; Schlessinger, J.; Hubbard, S. R.; Mohammadi, M.: Structural basis for FGF receptor dimerization and activation. Cell 98: 641–650, 1999.
- [26642] 194. Aerssens, J.; Chaffanet, M.; Baens, M.; Matthijs, G.; Van Den Berghe, H.; Cassiman, J.-J.; Marynen, P.: Regional assignment of seven loci to 12p13.2-pter by PCR analysis of somatic cell hybrids containing the der(12) or the der(X) chromosome from a mesothelioma showing t(X;12)(q22;p13). Genomics 20: 119–121, 1994.
- [26643] 195. Iida, S.; Yoshida, T.; Naito, K.; Sakamoto, H.; Katoh, O.; Hirohashi, S.; Sato, T.; Onda, M.; Sugimura, T.; Terada, M.: Human hst-2 (FGF-6) oncogene: cDNA cloning and

characterization. *Oncogene* 7: 303–309,1992.

- [26644] 196.Marics, I.; Adelaide, J.; Raybaud, F.; Mattei, M.–G.; Coulier,F.; Planche, J.; de Lapeyriere, O.; Birnbaum, D.: Characterizationof the HST–related FGF.6 gene, a new member of the fibroblast growthfactor gene family. *Onco–gene* 4: 335–340, 1989.
- [26645] 197.Avraham, K. B.; Givol, D.; Avivi, A.; Yayon, A.; Copeland, N. G.;Jenkins, N. A.: Mapping of murine fibrob–last growth factor receptorsrefines regions of homology between mouse and human chromosomes. *Genomics* 21:656–658, 1994.
- [26646] 198.Bota, D. A.; Davies, K. J. A.: Lon protease preferen–tially degradesoxidized mitochondrial aconitase by an ATP–stimulated mechanism. *NatureCell Biol.* 4: 674–680, 2002.
- [26647] 199.Slaughter, C. A.; Hopkinson, D. A.; Harris, H.: Aconi–tase polymorphismin man. *Ann. Hum. Genet.* 39: 193–202, 1975.
- [26648] 200.Azevedo, E. S.; Da Silva, M. C. B. O.; Lima, A. M. V.; Fonseca,E. F.; Conseicao, M. M.: Human aconitase poly–morphism in three samplesfrom northeastern Brazil. *Ann. Hum. Genet.* 43: 7–10, 1979.
- [26649] 201.Hentze, M. W.; Seuanez, H. N.; O'Brien, S. J.; Harford,

J. B.;Klausner, R. D.: Chromosomal localization of nucleic acid-bindingproteins by affinity mapping: assignment of the IRE-binding proteingene to human chromosome 9. Nucleic Acids Res. 17: 6103–6108, 1989.

[26650] 202.Mohandas, T.; Sparkes, R. S.; Sparkes, M. C.; Shulkin, J. D.; Toomey,K. E.; Funderburk, S. J.: Regional localization of human gene locion chromosome 9: studies of somatic cell hybrids containing humantranslocations. Am. J. Hum. Genet. 31: 586–600, 1979.

[26651] 203.Robson, E. B.; Cook, P. J. L.; Buckton, K. E.: Family studieswith the chromosome 9 markers ABO, AK–1, ACON–S and 9qh. Ann. Hum.Genet. 41: 53–60, 1977.

[26652] 204.Shay, J. W.; Werbin, H.: New evidence for the insertion of mitochondrialDNA into the human genome: signifi–cance for cancer and aging. Mutat.Res. 275: 227–235, 1992.

[26653] 205.Shows, T. B.; Brown, J. A.: Mapping AK–1, ACON–S, and AK–3 tochromosome 9 in man employing an X–9 translocation and somatic cellhybrids. Cytogenet. Cell Genet. 19: 26–37, 1977.

[26654] 206.Westerveld, A.; van Henegouwen, B. H. M. A.; Van Someren, H.:Evidence for synteny between the human loci for galactose–1–phosphateuridyl transferase and aconi–

tase in man–Chinese hamster somatic cellhybrids. Cyto-genet. Cell Genet. 14: 453–454, 1975.

- [26655] 207.Akkari, P. A.; Eyre, H. J.; Wilton, S. D.; Callen, D. F.; Lane,S. A.; Meredith, C.; Kedes, L.; Laing, N. G.: Assign-ment of the humanskeletal muscle alpha actin gene (ACTA1) to 1q42 by fluorescence insitu hybridisation. Cy-togenet. Cell Genet. 65: 265–267, 1994.
- [26656] 208.Alonso, S.; Montagutelli, X.; Simon–Chazottes, D.; Guenet, J.–L.;Buckingham, M.: Re–localization of Actsk–1 to mouse chromosome 8,a new region of homology with human chromosome 1. Mammalian Genome 4:15–20, 1993.
- [26657] 209.Czosnek, H.; Nudel, U.; Shani, M.; Barker, P. E.; Pravtcheva, D.D.; Ruddle, F. H.; Yaffe, D.: The genes cod-ing for the muscle contractileproteins, myosin heavy chain, myosin light chain 2, and skeletal muscleactin are located on three different mouse chromosomes. EMBO J. 1:1299–1305, 1982.
- [26658] 210.Drury, A. N.; Szent–Gyorgyi, A.: The physiological ac-tivity ofadenine compounds with especial reference to their action upon themammalian heart. J. Physiol. 68: 213–237, 1929.
- [26659] 211.Libert, F.; Passage, E.; Parmentier, M.; Simons, M.–J.;

Vassart,G.; Mattei, M.-G.: Chromosomal mapping of A1 and A2 adenosine receptors,VIP receptor, and a new subtype of serotonin receptor. *Genomics* 11:225–227, 1991.

[26660] 212.Stiles, G. L.: Adenosine receptors. *J. Biol. Chem.* 267: 6451–6454,1992.

[26661] 213.Sun, D.; Samuelson, L. C.; Yang, T.; Huang, Y.; Paliege, A.; Saunders,T.; Briggs, J.; Schnermann, J.: Mediation of tubuloglomerular feedbackby adenosine: evidence from mice lacking adenosine 1 receptors. *Proc.Nat. Acad. Sci.* 98: 9983–9988, 2001.

[26662] 214.Townsend–Nicholson, A.; Baker, E.; Schofield, P. R.; Sutherland,G. R.: Localization of the adenosine A1 receptor subtype gene (ADORA1)to chromosome 1q32.1. *Genomics* 26: 423–425, 1995.

[26663] 215.Chen, J.-F.; Huang, Z.; Ma, J.; Zhu, J.; Moratalla, R.; Standaert,D.; Moskowitz, M. A.; Fink, J. S.; Schwarzschild, M. A.: A2A adenosinereceptor deficiency attenuates brain injury induced by transient focalischemia in mice. *J. Neurosci.* 19: 9192–9200, 1999.

[26664] 216.Gaudray, P.: Personal Communication. Nice, France 6/1/1994.

[26665] 217.Gusella, J. F.: Personal Communication. Boston, Mass. 4/17/1994.

- [26666] 218. Le, F.; Townsend-Nicholson, A.; Baker, E.; Sutherland, G. R.; Schofield, P. R.: Characterization and chromosomal localization of the human A_{2a} adenosine receptor gene: ADORA2A. *Biochem. Biophys. Res. Commun.* 223:461–467, 1996.
- [26667] 219. Ledent, C.; Vaugeois, J.-M.; Schiffmann, S. N.; Pedrazzini, T.; El Yacoubi, M. E.; Vanderhaeghen, J.-J.; Costentin, J.; Heath, J.K.; Vassart, G.; Parmentier, M.: Aggressiveness, hypoalgesia and high blood pressure in mice lacking the adenosine A_{2a} receptor. *Nature* 388:674–678, 1997.
- [26668] 220. Libert, F.; Passage, E.; Parmentier, M.; Simons, M.-J.; Vassart, G.; Mattei, M.-G.: Chromosomal mapping of A₁ and A₂ adenosine receptors, VIP receptor, and a new subtype of serotonin receptor. *Genomics* 11:225–227, 1991. Note: Erratum: *Genomics* 23: 305 only, 1994.
- [26669] 221. MacCollin, M.; Peterfreund, R.; MacDonald, M.; Fink, J. S.; Gusella, J.: Mapping of a human A_{2a} adenosine receptor (ADORA2) to chromosome 22. *Genomics* 20: 332–333, 1994.
- [26670] 222. Ohta, A.; Sitkovsky, M.: Role of G-protein-coupled adenosine receptors in downregulation of inflammation and protection from tissue damage. *Nature*

916–920,2001.

- [26671] 223.Szepietowski, P.; Perucca–Lostanlen, D.; Gaudray, P.: Mapping genes according to their amplification status in tumor cells: contribution to the map of 11q13. *Genomics* 16: 745–750, 1993.
- [26672] 224.Minghetti, P. P.; Law, S. W.; Dugaiczyk, A.: The rate of molecular evolution of alpha–fetoprotein approaches that of pseudogenes. *Molec.Biol. Evol.* 2: 347–358, 1985.
- [26673] 225.Barbry, P.; Champe, M.; Chassande, O.; Munemitsu, S.; Champigny, G.; Lingueglia, E.; Maes, P.; Frelin, C.; Tartar, A.; Ullrich, A.; Lazdunski, M.: Human kidney amiloride–binding protein: cDNA structure and functional expression. *Proc. Nat. Acad. Sci.* 87: 7347–7351, 1990.
- [26674] 226.Barbry, P.; Simon–Bouy, B.; Mattei, M.–G.; Le Guern, E.; Jaume–Roig, B.; Chassande, O.; Ullrich, A.; Lazdunski, M.: Localization of the gene for amiloride binding protein on chromosome 7 and RFLP analysis in cystic fibrosis families. *Hum. Genet.* 85: 587–589, 1990.
- [26675] 227.Chassande, O.; Renard, S.; Barbry, P.; Lazdunski, M.: The human gene for diamine oxidase, an amiloride binding protein: molecular cloning, sequencing, and characterization of the promoter. *J. Biol.Chem.* 269: 14484–14489, 1994.

- [26676] 228. Novotny, W. F.; Chassande, O.; Baker, M.; Lazdunski, M.; Barbry, P.: Diamine oxidase is the amiloride-binding protein and is inhibited by amiloride analogues. *J. Biol. Chem.* 269: 9921–9925, 1994.
- [26677] 229. Brinkman–Van der Linden, E. C. M.; Sjöberg, E. R.; Juneja, L. R.; Crocker, P. R.; Varki, N.; Varki, A.: Loss of N-glycolylneuraminic acid in human evolution: implications for sialic acid recognition by siglecs. *J. Biol. Chem.* 275: 8633–8640, 2000.
- [26678] 230. O’Keefe, T. L.; Williams, G. T.; Davies, S. L.; Neuberger, M. S.: Hyperresponsive B cells in CD22-deficient mice. *Science* 274: 798–801, 1996.
- [26679] 231. Wilson, G. L.; Fox, C. H.; Fauchi, A. S.; Kehrl, J. H.: cDNA cloning of the B cell membrane protein CD22: a mediator of B–B cell interactions. *J. Exp. Med.* 173: 137–146, 1991.
- [26680] 232. Wilson, G. L.; Najfeld, V.; Kozlow, E.; Menniger, J.; Ward, D.; Kehrl, J. H.: Genomic structure and chromosomal mapping of the human CD22 gene. *J. Immun.* 150: 5013–5024, 1993.
- [26681] 233. Krainer, A. R.; Mayeda, A.; Kozak, D.; Binns, G.: Functional expression of cloned human splicing factor SF2: homology to RNA-binding proteins, U1 70K, and *Drosophila*

splicing regulators. *Cell* 66: 383–394, 1991.

[26682] 234. Schlossman, S. F.; Boumsell, L.; Gilks, W.; Harlan, J. M.; Kishimoto, T.; Morimoto, C.; Ritz, J.; Shaw, S.; Silverstein, R. L.; Springer, T. A.; Tedder, T. F.; Todd, R. F.: CD antigens 1993. *Immun. Today* 15:98–99, 1994.

[26683] 235. Spring, F. A.; Holmes, C. H.; Simpson, K. L.; Mawby, W. J.; Mattes, M. J.; Okubo, Y.; Parsons, S. F.: The Ok(a) blood group antigen is a marker for the M6 leukocyte activation antigen, the human homolog of OX-47 antigen, basigin and neurothelin, an immunoglobulin superfamily-molecule that is widely expressed in human cells and tissues. *Europ. J. Immun.* 27: 891–897, 1997.

[26684] 236. Brown, M. H.; Boles, K.; van der Merwe, P. A.; Kumar, V.; Mathew, P. A.; Barclay, A. N.: 2B4, the natural killer and T cell immunoglobulin superfamily surface protein, is a ligand for CD48. *J. Exp. Med.* 188:2083–2093, 1998.

[26685] 237. Olives, B.; Neau, P.; Bailly, P.; Hediger, M. A.; Rousset, G.; Cartron, J.-P.; Ripoche, P.: Cloning and functional expression of a urea transporter from human bone marrow cells. *J. Biol. Chem.* 269:31649–31652, 1994.

[26686] 238. Pausch, V.; Mayr, W. R.: Analysis of the linkage JK-IGK, MNS-GC and of two other possible linkage groups. *Hum. Hered.* 37: 260–262, 1987.

- [26687] 239.Promeneur, D.; Rousselet, G.; Bankir, L.; et al: Evidence for distinct vascular and tubular urea transporters in the rat kidney. *J.Am. Soc. Nephrol.* 7: 852–860, 1996.
- [26688] 240.Sands, J. M.; Gargus, J. J.; Frohlich, O.; Gunn, R. B.; Kokko, J. P.: Urinary concentrating ability in patients with Jk(a/b) bloodtype who lack carrier-mediated urea transport. *J. Am. Soc. Nephrol.* 2:1689–1696, 1992.
- [26689] 241.Sherman, S. L.; Simpson, S. P.: Evidence for the location of JK and CO on chromosome 2 based on family studies. (Abstract) *Cytogenet.Cell Genet.* 40: 743, 1985.
- [26690] 242.Shokeir, M. H. K.; Ying, K. L.; Pabello, P.: Deletion of the long arm of chromosome no. 7: tentative assignment of the Kidd (Jk) locus. *Clin. Genet.* 4: 360–368, 1973.
- [26691] 243.Sidoux-Walter, F.; Lucien, N.; Nissinen, R.; Sistonen, P.; Henry, S.; Moulds, J.; Cartron, J.-P.; Bailly, P.: Molecular heterogeneity of the Jk-null phenotype: expression analysis of the Jk(S291P) mutation found in Finns. *Blood* 96: 1566–1573, 2000.
- [26692] 244.Tsukaguchi, H.; Shayakul, C.; Berger, U. V.; Tokui, T.; Brown, D.; Hediger, M. A.: Cloning and characterization of the urea transporter UT3: localization in rat kidney and testis. *J. Clin. Invest.* 99:1506–1515, 1997.
- [26693] 245.Xu, Y.; Olives, B.; Bailly, P.; et al: Endothelial cells of

the kidney vasa recta express the urea transporter HUT11. Kidney Int. 51:138–146, 1997.

[26694] 246. Grollman, E. F.; Kobata, A.; Ginsburg, V.: An enzymatic basis for Lewis blood types in man. J. Clin. Invest. 48: 1489–1494, 1969.

[26695] 247. Koda, Y.; Kimura, H.; Mekada, E.: Analysis of Lewis fucosyltransferase genes from the human gastric mucosa of Lewis-positive and -negative individuals. Blood 82: 2915–2919, 1993.

[26696] 248. Koprowski, H.; Blaszczyk, M.; Steplewski, Z.; Brockhaus, M.; Magnani, J.; Ginsburg, V.: Lewis blood-type may affect the incidence of gastrointestinal cancer. Lancet I: 1332–1333, 1982.

[26697] 249. Nishihara, S.; Narimatsu, H.; Iwasaki, H.; Yazawa, S.; Akamatsu, S.; Ando, T.; Seno, T.; Narimatsu, I.: Molecular genetic analysis of the human Lewis histo-blood group system. J. Biol. Chem. 269:29271–29278, 1994.

[26698] 250. Orntoft, T. F.; Vestergaard, E. M.; Holmes, E.; Jakobsen, J. S.; Grunnet, N.; Mortensen, M.; Johnson, P.; Bross, P.; Gregersen, N.; Skorstengaard, K.; Jensen, U. B.; Bolund, L.; Wolf, H.: Influence of Lewis alpha-1-3/4-L-fucosyltransferase (FUT3) gene mutations on enzyme activity, erythrocyte phenotyping, and circulating

tumor markersialyl–Lewis a levels. J. Biol. Chem. 271: 32260–32268, 1996.

- [26699] 251.Pang, H.; Liu, Y.; Koda, Y.; Soejima, M.; Jia, J.; Schlaphoff,T.; du Toit, E. D.; Kimura, H.: Five novel mis-sense mutations ofthe Lewis gene (FUT3) in African (Xhosa) and Caucasian populationsin South Africa. Hum. Genet. 102: 675–680, 1998.
- [26700] 252.Reguigne–Arnould, I.; Couillin, P.; Mollicone, R.; Faure, S.; Fletcher,A.; Kelly, R. J.; Lowe, J. B.; Oriol, R.: Relative positions of twoclusters of human alpha–L–fucosyltransferases in 19q (FUT1–FUT2) and19p (FUT6–FUT3–FUT5) within the microsatellite genetic map of chromosome19. Cytogenet. Cell Genet. 71: 158–162, 1995.
- [26701] 253.Sheinfeld, J.; Schaeffer, A. J.; Cordon–Cardo, C.; Rogatko, A.;Fair, W. R.: Association of the Lewis blood–group phenotype withrecurrent urinary tract infections in women. New Eng. J. Med. 320:773–777, 1989.
- [26702] 254.Weitkamp, L. R.; Johnston, E.; Guttormsen, S. A.: Probable geneticlinkage between the loci for the Lewis blood group and complementC3. Cytogenet. Cell Genet. 13: 183–184, 1974.
- [26703] 255.Yazawa, S.; Oh–Kawara, H.; Nakajima, T.; Hosomi, O.;

Akamatsu,S.; Kishi, K.: Histo-blood group Lewis genotyping from human hairsand blood. *Jpn. J. Hum. Genet.* 41: 177–188, 1996.

[26704] 256.Whitehouse, D. B.; Attwood, J.; Green, C.; Bruce, M.; McQuade,M.; Tippett, P.: Inheritance and linkage data for an unusual combinationof genes (at the LKE, PI and C6 loci) in a single large sibship. *Ann.Hum. Genet.* 52: 197–201, 1988.

[26705] 257.Daniels, G. L.; Le Pennec, P. Y.; Rouger, P.; Salmon, C.; Tippett,P.: The red cell antigens Au(a) and Au(b) belong to the Lutheransystem. *Vox Sang.* 60: 191–192, 1991.

[26706] 258.Campbell, I. G.; Foulkes, W. D.; Senger, G.; Trowsdale, J.; Garin-Chesa,P.; Rettig, W. J.: Molecular cloning of the B-CAM cell surface glycoproteinof epithelial cancers: a novel member of the immunoglobulin superfamily. *CancerRes.* 54: 5761–5765, 1994.

[26707] 259.Cook, P. J. L.: The Lutheran-secretor recombination fraction inman: a possible sex difference. *Ann. Hum. Genet.* 28: 393–401, 1965.

[26708] 260.Brahe, C.; Bannetta, P.; Meera Khan, P.; Arwert, F.; Serra, A.: Assignment of the catechol-O-methyltransferase gene to human chromosome22 in

somatic cell hybrids. Hum. Genet. 74: 230–234, 1986.

- [26709] 261. Brahe, C.; Bannetta, P.; Serra, A.; Arwert, F.: The increased COMT activity in Down syndrome patients is not a consequence of dosage effect owing to location of the gene on chromosome 21: further evidence. (Letter) Am. J. Med. Genet. 24: 203–204, 1986.
- [26710] 262. Masuno, M.; Fukao, T.; Song, X.-Q.; Yamaguchi, S.; Orii, T.; Kondo, N.; Imaizumi, K.; Kuroki, Y.: Assignment of the human cytosolic acetoacetyl-coenzyme A thiolase (ACAT2) gene to chromosome 6q25.3–q26. Genomics 36: 217–218, 1996.
- [26711] 263. Willison, K.; Kelly, A.; Dudley, K.; Goodfellow, P.; Spurr, N.; Groves, V.; Gorman, P.; Sheer, D.; Trowsdale, J.: The human homologue of the mouse t-complex gene, TCP1, is located on chromosome 6 but is not near the HLA region. EMBO J. 6: 1967–1974, 1987.
- [26712] 264. Engel, A. G.; Ohno, K.; Milone, M.; Wang, H.-L.; Nakano, S.; Bouzat, C.; Pruitt, J. N., II; Hutchinson, D. O.; Brengman, J. M.; Bren, N.; Sieb, J. P.; Sine, S. M.: New mutations in acetylcholine receptor subunit genes reveal heterogeneity in the slow-channel congenital myasthenic syndrome. Hum. Molec. Genet. 5: 1217–1227, 1996.
- [26713] 265. Lobos, E. A.: Five subunit genes of the human muscle

nicotinicacetylcholine receptor are mapped to two linkage groups on chromosomes 2 and 17. *Genomics* 17: 642–650, 1993.

- [26714] 266. Deftos, L. J.; Murray, S. S.; Burton, D. W.; Parmer, R. J.; O'Connor, D. T.; Deleage, A. M.; Mellon, P. L.: A cloned chromogranin A (CgA) cDNA detects a 2.3 kb mRNA in diverse neuroendocrine tissues. *Biochem. Biophys. Res. Commun.* 137: 418–423, 1986.
- [26715] 267. Granberg, D.; Stridsberg, M.; Seensalu, R.; Eriksson, B.; Lundqvist, G.; Oberg, K.; Skogseid, B.: Plasma chromogranin A in patients with multiple endocrine neoplasia type 1. *J. Clin. Endocr. Metab.* 84: 2712–2717, 1999.
- [26716] 268. Hagn, C.; Schmid, K. W.; Fischer-Colbrie, R.; Winkler, H.: Chromogranin A, B, and C in human adrenal medulla and endocrine tissues. *Lab. Invest.* 55: 405–411, 1986.
- [26717] 269. Kim, T.; Tao-Cheng, J.-H.; Eiden, L. E.; Loh, Y. P.: Chromogranin A, an 'on/off' switch controlling dense-core secretory granule biogenesis. *Cell* 106: 499–509, 2001.
- [26718] 270. Konecki, D. S.; Benedum, U. M.; Gerdes, H.-H.; Huttner, W. B.: The primary structure of human chromogranin A and pancreastatin. *J. Biol. Chem.* 262: 17026–17030, 1987.
- [26719] 271. Kruggel, W.; O'Connor, D. T.; Lewis, R. V.: The amino

terminal sequences of bovine and human chromogranin A and secretory protein I are identical. *Biochem. Biophys. Res. Commun.* 127: 380–383, 1985.

[26720] 272. Modi, W. S.; Levine, M. A.; Dean, M.; Seuanez, H.; O'Brien, S. J.: The chromogranin A gene: chromosome assignment and RFLP analysis. (Abstract) *Cytogenet. Cell Genet.* 51: 1046 only, 1989.

[26721] 273. Modi, W. S.; Levine, M. A.; Seuanez, H. N.; Dean, M.; O'Brien, S. J.: The human chromogranin A gene: chromosome assignment and RFLP analysis. *Am. J. Hum. Genet.* 45: 814–818, 1989.

[26722] 274. Murray, S. S.; Deaven, L. L.; Burton, D. W.; O'Connor, D. T.; Mellon, P. L.; Deftos, L. J.: The gene for human chromogranin A (CgA) is located on chromosome 14. *Biochem. Biophys. Res. Commun.* 142: 141–146, 1987.

[26723] 275. Nobels, F. R. E.; Kwekkeboom, D. J.; Coopmans, W.; Schoenmakers, C. H. H.; Lindemans, J.; De Herder, W. W.; Krenning, E. P.; Bouillon, R.; Lamberts, S. W. J.: Chromogranin A as serum marker for neuroendocrine neoplasia: comparison with neuron-specific enolase and the alpha-subunit of glycoprotein hormones. *J. Clin. Endocr. Metab.* 82: 2622–2628, 1997.

[26724] 276. O'Connor, D. T.; Deftos, L. J.: Secretion of chromo-

granin A bypeptide-producing endocrine neoplasms. New Eng. J. Med. 314: 1145–1151,1986.

[26725] 277.Simon–Chazottes, D.; Wu, H.; Parmer, R. J.; Rozansky, D. J.; Szpirer,J.; Levan, G.; Kurtz, T. W.; Szpirer, C.; Guenet, J. L.; O'Connor,D. T.: Assignment of the chromogranin A (Chga) locus to homologousregions on mouse chromo–some 12 and rat chromosome 6. Genomics 17:252–255, 1993.

[26726] 278.Wu, H.–J.; Rozansky, D. J.; Parmer, R. J.; Gill, B. M.; O'Connor,D. T.: Structure and function of the chromo–granin A gene: clues toevolution and tissue–specific ex–pression. J. Biol. Chem. 266: 13130–13134,1991.

[26727] 279.Higashiyama, S.; Lau, K.; Besner, G. E.; Abraham, J. A.; Klagsbrun,M.: Structure of heparin–binding EGF–like growth factor: multipleforms, primary structure, and gly–cosylation of the mature protein. J.Biol. Chem. 267: 6205–6212, 1992.

[26728] 280.Naglich, J. G.; Metherall, J. E.; Russell, D. W.; Eidels, L.:Expression cloning of a diphtheria toxin receptor: iden–tity with aheparin–binding EGF–like growth factor precur–sor. Cell 69: 1051–1061,1992.

[26729] 281.Pappenheimer, A. M., Jr.: Diphtheria toxin. Ann. Rev. Biochem. 46:69–94, 1977.

- [26730] 282.Pappenheimer, A. M., Jr.; Gill, D. M.: Diphtheria. *Science* 182:353–358, 1973.
- [26731] 283.Pathak, B. G.; Gilbert, D. J.; Harrison, C. A.; Luetke, N. C.; Chen, X.; Klagsbrun, M.; Plowman, G. D.; Copeland, N. G.; Jenkins, N. A.; Lee, D. C.: Mouse chromosomal location of three EGF receptorligands: amphiregulin (Areg), betacellulin (Btc), and heparin-bindingEGF (Hegfl). *Genomics* 28: 116–118, 1995.
- [26732] 284.Roberts, M.; Ruddle, F. H.: The Chinese hamster gene map: assignment of four genes (DTS, PGM2, 6PGD, Eno1) to chromosome 2. *Exp. Cell Res.* 127: 47–54, 1980.
- [26733] 285.Chabas, D.; Baranzini, S. E.; Mitchell, D.; Bernard, C. C. A.; Rittling, S. R.; Denhardt, D. T.; Sobel, R. A.; Lock, C.; Karpus, M.; Pedotti, R.; Heller, R.; Oksenberg, J. R.; Steinman, L.: The influence of the proinflammatory cytokine, osteopontin, on autoimmune demyelinating disease. *Science* 294: 1731–1735, 2001.
- [26734] 286.Bowcock, A. M.; Hebert, J. M.; Christiano, A. M.; Wijsman, E.; Cavalli-Sforza, L. L.; Boyd, C. D.: The pro alpha 1 (IV) collagen gene is linked to the D13S3 locus at the distal end of human chromosome 13q. *Cytogenet. Cell Genet.* 45: 234–236, 1987.
- [26735] 287.Bowcock, A. M.; Hebert, J. M.; Wijsman, E.; Gadi, I.;

Cavalli-Sforza, L. L.; Boyd, C. D.: High recombination between two physically close human basement membrane collagen genes at the distal end of chromosome 13q. *Proc. Nat. Acad. Sci.* 85: 2701–2705, 1988.

[26736] 288. Boyd, C. D.; Weliky, K.; Toth-Fejel, S.; Deak, S. B.; Christiano, A. M.; Mackenzie, J. W.; Sandell, L. J.; Tryggvason, K.; Magenis, E.: The single copy gene coding for human alpha-1(IV) procollagen is located at the terminal end of the long arm of chromosome 13. *Hum. Genet.* 74: 121–125, 1986.

[26737] 289. Brinker, J. M.; Gudas, L. J.; Loidl, H. R.; Wang, S.-Y.; Rosenbloom, J.; Kefalides, N. A.; Myers, J. C.: Restricted homology between human alpha-1 type IV and other procollagen chains. *Proc. Nat. Acad. Sci.* 82: 3649–3653, 1985.

[26738] 290. Burbelo, P. D.; Martin, G. R.; Yamada, Y.: Alpha-1(IV) and alpha-2(IV) collagen genes are regulated by a bidirectional promoter and a shared enhancer. *Proc. Nat. Acad. Sci.* 85: 9679–9682, 1988.

[26739] 291. Crouch, E.; Sage, H.; Bornstein, P.: Structural basis for apparent heterogeneity of collagens in human basement membranes: type IV procollagen contains two distinct chains. *Proc. Nat. Acad. Sci.* 77: 745–749, 1980.

- [26740] 292.Cutting, G. R.; Kazazian, H. H., Jr.; Antonarakis, S. E.; Killen,P. D.; Yamada, Y.; Francomano, C. A.: Macrorestriction analysis mapsCOL4A1 and COL4A2 collagen genes within a 400 kb region on chromosome13q34. (Abstract) Am. J. Hum. Genet. 41: A163, 1987.
- [26741] 293.Cutting, G. R.; Kazazian, H. H., Jr.; Antonarakis, S. E.; Killen,P. D.; Yamada, Y.; Francomano, C. A.: Macrorestriction mapping ofCOL4A1 and COL4A2 collagen genes on human chromosome 13q34. Genomics 3:256–263, 1988.
- [26742] 294.Emanuel, B. S.; Sellinger, B. T.; Gudas, L. J.; Myers, J. C.:Localization of the human procollagen alpha–1(IV) gene to chromosome13q34 by in situ hybridization. Am. J. Hum. Genet. 38: 38–44, 1986.
- [26743] 295.Hebert, J. M.; Bowcock, A. M.; Wijsman, E.; Gadi, I.; Boyd, C.;Cavalli–Sforza, L. L.: The genes for pro–alpha–1 (IV) collagen, pro–alpha–2(IV) collagen and the D13S3 locus are linked at 13q34. (Abstract) Am.J. Hum. Genet. 41: A169, 1987.
- [26744] 296.Kuhn, K.: Personal Communication. Munich, Germany 1/7/1982.
- [26745] 297.Mayne, R.; Wiedemann, H.; Irwin, M. H.; Sanderson, R. D.; Fitch,J. M.; Linsenmayer, T. F.; Kuhn, K.: Monoclonal antibodies againstchicken type IV and V collagens: elec–

tron microscopic mapping of the epitopes after rotary shadowing. *J. Cell Biol.* 98: 1637–1644, 1984.

- [26746] 298. Pihlajaniemi, T.; Tryggvason, K.; Myers, J. C.; Kurkinen, M.; Lebo, R.; Cheung, M.-C.; Prockop, D. J.; Boyd, C. D.: cDNA clones coding for the pro- α -1(IV) chain of human type IV procollagen reveal an unusual homology of amino acid sequences in two halves of the carboxyl terminal domain. *J. Biol. Chem.* 260: 7681–7687, 1985.
- [26747] 299. Poschl, E.; Pollner, R.; Kuhn, K.: The genes for the α -1(IV) and α -2(IV) chains of human basement membrane collagen type IV are arranged head-to-head and separated by a bidirectional promoter of unique structure. *EMBO J.* 7: 2687–2695, 1988.
- [26748] 300. Soininen, R.; Chow, L.; Kurkinen, M.; Tryggvason, K.; Prockop, D. J.: The gene for the α -1(IV) chain of human type IV procollagen: the exon structures do not coincide with the two structural subdomains in the globular carboxy-terminus of the protein. *EMBO J.* 5: 2821–2823, 1986.
- [26749] 301. Soininen, R.; Huotari, M.; Hostikka, S. L.; Prockop, D. J.; Tryggvason, K.: The structural genes for α -1 and α -2 chains of human type IV collagen are divergently encoded on opposite DNA strands and have an overlapping

promoter region. J. Biol. Chem. 263: 17217–17220,1988.

- [26750] 302.Soininen, R.; Tikka, L.; Chow, L.; Pihlajaniemi, T.; Kurkinen,M.; Prockop, D. J.; Boyd, C. D.; Tryggvason, K.: Large introns inthe 3–prime end of the gene for the pro–alpha1(IV) chain of humanbasement membrane collagen. Proc. Nat. Acad. Sci. 83: 1568–1572,1986.
- [26751] 303.Keegan, K.; Johnson, D. E.; Williams, L. T.; Hayman, M. J.: Isolationof an additional member of the fibroblast growth factor receptor family,FGFR–3. Proc. Nat. Acad. Sci. 88: 1095–1099, 1991.
- [26752] 304.Sorrentino, R.; Corte, G.; Calabi, F.; Tanigaki, N.; Tosi, R.: Microfingerprinting analysis of human Ia molecules favours a threeloci model. Molec. Immun. 20: 333–343, 1983.
- [26753] 305.Jutel, M.; Watanabe, T.; Klunker, S.; Akdis, M.; Thomet, O. A.R.; Malolepszy, J.; Zak–Nejmark, T.; Koga, R.; Kobayashi, T.; Blaser,K.; Akdis, C. A.: Histamine regulates T–cell and antibody responsesby differential expression of H1 and H2 receptors. Nature 413: 420–425,2001.
- [26754] 306.Brown, S. D. M.; Chartier, F.; Johnson, K.; Cavanna, J. S.: Mappingthe Hrc gene to proximal mouse chromosome 7: delineation of a conservedlinkage group with human 19q. Genomics 18: 459–461, 1993.

- [26755] 307. Hofmann, S. L.; Brown, M. S.; Lee, E.; Pathak, R. K.; Anderson, R. G. W.; Goldstein, J. L.: Purification of a sarcoplasmic reticulum protein that binds Ca^{2+} and plasma lipoproteins. *J. Biol. Chem.* 264:8260–8270, 1989.
- [26756] 308. Hofmann, S. L.; Topham, M.; Hsieh, C.-L.; Francke, U.: cDNA and genomic cloning of HRC, a human sarcoplasmic reticulum protein, and localization of the gene to human chromosome 19 and mouse chromosome 7. *Genomics* 9: 656–669, 1991.
- [26757] 309. Doenecke, D.; Tonjes, R.: Differential distribution of lysine and arginine residues in the closely related histones H1 and H5. Analysis of a human H1 gene. *J. Molec. Biol.* 187: 461–464, 1986.
- [26758] 310. Albig, W.; Doenecke, D.: The human histone gene cluster at the D6S105 locus. *Hum. Genet.* 101: 284–294, 1997.
- [26759] 311. Nemergut, M. E.; Mizzen, C. A.; Stukenberg, T.; Allis, C. D.; Macara, I. G.: Chromatin docking and exchange activity enhancement of RCC1 by histones H2A and H2B. *Science* 292: 1540–1543, 2001.
- [26760] 312. Castellani, L. W.; Weinreb, A.; Bodnar, J.; Goto, A. M.; Doolittle, M.; Mehrabian, M.; Demant, P.; Lusk, A. J.: Mapping a gene for combined hyperlipidaemia in a mutant

mouse strain. *Nature Genet.* 18: 374–377,1998.

- [26761] 313. Geurts, J. M. W.; Janssen, R. G. J. H.; van Greevenbroek, M. M. J.; van der Kallen, C. J. H.; Cantor, R. M.; Bu, X.; Aouizerat, B. E.; Allayee, H.; Rotter, J. I.; de Bruin, T. W. A.: Identification of TNFRSF1B as a novel modifier gene in familial combined hyperlipidemia. *Hum. Molec. Genet.* 9: 2067–2074, 2000.
- [26762] 314. Suzuki, Y.; Wong, S.-Y.; Grumet, F. C.; Fessel, J.; Montoya, J. G.; Zolopa, A. R.; Portmore, A.; Schumacher-Perdreau, F.; Schrappe, M.; Koppen, S.; Ruf, B.; Brown, B. W.; Remington, J. S.: Evidence for genetic regulation of susceptibility to toxoplasmic encephalitis in AIDS patients. *J. Infect. Dis.* 173: 265–268, 1996.
- [26763] 315. Tanigaki, N.; Tosi, R.; Pressman, D.; Ferrara, G. B.: Molecular identification of human Ia antigens coded for by a gene closely linked to HLA-DR locus. *Immunogenetics* 10: 151–167, 1980.
- [26764] 316. Todd, J. A.; Bell, J. I.; McDevitt, H. O.: HLA-DQ(beta) gene contributes to susceptibility and resistance to insulin-dependent diabetes mellitus. *Nature* 329: 599–604, 1987.
- [26765] 317. Todd, J. A.; Fukui, Y.; Kitagawa, T.; Sasazuki, T.: The A3 allele of the HLA-DQA1 locus is associated with sus-

ceptibility to type 1 diabetes in Japanese. *Proc. Nat. Acad. Sci.* 87: 1094–1098, 1990.

[26766] 318. Tosi, R.; Tanigaki, N.; Cantis, D.; Ferrara, G. B.; Pressman, D.: Immunological dissection of human Ia molecules. *J. Exp. Med.* 148:1592–1611, 1978.

[26767] 319. Wen, L.; Wong, F. S.; Tang, J.; Chen, N.-Y.; Altieri, M.; David, C.; Flavell, R.; Sherwin, R.: In vivo evidence for the contribution of human histocompatibility leukocyte antigen (HLA)-DQ molecules to the development of diabetes. *J. Exp. Med.* 191: 97–104, 2000.

[26768] 320. Hatzivassiliou, G.; Miller, I.; Takizawa, J.; Palanisamy, N.; Rao, P. H.; Iida, S.; Tagawa, S.; Taniwaki, M.; Russo, J.; Neri, A.; Cattoretti, G.; Clynes, R.; Mendelsohn, C.; Chaganti, R. S. K.; Dalla-Favera, R.: IRTA1 and IRTA2, novel immunoglobulin superfamily receptors expressed in B cells and involved in chromosome 1q21 abnormalities in B cell-malignancy. *Immunity* 14: 277–289, 2001.

[26769] 321. Mollereau, C.; Muscatelli, F.; Mattei, M.-G.; Vassart, G.; Parmentier, M.: The high-affinity interleukin 8 receptor gene (IL8RA) maps to the 2q33–q36 region of the human genome: cloning of a pseudogene (IL8RBP) for the low-affinity receptor. *Genomics* 16: 248–251, 1993.

[26770] 322. Morris, S. W.; Nelson, N.; Valentine, M. B.; Shapiro, D.

N.; Look, A. T.; Kozlosky, C. J.; Beckmann, M. P.; Cerretti, D. P.: Assignment of the genes encoding human interleukin-8 receptor types 1 and 2 and an interleukin-8 receptor pseudogene to chromosome 2q35. *Genomics* 14:685-691, 1992.

[26771] 323. Palter, S. F.; Mulayim, N.; Senturk, L.; Arici, A.: Interleukin-8 in the human fallopian tube. *J. Clin. Endocr. Metab.* 86: 2660-2667, 2001.

[26772] 324. Tsai, H.-H.; Frost, E.; To, V.; Robinson, S.; French-Constant, C.; Geertman, R.; Ransohoff, R. M.; Miller, R. H.: The chemokine receptor CXCR2 controls positioning of oligodendrocyte precursors in developing spinal cord by arresting their migration. *Cell* 110:373-383, 2002.

[26773] 325. Bass, H. N.; Sparkes, R. S.; Crandall, B. F.; Marcy, S. M.: Congenital contractural arachnodactyly, keratoconus, and probable Marfan syndrome in the same pedigree. *J. Pediatr.* 98: 591-593, 1981.

[26774] 326. Bawle, E.; Quigg, M. H.: Ectopia lentis and aortic root dilatation in congenital contractural arachnodactyly. *Am. J. Med. Genet.* 42:19-21, 1992.

[26775] 327. Beals, R. K.; Hecht, F.: Congenital contractural arachnodactyly: a heritable disorder of connective tissue. *J. Bone Joint Surg.* 53A:987-993, 1971.

- [26776] 328.Belleh, S.; Zhou, G.; Wang, M.; Der Kaloustian, V. M.; Pagon, R.A.; Godfrey, M.: Two novel fibrillin-2 mutations in congenital contractural arachnodactyly. *Am. J. Med. Genet.* 92: 7-12, 2000.
- [26777] 329.Beyer, P.; Klein, M. L.; Iszepy, E.: Maladie de Marfan avec raideurs articulaires importantes atteignant les quatre enfants de la même fratrie et leur mère. *Arch. Franc. Pédiat.* 22: 210-216, 1965.
- [26778] 330.Bistritzer, T.; Fried, K.; Lahat, E.; Dvir, M.; Goldberg, M.: Congenital contractural arachnodactyly in two double second cousins: possible homozygosity. *Clin. Genet.* 44: 15-19, 1993.
- [26779] 331.Chaudhry, S. S.; Gazzard, J.; Baldock, C.; Dixon, J.; Rock, M.J.; Skinner, G. C.; Steel, K. P.; Kielty, C. M.; Dixon, M. J.: Mutation of the gene encoding fibrillin-2 results in syndactyly in mice. *Hum. Molec. Genet.* 10: 835-843, 2001.
- [26780] 332.Cole, T. R. P.; Hughes, H. E.: Congenital contractural arachnodactyly with unilateral lower limb deficiency. *Am. J. Med. Genet.* 44: 72-74, 1992.
- [26781] 333.Currarino, G.; Friedman, J. M.: A severe form of congenital contractural arachnodactyly in two newborn infants. *Am. J. Med. Genet.* 25: 763-773, 1986.

- [26782] 334.Delemarre–van de Waal, H. A.; van Benthem, L. H. B. M.; Bleeker–Wagemakers,E. M.: Congenitale contracturele arachnodactylie. Ned. Tijdschr.Geneeskd. 124: 348–351, 1980.
- [26783] 335.Dixon, M. J.; Gazzard, J.; Chaudhry, S. S.; Sampson, N.; Schulte,B. A.; Steel, K. P.: Mutation of the Na–K–Cl co–transporter geneSlc12a2 results in deafness in mice. Hum. Molec. Genet. 8: 1579–1584,1999.
- [26784] 336.Epstein, C. J.; Graham, C. B.; Hodgkin, W. E.; Hecht, F.; Motulsky,A. G.: Hereditary dysplasia of bone with kyphoscoliosis, contractures,and abnormally shaped ears. J. Pediat. 73: 379–386, 1968.
- [26785] 337.Gruber, M. A.; Graham, T. P., Jr.; Engel, E.; Smith, C.: Marfansyndrome with contractural arachnodactyly and severe mitral regurgitationin a premature infant. J. Pediat. 93: 80–82, 1978.
- [26786] 338.Hecht, F.; Beals, R. K.: 'New' syndrome of congenital contracturalarachnodactyly originally described by Marfan in 1896. Pediatrics 49:574–579, 1972.
- [26787] 339.Huggon, I. C.; Burke, J. P.; Talbot, J. F.: Contractural arachnodactylywith mitral regurgitation and iridodonesis. Arch. Dis. Child. 65:317–319, 1990.
- [26788] 340.Godfrey, M.; Raghunath, M.; Cisler, J.; Bevins, C. L.;

DePaepe,A.; Di Rocco, M.; Gregoritch, J.; Imaizumi, K.; Kaplan, P.; Kuroki,Y.; Silberbach, M.; Superti-Furga, A.; Van Thienen, M.-N.; Vetter,U.; Steinmann, B: Abnormal morphology of fibrillin microfibrils infibroblast cultures from patients with neonatal Marfan syndrome. *Am.J. Path.* 146: 1414–1421, 1995.

[26789] 341.Gupta, P. A.; Putnam, E. A.; Carmical, S. G.; Kaitila, I.; Steinmann,B.; Child, A.; Danesino, C.; Metcalfe, K.; Berry, S. A.; Chen, E.;Delorme, C. V.; Thong, M.-K.; Ades, L. C.; Milewicz, D. M.: Ten novelFBN2 mutations in congenital contractural arachnodactyly: delineationof the molecular pathogenesis and clinical phenotype. *Hum. Mutat.* 19:39–48, 2002.

[26790] 342.Kainulainen, K.; Karttunen, L.; Puhakka, L.; Sakai, L.; Peltonen,L.: Mutations in the fibrillin gene responsible for dominant ectopialentis and neonatal Marfan syndrome. *Nature Genet.* 6: 64–69, 1994.

[26791] 343.Kingsley–Pillers, E. M.: Arachnodactyly with amyoplasia congenita. *Proc.Roy. Soc. Med.* 39: 696–697, 1946.

[26792] 344.Langenskiold, A.: Congenital contractural arachnodactyly: reportof a case and of an operation for knee contracture. *J. Bone JointSurg.* 67: 44–46, 1985.

[26793] 345.Lee, B.; Godfrey, M.; Vitale, E.; Hori, H.; Mattei, M.-G.;

Sarfarazi,M.; Tsipouras, P.; Ramirez, F.; Hollister, D. W.:
Linkage of Marfansyndrome and a phenotypically related
disorder to two different fibrillingenes. Nature 352:
330–334, 1991.

[26794] 346.Li, X.; Pereira, L.; Zhang, H.; Sanguineti, C.; Ramirez,
F.; Bonadio,J.; Francke, U.: Fibrillin genes map to regions
of conserved mouse/humansynten on mouse chromo-
somes 2 and 18. Genomics 18: 667–672, 1993.

[26795] 347.Lipson, E. H.; Viseskul, C.; Herrmann, J.: The clinical
spectrumof congenital contractural arachnodactyly: a case
with congenitalheart disease. Z. Kinderheilk. 118: 1–8,
1974.

[26796] 348.Lowry, R. B.; Guichon, V. C.: Congenital contractural
arachnodactyly:a syndrome simulating Marfan's syndrome.
Canad. Med. Assoc. J. 107:531–533, 1972.

[26797] 349.Marfan, M. A. B.: Un cas de deformation congenitale
des quatremembres plus prononcee aux extremités, car-
acterisee par l'allongementdes os avec un certain degre
d'amincissement. Bull. Mem. Soc. Med.Hop. Paris 13:
220–226, 1896.

[26798] 350.Maslen, C.; Babcock, D.; Raghunath, M.; Steinmann,
B.: A rarebranch–point mutation is associated with miss-
plicing of fibrillin–2in a large family with congenital con–

tractural arachnodactyly. Am.J. Hum. Genet. 60:
1389–1398, 1997.

[26799] 351.Mirise, R. T.; Shear, S.: Congenital contractual arachnodactyly:description of a new kindred. Arthritis Rheum. 22: 542–546, 1979.

[26800] 352.Park, E.–S.; Putnam, E. A.; Chitayat, D.; Child, A.; Milewicz,D. M.: Clustering of FBN2 mutations in patients with congenital contractural arachnodactyly indicates an important role of the domains encoded by exons 24 through 34 during human development. Am. J. Med. Genet. 78:350–355, 1998.

[26801] 353.Putnam, E. A.; Milewicz, D. M.: A mutation in the FBN2 gene in dermal fibroblasts from a congenital contractural arachnodactyly patient.(Abstract) Am. J. Hum. Genet. 57: A225, 1995.

[26802] 354.Putnam, E. A.; Park, E.–S.; Aalfs, C. M.; Hennekam, R. C. M.;Milewicz, D. M.: Parental somatic and germ–line mosaicism for a FBN2 mutation and analysis of FBN2 transcript levels in dermal fibroblasts. Am.J. Hum. Genet. 60: 818–827, 1997.

[26803] 355.Putnam, E. A.; Zhang, H.; Ramirez, F.; Milewicz, D. M.: Fibrillin–2(FBN2) mutations result in the Marfan–like disorder, congenital contractural arachnodactyly. Nature

Genet. 11: 456–458, 1995.

[26804] 356.Pyeritz, R. E.: Personal Communication. Baltimore, Md. 4/28/1986.

[26805] 357.Ramos Arroyo, M. A.; Weaver, D. D.; Beals, R. K.: Congenitalcontractural arachnodactyly: report of four additional families andreview of literature. Clin. Genet. 27: 570–581, 1985.

[26806] 358.Krege, J. H.; John, S. W. M.; Langenbach, L. L.; Hodgins, J. B.;Hagaman, J. R.; Bachman, E. S.; Jennette, J. C.; O'Brien, D. A.; Smithies,O.: Male–female differences in fertility and blood pressure in ACE–deficientmice. Nature 375: 146–148, 1995.

[26807] 359.Dedhar, S.; Rennie, P. S.; Shago, M.; Hagesteijn, C.–Y. L.; Yang,H.; Filmus, J.; Hawley, R. G.; Bruchovsky, N.; Cheng, H.; Matusik,R. J.; Giguere, V.: Inhibition of nuclear hormone receptor activityby calreticulin. Nature 367: 480–483, 1994.

[26808] 360.Rose, P. M.; Fernandes, P.; Lynch, J. S.; Frazier, S. T.; Fisher,S. M.; Kodukula, K.; Kienzle, B.; Seethala, R.: Cloning and functionalexpression of a cDNA encoding a human type 2 neuropeptide Y receptor. J.Biol. Chem. 270: 22661–22664, 1995.

[26809] 361.Hagman, J.; Belanger, C.; Travis, A.; Turck, C. W.;

Grosschedl, R.: Cloning and functional characterization of early B-cell factor, a regulator of lymphocyte-specific gene expression. *Genes Dev.* 7:760–773, 1993.

[26810] 362. Milatovich, A.; Qiu, R.-G.; Grosschedl, R.; Francke, U.: Gene for a tissue-specific transcriptional activator (EBF or Olf-1), expressed in early B lymphocytes, adipocytes, and olfactory neurons, is located on human chromosome 5, band q34, and proximal mouse chromosome 11. *Mammalian Genome* 5: 211–215, 1994.

[26811] 363. Travis, A.; Hagman, J.; Hwang, L.; Grosschedl, R.: Purification of early-B-cell factor and characterization of its DNA-binding specificity. *Molec. Cell. Biol.* 13: 3392–3400, 1993.

[26812] 364. Wang, M. M.; Reed, R. R.: Molecular cloning of the olfactory neuronal transcription factor Olf-1 by genetic selection in yeast. *Nature* 364:121–126, 1993.

[26813] 365. Wang, K. C.; Koprivica, V.; Kim, J. A.; Sivasankaran, R.; Guo, Y.; Neve, R. L.; He, Z.: Oligodendrocyte-myelin glycoprotein is a Nogo receptor ligand that inhibits neurite outgrowth. *Nature* 417:941–944, 2002.

[26814] 366. Hovnanian, A.; Rebouillat, D.; Mattei, M.-G.; Levy, E. R.; Marie, I.; Monaco, A. P.; Hovanesian, A. G.: The human 2'-5'-oligoadenylate synthetase locus is com-

posed of three distinct genes clustered on chromosome 12q24.2 encoding the 100-, 69-, and 40-kDa forms. *Genomics* 52:267–277, 1998.

[26815] 367. Frank, M. B.: Personal Communication. Oklahoma City, Okla. 6/3/1994.

[26816] 368. Itoh, K.; Itoh, Y.; Frank, M. B.: Protein heterogeneity in the human Ro/SSA ribonucleoproteins: the 52- and 60-kD Ro/SSA autoantigens are encoded by separate genes. *J. Clin. Invest.* 87: 177–186, 1991.

[26817] 369. McCauliffe, D. P.; Lux, F. A.; Lieu, T.-S.; Sanz, I.; Hanke, J.; Newkirk, M. M.; Bachinski, L. L.; Itoh, Y.; Siciliano, M. J.; Reichlin, M.; Sontheimer, R. D.; Capra, J. D.: Molecular cloning, expression, and chromosome 19 localization of a human Ro/SS-A autoantigen. *J. Clin. Invest.* 85: 1379–1391, 1990.

[26818] 370. McCauliffe, D. P.; Zappi, E.; Lieu, T.-S.; Michalak, M.; Sontheimer, R. D.; Capra, J. D.: A human Ro/SS-A autoantigen is the homologue of calreticulin and is highly homologous with onchocercal RAL-1 antigen and an aplysia 'memory molecule.'. *J. Clin. Invest.* 86: 332–335, 1990.

[26819] 371. Orth, T.; Dorner, T.; Meyer Zum Buschenfelde, K.-H.; Mayet, W.-J.: Complete congenital heart block is associ-

ated with increased autoantibody titers against calreticulin.
Europ. J. Clin. Invest. 26: 205–215, 1996.

- [26820] 372. Rooke, K.; Briquet–Laugier, V.; Xia, Y.–R.; Lysis, A. J.; Doolittle, M. H.: Mapping of the gene for calreticulin (Calr) to mouse chromosome 8. Mammalian Genome 8: 870–871, 1997.
- [26821] 373. Frank, M. B.; Itoh, K.; Fujisaku, A.; Pontarotti, P.; Mattei, M.–G.; Neas, B. R.: The mapping of the human 52–kD Ro/SSA autoantigen gene to human chromosome 11, and its polymorphisms. Am. J. Hum. Genet. 52: 183–191, 1993.
- [26822] 374. Frank, M. B.; Mattei, M.–G.: Mapping of the human 60000 M(r) Ro/SSA locus: the genes for three Ro/SSA autoantigens are located on separate chromosomes. Immunogenetics 39: 428–431, 1994.
- [26823] 375. Schoenlebe, J.; Buyon, J. P.; Zitelli, B. J.; Friedman, D.; Greco, M. A.; Knisely, A. S.: Neonatal hemochromatosis associated with maternal autoantibodies against Ro/SS–A and La/SS–B ribonucleoproteins. Am. J. Dis. Child. 147: 1072–1075, 1993.
- [26824] 376. Maclaren, N. K.; Riley, W. J.: Inherited susceptibility to autoimmune Addison's disease is linked to human leukocyte antigens–DR3 and/or DR4, except when associated

with type I autoimmune polyglandular syndrome. *J.Clin. Endocr. Metab.* 62: 455–459, 1986.

- [26825] 377.Wang, Y. A.; Elson, A.; Leder, P.: Loss of p21 increases sensitivity to ionizing radiation and delays the onset of lymphoma in atm-deficient mice. *Proc. Nat. Acad. Sci.* 94: 14590–14595, 1997.
- [26826] 378.Zakut, R.; Givol, D.: The tumor suppression function of p21(Waf) is contained in its N-terminal half ('half-WAF'). *Oncogene* 11: 393–395, 1995.
- [26827] 379.Bourne, Y.; Watson, M. H.; Hickey, M. J.; Holmes, W.; Rocque, W.; Reed, S. I.; Turner, J. A.: Crystal structure and mutational analysis of the human CDK2 kinase complex with cell cycle-regulatory protein CksHs1. *Cell* 84: 863–874, 1996.
- [26828] 380.Boden, P.; Hall, M. D.; Hughes, J.: Cholecystokinin receptors. *Cell.Molec. Neurobiol.* 15: 545–559, 1995.
- [26829] 381.de Weerth, A.; Pisegna, J. R.; Huppi, K.; Wank, S. A.: Molecular cloning, functional expression and chromosomal localization of the human cholecystokinin type A receptor. *Biochem. Biophys. Res. Commun.* 194:811–818, 1993.
- [26830] 382.Funakoshi, A.; Miyasaka, K.; Shinozaki, H.; Masuda, M.; Kawanami, T.; Takata, Y.; Kono, A.: An animal model of congenital defect of gene expression of cholecystokinin

(CCK)–A receptor. *Biochem. Biophys. Res. Commun.* 210: 787–796, 1995.

- [26831] 383. Hamann, A.; Busing, B.; Munzberg, H.; de Weerth, A.; Hinney, A.; Mayer, H.; Siegfried, W.; Hebebrand, J.; Greten, H.: Missense variants in the human cholecystokinin type A receptor gene: no evidence for association with early-onset obesity. *Horm. Metab. Res.* 31: 287–288, 1999.
- [26832] 384. Huppi, K.; Siwarski, D.; Pisegna, J. R.; Wank, S.: Chromosomal localization of the gastric and brain receptors for cholecystokinin (CCKAR and CCKBR) in human and mouse. *Genomics* 25: 727–729, 1995.
- [26833] 385. Inoue, H.; Iannotti, C. A.; Welling, C. M.; Veile, R.; Donis-Keller, H.; Permutt, M. A.: Human cholecystokinin type A receptor gene: cytogenetic localization, physical mapping, and identification of two missense variants in patients with obesity and non-insulin-dependent diabetes mellitus (NIDDM). *Genomics* 42: 331–335, 1997.
- [26834] 386. Marchal-Victorion, S.; Vionnet, N.; Escrieut, C.; Dematos, F.; Dina, C.; Dufresne, M.; Vaysse, N.; Pradayrol, L.; Froguel, P.; Fourmy, D.: Genetic, pharmacological and functional analysis of cholecystokinin-1 and cholecystokinin-2 receptor polymorphism in type 2 diabetes and obese patients. *Pharmacogenetics* 12: 23–30, 2002.

- [26835] 387. Miller, L. J.; Holicky, E. L.; Ulrich, C. D.; Wieben, E. D.: Abnormal processing of the human cholecystokinin receptor gene in association with gallstones and obesity. *Gastroenterology* 109: 1375–1380, 1995.
- [26836] 388. Samuelson, L. C.; Isakoff, M. S.; Lacourse, K. A.: Localization of the murine cholecystokinin A and B receptor genes. *Mammalian Genome* 6:242–246, 1995.
- [26837] 389. Ulrich, C. D.; Ferber, I.; Holicky, E.; Hadac, E.; Buell, G.; Miller, L. J.: Molecular cloning and functional expression of the human gallbladder cholecystokinin A receptor. *Biochem. Biophys. Res. Commun.* 193: 204–211, 1993.
- [26838] 390. Butkowski, R. J.; Langeveld, J. P. M.; Wieslander, J.; Hamilton, J.; Hudson, B. G.: Localization of the Goodpasture epitope to a novel chain of basement membrane collagen. *J. Biol. Chem.* 262: 7874–7877, 1987.
- [26839] 391. Bora, N. S.; Lublin, D. M.; Kumar, B. V.; Hockett, R. D.; Holers, V. M.; Atkinson, J. P.: Structural gene for human membrane cofactor protein (MCP) of complement maps to within 100 kb of the 3-prime end of the C3b/C4b receptor gene. *J. Exp. Med.* 169: 597–602, 1989.
- [26840] 392. Cui, W.; Hourcade, D.; Post, T.; Greenlund, A. C.; Atkinson, J. P.; Kumar, V.: Characterization of the promoter region of the membrane cofactor protein (CD46) gene of

the human complement system and comparison to a membrane cofactor protein-like genetic element. *J. Immun.* 151:4137–4146, 1993.

[26841] 393. Dorig, R. E.; Marcil, A.; Chopra, A.; Richardson, C. D.: The human CD46 molecule is a receptor for measles virus (Edmonston strain). *Cell* 75:295–305, 1993.

[26842] 394. Kallstrom, H.; Gill, D. B.; Albiger, B.; Liszewski, M. K.; Atkinson, J. P.; Jonsson, A.-B.: Attachment of *Neisseria gonorrhoeae* to the cellular pilus receptor CD46: identification of domains important for bacterial adherence. *Cell. Microbiol.* 3: 133–143, 2001.

[26843] 395. Lublin, D. M.; Liszewski, M. K.; Post, T. W.; Arce, M. A.; LeBeau, M. M.; Rebentisch, M. B.; Lemons, R. S.; Seya, T.; Atkinson, J. P.: Molecular cloning and chromosomal localization of human complement membrane cofactor protein (MCP): evidence for inclusion in the multigene family of complement-regulatory proteins. *J. Exp. Med.* 168: 181–194, 1988.

[26844] 396. Marie, J. C.; Astier, A. L.; Rivaller, P.; Rabourdin-Combe, C.; Wild, T. F.; Horvat, B.: Linking innate and acquired immunity: divergent role of CD46 cytoplasmic domains in T cell-induced inflammation. *Nature Immun.* 3: 659–666, 2002.

- [26845] 397. Post, T. W.; Liszewski, M. K.; Adams, E. M.; Tedja, I.; Miller, E. A.; Atkinson, J. P.: Membrane cofactor protein of the complement system: alternative splicing of serine/threonine/proline-rich exons and cytoplasmic tails produces multiple isoforms that correlate with protein phenotype. *J. Exp. Med.* 174: 93–102, 1991.
- [26846] 398. Purcell, D. F. J.; Johnstone, R. W.; McKenzie, I. F. C.: Identification of four different CD46 (MCP) molecules with anti-peptide antibodies. *Biochem. Biophys. Res. Commun.* 180: 1091–1097, 1991.
- [26847] 399. Santoro, F.; Kennedy, P. E.; Locatelli, G.; Malnati, M. S.; Berger, E. A.; Lusso, P.: CD46 is a cellular receptor for human herpesvirus 6. *Cell* 99: 817–827, 1999.
- [26848] 400. Tatsuo, H.; Ono, N.; Tanaka, K.; Yanagi, Y.: SLAM (CDw150) is a cellular receptor for measles virus. *Nature* 406: 893–897, 2000.
- [26849] 401. Chan, P.; Simon-Chazottes, D.; Mattei, M. G.; Guenet, J. L.; Salier, J. P.: Comparative mapping of lipocalin genes in human and mouse: the four genes for complement C8 gamma chain, prostaglandin-D-synthase, oncogene-24P3, and progesterone-associated endometrial protein map to HSA9 and MMU2. *Genomics* 23: 145–150, 1994.
- [26850] 402. King, I. A.; Arnemann, J.; Spurr, N. K.; Buxton, R. S.:

Cloning of the cDNA (DSC1) coding for human type 1 desmocollin and its assignment to chromosome 18. *Genomics* 18: 185–194, 1993.

[26851] 403. Troyanovsky, S. M.; Eshkind, L. G.; Troyanovsky, R. B.; Leube, R. E.; Franke, W. W.: Contributions of cytoplasmic domains of desmosomal cadherins to desmosome assembly and intermediate filament anchorage. *Cell* 72:561–574, 1993.

[26852] 404. Arnemann, J.; Spurr, N. K.; Wheeler, G. N.; Parker, A. E.; Buxton, R. S.: Chromosomal assignment of the human genes coding for the major proteins of the desmosome junction, desmoglein DGI (DSG), desmocollins DGII/III (DSC), desmoplakins DPI/II (DSP), and plakoglobin DPIII (JUP). *Genomics* 10:640–645, 1991.

[26853] 405. Buxton, R. S.; Cowin, P.; Franke, W. W.; Garrod, D. R.; Green, K. J.; King, I. A.; Koch, P. J.; Magee, A. I.; Rees, D. A.; Stanley, J. R.; Steinberg, M. S.: Nomenclature of the desmosomal cadherins. *J. Cell Biol.* 121: 481–483, 1993.

[26854] 406. Buxton, R. S.; Wheeler, G. N.; Pidsley, S. C.; Marsden, M. D.; Adams, M. J.; Jenkins, N. A.; Gilbert, D. J.; Copeland, N. G.: Mouse desmocollin (Dsc3) and desmoglein (Dsg1) genes are closely linked in the proximal region of chromosome 18. *Genomics* 21: 510–516, 1994.

- [26855] 407.Greenwood, M. D.; Marsden, M. D.; Cowley, C. M. E.; Sahota, V.K.; Buxton, R. S.: Exon–intron organization of the human type 2 desmocollingene (DSC2): desmocollin gene structure is closer to 'classical' cadherin than to desmogleins. *Genomics* 44: 330–335, 1997.
- [26856] 408.Dale, D. C.; Person, R. E.; Bolyard, A. A.; Aprikyan, A. G.; Bos, C.; Bonilla, M. A.; Boxer, L. A.; Kannourakis, G.; Zeidler, C.; Welte, K.; Benson, K. F.; Horwitz, M.: Mutations in the gene encoding neutrophil elastase in congenital and cyclic neutropenia. *Blood* 96: 2317–2322, 2000.
- [26857] 409.Evans, E.; Cooley, J.; Remold–O'Donnell, E.: Characterization and chromosomal localization of ELANH2, the gene encoding human monocyte/neutrophil elastase inhibitor. *Genomics* 28: 235–240, 1995.
- [26858] 410.Schneider, S. S.; Schick, C.; Fish, K. E.; Miller, E.; Pena, J.C.; Treter, S. D.; Hui, S. M.; Silverman, G. A.: A serine proteinase inhibitor locus at 18q21.3 contains a tandem duplication of the human squamous cell carcinoma antigen gene. *Proc. Nat. Acad. Sci.* 92:3147–3151, 1995.
- [26859] 411.Ninomiya–Tsuji, J.; Nomoto, S.; Yasuda, H.; Reed, S. I.; Matsumoto, K.: Cloning of a human cDNA encoding a CDC2–related kinase by complementation of a budding yeast *cdc28* mutation. *Proc. Nat. Acad. Sci.* 88:

9006–9010,1991.

- [26860] 412.Shiffman, D.; Brooks, E. E.; Brooks, A. R.; Chan, C. S.; Milner,P. G.: Characterization of the human cyclin-dependent kinase 2 gene:promoter analysis and gene structure. *J. Biol. Chem.* 271: 12199–12204,1996.
- [26861] 413.Tsai, L.–H.; Harlow, E.; Meyerson, M.: Isolation of the human cdk2 gene that encodes the cyclin A- and adenovirus E1A-associated p33 kinase. *Nature* 353: 174–177, 1991.
- [26862] 414.Davies, R. L.; Grosse, V. A.; Kucherlapati, R.; Bothwell, M.:Genetic analysis of epidermal growth factor action: assignment of human epidermal growth factor receptor gene to chromosome 7. *Proc.Nat. Acad. Sci.* 77: 4188–4192, 1980.
- [26863] 415.Downward, J.; Yarden, Y.; Mayes, E.; Scrace, G.; Totty, N.; Stockwell,P.; Ullrich, A.; Schlessinger, J.; Waterfield, M. D.: Close similarity of epidermal growth factor receptor and v-erb-B oncogene protein sequences. *Nature* 307:521–527, 1984.
- [26864] 416.Haley, J.; Whittle, N.; Bennett, P.; Kinchington, D.; Ullrich,A.; Waterfield, M.: The human EGF receptor gene: structure of the 110 kb locus and identification of sequences regulating its transcription. *Oncogene Res.* 1:

375–396, 1987.

- [26865] 417.Henn, W.; Blin, N.; Zang, K. D.: Polysomy of chromosome 7 is correlated with overexpression of the erbB oncogene in human glioblastoma celllines. Hum. Genet. 74: 104–106, 1986.
- [26866] 418.Kondo, I.; Shimizu, N.: Mapping of the human gene for epidermal growth factor receptor (EGFR) on the p13–q22 region of chromosome 7. Cytogenet. Cell Genet. 35: 9–14, 1983.
- [26867] 419.Kramer, A.; Yang, F.–C.; Snodgrass, P.; Li, X.; Scammell, T. E.; Davis, F. C.; Weitz, C. J.: Regulation of daily locomotor activity and sleep by hypothalamic EGF receptor signaling. Science 294: 2511–2515, 2001.
- [26868] 420.Lanzetti, L.; Rybin, V.; Malabarba, M. G.; Christoforidis, S.; Scita, G.; Zerial, M.; Di Fiore, P. P.: The Eps8 protein coordinates EGF receptor signalling through Rac and trafficking through Rab5. Nature 408:374–377, 2000.
- [26869] 421.Maciag, T.: The human epidermal growth factor receptor–kinase complex. Trends Biochem. Sci. 7: 1–2, 1982.
- [26870] 422.Pai, R.; Soreghan, B.; Szabo, I. L.; Pavelka, M.; Baatar, D.; Tarnawski, A. S.: Prostaglandin E2 transactivates EGF receptor: a novel mechanism for promoting colon cancer growth and gastrointestinal hypertrophy. Nature Med. 8:

289–293, 2002.

- [26871] 423. Mannick, J. B.; Hausladen, A.; Liu, L.; Hess, D. T.; Zeng, M.; Miao, Q. X.; Kane, L. S.; Gow, A. J.; Stamler, J. S.: Fas-induced caspase denitrosylation. *Science* 284: 651–654, 1999.
- [26872] 424. Volpert, O. V.; Zaichuk, T.; Zhou, W.; Reiher, F.; Ferguson, T. A.; Stuart, P. M.; Amin, M.; Bouck, N. P.: Inducer-stimulated Fas targets activated endothelium for destruction by anti-angiogenic thrombospondin-1 and pigment epithelium-derived factor. *Nature Med.* 8: 349–357, 2002.
- [26873] 425. Scott, E. M.; Wright, R. C.: Variability of glutathione S-transferase of human erythrocytes. *Am. J. Hum. Genet.* 32: 115–117, 1980.
- [26874] 426. Seidegard, J.; Pero, R. W.; Markowitz, M. M.; Roush, G.; Miller, D. G.; Beattie, E. J.: Isoenzyme(s) of glutathione transferase (class mu) as a marker for the susceptibility to lung cancer: a follow up study. *Carcinogenesis* 11: 33–36, 1990.
- [26875] 427. Seidegard, J.; Pero, R. W.; Miller, D. G.; Beattie, E. J.: Glutathione transferase in human leukocytes as a marker for the susceptibility to lung cancer. *Carcinogenesis* 7: 751–753, 1986.
- [26876] 428. Seidegard, J.; Vorachek, W. R.; Pero, R. W.; Pearson,

W. R.:Hereditary differences in the expression of the human glutathione transferase active on trans-stilbene oxide are due to a gene deletion. *Proc.Nat. Acad. Sci.* 85: 7293–7297, 1988.

[26877] 429.Strange, R. C.; Davis, B. A.; Faulder, C. G.; Cotton, W.; Bain,A. D.; Hopkinson, D. A.; Hume, R.: The human glutathione S-transferases:developmental aspects of the GST1, GST2, and GST3 loci. *Biochem.Genet.* 23: 1011–1028, 1985.

[26878] 430.Strange, R. C.; Faulder, C. G.; Davis, B. A.; Hume, R.; Brown,J. A. H.; Cotton, W.; Hopkinson, D. A.: The human glutathione S-transferases:studies on the tissue distribution and genetic variation of the GST1,GST2 and GST3 isozymes. *Ann. Hum. Genet.* 48: 11–20, 1984.

[26879] 431.Strange, R. C.; Matharoo, B.; Faulder, G. C.; Jones, P.; Cotton,W.; Elder, J. B.; Deakin, M.: The human glutathione S-transferases:a case-control study of the incidence of the GST1 0 phenotype in patientswith adenocarcinoma. *Carcinogenesis* 12: 25–28, 1991.

[26880] 432.van Poppel, G.; de Vogel, N.; van Bladeren, P. J.; Kok, F. J.: Increased cytogenetic damage in smokers deficient in glutathioneS-transferase isozyme mu. *Carcinogenesis* 13: 303–305, 1992.

- [26881] 433. Xu, S.; Wang, Y.; Roe, B.; Pearson, W. R.: Characterization of the human class mu glutathione S-transferase gene cluster and the GSTM1 deletion. *J. Biol. Chem.* 273: 3517–3527, 1998.
- [26882] 434. Zhong, S.; Wolf, C. R.; Spurr, N. K.: Chromosomal assignment and linkage analysis of the human glutathione S-transferase mu-gene (GSTM1) using intron specific polymerase chain reaction. *Hum. Genet.* 90: 435–439, 1992.
- [26883] 435. Zhong, S.; Wyllie, A. H.; Barnes, D.; Wolf, C. R.; Spurr, N. K.: Relationship between the GSTM1 genetic polymorphism and susceptibility to bladder, breast and colon cancer. *Carcinogenesis* 14: 1821–1824, 1993.
- [26884] 436. Takahashi, Y.; Campbell, E. A.; Hirata, Y.; Takayama, T.; Listowsky, I.: A basis for differentiating among the multiple human mu-glutathione S-transferases and molecular cloning of brain GSTM5. *J. Biol. Chem.* 268: 8893–8898, 1993.
- [26885] 437. Campbell, E.; Takahashi, Y.; Abramovitz, M.; Peretz, M.; Listowsky, I.: A distinct human testis and brain mu-class glutathione S-transferase: molecular cloning and characterization of a form present even in individuals lacking hepatic type mu isoenzymes. *J. Biol. Chem.* 265: 9188–9193, 1990.

- [26886] 438.Elliott, K. J.; Ellis, S. B.; Berckhan, K. J.; Urrutia, A.; Chavez–Noriega,L. E.; Johnson, E. C.; Velicelebi, G.; Harpold, M. M.: Comparative structure of human neuronal alpha(2)–alpha(7) and beta(2)–beta(4)nicotinic acetylcholine receptor subunits and functional expression of the alpha(2), alpha(3), alpha(4), alpha(7), beta(2), and beta(4)subunits. *J. Molec. Neurosci.* 7: 217–228, 1996.
- [26887] 439.Solomon, E.; Hiorns, L. R.; Spurr, N.; Kurkinen, M.; Barlow, D.;Hogan, B. L. M.; Dalglish, R.: Chromosomal assignments of the genes coding for human types II, III and IV collagen: a dispersed gene family. *Proc.Nat. Acad. Sci.* 82: 3330–3334, 1985.
- [26888] 440.Reeve, R.; Silver, H. K.; Ferrier, P.: Marfan's syndrome (arachnodactyly)with arthrogryposis (amyoplasia congenita). *Am. J. Dis. Child.* 99:101–106, 1960.
- [26889] 441.Haddad, P.; Clement, M.–V.; Bernard, O.; Larsen, C.–J.; Degos,L.; Sasportes, M.; Mathieu–Mahul, D.: Structural organization of the CTLA–1 gene encoding human granzyme B. *Gene* 87: 265–271, 1990.
- [26890] 442.Kaplan, F. S.; McCluskey, W.; Hahn, G.; Tabas, J. A.; Muenke,M.; Zasloff, M. A.: Genetic transmission of fibrodysplasia ossificansprogressiva: report of a family. *J. Bone Joint Surg.* 75A: 1214–1220,1993.

- [26891] 443. Anisowicz, A.; Bardwell, L.; Sager, R.: Constitutive overexpression of a growth-regulated gene in transformed Chinese hamster and human cells. *Proc. Nat. Acad. Sci.* 84: 7188–7192, 1987.
- [26892] 444. Nieuwenhuis, E. E. S.; Matsumoto, T.; Exley, M.; Schleipman, R.A.; Glickman, J.; Bailey, D. T.; Corazza, N.; Colgan, S. P.; Onderdonk, A. B.; Blumberg, R. S.: CD1d-dependent macrophage-mediated clearance of *Pseudomonas aeruginosa* from lung. *Nature Med.* 8: 588–593, 2002.
- [26893] 445. Yang, J.; Patil, R. V.; Yu, H.; Gordon, M.; Wax, M. B.: T cell subsets and sIL-2R/IL-2 levels in patients with glaucoma. *Am. J. Ophthalmol.* 131: 421–426, 2001.
- [26894] 446. Eng, C. M.; Kozak, C. A.; Beaudet, A. L.; Zoghbi, H. Y.: Mapping of multiple subunits of the neuronal nicotinic acetylcholine receptor to chromosome 15 in man and chromosome 9 in mouse. *Genomics* 9: 278–282, 1991.
- [26895] 447. Flora, A.; Schulz, R.; Benfante, R.; Battaglioli, E.; Terzano, S.; Clementi, F.; Fornasari, D.: Transcriptional regulation of the human $\alpha 5$ nicotinic receptor subunit gene in neuronal and non-neuronal tissues. *Europ. J. Pharm.* 393: 85–95, 2000.
- [26896] 448. Fornasari, D.; Chini, B.; Tarroni, P.; Clementi, F.:

Molecular cloning of human neuronal nicotinic receptor α -3-subunit. *Neurosci.Lett.* 111: 351–356, 1990.

- [26897] 449. Groot Kormelink, P. J.; Luyten, W. H. M. L.: Cloning and sequence of full-length cDNAs encoding the human neuronal nicotinic acetylcholine receptor (nAChR) subunits β -3 and β -4 and expression of seven nAChR subunits in the human neuroblastoma cell line SH-SY5Y and/or IMR-32. *FEBS Lett.* 400: 309–314, 1997.
- [26898] 450. Mihovilovic, M.; Roses, A. D.: Expression of mRNAs in human thymus coding for the α -3 subunit of a neuronal acetylcholine receptor. *Exp.Neurol.* 111: 175–180, 1991.
- [26899] 451. Raimondi, E.; Rubboli, F.; Moralli, D.; Chini, B.; Fornasari, D.; Tarroni, P.; De Carli, L.; Clementi, F.: Chromosomal localization and physical linkage of the genes encoding the human α -3, α -5, and β -4 neuronal nicotinic receptor subunits. *Genomics* 12: 849–850, 1992.
- [26900] 452. Rempel, N.; Heyers, S.; Engels, H.; Slegers, E.; Steinlein, O.K.: The structures of the human neuronal nicotinic acetylcholine receptor β -2- and α -3-subunit genes (CHRNA2 and CHRNA3). *Hum.Genet.* 103: 645–653, 1998.
- [26901] 453. Forman, S. A.; Miller, K. W.; Yellen, G.: A discrete site for general anesthetics on a postsynaptic receptor. *Molec.*

Pharm. 48:574–581, 1995.

- [26902] 454. Forman, S. A.; Yellen, G.; Thiele, E. A.: Alternative mechanism for pathogenesis of an inherited epilepsy by a nicotinic AChR mutation. (Letter) *Nature Genet.* 13: 396–397, 1996.
- [26903] 455. Hirose, S.; Iwata, H.; Akiyoshi, H.; Kobayashi, K.; Ito, M.; Wada, K.; Kaneko, S.; Mitsudome, A.: A novel mutation of CHRNA4 responsible for autosomal dominant nocturnal frontal lobe epilepsy. *Neurology* 53:1749–1753, 1999.
- [26904] 456. Marubio, L. M.; del Mar Arroyo-Jimenez, M.; Cordero-Erausquin, M.; Lena, C.; Le Novere, N.; de Kerchove d'Exaerde, A.; Huchet, M.; Damaj, M. I.; Changeux, J.-P.: Reduced antinociception in mice lacking neuronal nicotinic receptor subunits. *Nature* 398: 805–810, 1999.
- [26905] 457. Monteggia, L. M.; Gopalakrishnan, M.; Touma, E.; Idler, K. B.; Nash, N.; Arneric, S. P.; Sullivan, J. P.; Giordano, T.: Cloning and transient expression of genes encoding the human alpha-4 and beta-2 neuronal nicotinic acetylcholine receptor (nAChR) subunits. *Gene* 155:189–193, 1995.
- [26906] 458. Pilz, A. J.; Willer, E.; Povey, S.; Abbott, C. M.: The genes coding for phosphoenolpyruvate carboxykinase-1 (PCK1) and neuronal nicotinic acetylcholine receptor al-

pha-4 subunit (CHRNA4) map to human chromosome 20, extending the known region of homology with mouse chromosome 2. *Ann. Hum. Genet.* 56: 289–293, 1992.

[26907] 459. Steinlein, O.; Smigrodzki, R.; Lindstrom, J.; Anand, R.; Kohler, M.; Tocharoentanaphol, C.; Vogel, F.: Refinement of the localization of the gene for neuronal nicotinic acetylcholine receptor alpha-4 subunit (CHRNA4) to human chromosome 20q13.2–q13.3. *Genomics* 22:493–495, 1994.

[26908] 460. Steinlein, O.; Weiland, S.; Stoodt, J.; Propping, P.: Exon–intron structure of the human neuronal nicotinic acetylcholine receptor alpha-4 subunit (CHRNA4). *Genomics* 32: 289–294, 1996.

[26909] 461. Steinlein, O. K.; Magnusson, A.; Stoodt, J.; Bertrand, S.; Weiland, S.; Berkovic, S. F.; Nakken, K. O.; Propping, P.; Bertrand, D.: An insertion mutation of the CHRNA4 gene in a family with autosomal dominant nocturnal frontal lobe epilepsy. *Hum. Molec. Genet.* 6: 943–947, 1997.

[26910] 462. Steinlein, O. K.; Mulley, J. C.; Propping, P.; Wallace, R. H.; Phillips, H. A.; Sutherland, G. R.; Scheffer, I. E.; Berkovic, S. F.: A missense mutation in the neuronal nicotinic acetylcholine receptor alpha-4 subunit is associated with autosomal dominant nocturnal frontal lobe epilepsy.

Nature Genet. 11: 201–203, 1995.

- [26911] 463. Tsonis, P.; Goetinck, P. F.: The *Drosophila* homoeotic gene *spaltis* structurally related to collagen alpha-1(IV) chain. (Letter) *Collagen Rel. Res.* 8: 451–452, 1988.
- [26912] 464. Wieslander, J.; Barr, J. F.; Butkowski, R. J.; Edwards, S. J.; Bygren, P.; Heinegard, D.; Hudson, B. G.: Goodpasture antigen of the glomerular basement membrane: localization to noncollagenous regions of type IV collagen. *Proc. Nat. Acad. Sci.* 81: 3838–3842, 1984.
- [26913] 465. Wieslander, J.; Langeveld, J.; Butkowski, R.; Jodlowski, M.; Noelken, M.; Hudson, B. G.: Physical and immunochemical studies of the globular domain of type IV collagen: cryptic properties of the Goodpasture antigen. *J. Biol. Chem.* 260: 8564–8570, 1985.
- [26914] 466. Kaplan, F. S.; Tabas, J. A.; Gannon, F. H.; Finkel, G.; Hahn, G. V.; Zasloff, M. A.: The histopathology of fibrodysplasia ossificans progressiva: an endochondral process. *J. Bone Joint Surg.* 75A: 220–230, 1993.
- [26915] 467. Lucotte, G.; Bathelier, C.; Mercier, G.; Gerard, N.; Lenoir, G.; Semonin, O.; Fontaine, K.; FOP Consortium: Localization of the gene for fibrodysplasia ossificans progressiva (FOP) to chromosome 17q21–22. *Genet. Counsel.* 11: 329–334, 2000.

- [26916] 468. Lucotte, G.; Semonin, O.; Lutz, P.: A de novo heterozygous deletion of 42 base-pairs in the *noggin* gene of a fibrodysplasia ossificans progressiva patient. (Letter) *Clin. Genet.* 56: 469–470, 1999.
- [26917] 469. Maxwell, W. A.; Spicer, S. S.; Miller, R. L.; Halushka, P. V.; Westphal, M. C.; Setser, M. E.: Histochemical and ultrastructural studies in fibrodysplasia ossificans progressiva (myositis ossificans progressiva). *Am. J. Path.* 87: 483–498, 1977.
- [26918] 470. Rogers, J. G.; Chase, G. A.: Paternal age effect in fibrodysplasia ossificans progressiva. *J. Med. Genet.* 16: 147–148, 1979.
- [26919] 471. Rogers, J. G.; Geho, W. B.: Fibrodysplasia ossificans progressiva: a survey of forty-two cases. *J. Bone Joint Surg.* 61A: 909–914, 1979.
- [26920] 472. Schroeder, H. W., Jr.; Zasloff, M.: The hand and foot malformations in fibrodysplasia ossificans progressiva. *Johns Hopkins Med. J.* 147: 73–78, 1980.
- [26921] 473. Shafritz, A. B.; Shore, E. M.; Gannon, F. H.; Zasloff, M. A.; Taub, R.; Muenke, M.; Kaplan, F. S.: Overexpression of an osteogenic morphogen in fibrodysplasia ossificans progressiva. *New Eng. J. Med.* 335: 555–561, 1996.
- [26922] 474. Smith, R.; Athanasou, N. A.; Vipond, S. E.: Fibrodys-

plasia (myositis)ossificans progressiva: clinicopathological features and natural history. Quart.J. Med. 89: 445–456, 1996.

[26923] 475.Tuente, W.; Becker, P. E.; Von Knorre, G. V.: Zur Genetik derMyositis ossificans progressiva. Humangenetik 4: 320–351, 1967.

[26924] 476.Viparelli, V.: La miosite ossificante progressiva. Ann. Neuropsychiat.Psicoanal. 9: 297–324, 1962.

[26925] 477.Xu, M.–Q.; Feldman, G.; Le Merrer, M.; Shugart, Y. Y.; Glaser,D. L.; Urtizberea, J. A.; Fardeau, M.; Connor, J. M.; Triffitt, J.;Smith, R.; Shore, E. M.; Kaplan, F. S.: Linkage exclusion and mutationalanalysis of the noggin gene in patients with fibrodysplasia ossificansprogressiva (FOP). Clin. Genet. 58: 291–298, 2000.

[26926] 478.Allende, M. L.; Amsterdam, A.; Becker, T.; Kawakami, K.; Gaiano,N.; Hopkins, N.: Insertional mutagenesis in zebrafish identifiestwo novel genes, pescadillo and dead eye, essential for embryonicdevelopment. Genes Dev. 10: 3141–3155, 1996.

[26927] 479.Haque, J.; Boger, S.; Li, J.; Duncan, S. A.: The murine Pes1 geneencodes a nuclear protein containing a BRCT domain. Genomics 70:201–210, 2000.

[26928] 480.Kinoshita, Y.; Jarell, A. D.; Flaman, J. M.; Foltz, G.;

Schuster, J.; Sopher, B. L.; Irvin, D. K.; Kanning, K.; Kornblum, H. I.; Nelson, P. S.; Hieter, P.; Morrison, R. S.:

Pescadillo, a novel cell cycleregulatory protein abnormally expressed in malignant cells. *J. Biol.Chem.* 276: 6656–6665, 2001.

[26929] 481. Sumoy, L.; Carim, L.; Escarceller, M.; Nadal, M.; Gratacos, M.; Pujana, M. A.; Estivill, X.; Peral, B.: HMG20A and HMG20B map to human chromosomes 15q24 and 19p13.3 and constitute a distinct class of HMG-box genes with ubiquitous expression. *Cytogenet. Cell Genet.* 88:62–67, 2000.

[26930] 482. Marmorstein, L. Y.; Kinev, A. V.; Chan, G. K. T.; Bochar, D. A.; Beniya, H.; Epstein, J. A.; Yen, T. J.; Shiekhattar, R.: A human BRCA2 complex containing a structural DNA binding component influences cell cycle progression. *Cell* 104: 247–257, 2001.

[26931] 483. Prekeris, R.; Klumperman, J.; Scheller, R. H.: A Rab11/Rip11 protein complex regulates apical membrane trafficking via recycling endosomes. *Molec. Cell* 6: 1437–1448, 2000.

[26932] 484. Hai, T. W.; Liu, F.; Coukos, W. J.; Green, M. R.: Transcription factor ATF cDNA clones: an extensive family of leucine zipper proteins able to selectively form DNA–

binding heterodimers. *Genes Dev.* 3:2083–2090, 1989.

[26933] 485.Hai, T. W.; Liu, F.; Coukos, W. J.; Green, M. R. :*Genes Dev.* 3:2083–2090, 1989.

[26934] 486.Haze, K.; Yoshida, H.; Yanagi, H.; Yura, T.; Mori, K.: Mammalian transcription factor ATF6 is synthesized as a transmembrane protein and activated by proteolysis in response to endoplasmic reticulum stress. *Molec. Biol. Cell* 10: 3787–3799, 1999.

[26935] 487.Li, M.; Baumeister, P.; Roy, B.; Phan, T.; Foti, D.; Luo, S.; Lee, A. S.: ATF6 as a transcription activator of the endoplasmic reticulum stress element: thapsigargin stress-induced changes and synergistic interactions with NF- κ B and YY1. *Molec. Cell. Biol.* 20: 5096–5106, 2000.

[26936] 488.Scott, A. F.: Personal Communication. Baltimore, Md. 2/26/2001.

[26937] 489.Yoshida, H.; Haze, K.; Yanagi, H.; Yura, T.; Mori, K.: Identification of the cis-acting endoplasmic reticulum stress response element responsible for transcriptional induction of mammalian glucose-regulated proteins: involvement of basic leucine zipper transcription factors. *J. Biol.Chem.* 273: 33741–33749, 1998.

[26938] 490.Zhu, C.; Johansen, F.-E.; Prywes, R.: Interaction of ATF6 and serum response factor. *Molec. Cell. Biol.* 17:

4957–4966, 1997.

- [26939] 491.Cenciarelli, C.; Chiaur, D. S.; Guardavaccaro, D.; Parks, W.; Vidal,M.; Pagano, M.: Identification of a family of human F-box proteins. *Curr.Biol.* 9: 1177–1179, 1999.
- [26940] 492.Winston, J. T.; Koepp, D. M.; Zhu, C.; Elledge, S. J.; Harper,J. W.: A family of mammalian F-box proteins. *Curr. Biol.* 9: 1180–1182,1999.
- [26941] 493.Gerlach, V. L.; Aravind, L.; Gotway, G.; Schultz, R. A.; Koonin,E. V.; Friedberg, E. C.: Human and mouse homologs of Escherichiacoli DinB (DNA polymerase IV), members of the UmuC/DinB superfamily. *Proc.Nat. Acad. Sci.* 96: 11922–11927, 1999.
- [26942] 494.Johnson, R. E.; Prakash, S.; Prakash, L.: The human DINB1 geneencodes the DNA polymerase Pol-theta. *Proc. Nat. Acad. Sci.* 97:3838–3843, 2000.
- [26943] 495.Ogi, T.; Kato, T., Jr.; Kato, T.; Ohmori, H.: Mutation enhancementby DINB1, a mammalian homologue of the Escherichia coli mutagenesisprotein dinB. *Genes Cells* 4: 607–618, 1999.
- [26944] 496.Ohashi, E.; Ogi, T.; Kusumoto, R.; Iwai, S.; Masutani, C.; Hanaoka,F.; Ohmori, H.: Error-prone bypass of certain DNA lesions by thehuman DNA polymerase kappa. *Genes Dev.* 14: 1589–1594, 2000.

- [26945] 497.Koike, J.; Sagara, N.; Kirikoshi, H.; Takagi, A.; Miwa, T.; Hirai,M.; Katoh, M.: Molecular cloning and genomic structure of the beta-TRCP2gene on chromosome 5q35.1. Biochem. Biophys. Res. Commun. 269: 103–109,2000.
- [26946] 498.Chiaur, D. S.; Murthy, S.; Cenciarelli, C.; Parks, W.; Loda, M.;Inghirami, G.; Demetrick, D.; Pagano, M.: Five human genes encodingF-box proteins: chromosome mapping and analysis in human tumors. Cytogenet.Cell Genet. 88: 255–258, 2000.
- [26947] 499.Fanciulli, M.; Bruno, T.; Cerboni, C.; Bonetto, F.; Iacobini, C.;Fрати, L.; Piccoli, M.; Floridi, A.; Santoni, A.; Punturieri, A.:Cloning of a novel human RNA polymerase II subunit downregulated bydoxorubicin: new potential mechanisms of drug related toxicity. FEBSLett. 384: 48–52, 1996.
- [26948] 500.Pati, U. K.: Human RNA polymerase II subunit hRPB14 is homologousto yeast RNA polymerase I, II, and III subunits (AC19 and RPB11) andis similar to a portion of the bacterial RNA polymerase alpha subunit. Gene 145:289–292, 1994.
- [26949] 501.Ito, K.; Kato, S.; Matsuda, Y.; Kimura, M.; Okano, Y.: cDNA cloning,characterization, and chromosome mapping of UBE2E3 (alias Ubch9),encoding an N-terminally ex-

tended human ubiquitin-conjugating enzyme. *Cytogenet. Cell Genet.* 84: 99–104, 1999.

[26950] 502. Matuschewski, K.; Hauser, H.-P.; Treier, M.; Jentsch, S.: Identification of a novel family of ubiquitin-conjugating enzymes with distinct amino-terminal extensions. *J. Biol. Chem.* 271: 2789–2794, 1996.

[26951] 503. Bauer, H.; Mayer, H.; Marchler-Bauer, A.; Salzer, U.; Prohaska, R.: Characterization of p40/GPR69A as a peripheral membrane protein related to the lantibiotic synthetase component C. *Biochem. Biophys. Res. Commun.* 275: 69–74, 2000.

[26952] 504. Mayer, H.; Bauer, H.; Prohaska, R.: Organization and chromosomal localization of the human and mouse genes coding for LanC-like protein 1 (LANCL1). *Cytogenet. Cell Genet.* 93: 100–104, 2001.

[26953] 505. Mayer, H.; Salzer, U.; Breuss, J.; Ziegler, S.; Marchler-Bauer, A.; Prohaska, R.: Isolation, molecular characterization, and tissue-specific expression of a novel putative G protein-coupled receptor. *Biochim. Biophys. Acta* 1395: 301–308, 1998.

[26954] 506. Baylin, S. B.; Herman, J. G.: DNA hypermethylation in tumorigenesis: epigenetics joins genetics. *Trends Genet.* 16: 168–174, 2000.

- [26955] 507.Cameron, E. E.; Bachman, K. E.; Myohanen, S.; Herman, J. G.; Baylin, S. B.: Synergy of demethylation and histone deacetylase inhibition in the re-expression of genes silenced in cancer. *Nature Genet.* 21:103–107, 1999.
- [26956] 508.Finch, P. W.; He, X.; Kelley, M. J.; Uren, A.; Schaudies, R. P.; Popescu, N. C.; Rudikoff, S.; Aaronson, S. A.; Varmus, H. E.; Rubin, J. S.: Purification and molecular cloning of a secreted, frizzled-related antagonist of Wnt action. *Proc. Nat. Acad. Sci.* 94: 6770–6775, 1997.
- [26957] 509.Fukuhara, K.; Kariya, M.; Kita, M.; Shime, H.; Kanamori, T.; Kosaka, C.; Orii, A.; Fujita, J.; Fujii, S.: Secreted frizzled related protein1 is overexpressed in uterine leiomyomas, associated with a high estrogenic environment and unrelated to proliferative activity. *J. Clin. Endocr. Metab.* 87: 1729–1736, 2002.
- [26958] 510.Melkonyan, H. S.; Chang, W. C.; Shapiro, J. P.; Mahadevappa, M.; Fitzpatrick, P. A.; Kiefer, M. C.; Tomei, L. D.; Umansky, S. R.: SARPs: a family of secreted apoptosis-related proteins. *Proc. Nat. Acad. Sci.* 94: 13636–13641, 1997.
- [26959] 511.Suzuki, H.; Gabrielson, E.; Chen, W.; Anbazhagan, R.; van Engeland, M.; Weijnenberg, M. P.; Herman, J. G.; Baylin, S. B.: A genomic screen for genes upregulated by

demethylation and histone deacetylase inhibition in human colorectal cancer. *Nature Genet.* 31: 141–149, 2002.

[26960] 512. Rattner, A.; Hsieh, J.-C.; Smallwood, P. M.; Gilbert, D. J.; Copeland, N. G.; Jenkins, N. A.; Nathans, J.: A family of secreted proteins contains homology to the cysteine-rich ligand-binding domain of frizzled receptors. *Proc. Nat. Acad. Sci.* 94: 2859–2863, 1997.

[26961] 513. Olaisen, B.; Gedde-Dahl, T., Jr.: GPT-epidermolysis bullosa simplex (EBS Ogna) linkage in man. *Hum. Hered.* 23: 189–196, 1973.

[26962] 514. Olaisen, B.; Gedde-Dahl, T., Jr.: GPT-EBS(1) linkage group: general linkage relations. *Hum. Hered.* 24: 178–185, 1974.

[26963] 515. Bach, I.; Galcheva-Gargova, Z.; Mattei, M.-G.; Simon-Chazottes, D.; Guenet, J.-L.; Cereghini, S.; Yaniv, M.: Cloning of human hepatic nuclear factor 1 (HNF1) and chromosomal localization of its gene in man and mouse. *Genomics* 8: 155–164, 1990.

[26964] 516. Byrne, M. M.; Sturis, J.; Menzel, S.; Yamagata, K.; Fajans, S.S.; Dronsfield, M. J.; Bain, S. C.; Hattersley, A. T.; Velho, G.; Froguel, P.; Bell, G. I.; Polonsky, K. S.: Altered insulin secretory responses to glucose in diabetic and nondiabetic subjects with mutations in the diabetes susceptibil-

ity gene MODY3 on chromosome 12. Diabetes 45:1503–1510, 1996.

- [26965] 517. Chiu, K. C.; Chuang, L.-M.; Ryu, J. M.; Tsai, G. P.; Saad, M. F.: The I27L amino acid polymorphism of hepatic nuclear factor-1-alpha is associated with insulin resistance. J. Clin. Endocr. Metab. 85:2178–2183, 2000.
- [26966] 518. Collet, C.; et al.; et al.: Prevalence of the missense mutation Gly574Ser in the hepatocyte nuclear factor-1-alpha in Africans with diabetes. Diabetes Metab. 28: 39–44, 2002.
- [26967] 519. Courtois, G.; Morgan, J. G.; Campbell, L. A.; Fourel, G.; Crabtree, G. R.: Interaction of a liver-specific nuclear factor with the fibrinogen and alpha-1-antitrypsin promoters. Science 238: 688–692, 1987.
- [26968] 520. De Simone, V.; De Magistris, L.; Lazzaro, D.; Gerstner, J.; Monaci, P.; Nicosia, A.; Cortese, R.: LFB3, a heterodimer-forming homeoprotein of the LFB1 family, is expressed in specialized epithelia. EMBO J. 10:1435–1443, 1991.
- [26969] 521. Ellard, S.: Hepatocyte nuclear factor 1 alpha (HNF-1-alpha) mutations in maturity-onset diabetes of the young. Hum. Mutat. 16: 377–385, 2000.
- [26970] 522. Frayling, T. M.; Bulman, M. P.; Appleton, M.; Hattersley, A. T.; Ellard, S.: A rapid screening method for hepato-

cyte nuclear factor1 alpha frameshift mutations; prevalence in maturity-onset diabetes of the young and late-onset non-insulin dependent diabetes. Hum.Genet. 101: 351–354, 1997.

[26971] 523.Frayling, T. M.; Bulman, M. P.; Ellard, S.; Appleton, M.; Dronsfield, M. J.; Mackie, A. D. R.; Baird, J. D.; Kaisaki, P. J.; Yamagata, K.; Bell, G. I.; Bain, S. C.; Hattersley, A. T.: Mutations in the hepatocyte nuclear factor-1-alpha gene are a common cause of maturity-onset diabetes of the young in the U.K. Diabetes 46: 720–725, 1997.

[26972] 524.Godart, F.; Bellanne-Chantelot, C.; Clauin, S.; Gragnoli, C.; Abderrahmani, A.; Blanche, H.; Boutin, P.; Chevre, J. C.; Froguel, P.; Bailleul, B.: Identification of seven novel nucleotide variants in the hepatocyte nuclear factor-1-alpha (TCF1) promoter region in MODY patients. Hum. Mutat. 15: 173–180, 2000.

[26973] 525.Gonzalez, F. J.; Liu, S.-Y.; Kozak, C. A.; Nebert, D. W.: Decreased Hnf-1 gene expression in mice homozygous for a 1.2-centimorgan deletion on chromosome 7. DNA Cell Biol. 9: 771–776, 1990.

[26974] 526.Gragnoli, C.; Lindner, T.; Cockburn, B. N.; Kaisaki, P. J.; Gragnoli, F.; Marozzi, G.; Bell, G. I.: Maturity-onset diabetes of the young due to a mutation in the hepatocyte

nuclear factor-4-alpha binding site in the promoter of the hepatocyte nuclear factor-1-alpha gene. *Diabetes* 46:1648-1651, 1997.

[26975] 527. Hansen, T.; Eiberg, H.; Rouard, M.; Vaxillaire, M.; Moller, A.M.; Rasmussen, S. K.; Fridberg, M.; Urhammer, S. A.; Holst, J. J.; Almind, K.; Echwald, S. M.; Hansen, L.; Bell, G. I.; Pedersen, O.: Novel MODY3 mutations in the hepatocyte nuclear factor-1-alpha gene: evidence for a hyperexcitability of pancreatic beta-cells to intravenous secretagogues in a glucose-tolerant carrier of a P447L mutation. *Diabetes* 46:726-730, 1997.

[26976] 528. Hegele, R. A.; Cao, H.; Harris, S. B.; Hanley, A. J. G.; Zinman, B.: The hepatic nuclear factor-1-alpha G319S variant is associated with early-onset type 2 diabetes in Canadian Oji-Cree. *J. Clin. Endocr. Metab.* 84: 1077-1082, 1999.

[26977] 529. Hegele, R. A.; Cao, H.; Harris, S. B.; Zinman, B.; Hanley, A. J. G.; Anderson, C. M.: Peroxisome proliferator-activated receptor-gamma-2P12A and type 2 diabetes in Canadian Oji-Cree. *J. Clin. Endocr. Metab.* 85:2014-2019, 2000.

[26978] 530. Hiraiwa, H.; Pan, C.-J.; Lin, B.; Akiyama, T. E.; Gonzalez, F. J.; Chou, J. Y.: A molecular link between the common

phenotypes of type 1 glycogen storage disease and HNF1- α -null mice. *J. Biol. Chem.* 276: 7963–7967, 2001.

[26979] 531. Hua, Q.-X.; Zhao, M.; Narayana, N.; Nakagawa, S. H.; Jia, W.; Weiss, M. A.: Diabetes-associated mutations in a beta-cell transcription factor destabilize an antiparallel 'mini-zipper' in a dimerization interface. *Proc. Nat. Acad. Sci.* 97: 1999–2004, 2000.

[26980] 532. Kaisaki, P. J.; Menzel, S.; Lindner, T.; Oda, N.; Rjasanowski, I.; Sahm, J.; Meincke, G.; Schulze, J.; Schmechel, H.; Petzold, C.; Ledermann, H. M.; Sachse, G.; Boriraj, V. V.; Menzel, R.; Kerner, W.; Turner, R. C.; Yamagata, K.; Bell, G. I.: Mutations in the hepatocyte nuclear factor-1- α gene in MODY and early-onset NIDDM: evidence for a mutational hotspot in exon 4. *Diabetes* 46: 528–535, 1997.

[26981] 533. Kuo, C. J.; Conley, P. B.; Hsieh, C.-L.; Francke, U.; Crabtree, G. R.: Molecular cloning, functional expression, and chromosomal localization of mouse hepatocyte nuclear factor 1. *Proc. Nat. Acad. Sci.* 87: 9838–9842, 1990.

[26982] 534. Mendel, D. B.; Hansen, L. P.; Graves, M. K.; Conley, P. B.; Crabtree, G. R.: HNF-1- α and HNF-1- β (vHNF-1) share dimerization and homeo domains, but not activation

domains, and form heterodimers invitro. *Genes Dev.* 5: 1042–1056, 1991.

- [26983] 535. Miedzybrodzka, Z.; Hattersley, A. T.; Ellard, S.; Pearson, D.; de Silva, D.; Harvey, R.; Haites, N.: Non-penetrance in a MODY 3 family with a mutation in the hepatic nuclear factor 1a gene: implications for predictive testing. *Europ. J. Hum. Genet.* 7: 729–732, 1999.
- [26984] 536. Pontoglio, M.; Barra, J.; Hadchouel, M.; Doyen, A.; Kress, C.; Bach, J. P.; Babinet, C.; Yaniv, M.: Hepatocyte nuclear factor 1 inactivation results in hepatic dysfunction, phenylketonuria, and renal Fanconi syndrome. *Cell* 84: 575–585, 1996.
- [26985] 537. Braud, V. M.; Allan, D. S. J.; O'Callaghan, C. A.; Soderstrom, K.; D'Andrea, A.; Ogg, G. S.; Lazetic, S.; Young, N. T.; Bell, J. I.; Phillips, J. H.; Lanier, L. L.; McMichael, A. J.: HLA-E binds to natural killer cell receptors CD94/NKG2A, B and C. *Nature* 391: 795–799, 1998.
- [26986] 538. Carroll, M. C.; Katzman, P.; Alicot, E. M.; Koller, B. H.; Geraghty, D. E.; Orr, H. T.; Strominger, J. L.; Spies, T.: Linkage map of the human major histocompatibility complex including the tumor necrosis factor genes. *Proc. Nat. Acad. Sci.* 84: 8535–8539, 1987.
- [26987] 539. Andrews, P. W.; Knowles, B. B.; Goodfellow, P. N.: A

human cell-surface antigen defined by a monoclonal antibody and controlled by a gene on chromosome 12. *Somat. Cell Genet.* 7: 435–443, 1981.

[26988] 540. Andrews, P. W.; Knowles, B. B.; Goodfellow, P. N.: A chromosome 12-controlled cell surface antigen defined by a monoclonal antibody. (Abstract) *Cytogenet. Cell Genet.* 32: 249 only, 1982.

[26989] 541. Benoit, P.; Gross, M. S.; Frachet, P.; Frezal, J.; Uzan, G.; Boucheix, C.; Van Cong, N.: Assignment of the human CD9 gene to chromosome 12 (region p13) by use of human specific DNA probes. *Hum. Genet.* 86:268–272, 1991.

[26990] 542. Boucheix, C.; Benoit, P.; Frachet, P.; Billard, M.; Worthington, R. E.; Gagnon, J.; Uzan, G.: Molecular cloning of the CD9 antigen: a new family of cell surface proteins. *J. Biol. Chem.* 266: 117–122, 1991.

[26991] 543. Goodfellow, P. N.: Personal Communication. London, England 1982.

[26992] 544. Kaji, K.; Oda, S.; Shikano, T.; Ohnuki, T.; Uematsu, Y.; Sakagami, J.; Tada, N.; Miyazaki, S.; Kudo, A.: The gamete fusion process is defective in eggs of Cd9-deficient mice. *Nature Genet.* 24: 279–282, 2000.

[26993] 545. Katz, F.; Povey, S.; Parkar, M.; Schneider, C.; Suther-

land, R.; Stanley, K.; Solomon, E.; Greaves, M.: Chromosome assignment of monoclonal antibody-defined determinants on human leukemic cells. *Europ. J. Immun.* 13: 1008–1013, 1983.

[26994] 546. Le Naour, F.; Rubinstein, E.; Jasmin, C.; Prenant, M.; Boucheix, C.: Severely reduced female fertility in CD9-deficient mice. *Science* 287:319–321, 2000.

[26995] 547. Miyado, K.; Yamada, G.; Yamada, S.; Hasuwa, H.; Nakamura, Y.; Ryu, F.; Suzuki, K.; Kosai, K.; Inoue, K.; Ogura, A.; Okabe, M.; Mekada, E.: Requirement of CD9 on the egg plasma membrane for fertilization. *Science* 287:321–324, 2000.

[26996] 548. Tachibana, I.; Hemler, M. E.: Role of transmembrane 4 superfamily (TM4SF) proteins CD9 and CD81 in muscle cell fusion and myotube maintenance. *J. Cell Biol.* 146: 893–904, 1999.

[26997] 549. Van Cong, N.; Benoit, P.; Gross, M. S.; Uzan, G.; Frachet, P.; Marguerie, G.; Frezal, J.; Boucheix, C.: Assignment of the gene for CD9 (p24) antigen to 12p13. (Abstract) *Cytogenet. Cell Genet.* 51:1096 only, 1989.

[26998] 550. Waterhouse, R.; Ha, C.; Dveksler, G. S.: Murine CD9 is the receptor for pregnancy-specific glycoprotein 17. *J. Exp. Med.* 195: 277–282, 2002.

- [26999] 551. Mullis, P. E.; Patel, M. S.; Brickell, P. M.; Hindmarsh, P. C.; Brook, C. G. D.: Growth characteristics and response to growth hormone therapy in patients with hypochondroplasia: genetic linkage of the insulin-like growth factor I gene at chromosome 12q23 to the disease in a subgroup of these patients. *Clin. Endocr.* 34: 265–274, 1991.
- [27000] 552. Miki, T.; Kawamata, N.; Arai, A.; Ohashi, K.; Nakamura, Y.; Kato, A.; Hirosawa, S.; Aoki, N.: Molecular cloning of the breakpoint for 3q27 translocation in B-cell lymphomas and leukemias. *Blood* 83: 217–222, 1994.
- [27001] 553. Ferrari, A. C.; Seunemann, H. N.; Hanash, S. M.; Atweh, G. F.: A gene that encodes for a leukemia-associated phosphoprotein (p18) maps to chromosome bands 1p35–36.1. *Genes Chromosomes Cancer* 2: 125–129, 1990.
- [27002] 554. Hanash, S. M.; Strahler, J. R.; Kuick, R.; Chu, E. H. Y.; Nichols, D.: Identification of a polypeptide associated with the malignant phenotype in acute leukemia. *J. Biol. Chem.* 263: 12813–12815, 1988.
- [27003] 555. Kumar, R.; Haugen, J. D.: Human and rat osteoblast-like cells express stathmin, a growth-regulatory protein. *Biochem. Biophys. Res. Commun.* 201: 861–865, 1994.
- [27004] 556. Maucuer, A.; Camonis, J. H.; Sobel, A.: Stathmin inter-

action with a putative kinase and coiled-coil-forming protein domains. *Proc. Nat. Acad. Sci.* 92: 3100–3104, 1995.

- [27005] 557. Mock, B. A.; Krall, M. M.; Padlan, C.; Dosik, J. K.; Schubart, U. K.: The gene for Lap18, leukemia-associated phosphoprotein p18(metablastin), maps to distal mouse chromosome 4. *Mammalian Genome* 4:461–462, 1993.
- [27006] 558. Okazaki, T.; Yoshida, B. N.; Avraham, K. B.; Wang, H.; Wuenschell, C. W.; Jenkins, N. A.; Copeland, N. G.; Anderson, D. J.; Mori, N.: Molecular diversity of the SCG10/stathmin gene family in the mouse. *Genomics* 18:360–373, 1993. Note: Erratum: *Genomics* 21: 298 only, 1994.
- [27007] 559. Sobel, A.: Stathmin: a relay phosphoprotein for multiple signal transduction? *Trends Biochem. Sci.* 16: 301–305, 1991.
- [27008] 560. Sobel, A.; Bouterin, M.-C.; Beretta, L.; Chneiweiss, H.; Doye, V.; Peyro-Saint-Paul, H.: Intracellular substrates for extracellular signaling: characterization of a ubiquitous, neuron-enriched phosphoprotein (stathmin). *J. Biol. Chem.* 264: 3765–3772, 1989.
- [27009] 561. Zhu, X. X.; Kozarsky, K.; Strahler, J. R.; Eckerskorn, C.; Lottspeich, F.; Melhem, R.; Lowe, J.; Fox, D. A.; Hanash, S. M.; Atweh, G. F.: Molecular cloning of a novel human

leukemia-associated gene: evidence of conservation in animal species. *J. Biol. Chem.* 264: 14556–14560, 1989.

[27010] 562. Delespesse, G.; Sarfati, M.; Peleman, R.: Influence of recombinant IL-4, IFN- α , and IFN- γ on the production of human IgE-binding factor (soluble CD23). *J. Immun.* 142: 134–138, 1989.

[27011] 563. Ludin, C.; Hofstetter, H.; Sarfati, M.; Levy, C. A.; Suter, U.; Alaimo, D.; Kilchherr, E.; Frost, H.; Delespesse, G.: Cloning and expression of the cDNA coding for a human lymphocyte IgE receptor. *EMBO J.* 6: 109–114, 1987.

[27012] 564. Wendel-Hansen, V.; Riviere, M.; Uno, M.; Jansson, I.; Szpirer, J.; Islam, M. Q.; Levan, G.; Klein, G.; Yodoi, J.; Rosen, A.; Szpirer, C.: The gene encoding CD23 leukocyte antigen (FCE2) is located on human chromosome 19. *Somat. Cell Molec. Genet.* 16: 283–286, 1990.

[27013] 565. Li, P.; Allen, H.; Banerjee, S.; Franklin, S.; Herzog, L.; Johnston, C.; McDowell, J.; Paskind, M.; Rodman, L.; Salfeld, J.; Towne, E.; Tracey, D.; Wardwell, S.; Wei, F.-Y.; Wong, W.; Kamen, R.; Seshadri, T.: Mice deficient in IL-1- β -converting enzyme are defective in production of mature IL-1- β and resistant to endotoxic shock. *Cell* 80:401–411, 1995.

[27014] 566. Li, P.; Allen, H.; Banerjee, S.; Seshadri, T.: Characteri-

zation of mice deficient in interleukin-1-beta converting enzyme. *J. Cell. Biochem.* 64: 27-32, 1997.

- [27015] 567. Li, S.-H.; Lam, S.; Cheng, A. L.; Li, X.-J.: Intracellular huntingtin increases the expression of caspase-1 and induces apoptosis. *Hum. Molec. Genet.* 9: 2859-2867, 2000.
- [27016] 568. Mangiarini, L.; Sathasivam, K.; Seller, M.; Cozens, B.; Harper, A.; Hetherington, C.; Lawton, M.; Trotter, Y.; Leach, H.; Davies, S. W.; Bates, G. P.: Exon 1 of the HD gene with an expanded CAG repeat is sufficient to cause a progressive neurological phenotype in transgenic mice. *Cell* 87: 493-506, 1996.
- [27017] 569. Miura, M.; Zhu, H.; Rotello, R.; Hartwig, E. A.; Yuan, J.: Induction of apoptosis in fibroblasts by IL-1-beta-converting enzyme, a mammalian homolog of the *C. elegans* cell death gene *ced-3*. *Cell* 75: 653-660, 1993.
- [27018] 570. Nett, M. A.; Cerretti, D. P.; Berson, D. R.; Seavitt, J.; Gilbert, D. J.; Jenkins, N. A.; Copeland, N. G.; Black, R. A.; Chaplin, D. D.: Molecular cloning of the murine IL-1-beta converting enzyme cDNA. *J. Immunol.* 149: 3254-3259, 1992.
- [27019] 571. Ona, V. O.; Li, M.; Vonsattel, J. P. G.; Andrews, L. J.; Khan, S. Q.; Chung, W. M.; Frey, A. S.; Menon, A. S.; Li, X.-J.; Stieg, P. E.; Yuan, J.; Penney, J. B.; Young, A. B.; Cha,

J.-H. J.; Friedlander, R. M.: Inhibition of caspase-1 slows disease progression in a mouse model of Huntington's disease. *Nature* 399: 263–267, 1999.

[27020] 572. Wilson, K. P.; Black, J.-A. F.; Thomson, J. A.; Kim, E. E.; Griffith, J. P.; Navia, M. A.; Murcko, M. A.; Chambers, S. P.; Aldape, R. A.; Raybuck, S. A.; Livingston, D. J.: Structure and mechanism of interleukin-1-beta converting enzyme. *Nature* 370: 270–275, 1994.

[27021] 573. Yuan, J.; Shaham, S.; Ledoux, S.; Ellis, H. M.; Horvitz, H. R.: The *C. elegans* cell death gene *ced-3* encodes a protein similar to mammalian interleukin-1-beta-converting enzyme. *Cell* 75: 641–652, 1993.

[27022] 574. Arend, W. P.: Interleukin 1 receptor antagonist: a new member of the interleukin 1 family. *J. Clin. Invest.* 88: 1445–1451, 1991.

[27023] 575. Thornberry, N. A.; Bull, H. G.; Calaycay, J. R.; Chapman, K. T.; Howard, A. D.; Kostura, M. J.; Miller, D. K.; Molineaux, S. M.; Weidner, J. R.; Aunins, J.; Elliston, K. O.; Ayala, J. M.; Casano, F. J.; Chin, J.; Ding, G. J.-F.; Egger, L. A.; Gaffney, E. P.; Limjoco, G.; Palyha, O. C.; Raju, S. M.; Rolando, A. M.; Salley, J. P.; Yamin, T.-T.; Lee, T. D.; Shively, J. E.; MacCross, M.; Mumford, R. A.; Schmidt, J. A.; Tocci, M. J.: A novel heterodimeric cysteine protease is

required for interleukin-1-beta processing in monocytes.
Nature 356: 768-774, 1992.

[27024] 576. Blakemore, A. I. F.; Cox, A.; Gonzalez, A.-M.; Maskill, J. K.; Hughes, M. E.; Wilson, R. M.; Ward, J. D.; Duff, G. W.: Interleukin-1 receptor antagonist allele (IL1RN*2) associated with nephropathy in diabetes mellitus. Hum. Genet. 97: 369-374, 1996.

[27025] 577. Blakemore, A. I. F.; Tarlow, J. K.; Cork, M. J.; Gordon, C.; Emery, P.; Duff, G. W.: Interleukin-1 receptor antagonist gene polymorphism as a disease severity factor in systemic lupus erythematosus. Arthritis Rheum. 37: 1380-1385, 1994.

[27026] 578. Carter, D. B.; Deibel, M. R., Jr.; Dunn, C. J.; Tomich, C.-S. C.; Laborde, A. L.; Slightom, J. L.; Berger, A. E.; Bienkowski, M. J.; Sun, F. F.; McEwan, R. N.; Harris, P. K. W.; Yem, A. W.; Waszak, G. A.; Chosay, J. G.; Sieu, L. C.; Hardee, M. M.; Zurcher-Neely, H. A.; Reardon, I. M.; Henrikson, R. L.; Truesdell, S. E.; Shelly, J. A.; Eessalu, T. E.; Taylor, B. M.; Tracey, D. E.: Purification, cloning, expression and biological characterization of an interleukin-1 receptor antagonist protein. Nature 344: 633-638, 1990.

[27027] 579. Dinarello, C. A.; Wolff, S. M.: The role of interleukin-1

in disease. *NewEng. J. Med.* 328: 106–113, 1993.

- [27028] 580.El-Omar, E. M.; Carrington, M.; Chow, W.-H.; McColl, K. E. L.;Bream, J. H.; Young, H. A.; Herrera, J.; Lissowska, J.; Yuan, C.-C.;Rothman, N.; Lanyon, G.; Martin, M.; Fraumeni, J. F., Jr.; Rabkin,C. S.: Interleukin-1 polymorphisms associated with increased riskof gastric cancer. *Nature* 404: 398–402, 2000.
- [27029] 581.Gabay, C.; Smith, M. F., Jr.; Eidlen, D.; Arend, W. P.: Interleukin1 receptor antagonist (IL-1Ra) is an acute-phase protein. *J. Clin.Invest.* 99: 2930–2940, 1997.
- [27030] 582.Langdahl, B. L.; Lokke, E.; Carstens, M.; Stenkjaer, L. L.; Eriksen,E. F.: Osteoporotic fractures are associated with an 86-base pairrepeat polymorphism in the interleukin-1-receptor antagonist genebut not with polymorphisms in the interleukin-1 beta gene. *J. BoneMiner. Res.* 15: 402–414, 2000.
- [27031] 583.Fischer-Colbrie, R.; Hagn, C.; Schober, M.: Chromogranins A, B,and C: widespread constituents of secretory vesicles. *Ann. N.Y. Acad.Sci.* 493: 120–134, 1987.
- [27032] 584.Mahata, S. K.; Kozak, C. A.; Szpirer, J.; Szpirer, C.; Modi, W.S.; Gerdes, H.-H.; Huttner, W. B.; O'Connor, D. T.: Dispersion ofchromogranin/secretogranin secretory protein family loci in mammaliangenomes. *Genomics* 33:

135–139, 1996.

- [27033] 585. Rosa, P.; Zanini, A.: Characterization of adeno-physiologic polypeptides by two-dimensional gel electrophoresis. II. Sulfated and glycosylated polypeptides. *Molec. Cell. Endocr.* 24: 181–193, 1981.
- [27034] 586. Rosa, P.; Zanini, A.: Purification of a sulfated secretory protein from the adenohypophysis: immunochemical evidence that similar macromolecules are present in other glands. *Europ. J. Cell Biol.* 31: 94–98, 1983.
- [27035] 587. Caughey, G. H.; Schaumberg, T. H.; Zerweck, E. H.; Butterfield, J. H.; Hanson, R. D.; Silverman, G. A.; Ley, T. J.: The human mast cell chymase gene (CMA1): mapping to the cathepsin G/granzyme gene cluster and lineage-restricted expression. *Genomics* 15: 614–620, 1993.
- [27036] 588. Caughey, G. H.; Zerweck, E. H.; Vanderslice, P.: Structure, chromosomal assignment, and deduced amino acid sequence of a human gene for mast cell chymase. *J. Biol. Chem.* 266: 12956–12963, 1991.
- [27037] 589. Ju, H.; Gros, R.; You, X.; Tsang, S.; Husain, M.; Rabinovitch, M.: Conditional and targeted overexpression of vascular chymase causes hypertension in transgenic mice. *Proc. Nat. Acad. Sci.* 98: 7469–7474, 2001.
- [27038] 590. Urata, H.; Boehm, K. D.; Philip, A.; Kinoshita, A.;

Gabrovsek,J.; Bumpus, F. M.; Husain, A.: Cellular localization and regional distribution of an angiotensin II-forming chymase in the heart. J.Clin. Invest. 91: 1269–1281, 1993.

[27039] 591.Anzick, S. L.; Kononen, J.; Walker, R. L.; Azorsa, D. O.; Tanner,M. M.; Guan, X.–Y.; Sauter, G.; Kallioniemi, O.–P.; Trent, J. M.;Meltzer, P. S.: ALB1, a steroid receptor coactivator amplified inbreast and ovarian cancer. Science 277: 965–968, 1997.

[27040] 592.Bulavin, D. V.; Demidov, O. N.; Saito, S.; Kauraniemi, P.; Phillips,C.; Amundson, S. A.; Ambrosino, C.; Sauter, G.; Nebreda, A. R.; Anderson,C. W.; Kallioniemi, A.; Fornace, A. J., Jr.; Appella, E.: Amplification of PPM1D in human tumors abrogates p53 tumor-suppressor activity. Nature-Genet. 31: 210–215, 2002.

[27041] 593.Reguigne, I.; James, M. R.; Richard, C. W., III; Mollicone, R.;Seawright, A.; Lowe, J. B.; Oriol, R.; Couillin, P.: The gene encoding myeloid alpha-3-fucosyltransferase (FUT4) is located between D11S388 and D11S919 on 11q21. Cytogenet. Cell Genet. 66: 104–106, 1994.

[27042] 594.Tetteroo, P. A. T.; de Heij, H. T.; Van den Eijnden, D. H.; Visser,F. J.; Schoenmaker, E.; Geurts van Kessel, A. H. M.: A GDP-fucose:(Gal-beta-1-to-4)GlcNAc alpha-1-to-3-fucosyltransf

erase activity is correlated with the presence of human chromosome 11 and the expression of the Le(x), Le(y), and sialyl-Le(x) antigens in human-mouse cell hybrids. J. Biol. Chem. 262:15984-15989, 1987.

- [27043] 595. Chang, M.-L.; Eddy, R. L.; Shows, T. B.; Lau, J. T. Y.: Three genes that encode human beta-galactoside alpha-2,3-sialyltransferases. Structural analysis and chromosomal mapping studies. Glycobiology 5:319-325, 1995.
- [27044] 596. de Heij, H. T.; Tetteroo, P. A. T.; Geurts van Kessel, A. H. M.; Schoenmaker, E.; Visser, F. J.; van den Eijnden, D. H.: Specific expression of a myeloid-associated CMP-NeuAc:Gal-beta-1-3GalNAc-alpha-R-alpha-2-3-sialyltransferase and the sialyl-X determinant in myeloid human-mouse cell hybrids containing human chromosome 11. Cancer Res. 48: 1489-1493, 1988.
- [27045] 597. Kitagawa, H.; Mattei, M.-G.; Paulson, J. C.: Genomic organization and chromosomal mapping of the Gal-beta-1,3GalNAc/Gal-beta-1,4GlcNAc alpha-2,3-sialyltransferase. J. Biol. Chem. 271: 931-938, 1996.
- [27046] 598. Regan, J. W.; Kobilka, T. S.; Yang-Feng, T. L.; Caron, M. G.; Lefkowitz, R. J.; Kobilka, B. K.: Cloning and expression of a human kidney cDNA for an alpha-2-adrenergic

receptor subtype. *Proc. Nat. Acad. Sci.* 85:6301–6305, 1988.

[27047] 599. Heinonen, P.; Koulu, M.; Pesonen, U.; Karvonen, M. K.; Rissanen, A.; Laakso, M.; Valve, R.; Uusitupa, M.; Scheinin, M.: Identification of a three-amino acid deletion in the alpha-2B-adrenergic receptor that is associated with reduced basal metabolic rate in obese subjects. *J. Clin. Endocr. Metab.* 84: 2429–2433, 1999.

[27048] 600. Lomasney, J. W.; Lorenz, W.; Allen, L. F.; King, K.; Regan, J. W.; Yang-Feng, T. L.; Caron, M. G.; Lefkowitz, R. J.: Expansion of the alpha-2-adrenergic receptor family: cloning and characterization of a human alpha-2-adrenergic receptor subtype, the gene for which is located on chromosome 2. *Proc. Nat. Acad. Sci.* 87: 5094–5098, 1990.

[27049] 601. Sellar, G. C.; Jordon, S. A.; Bickmore, W. A.; Fantes, J. A.; van Heyningen, V.; Whitehead, A. S.: The human serum amyloid A protein (SAA) superfamily gene cluster: mapping to chromosome 11p15.1 by physical and genetic linkage analysis. *Genomics* 19: 221–227, 1994.

[27050] 602. Steel, D. M.; Sellar, G. C.; Uhlar, C. M.; Simon, S.; DeBeer, F. C.; Whitehead, A. S.: A constitutively expressed serum amyloid A protein gene (SAA4) is closely linked to,

and shares structural similarities with, an acute-phase serum amyloid A protein gene (SAA1). *Genomics* 16:447–454, 1993.

- [27051] 603. de Beer, M. C.; de Beer, F. C.; Gerardot, C. J.; Cecil, D. R.; Webb, N. R.; Goodson M. L.; Kindy, M. S.: Structure of the mouse Saa4 gene and its linkage to the serum amyloid A gene family. *Genomics* 34:139–142, 1996.
- [27052] 604. Watson, G.; Coade, S.; Woo, P.: Analysis of the genomic and derived protein structure of a novel human serum amyloid A gene, SAA4. *Scand.J. Immun.* 36: 703–712, 1992.
- [27053] 605. Whitehead, A. S.; DeBeer, M. C.; Steel, D. M.; Rits, M.; Lelias, J. M.; Lane, W. S.; DeBeer, F. C.: Identification of novel members of the serum amyloid A protein superfamily as constitutive apolipoproteins of high density lipoprotein. *J. Biol. Chem.* 267: 3862–3867, 1992.
- [27054] 606. Baek, S. H.; Ohgi, K. A.; Rose, D. W.; Koo, E. H.; Glass, C. K.; Rosenfeld, M. G.: Exchange of N-CoR corepressor and Tip60 coactivator complexes links gene expression by NF-kappa-B and beta-amyloid precursor protein. *Cell* 110: 55–67, 2002.
- [27055] 607. Cao, X.; Sudhof, T. C.: A transcriptionally active complex of APP with Fe65 and histone acetyltransferase Tip60.

Science 293: 115–120,2001.

- [27056] 608.Gunning, P.; Ponte, P.; Kedes, L.; Eddy, R.; Shows, T.: Chromosomallocation of the co-expressed human skeletal and cardiac actin genes. Proc.Nat. Acad. Sci. 81: 1813–1817, 1984.
- [27057] 609.Shows, T.; Eddy, R. L.; Haley, L.; Byers, M.; Henry, M.; Gunning,P.; Ponte, P.; Kedes, L.: The coexpressed genes for human alpha (ACTA)and cardiac actin (ACTC) are on chromosomes 1 and 15, respectively.(Abstract) Cytogenet. Cell Genet. 37: 583 only, 1984.
- [27058] 610.Ueyama, H.; Inazawa, J.; Ariyama, T.; Nishino, H.; Ochiai, Y.;Ohkubo, I.; Miwa, T.: Reexamination of chromosomal loci of humanmuscle actin genes by fluorescence in situ hybridization. Jpn. J.Hum. Genet. 40: 145–148, 1995.
- [27059] 611.Nasir, J.; Lin, B.; Bucan, M.; Koizumi, T.; Nadeau, J. H.; Hayden,M. R.: The murine homologues of the Huntington disease gene (Hdh)and the alpha-adducin gene (Add1) map to mouse chromosome 5 within a region of conserved synteny with human chromosome 4p16.3. Genomics 22:198–201, 1994.
- [27060] 612.Peters, L. L.; Birkenmeier, C. S.; Bronson, R. T.; White, R. A.;Lux, S. E.; Otto, E.; Bennett, V.; Higgins, A.; Barker, J. E.: Purkinjecell degeneration associated with erythroid

ankyrin deficiency innb/nb mice. J. Cell Biol. 114:
1233–1241, 1991.

[27061] 613. Taylor, S. A. M.; Snell, R. G.; Buckler, A.; Ambrose, C.; Duyao, M.; Church, D.; Lin, C. S.; Altherr, M.; Bates, G. P.; Groot, N.; Barnes, G.; Shaw, D. J.; Lehrach, H.; Wasmuth, J. J.; Harper, P. S.; Housman, D. E.; MacDonald, M. E.; Gusella, J. F.: Cloning of the α -adducin gene from the Huntington's disease candidate region of chromosome 4 by exon amplification. Nature Genet. 2: 223–227, 1992.

[27062] 614. Gilligan, D. M.; Lieman, J.; Bennett, V.: Assignment of the human β -adducin gene (ADD2) to 2p13–p14 by in situ hybridization. Genomics 28: 610–612, 1995.

[27063] 615. Muro, A. F.; Marro, M. L.; Gajovic, S.; Porro, F.; Luzzatto, L.; Baralle, F. E.: Mild spherocytic hereditary elliptocytosis and altered levels of α - and γ -adducins in β -adducin-deficient mice. Blood 95: 3978–3985, 2000.

[27064] 616. Tisminetzky, S.; Devescovi, G.; Tripodi, G.; Muro, A.; Bianchi, G.; Colombi, M.; Moro, L.; Barlati, S.; Tuteja, R.; Baralle, F. E.: Genomic organisation and chromosomal localisation of the gene encoding human β adducin. Gene 167: 313–316, 1995.

[27065] 617. White, R. A.; Angeloni, S. V.; Pasztor, L. M.: Chromo-

somal localization of the beta-adducin gene to mouse chromosome 6 and human chromosome 2. *Mammalian Genome* 6: 741–743, 1995.

- [27066] 618. Abbott, C. M.; Skidmore, C. J.; Searle, A. G.; Peters, J.: Deficiency of adenosine deaminase in the wasted mouse. *Proc. Nat. Acad. Sci.* 83:693–695, 1986.
- [27067] 619. Adrian, G. S.; Wiginton, D. A.; Hutton, J. J.: Characterization of normal and mutant adenosine deaminase messenger RNAs by translation and hybridization to a cDNA probe. *Hum. Genet.* 68: 169–172, 1984.
- [27068] 620. Adrian, G. S.; Wiginton, D. A.; Hutton, J. J.: Structure of adenosine deaminase mRNAs from normal and adenosine deaminase-deficient human cell lines. *Molec. Cell. Biol.* 4: 1712–1717, 1984.
- [27069] 621. Aitken, D. A.; Ferguson-Smith, M. A.: Investigation of the intrachromosomal position of the ADA locus on chromosome 20 by gene dosage studies. *Cytogenet. Cell Genet.* 22: 514–517, 1978.
- [27070] 622. Aitken, D. A.; Kleijer, W. J.; Niermeijer, M. F.; Herbschleb-Voogt, E.; Galjaard, H.: Prenatal detection of a probable heterozygote for ADA deficiency and severe combined immunodeficiency disease using a microradioassay. *Clin. Genet.* 17: 293–298, 1980.

- [27071] 623. Aiuti, A.; Slavin, S.; Aker, M.; Ficara, F.; Deola, S.; Mortellaro, A.; Morecki, S.; Andolfi, G.; Tabucchi, A.; Carlucci, F.; Marinello, E.; Cattaneo, F.; Vai, S.; Servida, P.; Miniero, R.; Roncarolo, M.G.; Bordignon, C.: Correction of ADA-SCID by stem cell gene therapy combined with non-myeloablative conditioning. *Science* 296: 2410-2413, 2002.
- [27072] 624. Akeson, A. L.; Wiginton, D. A.; Dusing, M. R.; States, J. C.; Hutton, J. J.: Mutant human adenosine deaminase alleles and their expression by transfection into fibroblasts. *J. Biol. Chem.* 263: 16291-16296, 1988.
- [27073] 625. Akeson, A. L.; Wiginton, D. A.; Hutton, J. J.: Normal and mutant human adenosine deaminase genes. *J. Cell. Biochem.* 39: 217-228, 1989.
- [27074] 626. Akeson, A. L.; Wiginton, D. A.; States, J. C.; Perme, C. M.; Dusing, M. R.; Hutton, J. J.: Mutations in the human adenosine deaminase gene that affect protein structure and RNA splicing. *Proc. Nat. Acad. Sci.* 84: 5947-5951, 1987.
- [27075] 627. Arredondo-Vega, F. X.; Kurtzberg, J.; Chaffee, S.; Santisteban, I.; Reisner, E.; Povey, M. S.; Hershfield, M. S.: Paradoxical expression of adenosine deaminase in T cells cultured from a patient with adenosine deaminase defi-

ciency and combined immunodeficiency. *J. Clin. Invest.* 86:444–452, 1990.

- [27076] 628. Arredondo-Vega, F. X.; Santisteban, I.; Kelly, S.; Schlossman, C. M.; Umetsu, D. T.; Hershfield, M. S.: Correct splicing despite mutation of the invariant first nucleotide of a 5-prime splice site: a possible basis for disparate clinical phenotypes in siblings with adenosine deaminase deficiency. *Am. J. Hum. Genet.* 54: 820–830, 1994.
- [27077] 629. Arredondo-Vega, F. X.; Santisteban, I.; Richard, E.; Bali, P.; Koleilat, M.; Loubser, M.; Al-Ghonaïm, A.; Al-Helali, M.; Hershfield, M. S.: Adenosine deaminase deficiency with mosaicism for a 'second-site suppressor' of a splicing mutation: decline in revertant T lymphocytes during enzyme replacement therapy. *Blood* 99: 1005–1013, 2002.
- [27078] 630. Berkvens, T. M.; Gerritsen, E. J. A.; Oldenburg, M.; Breukel, C.; Wijnen, J. T.; van Ormondt, H.; Vossen, J. M.; van der Eb, A. J.; Meera Khan, P.: Severe combined immune deficiency due to a homozygous 3.2-kb deletion spanning the promoter and first exon of the adenosine deaminase gene. *Nucleic Acids Res.* 15: 9365–9378, 1987.
- [27079] 631. Lai, L.; Hart, I.; Patterson, D.: Human chromosome 1

corrects the defect in the CHO mutant (Ade⁻H) deficient in a branch point enzyme in purine de novo biosynthesis.

(Abstract) Cytogenet. Cell Genet. 51:1028 only, 1989.

[27080] 632. Lai, L.-W.; Hart, I. M.; Patterson, D.: A gene correcting the defect in the CHO mutant Ade⁽⁻⁾H, deficient in a branch point enzyme (adenylosuccinate synthetase) of de novo purine biosynthesis, is located on the long arm of chromosome 1. Genomics 9: 322–328, 1991.

[27081] 633. Powell, S. M.; Zalkin, H.; Dixon, J. E.: Cloning and characterization of the cDNA encoding human adenylosuccinate synthetase. FEBS Lett. 303:4–10, 1992.

[27082] 634. Parma, J.; Stengel, D.; Gannage, M.-H.; Poyard, M.; Barouki, R.; Hanoune, J.: Sequence of a human brain adenylyl cyclase partial cDNA: evidence for a consensus cyclase domain. Biochem. Biophys. Res. Commun. 179:455–462, 1991.

[27083] 635. Stengel, D.; Parma, J.; Gannage, M.-H.; Roeckel, N.; Mattei, M.-G.; Barouki, R.; Hanoune, J.: Different chromosomal localization of two adenylyl cyclase genes expressed in human brain. Hum. Genet. 90:126–130, 1992.

[27084] 636. Edelhoff, S.; Villacres, E. C.; Storm, D. R.; Disteche, C. M.: Mapping of adenylyl cyclase genes type I, II, III, IV, V, and VI in mouse. Mammalian Genome 6: 111–113, 1995.

- [27085] 637. Gaudin, C.; Homcy, C. J.; Ishikawa, Y.: Mammalian adenylyl cyclase family members are randomly located on different chromosomes. *Hum. Genet.* 94: 527–529, 1994.
- [27086] 638. Wong, S. T.; Trinh, K.; Hacker, B.; Chan, G. C. K.; Lowe, G.; Gaggari, A.; Xia, Z.; Gold, G. H.; Storm, D. R.: Disruption of the type III adenylyl cyclase gene leads to peripheral and behavioral anosmia in transgenic mice. *Neuron* 27: 487–497, 2000.
- [27087] 639. Abdel-Majid, R. M.; Leong, W. L.; Schalkwyk, L. C.; Smallman, D. S.; Wong, S. T.; Storm, D. R.; Fine, A.; Dobson, M. J.; Guernsey, D. L.; Neumann, P. E.: Loss of adenylyl cyclase I activity disrupts patterning of mouse somatosensory cortex. *Nature Genet.* 19: 289–291, 1998.
- [27088] 640. Villacres, E. C.; Xia, Z.; Bookbinder, L. H.; Edelhoff, S.; Distèche, C. M.; Storm, D. R.: Cloning, chromosomal mapping, and expression of human fetal brain type I adenylyl cyclase. *Genomics* 16: 473–478, 1993.
- [27089] 641. Welker, E.; Armstrong-James, M.; Bronchti, G.; Ourednik, W.; Gheorghita-Baechler, F.; Dubois, R.; Guernsey, D. L.; Van der Loos, H.; Neumann, P. E.: Altered sensory processing in the somatosensory cortex of the mutant mouse barrelless. *Science* 271: 1864–1867, 1996.
- [27090] 642. Xu, Y.; Carr, L. G.; Bosron, W. F.; Li, T.-K.; Edenberg,

H. J.: Genotyping of human alcohol dehydrogenases at the ADH2 and ADH3 loci following DNA sequence amplification. *Genomics* 2: 209–214, 1988.

[27091] 643. Azevedo, E. S.; Da Silva, M. C. B. O.; Tavares-Neto, J.: Human alcohol dehydrogenase ADH 1, ADH 2 and ADH 3 loci in a mixed population of Bahia, Brazil. *Ann. Hum. Genet.* 39: 321–327, 1976.

[27092] 644. Hines, L. M.; Stampfer, M. J.; Ma, J.; Gaziano, J. M.; Ridker, P. M.; Hankinson, S. E.; Sacks, F.; Rimm, E. B.; Hunter, D. J.: Genetic variation in alcohol dehydrogenase and the beneficial effect of moderate alcohol consumption on myocardial infarction. *New Eng. J. Med.* 344:549–555, 2001.

[27093] 645. Holmes, R. S.: Genetics and ontogeny of alcohol dehydrogenase isozymes in the mouse: evidence for a cis-acting regulator gene (Adt-I) controlling C(2) isozyme expression in reproductive tissues and close linkage of Adh-3 and Adt-I on chromosome 3. *Biochem. Genet.* 17:461–472, 1979.

[27094] 646. Hoog, J.-O.; Heden, L.-O.; Larsson, K.; Jornvall, H.; von Bahr-Lindstrom, H.: The gamma-1 and gamma-2 subunits of human liver alcohol dehydrogenase: cDNA structures, two amino acid replacements, and compatibil-

ity with changes in the enzymatic properties. *Europ. J. Biochem.* 159: 215–218, 1986.

- [27095] 647. Morris, D. J.; Willem, P.; dos Santos, M.; Povey, S.; Jenkins, T.: A new chromosome 4q marker, D4S138, closely linked to the ADH3 locus. (Abstract) *Cytogenet. Cell Genet.* 51: 1047–1048, 1989.
- [27096] 648. Zgombic-Knight, M.; Deltour, L.; Haselbeck, R. J.; Foglio, M. H.; Duester, G.: Gene structure and promoter for Adh3 encoding mouse class IV alcohol dehydrogenase (retinol dehydrogenase). *Genomics* 41: 105–109, 1997.
- [27097] 649. Badawi, A. F.; Hirvonen, A.; Bell, D. A.; Lang, N. P.; Kadlubar, F. F.: Role of aromatic amine acetyltransferases, NAT1 and NAT2, in carcinogen–DNA adduct formation in the human urinary bladder. *Cancer Res.* 55: 5230–5237, 1995.
- [27098] 650. Bell, D. A.; Badawi, A. F.; Lang, N. P.; Ilett, K. P.; Kadlubar, F. F.; Hirvonen, A.: Polymorphism in the N-acetyltransferase 1 (NAT1) polyadenylation signal: association of NAT1*10 allele with higher N-acetylation activity in bladder and colon tissue. *Cancer Res.* 55: 5226–5229, 1995.
- [27099] 651. Bell, D. A.; Stephens, E. A.; Castranio, T.; Umbach, D. M.; Watson, M.; Deakin, M.; Elder, J.; Hendrickse, C.; Dun-

can, H.; Strange, R.C.: Polyadenylation polymorphism in the acetyltransferase 1 gene(NAT1) increases risk of colorectal cancer. *Cancer Res.* 55: 3537–3542,1995.

[27100] 652.Blum, M.; Grant, D. M.; McBride, W.; Heim, M.; Meyer, U. A.: Human arylamine N-acetyltransferase genes: isolation, chromosomal localization, and functional expression. *DNA Cell Biol.* 9: 193–203, 1990.

[27101] 653.Bouchardy, C.; Mitrunen, K.; Wikman, H.; Husgafvel-Pursiainen, K.; Dayer, P.; Benhamou, S.; Hirvonen, A.: N-acetyltransferase NAT1 and NAT2 genotypes and lung cancer risk. *Pharmacogenetics* 8: 291–298,1998.

[27102] 654.Butcher, N. J.; Ilett, K. F.; Minchin, R. F.: Functional polymorphism of the human arylamine N-acetyltransferase type 1 gene caused by C190T and G560A mutations. *Pharmacogenetics* 8: 67–72, 1998.

[27103] 655.Doll, M. A.; Jiang, W.; Deitz, A. C.; Rustan, T. D.; Hein, D. W.: Identification of a novel allele at the human NAT1 acetyltransferase locus. *Biochem. Biophys. Res. Commun.* 233: 584–591, 1997.

[27104] 656.Evans, D. A. P.: Personal Communication. Saudi Arabia 6/1/1998.

[27105] 657.Evans, D. A. P.: *Genetic Factors in Drug Therapy: Clinical and Molecular Pharmacogenetics*. Cambridge: Cam-

bridge Univ. Press , 1993.Pp. 211–305.

- [27106] 658.Hickman, D.; Risch, A.; Buckle, V.; Spurr, N. K.; Jeremiah, S.J.; McCarthy, A.; Sim, E.: Chromosomal localization of human genesfor arylamine N–acetyltransferase. *Biochem. J.* 297: 441–445, 1994.
- [27107] 659.Matas, N.; Thygesen, P.; Stacey, M.; Risch, A.; Sim, E.: MappingAAC1, AAC2 and AACP, the genes for arylamine N–acetyltransferases,carcinogen metabolising enzymes on human chromosome 8p22, a regionfrequently deleted in tumours. *Cytogenet. Cell Genet.* 77: 290–295,1997.
- [27108] 660.Mattano, S. S.; Erickson, R. P.; Nesbitt, M. N.; Weber, W. W.: Linkage of Nat and Es–1 in the mouse and development of strainscongenic for N–acetyltransferase. *J. Hered.* 79: 430–433, 1988.
- [27109] 661.Moisio, A.–L.; Sistonen, P.; Mecklin, J.–P.; Jarvinen, H.; Peltomaki,P.: Genetic polymorphisms in carcinogen metabolism and their associationto hereditary nonpolypoid colon cancer. *Gastroenterology* 115: 1387–1394,1998.
- [27110] 662.Sim, E.; Payton, M.; Noble, M.; Minchin, R.: An update on genetic,structural and functional studies of arylamine N–acetyltransferasesin eucaryotes and procaryotes. *Hum. Molec. Genet.* 9: 2435–2441,2000.
- [27111] 663.Smelt, V. A.; Upton, A.; Adjaye, J.; Payton, M. A.;

Boukouvala, S.; Johnson, N.; Mardon, H. J.; Sim, E.: Expression of arylamine N-acetyltransferases in pre-term placentas and in human pre-implantation embryos. *Hum. Molec. Genet.* 9: 1101–1107, 2000.

[27112] 664. Vatsis, K. P.; Martell, K. J.; Weber, W. W.: Diverse point mutations in the human gene for polymorphic N-acetyltransferase. *Proc. Nat. Acad. Sci.* 88: 6333–6337, 1991.

[27113] 665. Vatsis, K. P.; Weber, W. W.: Structural heterogeneity of Caucasian N-acetyltransferase at the NAT1 gene locus. *Arch. Biochem. Biophys.* 301: 71–76, 1993.

[27114] 666. Vatsis, K. P.; Weber, W. W.; Bell, D. A.; Dupret, J.-M.; Price-Evans, D. A.; Grant, D. M.; Hein, D. W.; Lin, H. J.; Meyer, U. A.; Relling, M. V.; Sim, E.; Suzuki, T.; Yamazoe, Y.: Nomenclature for N-acetyltransferases. *Pharmacogenetics* 5: 1–17, 1995.

[27115] 667. Cheng, A. M.; Saxton, T. M.; Sakai, R.; Kulkarni, S.; Mbamalu, G.; Vogel, W.; Tortorice, C. G.; Cardiff, R. D.; Cross, J. C.; Muller, W. J.; Pawson, T.: Mammalian Grb2 regulates multiple steps in embryonic development and malignant transformation. *Cell* 95: 793–803, 1998.

[27116] 668. Clark, S. G.; Stern, M. J.; Horvitz, H. R.: *C. elegans* cell-signalling gene *sem-5* encodes a protein with SH2 and

SH3 domains. *Nature* 356:340–344, 1992.

- [27117] 669. Hill, R. J.; Zozulya, S.; Lu, Y.-L.; Ward, K.; Gishizky, M.; Jallal, B.: The lymphoid protein tyrosine phosphatase Lyp interacts with the adaptor molecule Grb2 and functions as a negative regulator of T-cell activation. *Exp. Hemat.* 30: 237–244, 2002.
- [27118] 670. Huebner, K.; Kastury, K.; Druck, T.; Salcini, A. E.; Lanfrancone, L.; Pelicci, G.; Lowenstein, E.; Li, W.; Park, S.-H.; Cannizzaro, L.; Pelicci, P. G.; Schlessinger, J.: Chromosome locations of genes encoding human signal transduction adapter proteins, Nck (NCK), Shc (SHC1), and Grb2 (GRB2). *Genomics* 22: 281–287, 1994.
- [27119] 671. Lowenstein, E. J.; Daly, R. J.; Batzer, A. G.; Li, W.; Margolis, B.; Lammers, R.; Ullrich, A.; Skolnik, E. Y.; Bar-Sagi, D.; Schlessinger, J.: The SH2 and SH3 domain-containing protein GRB2 links receptor tyrosine kinases to ras signaling. *Cell* 70: 431–442, 1992.
- [27120] 672. Matuoka, K.; Shibasaki, F.; Shibata, M.; Takenawa, T.: Ash/Grb2, a SH2/SH3-containing protein, couples to signaling for mitogenesis and cytoskeletal reorganization by EGF and PDGF. *EMBO J.* 12: 3467–3473, 1993.
- [27121] 673. Matuoka, K.; Shibata, M.; Yamakawa, A.; Takenawa, T.: Cloning of ASH, a ubiquitous protein composed of one

Src homology region (SH)2 and two SH3 domains, from human and rat cDNA libraries. *Proc. Nat. Acad. Sci.* 89: 9015–9019, 1992.

- [27122] 674. Yulug, I. G.; Egan, S. E.; See, C. G.; Fisher, E. M. C.: Mapping GRB2, a signal transduction gene in the human and the mouse. *Genomics* 22:313–318, 1994.
- [27123] 675. Tangye, S. G.; Phillips, J. H.; Lanier, L. L.; Nichols, K. E.: Cutting edge: functional requirement for SAP in 2B4-mediated activation of human natural killer cells as revealed by the X-linked lymphoproliferative syndrome. *J. Immun.* 165: 2932–2936, 2000.
- [27124] 676. Gras, M. P.; Laabi, Y.; Linares-Cruz, G.; Blondel, M. O.; Rigaut, J. P.; Brouet, J. C.; Leca, G.; Haguenaue-Tsapis, R.; Tsapis, A.: BCMAp: an integral membrane protein in the Golgi apparatus of human mature B lymphocytes. *Int. Immun.* 7: 1093 only, 1995.
- [27125] 677. Hatzoglou, A.; Roussel, J.; Bourgeade, M.-F.; Rogier, E.; Madry, C.; Inoue, J.; Devergne, O.; Tsapis, A.: TNF receptor family member BCMA (B cell maturation) associates with TNF receptor-associated factor (TRAF) 1, TRAF2, and TRAF3 and activates NF- κ B, Elk-1, c-Jun N-terminal kinase, and p38 mitogen-activated protein kinase. *J. Immun.* 165:1322–1330, 2000.

- [27126] 678.Laabi, Y.; Gras, M. P.; Carbonnel, F.; Brouet, J. C.; Berger, R.;Larsen, C. J.; Tsapis, A.: A new gene, BCM, on chromosome 16 is fused to the interleukin 2 gene by a t(4;16)(q26;p13) translocation in a malignant T cell lymphoma. *EMBO J.* 11: 3897–3904, 1992.
- [27127] 679.Bhatia, K.; Huppi, K.; McKeithan, T.; Siwarski, D.; Mushinski, J. F.; Magrath, I.: Mouse bcl-3: cDNA structure, mapping and stage-dependent expression in B lymphocytes. *Oncogene* 6: 1569–1573, 1991.
- [27128] 680.Franzoso, G.; Bours, V.; Park, S.; Tomita-Yamaguchi, M.; Kelly, K.; Siebenlist, U.: The candidate oncoprotein Bcl-3 is an antagonist of p50/NF- κ B-mediated inhibition. *Nature* 359: 338–342, 1992.
- [27129] 681.Mitchell, T. C.; Hildeman, D.; Kedl, R. M.; Teague, T. K.; Schaefer, B. C.; White, J.; Zhu, Y.; Kappler, J.; Marrack, P.: Immunological adjuvants promote activated T cell survival via induction of Bcl-3. *Nature Immun.* 2: 397–402, 2001.
- [27130] 682.El Nemer, W.; Rahuel, C.; Colin, Y.; Gane, P.; Cartron, J. P.; Le Van Kim, C.: Organization of the human LU gene and molecular basis of the Lu(a)/Lu(b) blood group polymorphism. *Blood* 89: 4608–4616, 1997.
- [27131] 683.Frandson, S.; Atkins, C. J.; Moulds, M.; Poole, J.;

Crawford, M.N.; Tippett, P.: Anti-Au(b): the antithetical antibody to anti-Au(a). *VoxSang.* 56: 54–56, 1989.

[27132] 684.Lewis, M.; Kaita, H.; Chown, B.; Giblett, E. R.; Anderson, J.;Cote, G. B.: The Lutheran and Secretor loci: genetic linkage analysis. *Am.J. Hum. Genet.* 29: 101–106, 1977.

[27133] 685.Lewis, M.; Kaita, H.; Coghlan, G.; Philipps, S.; Belcher, E.; McAlpine,P. J.; Coopland, G. R.; Woods, R. A.: The chromosome 19 linkage groupLDLR, C3, LW, APOC2, LU, SE in man. *Ann. Hum. Genet.* 52: 137–144,1988.

[27134] 686.Lewis, M.; Kaita, H.; Giblett, E. R.; Anderson, J. E.: Lods forLu:Se and other loci. *Cytogenet. Cell Genet.* 22: 627–628, 1978.

[27135] 687.Mohr, J.: Search for linkage between Lutheran blood group andother hereditary characters. *Acta Path. Microbiol. Scand.* 28: 207–210,1951.

[27136] 688.Parsons, S. F.; Lee, G.; Spring, F. A.; Willig, T.–N.; Peters,L. L.; Gimm, J. A.; Tanner, M. J. A.; Mohandas, N.; Anstee, D. J.;Chasis, J. A.: Lutheran blood group glycoprotein and its newly characterizedmouse homologue specifically bind alpha–5 chain–containing human lamininwith high affinity. *Blood* 97: 312–320, 2001.

[27137] 689.Parsons, S. F.; Mallinson, G.; Daniels, G. L.; Green, C. A.; Smythe,J. S.; Anstee, D. J.: Use of domain–deletion mu–

tants to locate Lutheran blood group antigens to each of the five immunoglobulin superfamily domains of the Lutheran glycoprotein: elucidation of the molecular basis of the Lu(a)/Lu(b) and the Au(a)/Au(b) polymorphisms. Blood 89:4219–4225, 1997.

- [27138] 690. Parsons, S. F.; Mallinson, G.; Holmes, C. H.; Houlihan, J. M.; Simpson, K. L.; Mawby, W. J.; Spurr, N. K.; Warne, D.; Barclay, A.N.; Anstee, D. J.: The Lutheran blood group glycoprotein, another member of the immunoglobulin superfamily, is widely expressed in human tissues and is developmentally regulated in human liver. Proc. Nat. Acad. Sci. 92: 5496–5500, 1995.
- [27139] 691. Rahuel, C.; Le Van Kim, C.; Mattei, M. G.; Cartron, J. P.; Colin, Y.: A unique gene encodes splice forms of the B-cell adhesion molecule cell surface glycoprotein of epithelial cancer and of the Lutheran blood group glycoprotein. Blood 88: 1865–1872, 1996.
- [27140] 692. Salmon, C.; Salmon, D.; Liberge, G.; Andre, R.; Tippet, P.; Sanger, R.: Un nouvel antigene de groupe sanguin erythrocytaire present chez 80% des sujets de race blanche. Nouv. Rev. Franc. Hemat. 1: 649–661, 1961.
- [27141] 693. Zelinski, T.; Kaita, H.; Johnson, K.; Moulds, M.: Genetic evidence that the gene controlling Au(b) is located on

chromosome 19. *VoxSang.* 58: 126–128, 1990.

- [27142] 694. Anstee, D. J.: The blood group MNSs—active sialoglycoproteins. *Seminars Hemat.* 18: 13–31, 1981.
- [27143] 695. Bias, W. B.; Meyers, D. A.: Segregation and linkage analysis of the Stoltzfus blood group (SF). (Abstract) *Cytogenet. Cell Genet.* 25:137, 1979.
- [27144] 696. Blumenfeld, O. O.; Adamany, A. M.: Structural (glycophorins) of the human erythrocyte membrane. *Proc. Nat. Acad. Sci.* 75: 2727–2731, 1978.
- [27145] 697. Blumenfeld, O. O.; Huang, C.–H.: Molecular genetics of the glycophorin gene family, the antigens for MNSs blood groups: multiple gene rearrangements and modulation of splice site usage result in extensive diversification. *Hum. Mutat.* 6: 199–209, 1995.
- [27146] 698. Cook, P. J. L.; Lindenbaum, R. H.; Salonen, R.; de la Chapelle, A.; Daker, M. G.; Buckton, K. E.; Noades, J. E.; Tippett, P.: The MNSs blood groups of families with chromosome 4 rearrangements. *Ann. Hum. Genet.* 45: 39–47, 1981.
- [27147] 699. Cook, P. J. L.; Noades, J. E.; Lomas, C. G.; Buckton, K. E.; Robson, E. B.: Exclusion mapping illustrated by the MNSs blood group. *Ann. Hum. Genet.* 44: 61–73, 1980.
- [27148] 700. Cook, P. J. L.; Robson, E. B.; Buckton, K. E.; Slaughter,

C. A.; Gray, J. E.; Blank, C. E.; James, F. E.; Ridler, M. A. C.; Insley, J.; Hulten, M.: Segregation of ABO, AK-1 and ACON-S in families with abnormalities of chromosome 9. *Ann. Hum. Genet.* 41: 365-377, 1978.

[27149] 701. Divelbiss, J.; Shiang, R.; German, J.; Moore, J.; Murray, J. C.; Patil, S. R.: Refinement of the physical location of glycophorin A and beta fibrinogen using in situ hybridization and RFLP analysis. (Abstract) *Cytogenet. Cell Genet.* 51: 991, 1989.

[27150] 702. Evans, R. M.: The steroid and thyroid hormone receptor superfamily. *Science* 240:889-895, 1988.

[27151] 703. Bader, B. L.; Rayburn, H.; Crowley, D.; Hynes, R. O.: Extensive vasculogenesis, angiogenesis, and organogenesis precede lethality in mice lacking all alpha-V integrins. *Cell* 95: 507-519, 1998.

[27152] 704. Fernandez-Ruiz, E.; de Villena, F. P.-M.; Rodriguez de Cordoba, S.; Sanchez-Madrid, F.: Regional localization of the human vitronectin receptor alpha-subunit gene (VNRA) to chromosome 2q31-q32. *Cytogenet. Cell Genet.* 62: 26-28, 1993.

[27153] 705. Sims, M. A.; Field, S. D.; Barnes, M. R.; Shaikh, N.; Ellington, K.; Murphy, K. E.; Spurr, N.; Campbell, D. A.: Cloning and characterisation of ITGAV, the genomic se-

quence for human cell adhesion protein

(vitronectin)receptor alpha subunit, CD51. Cytogenet. Cell Genet. 89: 268–271,2000.

[27154] 706.Suzuki, S.; Argraves, W. S.; Pytela, R.; Arai, H.; Kru-
sius, T.;Pierschbacher, M. D.; Ruoslahti, E.: cDNA and
amino acid sequencesof the cell adhesion protein receptor
recognizing vitronectin reveala transmembrane domain
and homologies with other adhesion proteinreceptors.
Proc. Nat. Acad. Sci. 83: 8614–8618, 1986.

[27155] 707.Blachly–Dyson, E.; Baldini, A.; Litt, M.; McCabe, E. R.
B.; Forte,M.: Human genes encoding the voltage–de-
pendent anion channel (VDAC)of the outer mitochondrial
membrane: mapping and identification oftwo new iso-
forms. Genomics 20: 62–67, 1994.

[27156] 708.Blachly–Dyson, E.; Zambronicz, E. B.; Yu, W. H.;
Adams, V.; McCabe,E. R. B.; Adelman, J.; Colombini, M.;
Forte, M.: Cloning and functionalexpression in yeast of
two human isoforms of the outer mitochondrialmembrane
channel, the voltage–dependent anion channel. J. Biol.
Chem. 268:1835–1841, 1993.

[27157] 709.Huizing, M.; Ruitenbeek, W.; van den Heuvel, L. P.;
Dolce, V.;Iacobazzi, V.; Smeitink, J. A. M.; Palmieri, F.; Tri-
jbels, J. M. F.: Human mitochondrial transmembrane

metabolite carriers: tissue distribution and its implication for mitochondrial disorders. *J. Bioenerg. Biomembr.* 30:277–284, 1998.

- [27158] 710. Hromas, R.; Collins, S. J.; Hickstein, D.; Raskind, W.; Deaven, L. L.; O'Hara, P.; Hagen, F. S.; Kaushansky, K.: A retinoic acid-responsive human zinc finger gene, MZF-1, preferentially expressed in myeloid cells. *J. Biol. Chem.* 266: 14183–14187, 1991.
- [27159] 711. Morris, J. F.; Rauscher, F. J., III; Davis, B.; Klemsz, M.; Xu, D.; Tenen, D.; Hromas, R.: The myeloid zinc finger gene, MZF-1, regulates the CD34 promoter in vitro. *Blood* 86: 3640–3647, 1995.
- [27160] 712. Tommerup, N.; Aagaard, L.; Lund, C. L.; Boel, E.; Baxendale, S.; Bates, G. P.; Lehrach, H.; Vissing, H.: A zinc-finger gene ZNF141 mapping at 4p16.3/D4S90 is a candidate gene for the Wolf-Hirschhorn (4p-) syndrome. *Hum. Molec. Genet.* 2: 1571–1575, 1993.
- [27161] 713. Kromberg, J. G. R.; Jenkins, T.: Prevalence of albinism in the South African Negro. *S. Afr. Med. J.* 61: 383–386, 1982.
- [27162] 714. McAlpine, P. J.; Coopland, G.; Guy, C.; James, S.; Komarnicki, L.; MacDonald, M.; Stranc, L.; Lewis, M.; Philipps, S.; Coghlan, G.; Kaita, H.; Cox, D. W.; Guinto, E. R.;

MacGillivray, R.: Mapping the genes for erythrocytic alpha-spectrin 1 (SPTA1) and coagulation factor V (F5). (Abstract) Cytogenet. Cell Genet. 51: 1042, 1989.

[27163] 715. de Silva, H. V.; Harmony, J. A. K.; Stuart, W. D.; Gil, C. M.; Robbins, J.: Apolipoprotein J: structure and tissue distribution. Biochemistry 29:5380–5389, 1990.

[27164] 716. de Silva, H. V.; Stuart, W. D.; Park, Y. B.; Mao, S. J. T.; Gil, C. M.; Wetterau, J. R.; Busch, S. J.; Harmony, J. A. K.: Purification and characterization of apolipoprotein J. J. Biol. Chem. 265: 14292–14297, 1990.

[27165] 717. Dietzsch, E.; Murphy, B. F.; Kirschbaum, L.; Walker, I. D.; Garson, O. M.: Regional localization of the gene for clusterin (SP-40,40; gene symbol CLI) to human chromosome 8p12–p21. Cytogenet. Cell Genet. 61:178–179, 1992.

[27166] 718. Dragunow, M.; Preston, K.; Dodd, J.; Young, D.; Lawlor, P.; Christie, D.: Clusterin accumulates in dying neurons following status epilepticus. Molec. Brain Res. 32: 279–290, 1995.

[27167] 719. Duguid, J. R.; Bohmont, C. W.; Liu, N.; Tourtellotte, W. W.: Changes in brain gene expression shared by scrapie and Alzheimer disease. Proc. Nat. Acad. Sci. 86: 7260–7264, 1989.

- [27168] 720.Fink, T. M.; Zimmer, M.; Tschopp, J.; Etienne, J.; Jenne, D. E.;Lichter, P.: Human clusterin (CLI) maps to 8p21 in proximity to the lipoprotein lipase (LPL) gene. *Genomics* 16: 526–528, 1993.
- [27169] 721.Han, B. H.; DeMattos, R. B.; Dugan, L. L.; Kim–Han, J. S.; Brendza,R. P.; Fryer, J. D.; Kierson, M.; Cirrito, J.; Quick, K.; Harmony,J. A. K.; Aronow, B. J.; Holtzman, D. M.: Clusterin contributes to caspase–3–independent brain injury following neonatal hypoxia–ischemia. *Nature Med.* 7: 338–343, 2001.
- [27170] 722.Jenne, D. E.; Tschopp, J.: Clusterin: the intriguing guises of a widely expressed glycoprotein. *Trends Biochem. Sci.* 17: 154–159,1992.
- [27171] 723.Kamboh, M. I.; Harmony, J. A. K.; Sepehrnia, B.; Nwankwo, M.;Ferrell, R. E.: Genetic studies of human apolipoproteins. XX. Genetic polymorphism of apolipoprotein J and its impact on quantitative lipid traits in normolipidemic subjects. *Am. J. Hum. Genet.* 49: 1167–1173,1991.
- [27172] 724.Kirschbaum, L.; Sharpe, J. A.; Murphy, B.; d'Apice, A. J. F.; Classon,B.; Hudson, P.; Walker, I. D.: Molecular cloning and characterization of the novel, human complement–associated protein, SP–40,40: a link between the complement

and reproductive systems. EMBO J. 8: 711–718,1989.

[27173] 725.Murphy, B. F.; Kirszbaum, L.; Walker, I. D.; d'Apice, A. J. F.: SP-40,40, a newly identified normal human serum protein found inthe SC5b-9 complex of complement and in the immune deposits in glomerulonephritis. J.Clin. Invest. 81: 1858–1864, 1988.

[27174] 726.O'Bryan, M. K.; Baker, H. W. G.; Saunders, J. R.; Kirszbaum, L.;Walker, I. D.; Hudson, P.; Liu, D. Y.; Glew, M. D.; d'Apice, A. J.F.; Murphy, B. F.: Human seminal clusterin (SP-40,40): isolationand characterization. J. Clin. Invest. 85: 1477–1486, 1990.

[27175] 727.Purrello, M.; Bettuzzi, S.; Di Pietro, C.; Mirabile, E.; Di Blasi,M.; Rimini, R.; Grzeschik, K.-H.; Ingletti, C.; Corti, A.; Sichel,G.: The gene for SP-40,40, human homolog of rat sulfated glycoprotein2, rat clusterin, and rat testosterone-repressed prostate message2, maps to chromosome 8. Genomics 10: 151–156, 1991.

[27176] 728.Slawin, K.; Sawczuk, I. S.; Olsson, C. A.; Buttyan, R.: Chromosomalassignment of the human homologue encoding SGP-2. Biochem. Biophys.Res. Commun. 172: 160–164, 1990.

[27177] 729.Bachega, T. A. S. S.; Brenlha, E. M. L.; Billerbeck, A. E. C.;Marcondes, J. A. M.; Madureira, G.; Arnhold, I. J. P.;

Mendonca, B.B.: Variable ACTH-stimulated 17-hydroxyprogesterone values in 21-hydroxylase deficiency carriers are not related to the different CYP21 gene mutations. *J.Clin. Endocr. Metab.* 87: 786–790, 2002.

[27178] 730. Baumgartner-Parzer, S. M.; Schulze, E.; Waldhausl, W.; Pauschenwein, S.; Rondot, S.; Nowotny, P.; Meyer, K.; Frisch, H.; Waldhauser, F.; Vierhapper, H.: Mutational spectrum of the steroid 21-hydroxylase gene in Austria: identification of a novel missense mutation. *J.Clin. Endocr. Metab.* 86: 4771–4775, 2001.

[27179] 731. Beuschlein, F.; Schulze, E.; Mora, P.; Gensheimer, H.-P.; Maser-Gluth, C.; Allolio, B.; Reincke, M.: Steroid 21-hydroxylase mutations and 21-hydroxylase messenger ribonucleic acid expression in human adrenocortical tumors. *J. Clin. Endocr. Metab.* 83: 2585–2588, 1998.

[27180] 732. Bias, W. B.; Urban, M. D.; Migeon, C. J.; Hsu, S. H.; Lee, P. A.: Intra-HLA recombinations localizing the 21-hydroxylase deficiency gene within the HLA complex. *Hum. Immun.* 2: 139–145, 1981.

[27181] 733. Billerbeck, A. E. C.; Bachega, T. A. S. S.; Frazzatto, E. T.; Nishi, M. Y.; Goldberg, A. C.; Marin, M. L. C.; Madureira, G.; Monte, O.; Arnhold, I. J. P.; Mendonca, B. B.: A novel

missense mutation, GLY424SER, in Brazilian patients with 21-hydroxylase deficiency. *J. Clin. Endocr. Metab.* 84: 2870–2872, 1999.

- [27182] 734. Alves, S.; Prata, M.-J.; Ferreira, F.; Amorim, A.: Thiopurinemethyltransferase pharmacogenetics: alternative molecular diagnosis and preliminary data from Northern Portugal. *Pharmacogenetics* 9:257–261, 1999.
- [27183] 735. Ameyaw, M.-M.; Collie-Duguid, E. S. R.; Powrie, R. H.; Ofori-Adjei, D.; McLeod, H. L.: Thiopurine methyltransferase alleles in British and Ghanaian populations. *Hum. Molec. Genet.* 8: 367–370, 1999.
- [27184] 736. Collie-Duguid, E. S. R.; Pritchard, S. C.; Powrie, R. H.; Sludden, J.; Collier, D. A.; Li, T.; McLeod, H. L.: The frequency and distribution of thiopurine methyltransferase alleles in Caucasian and Asian populations. *Pharmacogenetics* 9:37–42, 1999.
- [27185] 737. Evans, W. E.; Horner, M.; Chu, Y. Q.; Kalwinsky, D.; Roberts, W. M.: Altered mercaptopurine metabolism, toxic effects, and dosage requirement in a thiopurine methyltransferase-deficient child with acute lymphocytic leukemia. *J. Pediatr.* 119: 985–989, 1991.
- [27186] 738. Hon, Y. Y.; Fessing, M. Y.; Pui, C.-H.; Relling, M. V.; Krynetski, E. Y.; Evans, W. E.: Polymorphism of the thiop-

urine S-methyltransferase gene in African-Americans.

Hum. Molec. Genet. 8: 371-376, 1999.

[27187] 739. Honchel, R.; Aksoy, I. A.; Szumlanski, C.; Wood, T. C.; Otterness, D. M.; Wieben, E. D.; Weinshilboum, R. M.: Human thiopurine methyltransferase: molecular cloning and expression of T84 colon carcinoma cell cDNA.

Molec. Pharm. 43: 878-887, 1993.

[27188] 740. Klemetsdal, B.; Wist, E.; Aarbakke, J.: Gender difference in red blood cell thiopurine methyltransferase activity. Scand. J. Clin. Lab. Invest. 53: 747-749, 1993.

[27189] 741. Krynetski, E. Y.; Schuetz, J. D.; Galpin, A. J.; Pui, C.-H.; Relling, M. V.; Evans, W. E.: A single point mutation leading to loss of catalytic activity in human thiopurine S-methyltransferase. Proc. Nat. Acad. Sci. 92: 949-953, 1995.

[27190] 742. Lennard, L.; Lilleyman, J. S.; Van Loon, J.; Weinshilboum, R. M.: Genetic variation in response to 6-mercaptopurine for childhood acute lymphoblastic leukaemia. Lancet 336: 225-229, 1990.

[27191] 743. Lennard, L.; Van Loon, J. A.; Weinshilboum, R. M.: Pharmacogenetics of acute azathioprine toxicity: relationship to thiopurine methyltransferase genetic polymorphism. Clin. Pharm. Therap. 46: 149-154, 1989.

- [27192] 744. McLeod, H. L.; Pritchard, S. C.; Githang'a, J.; Indalo, A.; Ameyaw, M.-M.; Powrie, R. H.; Booth, L.; Collie-Duguid, E. S. R.: Ethnic differences in thiopurine methyltransferase pharmacogenetics: evidence for allele specificity in Caucasian and Kenyan individuals. *Pharmacogenetics* 9:773-776, 1999.
- [27193] 745. Otterness, D.; Szumlanski, C.; Lennard, L. Klemetsdal, B.; Aarbakke, J.; Park-Hah, J. O.; Iven, H.; Schmiegelow, K.; Branum, E.; O'Brien, J.; Weinshilboum, R.: Human thiopurine methyltransferase pharmacogenetics: gene sequence polymorphisms. *Clin. Pharm. Therap.* 62: 60-73, 1997.
- [27194] 746. Otterness, D. M.; Szumlanski, C. L.; Wood, T. C.; Weinshilboum, R. M.: Human thiopurine methyltransferase pharmacogenetics: kindred with a terminal exon splice junction mutation that results in loss of activity. *J. Clin. Invest.* 101: 1036-1044, 1998.
- [27195] 747. Szumlanski, C.; Otterness, D.; Her, C.; Lee, D.; Brandriff, B.; Kelsell, D.; Spurr, N.; Lennard, L.; Wieben, E.; Weinshilboum, R.: Thiopurine methyltransferase pharmacogenetics: human gene cloning and characterization of a common polymorphism. *DNA Cell Biol.* 15:17-30, 1996.
- [27196] 748. Tai, H.-L.; Krynetski, E. Y.; Schuetz, E. G.; Yanishevski, Y.; Evans, W. E.: Enhanced proteolysis of thiop-

urine S-methyltransferase(TPMT) encoded by mutant alleles in humans (TPMT*3A, TPMT*2): mechanisms for the genetic polymorphism of TPMT activity. *Proc. Nat. Acad. Sci.* 94:6444–6449, 1997.

[27197] 749.Tai, H.-L.; Krynetski, E. Y.; Yates, C. R.; Loennechen, T.; Fessing, M. Y.; Krynetskaia, N. F.; Evans, W. E.: Thiopurine S-methyltransferase deficiency: two nucleotide transitions define the most prevalent mutant allele associated with loss of catalytic activity in Caucasians. *Am.J. Hum. Genet.* 58: 694–702, 1996.

[27198] 750.Weinshilboum, R. M.; Sladek, S. L.: Mercaptopurine pharmacogenetics: monogenic inheritance of erythrocyte thiopurine methyltransferase activity. *Am. J. Hum. Genet.* 32: 651–662, 1980.

[27199] 751.Reed, T. M.; Browning, J. E.; Blough, R. I.; Vorhees, C. V.; Repaske, D. R.: Genomic structure and chromosome location of the murine PDE1B phosphodiesterase gene. *Mammalian Genome* 9: 571–576, 1998.

[27200] 752.Repaske, D. R.; Swinnen, J. V.; Jin, S.-L. C.; Van Wyk, J. J.; Conti, M.: A polymerase chain reaction strategy to identify and clone cyclic nucleotide phosphodiesterase cDNAs: molecular cloning of the cDNA encoding the 63-kDa calmodulin-dependent phosphodiesterase. *J.Biol. Chem.*

267: 18683–18688, 1992.

- [27201] 753. Yu, J.; Wolda, S. L.; Frazier, A. L. B.; Florio, V. A.; Martins, T. J.; Snyder, P. B.; Harris, E. A. S.; McCaw, K. N.; Farrell, C. A.; Steiner, B.; Bentley, J. K.; Beavo, J. A.; Ferguson, K.; Gelinas, R.: Identification and characterisation of a human calmodulin-stimulated phosphodiesterase PDE1B1. *Cell. Signal.* 9: 519–529, 1997.
- [27202] 754. Bargagna, M.; Abbagnale, L.: Isoelectric focusing of human red cell phosphoglucomutase (PGM1): phenotype distribution in the population of Tuscany and two hereditary variants. *Hum. Genet.* 61: 242–245, 1982.
- [27203] 755. Chagnon, Y. C.; Bouchard, C.; Allard, C.: Isoelectric focusing of red cell phosphoglucomutase (E.C.:2.7.5.1) at the PGM-1 locus in a French-Canadian population. *Hum. Genet.* 59: 36–38, 1981.
- [27204] 756. Cook, P. J. L.; Noades, J.; Hopkinson, D. A.; Robson, E. B.; Cleghorn, T. E.: Demonstration of a sex difference in recombination fraction in the loose linkage, Rh and PGM(1). *Ann. Hum. Genet.* 35: 239–242, 1972.
- [27205] 757. Cook, P. J. L.; Noades, J. E.; Newton, M. S.; de Mey, R.: On the orientation of the Rh:E1 linkage group. *Ann. Hum. Genet.* 41: 157–162, 1977.
- [27206] 758. Cook, P. J. L.; Robson, E. B.; Buckton, K. E.; Jacobs, P.

A.; Polani, P. E.: Segregation of genetic markers in families with chromosome polymorphisms and structural rearrangements involving chromosome 1. *Ann. Hum. Genet.* 37: 261–274, 1974.

[27207] 759. Douglas, G. R.; McAlpine, P. J.; Hamerton, J. L.: Regional localization of loci for human PGM(1) and 6PGD on human chromosome one by use of hybrids of Chinese hamster–human somatic cells. *Proc. Nat. Acad. Sci.* 70: 2737–2740, 1973.

[27208] 760. Dykes, D. D.; Kuhl, P.; Martin, W.: PGM1 system: report on the International Workshop, October 10–11, 1983, Munich, West Germany. *Am. J. Hum. Genet.* 37: 1225–1231, 1985.

[27209] 761. Ferrell, R. E.; Escallon, M.; Aguilar, L.; Bertin, T.: Erythrocyte phosphoglucomutase: a family study of a PGM1 deficient allele. *Hum. Genet.* 67: 306–308, 1984.

[27210] 762. Francke, U.; George, D. L.: Precise mapping of genes for phosphoglucomutase-1 and uridine monophosphate kinase on the short arm of human chromosome 1. *Cytogenet. Cell Genet.* 22: 384–388, 1978.

[27211] 763. Gedde-Dahl, T., Jr.; Monn, E.: Linkage relations of the phosphoglucomutase PGM(1) locus in man. Probable linkage to phenylthiocarbamid (PTC) taster locus. *Acta Genet.*

Statist. Med. 17: 482–494, 1967.

- [27212] 764. Herbach, J.; Szilvassy, J.; Schnedl, W.: Gene localisation of the PGM-1 enzyme system and the Duffy blood groups on chromosome no. 1 by means of a new fragile site at 1p31. Hum. Genet. 70: 178–180, 1985.
- [27213] 765. Hopkinson, D. A.; Harris, H.: Rare phosphoglucomutase phenotypes. Ann. Hum. Genet. 30: 167–181, 1966.
- [27214] 766. Ishimoto, G.: Placental phosphoglucomutase in Japanese. Jpn. J. Hum. Genet. 14: 183–188, 1969.
- [27215] 767. Kamboh, M. I.; Kirk, R. L.: Investigation of PGM1(3), PGM1(6), and PGM1(7) variants by isoelectric focusing: evidence for new subtypes of the PGM1(3) and PGM1(7) alleles. Hum. Genet. 64: 58–60, 1983.
- [27216] 768. Kidd, J. R.; Matsubara, Y.; Castiglione, C. M.; Tanaka, K.; Kidd, K. K.: The locus for the medium-chain acyl-CoA dehydrogenase gene on chromosome 1 is highly polymorphic. Genomics 6: 89–93, 1990.
- [27217] 769. Kidd, K. K.; Kidd, J. R.; Castiglione, C. M.; Sparkes, R. S.; Egeland, J. A.; Bakker, E.: The anonymous RFLP locus D1S2 is close to PGM1 on chromosome 1. Hum. Hered. 38: 22–26, 1988.
- [27218] 770. March, R. E.; Putt, W.; Hollyoake, M.; Ives, J. H.; Lovegrove, J. U.; Hopkinson, D. A.; Edwards, Y. H.; Whitehouse,

D. B.: The classical human phosphoglucomutase (PGM1) isozyme polymorphism is generated by intragenic recombination. *Proc. Nat. Acad. Sci.* 90: 10730–10733, 1993.

[27219] 771. McAlpine, P. J.; Hopkinson, D. A.; Harris, H.: Thermostability studies on the isoenzymes of human phosphoglucomutase. *Ann. Hum. Genet.* 34: 61–71, 1970.

[27220] 772. Monn, E.: A new red cell phosphoglucomutase phenotype in man. *Acta Genet. Statist. Med.* 18: 123–127, 1967.

[27221] 773. Parrington, J. M.; Cruickshank, G.; Hopkinson, D. A.; Robson, E. B.; Harris, H.: Linkage relationships between the three phosphoglucomutase loci PGM(1), PGM(2) and PGM(3). *Ann. Hum. Genet.* 32: 27–34, 1968.

[27222] 774. Quick, C. B.; Fisher, R. A.; Harris, H.: Differentiation of the PGM(2) locus isozymes from those of PGM(1) and PGM(3) in terms of phosphopentomutase activity. *Ann. Hum. Genet.* 35: 445–454, 1972.

[27223] 775. Robson, E. B.; Cook, P. J. L.; Corney, G.; Hopkinson, D. A.; Noades, J.; Cleghorn, T. E.: Linkage data on Rh, PGM1, PGD, peptidase C and Fy from family studies. *Ann. Hum. Genet.* 36: 393–399, 1973.

[27224] 776. Sachs, V.; Siemsen, M.; Martin, W.; Vollert, B.: A new hereditary variant of the PGM(1) erythrocyte enzyme sys-

tem determined by isoelectric focusing. Hum. Genet. 58: 411–413, 1981.

[27225] 777.Santachiara-Benerecetti, A. S.; Cattaneo, A.; Meera Khan, P.:Rare phenotypes of the PGM(1) and PGM(2) loci and a new PGM(2) variant allele in the Indians. Am. J. Hum. Genet. 24: 680–685, 1972.

[27226] 778.Santachiara-Benerecetti, A. S.; Ranzani, G. N.; Antonini, G.:Subtyping of human red cell phosphoglucomutase locus 1 (PGM-1) polymorphism:a third PGM-1(1) allele common among Twa pygmies from North Rwanda. Am.J. Hum. Genet. 33: 817–822, 1981.

[27227] 779.Santachiara-Benerecetti, A. S.; Ranzani, G. N.; Antonini, G.;Beretta, M.: Subtyping of phosphoglucomutase locus 1 (PGM1) polymorphism in some populations of Rwanda: description of variant phenotypes,'haplotype' frequencies, and linkage disequilibrium data. Am. J.Hum. Genet. 34: 337–348, 1982.

[27228] 780.Sutherland, R.; Delia, D.; Schneider, C.; Newman, R.; Kemshead,J.; Greaves, M.: Ubiquitous cell-surface glycoprotein on tumor cells is proliferation-associated receptor for transferrin. Proc. Nat.Acad. Sci. 78: 4515–4519, 1981.

[27229] 781.Abraham, E.; Glauser, M. P.; Butler, T.; Garbino, J.; Gelmont,D.; Laterre, P. F.; Kudsk, K.; Bruining, H. A.; Otto,

C.; Tobin, E.; Zwingelstein, C.; Lesslauer, W.; Leighton, A.:
p55 tumor necrosis factor receptor fusion protein in the
treatment of patients with severe sepsis and septic shock.
A randomized controlled multicenter trial. J.A.M.A.
277:1531, 1997.

- [27230] 782. Carballo, E.; Lai, W. S.; Blackshear, P. J.: Evidence that
tristetraprolin is a physiological regulator of granulocyte-
macrophage colony-stimulating factor messenger RNA
deadenylation and stability. Blood 95: 1891-1899, 2000.
- [27231] 783. Carballo, E.; Lai, W. S.; Blackshear, P. J.: Feedback in-
hibition of macrophage tumor necrosis factor- α pro-
duction by tristetraprolin. Science 281:1001-1005, 1998.
- [27232] 784. DuBois, R. N.; McLane, M. W.; Ryder, K.; Lau, L. F.;
Nathans, D.: A growth factor-inducible nuclear protein
with a novel cysteine/histidine repetitive sequence. J. Biol.
Chem. 265: 19185-19191, 1990.
- [27233] 785. Hoovers, J. M. N.; Mannens, M.; John, R.; Blik, J.; van
Heyningen, V.; Porteous, D. J.; Leschot, N. J.; Westerveld,
A.; Little, P. F. R.: High-resolution localization of 69 poten-
tial human zinc finger protein genes: a number are clus-
tered. Genomics 12: 254-263, 1992.
- [27234] 786. Huebner, K.; Druck, T.; LaForgia, S.; Lasota, J.; Croce,
C. M.; Lanfrancone, L.; Donti, E.; Pengue, G.; La Mantia, G.;

Pelicci, P.-G.; Lania, L.: Chromosomal localization of four human zinc finger cDNAs. *Hum.Genet.* 91: 217–222, 1993.

[27235] 787.Lai, W. S.; Stumpo, D. J.; Blackshear, P. J.: Rapid insulin-stimulated accumulation of an mRNA encoding a proline-rich protein. *J. Biol.Chem.* 265: 16556–16563, 1990.

[27236] 788.Lorenz, H. M.; Antoni, C.; Valerius, T.; Repp, R.; Grunke, M.; Schwerdtner, N.; Nusslein, H.; Woody, J.; Kalden, J. R.; Manger, B.: In vivo blockade of TNF-alpha by intravenous infusion of a chimeric monoclonal TNF-alpha antibody in patients with rheumatoid arthritis. Short term cellular and molecular effects. *J. Immun.* 156: 1646–1653, 1996.

[27237] 789.Taylor, G. A.; Lai, W. S.; Oakey, R. J.; Seldin, M. F.; Shows, T. B.; Eddy, R. L., Jr.; Blackshear, P. J.: The human TTP protein: sequence, alignment with related proteins, and chromosomal localization of the mouse and human genes. *Nucleic Acids Res.* 19: 3454 only, 1991.

[27238] 790.Varnum, B. C.; Ma, Q.; Chi, T.; Fletcher, B.; Herschman, H. R.: The TIS11 primary response gene is a member of a gene family that encodes proteins with a highly conserved sequence containing an unusual cys-his

repeat. *Molec. Cell. Biol.* 11: 1754–1758, 1991.

- [27239] 791.Andersson, O.; Nordlund–Moller, L.; Bronnegard, M.; Sirzea, F.;Ripe, E.; Lund, J.: Purification and level of expression in bronchoalveolarlavage of a human polychlorinated biphenyl (PCB)–binding protein:evidence for a structural and functional kinship to the multihormonallyregulated protein uteroglobin. *Am. J. Resp. Cell Molec. Biol.* 5:6–12, 1991.
- [27240] 792.Assmann, K. J. M.; Koene, R. A. P.; Wetzels, J. F. M.: Familialglomerulonephritis characterized by massive deposits of fibronectin. *Am.J. Kidney Dis.* 25: 781–791, 1995.
- [27241] 793.Atger, M.; Atger, P.; Tiollais, P.; Milgrom, E.: Cloning of rabbitgenomic fragments containing the uteroglobin gene. *J. Biol. Chem.* 256:5970–5972, 1981.
- [27242] 794.Gemperle, O.; Neuweiler, J.; Reutter, F. W.; Hildebrandt, F.; Krapf,R.: Familial glomerulopathy with giant fibrillar (fibronectin–positive)deposits: 15–year follow–up in a large kindred. *Am. J. Kidney Dis.* 28:668–675, 1996.
- [27243] 795.Hagen, G.; Wolf, M.; Katyal, G.; Singh, G.; Beato, M.; Suske, G.: Tissue–specific expression, hormonal regulation and 5–prime–flankinggene region of the rat Clara cell 10 kDa protein: comparison to rabbituteroglobin. *Nucleic*

Acids Res. 18: 2939–2945, 1990.

[27244] 796.Hay, J. G.; Danel, C.; Chu, C.-S.; Crystal, R. G.: Human CC10gene expression in airway epithelium and subchromosomal locus suggest linkage to airway disease. Am. J. Physiol. 268: L565–L575, 1995.

[27245] 797.Laing, I. A.; Goldblatt, J.; Eber, E.; Hayden, C. M.; Rye, P. J.;Gibson, N. A.; Palmer, L. J.; Burton, P. R.; Le Souef, P. N.: A polymorphism of the CC16 gene is associated with an increased risk of asthma. J.Med. Genet. 35: 463–467, 1998.

[27246] 798.Mazzucco, G.; Maran, E.; Rollino, C.; Monga, G.: Glomerulonephritis with organized deposits: a mesangio-pathic, not immune complex-mediated disease: a pathologic study of two cases in the same family. Hum.Path. 23: 63–68, 1992.

[27247] 799.Menne, C.; Suske, G.; Arnemann, J.; Wenz, M.; Kato, C. B.; Beato, M.: Isolation and structure of the gene for the progesterone-inducible protein uteroglobin. Proc. Nat. Acad. Sci. 79: 4853–4857, 1982.

[27248] 800.Mihal, K.; Riedel, N.: One gene encoding three proteins with different functions. Am. J. Resp. Cell Molec. Biol. 5: 1–3, 1991.

[27249] 801.Singh, G.; Katyal, S. L.; Brown, W. E.; Kennedy, A. L.;

Singh,U.; Wong-Chong, M.-L.: Clara cell 10 kDa protein (CC10): comparison of structure and function to uteroglobin. *Biochim. Biophys. Acta* 1039:348–355, 1990.

- [27250] 802.Singh, G.; Singh, J.; Katyal, S. L.; Brown, W. E.; Kramps, J.A.; Paradis, I. L.; Dauber, J. H.; Macpherson, T. A.; Squeglia, N.: Identification, cellular localization, isolation, and characterization of human Clara cell-specific 10 kD protein. *J. Histochem. Cytochem.* 36:73–80, 1988.
- [27251] 803.Stohr, H.; Weber, B. H. F.: (ATTT)_n-tetranucleotide repeat polymorphism in the 5-prime-flanking region of the UGB gene. *Hum. Molec. Genet.* 3:2086 only, 1994.
- [27252] 804.Stripp, B. R.; Huffman, J. A.; Bohinski, R. J.: Structure and regulation of the murine Clara cell secretory protein gene. *Genomics* 20:27–35, 1994.
- [27253] 805.Strom, E. H.; Banfi, G.; Krapf, R.; Abt, A. B.; Mazzucco, G.; Monga, G.; Gloor, F.; Neuweiler, J.; Riess, R.; Stosiek, P.; Hebert, L. A.; Sedmak, D. D.; Gudat, F.; Mihatsch, M. J.: Glomerulopathy associated with predominant fibronectin deposits: a newly recognized hereditary disease. *Kidney Int.* 48: 163–170, 1995.
- [27254] 806.Wolf, M.; Klug, J.; Hackenberg, R.; Gessler, M.; Grzeschik, K.-H.; Beato, M.; Suske, G.: Human CC10, the homologue of rabbit uteroglobin: genomic cloning, chro-

mosomal localization and expression in endometrial cell lines. Hum. Molec. Genet. 1: 371–378, 1992.

- [27255] 807. Zhang, Z.; Kundu, G. C.; Yuan, C.-J.; Ward, J. M.; Lee, E. J.; DeMayo, F.; Westphal, H.; Mukherjee, A. B.: Severe fibronectin-deposit renal glomerular disease in mice lacking uteroglobin. Science 276:1408–1411, 1997.
- [27256] 808. Zhang, Z.; Zimonjic, D. B.; Popescu, N. C.; Wang, N.; Gerhard, D. S.; Stone, E. M.; Arbour, N. C.; De Vries, H. G.; Scheffer, H.; Gerritsen, J.; Colle'e, J. M.; Ten Kate, L. P.; Mukherjee, A. B.: Human uteroglobin gene: structure, sub-chromosomal localization, and polymorphism. DNA Cell Biol. 16: 73–83, 1997.
- [27257] 809. Erbe, R. W.: Cabot case. New Eng. J. Med. 298: 95–101, 1978.
- [27258] 810. Feldman, D. E.; Thulasiraman, V.; Ferreyra, R. G.; Frydman, J.: Formation of the VHL–elongin BC tumor suppressor complex is mediated by the chaperonin TRiC. Molec. Cell 1051–1061, 1999.
- [27259] 811. Fill, W. L.; Lamiell, J. M.; Polk, N. O.: The radiographic manifestations of von Hippel–Lindau disease. Radiology 133: 289–295, 1979.
- [27260] 812. Fishman, R. S.; Bartholomew, L. G.: Severe pancreatic involvement in three generations in von Hippel–Lindau

disease. Mayo Clin. Proc. 54:329–331, 1979.

- [27261] 813.Friedrich, C. A.: Genotype/phenotype correlation in von Hippel–Lindausyndrome. Hum. Molec. Genet. 10: 763–767, 2001.
- [27262] 814.Fukino, K.; Teramoto, A.; Adachi, K.; Takahashi, H.; Emi, M.:A family with hydrocephalus as a complication of cerebellar hemangioblastoma:identification of pro157leu mutation in the VHL gene. J. Hum. Genet. 45:47–51, 2000.
- [27263] 815.Fulton, J. F.: Harvey Cushing: A Biography. Springfield, Illinois:Charles C Thomas (pub.) 1946.
- [27264] 816.Funk, K. C.; Heiken, J. P.: Papillary cystadenoma of the broadligament in a patient with von Hippel–Lindau disease. Am. J. Radiol. 153:527–528, 1989.
- [27265] 817.Gaffey, M. J.; Mills, S. E.; Boyd, J. C.: Aggressive papillarytumor of middle ear/temporal bone and adnexal papillary cystadenoma. Am.J. Surg. Path. 18: 1254–1260, 1994.
- [27266] 818.Gallou, C.; Joly, D.; Mejean, A.; Staroz, F.; Martin, N.; Tarlet,G.; Orfanelli, M. T.; Bouvier, R.; Droz, D.; Chretien, Y.; Marechal,J. M.; Richard, S.; Junien, C.; Beroud, C.: Mutations of the VHLgene in sporadic renal cell carcinoma: definition of a risk factorfor VHL patients to develop an RCC. Hum. Mutat. 13: 464–475, 1999.

- [27267] 819.Garcia, A.; Matias-Guiu, X.; Cabezas, R.; Chico, A.; Prat, J.; Baiget, M.; De Leiva, A.: Molecular diagnosis of von Hippel-Lindau disease in a kindred with a predominance of familial pheochromocytoma. *Clin.Endocr.* 46: 359-363, 1997.
- [27268] 820.Gemmill, R. M.; Bemis, L. T.; Lee, J. P.; Sozen, M. A.; Baron, A.; Zeng, C.; Erickson, P. F.; Hooper, J. E.; Drabkin, H. A.: The TRC8 hereditary kidney cancer gene suppresses growth and functions with VHL in a common pathway. *Oncogene* 21: 3507-3516, 2002.
- [27269] 821.Gersell, D. J.; King, T. C.: Papillary cystadenoma of the mesosalpinx in von Hippel-Lindau disease. *Am. J. Surg. Path.* 12: 145-149, 1988.
- [27270] 822.Gilcrease, M. Z.; Schmidt, L.; Zbar, B.; Truong, L.; Rutledge, M.; Wheeler, T. M.: Somatic von Hippel-Lindau mutation in clear cell papillary cystadenoma of the epididymis. *Hum. Path.* 26: 1341-1346, 1995.
- [27271] 823.Glenn, G. M.; Daniel, L. N.; Choyke, P.; Linehan, W. M.; Oldfield, E.; Gorin, M. B.; Hosoe, S.; Latif, F.; Weiss, G.; Walther, M.; Lerman, M. I.; Zbar, B.: Von Hippel-Lindau (VHL) disease: distinct phenotypes suggest more than one mutant allele at the VHL locus. *Hum. Genet.* 87:207-210, 1991.

- [27272] 824. Glenn, G. M.; Linehan, W. M.; Hosoe, S.; Latif, F.; Yao, M.; Choyke, P.; Gorin, M. B.; Chew, E.; Oldfield, E.; Manolatos, C.; Orcutt, M. L.; Walther, M. M.; Weiss, G. H.; Tory, K.; Jensson, O.; Lerman, M. I.; Zbar, B.: Screening for von Hippel–Lindau disease by DNA polymorphism analysis. *J. A. M. A.* 267: 1226–1231, 1992.
- [27273] 825. Gnarr, J. R.; Ward, J. M.; Porter, F. D.; Wagner, J. R.; Devor, D. E.; Grinberg, A.; Emmert–Buck, M. R.; Westphal, H.; Klausner, R. D.; Marston Linehan, W.: Defective placental vasculogenesis causes embryonic lethality in VHL–deficient mice. *Proc. Nat. Acad. Sci.* 94: 9102–9107, 1997.
- [27274] 826. Go, R. C. P.; Lamiell, J. M.; Hsia, Y. E.; Yuen, J. W.–M.; Paik, Y.: Segregation and linkage analyses of von Hippel Lindau disease among 220 descendants from one kindred. *Am. J. Hum. Genet.* 36: 131–142, 1984.
- [27275] 827. Goldberg, M. F.; Duke, J. R.: Von Hippel–Lindau disease: histopathologic findings in a treated and untreated eye. *Am. J. Ophthalmol.* 66: 693–705, 1968.
- [27276] 828. Graff, J. W.: Personal Communication. Brookline, Mass. 10/4/1998.
- [27277] 829. Green, J. S.; Bowmer, M. I.; Johnson, G. J.: Von Hippel–Lindau disease in a Newfoundland kindred. *Canad. Med. Assoc. J.* 134: 133–138 and 146, 1986.

- [27278] 830. Gross, D. J.; Avishai, N.; Meiner, V.; Filon, D.; Zbar, B.; Abeliovich, D.: Familial pheochromocytoma associated with a novel mutation in the von Hippel–Lindau gene. *J. Clin. Endocr. Metab.* 81: 147–149, 1996.
- [27279] 831. Grossman, M.; Melmon, K. L.: Von Hippel–Lindau disease. In: Vinken, P. J.; Bruyn, G. W.: *Handbook of Clinical Neurology. The Phakomatoses*. Amsterdam: North Holland (pub.) 14: 1972. Pp. 241–259.
- [27280] 832. Haase, V. H.; Glickman, J. N.; Socolovsky, M.; Jaenisch, R.: Vascular tumors in livers with targeted inactivation of the von Hippel–Lindau tumor suppressor. *Proc. Nat. Acad. Sci.* 98: 1583–1588, 2001.
- [27281] 833. Hagler, W. S.; Hyman, B. N.; Waters, W. C., III: Von Hippel's angiomatosis retinae and pheochromocytoma. *Trans. Am. Acad. Ophthalmol. Otolaryng.* 75: 1022–1034, 1971.
- [27282] 834. Hennessy, T. G.; Stern, W. E.; Herrick, S. E.: Cerebellar hemangioblastoma: erythropoietic activity by radioimmunoassay. *J. Nucl. Med.* 8: 601–606, 1967.
- [27283] 835. Herman, J. G.; Latif, F.; Weng, Y.; Lerman, M. I.; Zbar, B.; Liu, S.; Samid, D.; Duan, D.–S. R.; Guarra, J. R.; Linehan, W. M.; Baylin, S. B.: Silencing of the VHL tumor-suppressor gene by DNA methylation in renal carcinomas. *Proc. Nat.*

Acad. Sci. 91: 9700–9704, 1994.

- [27284] 836.Hes, F.; Zewald, R.; Peeters, T.; Sijmons, R.; Links, T.; Verheij, J.; Matthijs, G.; Legius, E.; Mortier, G.; van der Torren, K.; Rosman, M.; Lips, C.; Pearson, P.; van der Luit, R.: Genotype–phenotype correlations in families with deletions in the von Hippel–Lindau (VHL) gene. Hum. Genet. 106: 425–431, 2000.
- [27285] 837.Hes, F. J.; McKee, S.; Taphoorn, M. J. B.; Rehal, P.; van der Luit, R. B.; McMahon, R.; van der Smagt, J. J.; Dow, D.; Zewald, R. A.; Whittaker, J.; Lips, C. J. M.; MacDonald, F.; Pearson, P. L.; Maher, E. R.: Cryptic von Hippel–Lindau disease: germline mutations in patients with haemangioblastoma only. J. Med. Genet. 37: 939–943, 2000.
- [27286] 838.Hoffman, M. A.; Ohh, M.; Yang, H. Kico, J. M.; Ivan, M.; Kaelin, W. G., Jr.: von Hippel–Lindau, protein mutants linked to type 2CVHL disease preserve the ability to downregulated HIF. Hum. Molec. Genet. 10: 1019–1027, 2001.
- [27287] 839.Rocchi, M.; Archidiacono, N.; Romeo, G.; Saginati, M.; Zardi, L.: Assignment of the gene for human tenascin to the region q32–q34 of chromosome 9. Hum. Genet. 86: 621–623, 1991.
- [27288] 840.Steindler, D. A.; Settles, D.; Erickson, H. P.; Laywell, E.

D.;Yoshiki, A.; Faissner, A.; Kusakabe, M.: Tenascin knockout mice:barrels, boundary molecules, and glial scars. J. Neurosci. 15: 1971–1983,1995.

[27289] 841.Bamford, R. N.; Roessler, E.; Burdine, R. D.; Saplakoglu, U.; delaCruz, J.; Splitt, M.; Towbin, J.; Bowers, P.; Marino, B.; Schier,A. F.; Shen, M. M.; Muenke, M.; Casey, B.: Loss-of-function mutationsin the EGF-CFC gene CFC1 are associated with human left-right laterality-defects. Nature Genet. 26: 365–369, 2000.

[27290] 842.Ciccodicola, A.; Dono, R.; Obici, S.; Simeone, A.; Zollo, M.; Persico,M. G.: Molecular characterization of a gene of the 'EGF family' expressedin undifferentiated human NTERA2 teratocarcinoma cells. EMBO J. 8:1987–1991, 1989.

[27291] 843.de la Cruz, J. M.; Bamford, R. N.; Burdine, R. D.; Roessler, E.;Barkovich, A. J.; Donnai, D.; Schier, A. F.; Muenke, M.: A loss-of-functionmutation in the CFC domain of TDGF1 is associated with human forebraindefects. Hum. Genet. 110: 422–428, 2002.

[27292] 844.Dono, R.; Montuori, N.; Rocchi, M.; De Ponti-Zilli, L.; Ciccodicola,A.; Persico, M. G.: Isolation and characteriza-tion of the CRIPTOautosomal gene and its X-linked related sequence. Am. J. Hum. Genet. 49:555–565, 1991.

- [27293] 845.Liguori, G.; De Gregorio, L.; Tucci, M.; Lago, C. T.; Barra, A.;Dragani, T. A.; Persico, M.: Mapping of the mouse Tdgf1 gene andTdgf pseudogenes. Mammalian Genome 8: 502–505, 1997.
- [27294] 846.Liguori, G.; Tucci, M.; Montuori, N.; Dono, R.; Lago, C. T.; Pacifico,F.; Armenante, F.; Persico, M. G.: Characterization of the mouseTdgf1 gene and Tdgf pseudogenes. Mammalian Genome 7: 344–348, 1996.
- [27295] 847.Saccone, S.; Rapisarda, A.; Motta, S.; Dono, R.; Persico, G. M.;Della Valle, G.: Regional localization of the human EGF–like growthfactor CRIPTO gene (TDGF–1) to chromosome 3p21. Hum. Genet. 95:229–230, 1995.
- [27296] 848.Shen, M. M.; Schier, A. F.: The EGF–CFC gene family in vertebratedevelopment. Trends Genet. 16: 303–309, 2000.
- [27297] 849.Chang, L. M. S.; Bollum, F. J.: Molecular biology of terminaltransferase. CRC Crit. Rev. Biochem. 21: 27–52, 1986.
- [27298] 850.Epplen, J. T.; Chluba, J.; Hardt, C.; Hinkkanen, A.; Steimle, V.;Stockinger, H.: Mammalian T–lymphocyte antigen receptor genes: geneticand nongenetic potential to generate variability. Hum. Genet. 75:300–310, 1987.
- [27299] 851.Isobe, M.; Huebner, K.; Erikson, J.; Peterson, R. C.;

Bollum, F.J.; Chang, L. M. S.; Croce, C. M.: Chromosome localization of the gene for human terminal deoxynucleotidyltransferase to region 10q23–q25. *Proc. Nat. Acad. Sci.* 82: 5836–5840, 1985.

[27300] 852. Landau, N. R.; St. John, T. P.; Weissman, I. L.; Wolf, S. C.; Silverstone, A. E.; Baltimore, D.: Cloning of terminal transferase cDNA by antibody screening. *Proc. Nat. Acad. Sci.* 81: 5836–5840, 1984.

[27301] 853. Riley, L. K.; Morrow, J. K.; Danton, M. J.; Coleman, M. S.: Human terminal deoxyribonucleotidyltransferase: molecular cloning and structural analysis of the gene and 5–prime flanking region. *Proc. Nat. Acad. Sci.* 85: 2489–2493, 1988.

[27302] 854. Thai, T.–H.; Purugganan, M. M.; Roth, D. B.; Kearney, J. F.: Distinct and opposite diversifying activities of terminal transferase splice variants. *Nature Immun.* 3: 457–462, 2002.

[27303] 855. Yang–Feng, T. L.; Landau, N. R.; Baltimore, D.; Francke, U.: The terminal deoxynucleotidyltransferase gene is located on human chromosome 10 (10q23–q24) and on mouse chromosome 19. *Cytogenet. Cell Genet.* 43:121–126, 1986.

[27304] 856. Kasahara, M.; Figueroa, F.; Klein, J.: Random cloning

of genes from mouse chromosome 17. *Proc. Nat. Acad. Sci.* 84: 3325–3328, 1987.

- [27305] 857. Kasahara, M.; Gutknecht, J.; Brew, K.; Spurr, N.; Goodfellow, P.N.: Cloning and mapping of a testis-specific gene with sequence similarity to a sperm-coating glycoprotein gene. *Genomics* 5: 527–534, 1989.
- [27306] 858. Kasahara, M.; Passmore, H. C.; Klein, J.: A testis-specific gene Tpx-1 maps between Pgk-2 and Mep-1 on mouse chromosome 17. *Immunogenetics* 29:61–63, 1989.
- [27307] 859. Gonzatti-Haces, M.; Seth, A.; Park, M.; Copeland, T.; Oroszlan, S.; Vande Woude, G. F.: Characterization of the TPR-MET oncogene p65 and the MET protooncogene p140 protein-tyrosine kinases. *Proc. Nat. Acad. Sci.* 85: 21–25, 1988.
- [27308] 860. Miranda, C.; Minoletti, F.; Greco, A.; Sozzi, G.; Pierotti, M.A.: Refined localization of the human TPR gene to chromosome 1q25 by *in situ* hybridization. *Genomics* 23: 714–715, 1994.
- [27309] 861. Jenkins, N. A.; Gilbert, D. J.; Cho, B. C.; Strobel, M. C.; Williams, S. C.; Copeland, N. G.; Johnson, P. F.: Mouse chromosomal location of the CCAAT/enhancer binding proteins C/EBP-beta (Cebpb), C/EBP-delta (Cebpd), and

CRP1 (Cebpe). Genomics 28: 333–336, 1995.

[27310] 862.Harding, D.; Jeremiah, S. J.; Povey, S.; Burchell, B.: Chromosomal mapping of a human phenol UDP–glucuronosyltransferase, GNT1. Ann.Hum. Genet. 54: 17–21, 1990.

[27311] 863.Harding, D.; Jeremiah, S. J.; Povey, S.; Burchell, B.: PhenolUDP–glucuronosyltransferase is coded by a gene on human chromosome2. (Abstract) Cytogenet. Cell Genet. 51: 1011, 1989.

[27312] 864.Jackson, M. R.; McCarthy, L. R.; Harding, D.; Wilson, S.; Coughtrie,M. W. H.; Burchell, B.: Cloning of a human liver microsomal UDP–glucuronosyltransferase cDNA. Biochem. J. 242: 581–588, 1987.

[27313] 865.Shprintzen, R. J.; Goldberg, R.; Golding–Kushner, K. J.; Marion,R.: Late–onset psychosis in the velo–cardio–facial syndrome. Am.J. Med. Genet. 42: 141–142, 1992.

[27314] 866.Sirotkin, H.; Morrow, B.; DasGupta, R.; Goldberg, R.; Patanjali,S. R.; Shi, G.; Cannizzaro, L.; Shprintzen, R.; Weissman, S. M.; Kucherlapati,R.: Isolation of a new clathrin heavy chain gene with muscle–specific expression from the region commonly deleted in velo–cardio–facial syndrome. Hum. Molec. Genet. 5: 617–624, 1996.

- [27315] 867.Sirotkin, H.; Morrow, B.; Saint-Jore, B.; Puech, A.; Das Gupta,R.; Patanjali, S. R.; Skoultchi, A.; Weissman, S. M.; Kucherlapati,R.: Identification, characterization, and precise mapping of a humangene encoding a novel membrane-spanning protein from the 22q11 regiondeleted in velo-cardio-facial syndrome. *Genomics* 42: 245-251, 1997.
- [27316] 868.Freund, C. L.; Valle, D.: Cloning and initial characterizationof the cDNA and gene for human recoverin. (Abstract) *Am. J. Hum.Genet.* 51 (supl.): A6 only, 1992.
- [27317] 869.Heckenlively, J. R.; Fawzi, A. A.; Oversier, J.; Jordan, B. L.;Aptsiauri, N.: Autoimmune retinopathy: patients with antirecoverinimmunoreactivity and panretinal degeneration. *Arch. Ophthal.* 118:1525-1533, 2000.
- [27318] 870.Hurley, J. B.; Dizhoor, A. M.; Ray, S.; Stryer, L.: Recoverin'srole: conclusion withdrawn. (Letter) *Lancet* 260: 740 only, 1993.
- [27319] 871.McGinnis, J. F.; Lerious, V.; Pazik, J.; Elliott, R. W.: Chromosomalassignment of the recoverin gene and cancer-associated retinopathy. *MammalianGenome* 4: 43-45, 1993.
- [27320] 872.McGinnis, J. F.; Leveille, P. J.: Soluble retinal proteins associatedwith photoreceptor cell death in the rd mouse.

Curr. Eye Res. 4:1127–1135, 1985.

- [27321] 873. McGinnis, J. F.; Stepanik, P. L.; Baehr, W.; Subbaraya, I.; Lerious, V.: Cloning and sequencing of the 23 kDa mouse photoreceptor cell-specific protein. FEBS Lett. 302: 172–176, 1992.
- [27322] 874. Murakami, A.; Yajima, T.; Inana, G.: Isolation of human retinal genes: recoverin cDNA and gene. Biochem. Biophys. Res. Commun. 187:234–244, 1992.
- [27323] 875. Polans, A. S.; Buczylo, J.; Crabb, J.; Palczewski, K.: A photoreceptor calcium binding protein is recognized by autoantibodies obtained from patients with cancer-associated retinopathy. J. Cell Biol. 112:981–989, 1991.
- [27324] 876. Polans, A. S.; Witkowska, D.; Haley, T. L.; Amundson, D.; Baizer, L.; Adamus, G.: Recoverin, a photoreceptor-specific calcium-binding protein, is expressed by the tumor of a patient with cancer-associated retinopathy. Proc. Nat. Acad. Sci. 92: 9176–9180, 1995.
- [27325] 877. Thirkill, C. E.; Roth, A. M.; Keltner, J. L.: Cancer-associated retinopathy. Arch. Ophthalmol. 105: 372–375, 1987.
- [27326] 878. Bischoff, F. R.; Maier, G.; Tilz, G.; Ponstingl, H.: A 47-kDa human nuclear protein recognized by anti-kinetochore autoimmune sera is homologous with the protein

encoded by RCC1, a gene implicated in onset of chromosome condensation. *Proc. Nat. Acad. Sci.* 87: 8617–8621, 1990.

- [27327] 879. Carazo-Salas, R. E.; Guarguaglini, G.; Gruss, O. J.; Segref, A.; Karsenti, E.; Mattaj, I. W.: Generation of GTP-bound Ran by RCC1 is required for chromatin-induced mitotic spindle formation. *Nature* 400:178–181, 1999.
- [27328] 880. Furuno, N.; Nakagawa, K.; Eguchi, U.; Ohtubo, M.; Nishimoto, T.; Soeda, E.: Complete nucleotide sequence of the human RCC1 gene involved in coupling between DNA replication and mitosis. *Genomics* 11: 459–461, 1991.
- [27329] 881. Stadler, H. S.; Padanilam, B. J.; Buetow, K.; Murray, J. C.; Solursh, M.: Identification and genetic mapping of a homeobox gene to the 4p16.1 region of human chromosome 4. *Proc. Nat. Acad. Sci.* 89: 11579–11583, 1992.
- [27330] 882. Wang, W.; Yoshiura, K.; Murray, J.; Lufkin, T.: Assignment of the murine Hmx1 homeobox gene to the proximal region of mouse chromosome 5. *Mammalian Genome* 8: 869–876, 1997.
- [27331] 883. Deguchi, Y.; Agus, D.; Kehrl, J. H.: A human homeobox gene, HB24, inhibits development of CD4⁺ T cells and impairs thymic involution in transgenic mice. *J. Biol. Chem.* 268: 3646–3653, 1993.

- [27332] 884. Deguchi, Y.; Moroney, J. F.; Wilson, G. L.; Fox, C. H.; Winter, H. S.; Kehrl, J. H.: Cloning of a human homeobox gene that resembles a diverged *Drosophila* homeobox gene and is expressed in activated lymphocytes. *New Biologist* 3: 353–363, 1991.
- [27333] 885. Kennedy, M. A.; Rayner, J. C.; Morris, C. M.: Genomic structure, promoter sequence, and revised translation of human homeobox gene HLX1. *Genomics* 22: 348–355, 1994.
- [27334] 886. Mullen, A. C.; Hutchins, A. S.; High, F. A.; Lee, H. W.; Sykes, K. J.; Chodosh, L. A.; Reiner, S. L.: Hlx is induced by and genetically interacts with T-bet to promote heritable T(H)1 gene induction. *Nature Immun.* 3: 652–658, 2002.
- [27335] 887. Nishimura, D. Y.; Purchio, A. F.; Murray, J. C.: Linkage localization of TGFB2 and the human homeobox gene HLX1 to chromosome 1q. *Genomics* 15: 357–364, 1993.
- [27336] 888. Raftery, M. J.; Schwab, M.; Eibert, S. M.; Samstag, Y.; Walczak, H.; Schonrich, G.: Targeting the function of mature dendritic cells by human cytomegalovirus: a multilayered viral defense strategy. *Immunity* 15: 997–1009, 2001.
- [27337] 889. Gough, A. C.; Zhong, S.; Wolf, C. R.; Spurr, N. K.: Chromosome assignment of the human glutathione S-transferase mu-3 gene (GSTM3) to chromosome 1 by gene

specific polymerase chain reaction. *Cytogenet. Cell Genet.* 65: 111–114, 1994.

- [27338] 890. Galiegue-Zouitina, S.; Quief, S.; Hildebrand, M.-P.; Denis, C.; Detourmignies, L.; Lai, J.-L.; Kerckaert, J.-P.: Nonrandom fusion of L-plastin (LCP1) and LAZ3 (BCL6) genes by t(3;13)(q27;q14) chromosomal translocation in two cases of B-cell non-Hodgkin lymphoma. *Genes Chromosomes Cancer* 26: 97–105, 1999.
- [27339] 891. Giometti, C. S.; Anderson, N. L.: A variant of human nonmuscle tropomyosin found in fibroblasts by using two-dimensional electrophoresis. *J. Biol. Chem.* 256: 11840–11846, 1981.
- [27340] 892. Hamaguchi, H.; Ohta, A.; Mukai, R.; Yabe, T.; Yamada, M.: Genetic analysis of human lymphocyte proteins by two-dimensional gel electrophoresis: 1. Detection of genetic variant polypeptides in PHA-stimulated peripheral-blood lymphocyte. *Hum. Genet.* 59: 215–220, 1981.
- [27341] 893. Hamaguchi, H.; Yamada, M.; Noguchi, A.; Fujii, K.; Shibasaki, M.; Mukai, R.; Yabe, T.; Kondo, I.: Genetic analysis of human lymphocyte proteins by two-dimensional gel electrophoresis: 2. Genetic polymorphism of lymphocyte cytosol 64K polypeptide. *Hum. Genet.* 60: 176–180, 1982.
- [27342] 894. Hamaguchi, H.; Yamada, M.; Shibasaki, M.; Kondo, I.:

Genetic analysis of human lymphocyte proteins by two-dimensional gel electrophoresis: 4. Genetic polymorphism of cytosol 100k polypeptide. Hum. Genet. 62:148–151, 1982.

[27343] 895. Hamaguchi, H.; Yamada, M.; Shibasaki, M.; Mukai, R.; Yabe, T.; Kondo, I.: Genetic analysis of human lymphocyte proteins by two-dimensional gel electrophoresis: 3. Frequent occurrence of genetic variants in some abundant polypeptides of PHA-stimulated peripheral blood lymphocytes. Hum. Genet. 62: 142–147, 1982.

[27344] 896. Klose, J.: Protein mapping by combined isoelectric focusing and electrophoresis of mouse tissue: a novel approach to testing for induced point mutations in mammals. Humangenetik 26: 231–243, 1975.

[27345] 897. Klose, J.; Feller, M.: Genetic variability of proteins from plasma membranes and cytosols of mouse organs. Biochem. Genet. 19: 859–870, 1981.

[27346] 898. Kondo, I.; Hamaguchi, H.: Evidence for the close linkage between lymphocyte cytosol polypeptide with molecular weight of 64,000 (LCP1) and esterase D. Am. J. Hum. Genet. 37: 1106–1111, 1985.

[27347] 899. Kondo, I.; Hamaguchi, H.: Study of the linkage relationship between LCP1 and ESD. (Abstract) Cytogenet. Cell

Genet. 40: 672 only, 1985.

- [27348] 900.Akashi, K.; Traver, D.; Miyamoto, T.; Weissman, I. L.: A clonogenic common myeloid progenitor that gives rise to all myeloid lineages. *Nature* 404:193–197, 2000.
- [27349] 901.Arcasoy, M. O.; Degar, B. A.; Harris, K. W.; Forget, B. G.: Familial erythrocytosis associated with a short deletion in the erythropoietin receptor gene. *Blood* 89: 4628–4635, 1997.
- [27350] 902.Budarf, M.; Huebner, K.; Emanuel, B.; Croce, C. M.; Copeland, N.G.; Jenkins, N. A.; D'Andrea, A. D.: Assignment of the erythropoietin receptor (EPOR) gene to mouse chromosome 9 and human chromosome 19. *Genomics* 8:575–578, 1990.
- [27351] 903.D'Andrea, A. D.; Yoshimura, A.; Youssoufian, H.; Zon, L. I.; Koo, J.-W.; Lodish, H. F.: The cytoplasmic region of the erythropoietin receptor contains nonoverlapping positive and negative growth-regulatory domains. *Molec. Cell. Biol.* 11: 1980–1987, 1991.
- [27352] 904.D'Andrea, A. D.; Zon, L. I.: Erythropoietin receptor: subunit structure and activation. *J. Clin. Invest.* 86: 681–687, 1990.
- [27353] 905.de la Chapelle, A.; Traskelin, A.-L.; Juvonen, E.: Truncated erythropoietin receptor causes dominantly inherited

benign human erythrocytosis. Proc.Nat. Acad. Sci. 90: 4495–4499, 1993.

- [27354] 906.Divoky, V.; Liu, Z.; Ryan, T. M.; Prchal, J. F.; Townes, T. M.;Prchal, J. T.: Mouse model of congenital polycythemia: homologousreplacement of murine gene by mutant human erythropoietin receptorgene. Proc. Nat. Acad. Sci. 98: 986–991, 2001.
- [27355] 907.Hess, G.; Rose, P.; Gamm, H.; Papadileris, S.; Huber, C.; Seliger,B.: Molecular analysis of the erythropoietin receptor system in patientswith polycythaemia vera. Brit. J. Haemat. 88: 794–802, 1994.
- [27356] 908.Jones, S. S.; D'Andrea, A. D.; Haines, L. L.; Wong, G. G.: Humanerythropoietin receptor: cloning, expression, and biologic characterization. Blood 76:31–35, 1990.
- [27357] 909.Kralovics, R.; Indrak, K.; Stopka, T.; Berman, B. W.; Prchal,J. F.; Prchal, J. T.: Two new EPO receptor mutations: truncated EPOreceptors are most frequently associated with primary familial andcongenital polycythemias. Blood 90: 2057–2061, 1997.
- [27358] 910.Kralovics, R.; Sokol, L.; Prchal, J. T.: Absence of polycythemiain a child with a unique erythropoietin receptor mutation in a familywith autosomal dominant primary polycythemia. J. Clin. Invest. 102:124–129, 1998.

- [27359] 911. Le Couedic, J.-P.; Mitjavila, M.-T.; Villeval, J.-L.; Feger, F.; Gobert, S.; Mayeux, P.; Casadevall, N.; Vainchenker, W.: Missense mutation of the erythropoietin receptor is a rare event in human erythroid malignancies. *Blood* 87: 1502–1511, 1996.
- [27360] 912. Longmore, G. D.; Lodish, H. F.: An activating mutation in the murine erythropoietin receptor induces erythroleukemia in mice: a cytokine receptor superfamily oncogene. *Cell* 67: 1089–1102, 1991.
- [27361] 913. Maouche, L.; Tournamille, C.; Hattab, C.; Boffa, G.; Cartron, J.-P.; Chretien, S.: Cloning of the gene encoding the human erythropoietin receptor. *Blood* 78: 2557–2563, 1991.
- [27362] 914. Noguchi, C. T.; Bae, K. S.; Chin, K.; Wada, Y.; Schechter, A. N.; Hankins, W. D.: Cloning of the human erythropoietin receptor gene. *Blood* 78: 2548–2556, 1991.
- [27363] 915. Penny, L. A.; Forget, B. G.: Genomic organization of the human erythropoietin receptor gene. *Genomics* 11: 974–980, 1991.
- [27364] 916. Percy, M. J.; McMullin, M. F.; Roques, A. W. W.; Westwood, N. B.; Acharya, J.; Hughes, A. E.; Lappin, T. R. J.; Pearson, T. C.: Erythrocytosis due to a mutation in the erythropoietin receptor gene. *Brit. J. Haemat.* 100: 407–410,

1998.

- [27365] 917. Sistonen, P.; Traskelin, A.-L.; Lehvaslaiho, H.; de la Chapelle, A.: Genetic mapping of the erythropoietin receptor gene. *Hum. Genet.* 92:299–301, 1993.
- [27366] 918. Sokol, L.; Luhovy, M.; Guan, Y.; Prchal, J. F.; Semenza, G. L.; Prchal, J. T.: Primary familial polycythemia: a frameshift mutation in the erythropoietin receptor gene and increased sensitivity of erythroid progenitors to erythropoietin. *Blood* 86: 15–22, 1995.
- [27367] 919. Ward, J. C.; Harris, K. W.; Penny, L. A.; Forget, B. G.; Kitamura, T.; Winkelmann, J. C.: A structurally abnormal erythropoietin receptor gene in a human erythroleukemia cell line. *Exp. Hemat.* 20: 371–373, 1992.
- [27368] 920. Winkelmann, J. C.; Penny, L. A.; Deaven, L. L.; Forget, B. G.; Jenkins, R. B.: The gene for the human erythropoietin receptor: analysis of the coding sequence and assignment to chromosome 19p. *Blood* 76:24–30, 1990.
- [27369] 921. Yu, X.; Lin, C.-S.; Costantini, F.; Noguchi, C. T.: The human erythropoietin receptor gene rescues erythropoiesis and developmental defects in the erythropoietin receptor null mouse. *Blood* 98: 475–477, 2001.
- [27370] 922. Baier, L. J.; Sacchettini, J. C.; Knowler, W. C.; Eads, J.; Paolisso, G.; Tataranni, P. A.; Mochizuki, H.; Bennett, P. H.;

Bogardus, C.; Prochazka, M.: An amino acid substitution in the human intestinal fatty acid binding protein is associated with increased fatty acid binding, increased fat oxidation, and insulin resistance. *J. Clin. Invest.* 95: 1281–1287, 1995.

[27371] 923. Carlsson, M.; Orho-Melander, M.; Hedenbro, J.; Almgren, P.; Groop, L. C.: The T54 allele of the intestinal fatty acid-binding protein 2 is associated with a parental history of stroke. *J. Clin. Endocr. Metab.* 85: 2801–2804, 2000.

[27372] 924. Georgopoulos, A.; Aras, O.; Tsai, M. Y.: Codon-54 polymorphism of the fatty acid-binding protein 2 gene is associated with elevation of fasting and postprandial triglyceride in type 2 diabetes. *J. Clin. Endocr. Metab.* 85: 3155–3160, 2000.

[27373] 925. Hegele, R. A.; Harris, S. B.; Hanley, A. J. G.; Sadikian, S.; Connelly, P. W.; Zinman, B.: Genetic variation of intestinal fatty acid-binding protein associated with variation in body mass in aboriginal Canadians. *J. Clin. Endocr. Metab.* 81: 4334–4337, 1996.

[27374] 926. Polymeropoulos, M. H.; Rath, D. S.; Xiao, H.; Merrill, C. R.: Trinucleotide repeat polymorphism at the human intestinal fatty acid binding protein gene (FABP2). *Nucleic Acids Res.* 18: 7198 only, 1991.

- [27375] 927.Prochazka, M.; Lillioja, S.; Tait, J. F.; Knowler, W. C.; Mott,D. M.; Spraul, M.; Bennett, P. H.; Bogardus, C.: Linkage of chromosomal markers on 4q with a putative gene determining maximal insulin action in Pima Indians. Diabetes 42: 514–519, 1993.
- [27376] 928.Sipilainen, R.; Uusitupa, M.; Heikkinen, S.; Rissanen, A.; Laakso,M.: Variants in the human intestinal fatty acid binding protein 2 gene in obese subjects. J. Clin. Endocr. Metab. 82: 2629–2632, 1997.
- [27377] 929.Sparkes, R. S.; Mohandas, T.; Heinzmann, C.; Gordon, J. I.; Klisak,I.; Zollman, S.; Sweetser, D. A.; Ragunathan, L.; Winokur, S.; Lusi,A. J.: Human fatty acid binding protein assignments intestinal to 4q28–4q31 and liver to 2p11. (Abstract) Cytogenet. Cell Genet. 46:697 only, 1987.
- [27378] 930.Sweetser, D. A.; Birkenmeier, E. H.; Klisak, I. J.; Zollman, S.;Sparkes, R. S.; Mohandas, T.; Lusi, A. J.; Gordon, J. I.: The human and rodent intestinal fatty acid binding protein genes: a comparative analysis of their structure, expression, and linkage relationships. J.Biol. Chem. 262: 16060–16071, 1987.
- [27379] 931.Weber, J. L.; Kwitek, A. E.; May, P. E.; Polymeropoulos, M.: Dinucleotide repeat polymorphism at the D12S43 locus. Nucleic Acids Res. 18: 4637 only, 1990.

- [27380] 932. Weber, J. L.; May, P. E.: Abundant class of human DNA polymorphisms which can be typed using the polymerase chain reaction. *Am. J. Hum. Genet.* 44: 388–396, 1989.
- [27381] 933. Buckle, V. J.; Fujita, N.; Bateson, A. N.; Darlison, M. G.; Barnard, E. A.: Localization of human GABA-A receptor subunit genes to chromosomes 4 and 5. (Abstract) *Cytogenet. Cell Genet.* 51: 972 only, 1989.
- [27382] 934. Danciger, M.; Farber, D. B.; Kozak, C. A.: Genetic mapping of three GABA-A receptor-subunit genes in the mouse. *Genomics* 16: 361–365, 1993.
- [27383] 935. Hadingham, K. L.; Wingrove, P.; Le Bourdelles, B.; Palmer, K. J.; Ragan, C. I.; Whiting, P. J.: Cloning of cDNA sequences encoding human alpha-2 and alpha-3 gamma-aminobutyric acid-A receptor subunits and characterization of the benzodiazepine pharmacology of recombinant alpha-1-, alpha-2-, alpha-3-, and alpha-5-containing human gamma-aminobutyric acid-A receptors. *Molec. Pharm.* 43: 970–975, 1993.
- [27384] 936. Low, K.; Crestani, F.; Keist, R.; Benke, D.; Brunig, I.; Benson, J. A.; Fritschy, J.-M.; Rulicke, T.; Bluethmann, H.; Mohler, H.; Rudolph, U.: Molecular and neuronal substrate for the selective attenuation of anxiety. *Science* 290: 131–134, 2000.

- [27385] 937.Glatt, K.; Glatt, H.; Lalande, M.: Structure and organization of GABRB3 and GABRA5. *Genomics* 41: 63–69, 1997.
- [27386] 938.Russek, S. J.; Farb, D. H.: Mapping of the beta-2 subunit gene(GABRB2) to microdissected human chromosome 5q34–q35 defines a genecluster for the most abundant GABA-A receptor isoform. *Genomics* 23:528–533, 1994.
- [27387] 939.Geiman, T. M.; Durum, S. K.; Muegge, K.: Characterization of geneexpression, genomic structure, and chromosomal localization of Hells(Lsh). *Genomics* 54: 477–483, 1998.
- [27388] 940.Geiman, T. M.; Muegge, K.: Lsh, an SNF2/helicase family member,is required for proliferation of mature T lymphocytes. *Proc. Nat.Acad. Sci.* 97: 4772–4777, 2000.
- [27389] 941.Jarvis, C. D.; Geiman, T.; Vila-Storm, M. P.; Osipovich, O.; Akella,U.; Candeias, S.; Nathan, I.; Durum, S. K.; Muegge, K.: A novel putativehelicase produced in early murine lymphocytes. *Gene* 169: 203–207,1996.
- [27390] 942.Futamura, M.; Nishimori, H.; Shiratsuchi, T.; Saji, S.; Nakamura,Y.; Tokino, T.: Molecular cloning, mapping, and characterization of a novel human gene, MTA1-L1, showing homology to a metastasis-associatedgene, MTA1. *J. Hum. Genet.* 44: 52–56, 1999.

- [27391] 943. Brenner, R.; Perez, G. J.; Bonev, A. D.; Eckman, D. M.; Kosek, J. C.; Wiler, S. W.; Patterson, A. J.; Nelson, M. T.; Aldrich, R. W.: Vasoregulation by the beta-1 subunit of the calcium-activated potassium channel. *Nature* 407: 870–876, 2000.
- [27392] 944. Jiang, Z.; Wallner, M.; Meera, P.; Toro, L.: Human and rodent MaxiK channel beta-subunit genes: cloning and characterization. *Genomics* 55:57–67, 1999.
- [27393] 945. Knaus, H.-G.; Folander, K.; Garcia-Calvo, M.; Garcia, M. L.; Kaczorowski, G. J.; Smith, M.; Swanson, R.: Primary sequence and immunological characterization of beta-subunit of high conductance Ca^{2+} -activated K^{+} channel from smooth muscle. *J. Biol. Chem.* 269: 17274–17278, 1994.
- [27394] 946. Meera, P.; Wallner, M.; Jiang, Z.; Toro, L.: A calcium switch for the functional coupling between alpha (hslo) and beta subunits (K^{+} , Ca -beta) of maxi K channels. *FEBS Lett.* 382: 84–88, 1996.
- [27395] 947. Tseng-Crank, J.; Godinot, N.; Johansen, T. E.; Ahring, P. K.; Strobaek, D.; Mertz, R.; Foster, C. D.; Olesen, S.-P.; Reinhart, P. H.: Cloning, expression, and distribution of a Ca^{2+} -activated K^{+} channel beta-subunit from human brain. *Proc. Nat. Acad. Sci.* 93: 9200–9205, 1996.

- [27396] 948.Xie, J.; Black, D. L.: A CaMK IV responsive RNA element mediates depolarization-induced alternative splicing of ion channels. *Nature* 410:936–439, 2001.
- [27397] 949.Tsujimoto, A.; Nyunoya, H.; Morita, T.; Sato, T.; Shimotohno, K.: Isolation of cDNAs for DNA-binding proteins which specifically bind to a tax-responsive enhancer element in the long terminal repeat of human T-cell leukemia virus type I. *J. Virol.* 65: 1420–1426, 1991.
- [27398] 950.Kim, D.; Song, I.; Keum, S.; Lee, T.; Jeong, M.-J.; Kim, S.-S.; McEnery, M. W.; Shin, H.-S.: Lack of the burst firing of thalamocortical relay neurons and resistance to absence seizures in mice lacking alpha-1G-type Ca^{2+} channels. *Neuron* 31: 35–45, 2001.
- [27399] 951.Perez-Reyes, E.; Cribbs, L. L.; Daud, A.; Lacerda, A. E.; Barclay, J.; Williamson, M. P.; Fox, M.; Rees, M.; Lee, J.-H.: Molecular characterization of a neuronal low-voltage-activated T-type calcium channel. *Nature* 391:896–900, 1998.
- [27400] 952.Goto, K.; Kondo, H.: Molecular cloning and expression of a 90-kDa diacylglycerol kinase that predominantly localizes in neurons. *Proc.Nat. Acad. Sci.* 90: 7598–7602, 1993.
- [27401] 953.Ding, L.; Traer, E.; McIntyre, T. M.; Zimmerman, G. A.;

Prescott, S. M.: The cloning and characterization of a novel human diacylglycerol kinase, DGK- ι . *J. Biol. Chem.* 273: 32746–32752, 1998.

[27402] 954. Nothwang, H. G.; Kim, H. G.; Aoki, J.; Geisterfer, M.; Kubart, S.; Wegner, R. D.; van Moers, A.; Ashworth, L. K.; Haaf, T.; Bell, J.; Arai, H.; Tommerup, N.; Ropers, H. H.; Wirth, J.: Functional hemizyosity of PAFAH1B3 due to a PAFAH1B3-CLK2 fusion gene in a female with mental retardation, ataxia and atrophy of the brain. *Hum. Molec. Genet.* 10: 797–806, 2001.

[27403] 955. Katsanis, N.; Fisher, E. M. C.: Identification, expression, and chromosomal localization of ubiquitin conjugating enzyme 7 (UBE2G2), a human homologue of the *Saccharomyces cerevisiae* Ubc7 gene. *Genomics* 51: 128–131, 1998.

[27404] 956. Rose, S. A.; Leek, J. P.; Moynihan, T. P.; Ardley, H. C.; Markham, A. F.; Robinson, P. A.: Assignment of the ubiquitin conjugating enzyme gene, UBE2G2, to human chromosome band 21q22.3 by in situ hybridization. *Cytogenet. Cell Genet.* 83: 98–99, 1998.

[27405] 957. Beisswanger, R.; Corbeil, D.; Vannier, C.; Thiele, C.; Dohrmann, U.; Kellner, R.; Ashman, K.; Niehrs, C.; Huttner, W. B.: Existence of distinct tyrosylprotein sulfotransferase

genes: molecular characterization of tyrosylprotein sulfotransferase-2. *Proc. Nat. Acad. Sci.* 95:11134–11139, 1998.

[27406] 958. Ouyang, Y.-B.; Lane, W. S.; Moore, K. L.: Tyrosylprotein sulfotransferase: purification and molecular cloning of an enzyme that catalyzes tyrosine O-sulfation, a common posttranslational modification of eukaryotic proteins *Proc. Nat. Acad. Sci.* 95: 2896–2901, 1998.

[27407] 959. Ju, Y.-T.; Chang, A. C. Y.; She, B.-R.; Tsaur, M.-L.; Hwang, H.-M.; Chao, C. C.-K.; Cohen, S. N.; Lin-Chao, S.: Gas7: a gene expressed preferentially in growth-arrested fibroblasts and terminally differentiated Purkinje neurons affects neurite formation. *Proc. Nat. Acad. Sci.* 95:11423–11428, 1998.

[27408] 960. Kurtz, A.; Zimmer, A.: Interspecies fluorescence in situ hybridization further defines synteny homology between mouse chromosome 11 and human chromosome 17. *Mammalian Genome* 6: 379–380, 1995.

[27409] 961. Megonigal, M. D.; Cheung, N.-K. V.; Rappaport, E. F.; Nowell, P. C.; Wilson, R. B.; Jones, D. H.; Addya, K.; Leonard, D. G. B.; Kushner, B. H.; Williams, T. M.; Lange, B. J.; Felix, C. A.: Detection of leukemia-associated MLL-GAS7 translocation early during chemotherapy with DNA

topoisomerase II inhibitors. *Proc. Nat. Acad. Sci.*
97:2814–2819, 2000.

- [27410] 962.Chrast, R.; Scott, H. S.; Chen, H.; Kudoh, J.; Rossier, C.; Minoshima,S.; Wang, Y.; Shimizu, N.; Antonarakis, S. E.: Cloning of two humanhomologs of the *Drosophila* single-minded gene SIM1 on chromosome 6qand SIM2 on 21q within the Down syndrome chromosomal region. *GenomeRes.* 7: 615–624, 1997.
- [27411] 963.Holder, J. L., Jr.; Butte, N. F.; Zinn, A. R.: Profound obesityassociated with a balanced translocation that disrupts the SIM1 gene. *Hum.Molec. Genet.* 9: 101–108, 2000.
- [27412] 964.Michaud, J. L.; Boucher, F.; Melnyk, A.; Gauthier, F.; Goshu, E.;Levy, E.; Mitchell, G. A.; Himms–Hagen, J.; Fan, C.–M.: Sim1 haploinsufficiencycauses hyperphagia, obesity and reduction of the paraventricular nucleusof the hypothalamus. *Hum. Molec. Genet.* 10: 1465–1473, 2001.
- [27413] 965.Fan, C. M.; Kuwana, E.; Bulfone, A.; Fletcher, C. F.; Copeland,N. G.; Jenkins, N. A.; Crews, S.; Martinez, S.; Puellas, L.; Rubenstein,J. L.; Tessier–Lavigne, M.: Expression patterns of two murine homologsof *Drosophila* single-minded suggest possible roles in embryonic patterningand in the pathogenesis of Down syndrome. *Molec.*

Cell. Neurosci. 7:1–16, 1996.

- [27414] 966. Duke-Cohan, J. S.; Gu, J.; McLaughlin, D. F.; Xu, Y.; Freeman, G. J.; Schlossman, S. F.: Attractin (DPPT-L), a member of the CUB family of cell adhesion and guidance proteins, is secreted by activated human T lymphocytes and modulates immune cell interactions. *Proc. Nat. Acad. Sci.* 95: 11336–11341, 1998.
- [27415] 967. Gunn, T. M.; Miller, K. A.; He, L.; Hyman, R. W.; Davis, R. W.; Azarani, A.; Schlessman, S. F.; Duke-Cohan, J. S.; Barsh, G. S.: The mouse mahogany locus encodes a trans-membrane form of human attractin. *Nature* 398:152–156, 1999.
- [27416] 968. He, L.; Gunn, T. M.; Bouley, D. M.; Lu, X.-Y.; Watson, S. J.; Schlossman, S. F.; Duke-Cohan, J. S.; Barsh, G. S.: A biochemical function for attractin in agouti-induced pigmentation and obesity. *Nature Genet.* 27:40–47, 2001.
- [27417] 969. Tang, W.; Gunn, T. M.; McLaughlin, D. F.; Barsh, G. S.; Schlossman, S. F.; Duke-Cohan, J. S.: Secreted and membrane attractin result from alternative splicing of the human ATRN gene. *Proc. Nat. Acad. Sci.* 97: 6025–6030, 2000.
- [27418] 970. Fields, S.; Song, O.: A novel genetic system to detect protein–protein interactions. (Letter) *Nature* 340: 245–246,

1989.

- [27419] 971. Gordon, D. M.; Shi, Q.; Dancis, A.; Pain, D.: Maturation of frataxin within mammalian and yeast mitochondria: one-step processing by matrix processing peptidase. *Hum. Molec. Genet.* 8: 2255–2262, 1999.
- [27420] 972. Koutnikova, H.; Campuzano, V.; Koenig, M.: Maturation of wild-type and mutated frataxin by the mitochondrial processing peptidase. *Hum. Molec. Genet.* 7: 1485–1489, 1998.
- [27421] 973. Lewis, T. M.; Roberts, M. L.; Bretag, A. H.: Immunolabeling for VDAC, the mitochondrial voltage-dependent anion channel, on sarcoplasmic reticulum from amphibian skeletal muscle. *Neurosci. Lett.* 181: 83–86, 1994.
- [27422] 974. Campbell, R. D.; Porter, R. R.: Molecular cloning and characterization of the gene coding for human complement protein factor B. *Proc. Nat. Acad. Sci.* 80: 4464–4468, 1983.
- [27423] 975. Akao, Y.; Utsumi, K. R.; Naito, K.; Ueda, R.; Takahashi, T.; Yamada, K.: Chromosomal assignments of genes coding for human leukocyte common antigen, T-200, and lymphocyte function-associated antigen 1, LFA-1 beta subunit. *Somat. Cell Molec. Genet.* 13: 273–278, 1987.
- [27424] 976. Kondo, I.; Ikeuchi, T.; Nishigaki, I.; Takita, H.; Fujiki,

K.;Takahashi, Y.; Hamaguchi, H.: Assignment of the gene for LCP1 on chromosome 13. (Abstract) Cytogenet. Cell Genet. 40: 673 only, 1985.

- [27425] 977.Xiao, H.; Neuveut, C.; Tiffany, H. L.; Benkirane, M.; Rich, E.A.; Murphy, P. M.; Jeang, K.-T.: Selective CXCR4 antagonism by Tat:implications for in vivo expansion of coreceptor use by HIV-1. Proc.Nat. Acad. Sci. 97: 11466-11471, 2000.
- [27426] 978.David, V.; Fauchet, R.; Phengsavath, H.; Guenet, L.; Le Gall, J. Y.: Properdin factor B (Bf) polymorphism: subtyping of SS phenotypes. Hum.Genet. 64: 189-190, 1983.
- [27427] 979.Davis, C. A.; Forristal, J.: Partial properdin deficiency. J.Clin. Lab. Med. 96: 633-639, 1980.
- [27428] 980.Davrinche, C.; Abbal, M.; Clerc, A.: Molecular characterization of human complement factor B subtypes. Immunogenetics 32: 309-312, 1990.
- [27429] 981.Dornan, J.; Allan, P.; Noel, E. P.; Larsen, B.; Farid, N. R.: Properdin factor B(Bf) allele Bf(F1) specifies an HLA-B18 diabetogenic haplotype. Diabetes 29: 423-427, 1980.
- [27430] 982.Dunham, I.; Sargent, C. A.; Trowsdale, J.; Campbell, R. D.: The orientation and location of the complement genes in the human MHC by pulsed field gel electrophoresis. (Abstract) Complement 4: 152-153, 1987.

- [27431] 983.Dykes, D. D.; DeFurio, C. M.; Polesky, H. F.: Five new rare variants of the properdin factor B (BF) locus. *Am. J. Hum. Genet.* 35: 652–655, 1983.
- [27432] 984.Hauptmann, G.; Tongio, M. M.; Klein, J.; Mayer, S.; Cinqualbre, J.; Jeanblanc, B.; Kieny, R.; Mauff, G.; Federmann, G.: Le facteur B de la properdine: polymorphism, lieu de synthèse et premier cas de déficit génétique. *Nouv. Presse Med.* 9: 45 only, 1980.
- [27433] 985.Malavasi, F.; Olivetti, E.; Milanese, C.; Carbonara, A. O.: Properdin factor B polymorphism in continental Italy and Sardinia. *Hum. Genet.* 58:209–212, 1981.
- [27434] 986.Mejia, J. E.; Jahn, I.; de la Salle, H.; Hauptmann, G.: Human factor B: complete cDNA sequence of the BF*S allele. *Hum. Immun.* 39:49–53, 1994.
- [27435] 987.Nerl, C.; O'Neill, G. J.: Factor B polymorphism in North American blacks: study of a new variant Bf F1.35. *Hum. Genet.* 61: 357–359, 1982.
- [27436] 988.Ohayon, E.; de Mouzon, A.; Hauptmann, G.; Klein, J.; Ducos, J.: Genetic linkage between Bf S0.7 (Bf S1) and HLA-Bw50. *Hum. Genet.* 54:417–418, 1980.
- [27437] 989.Raum, D.; Alper, C. A.; Stein, R.; Gabbay, K. H.: Genetic marker for insulin-dependent diabetes mellitus. *Lancet* II: 1208–1210, 1979.

- [27438] 990.Raum, D.; Balner, H.; Petersen, B. H.; Alper, C. A.: Genetic polymorphism of serum complement components in the chimpanzee. *Immunogenetics* 10:455–468, 1980.
- [27439] 991.Raum, D.; Glass, D.; Carpenter, C. B.; Alper, C. A.; Schur, P.H.: The chromosomal order of genes controlling the major histocompatibility complex, properdin factor B, and deficiency of the second component of complement. *J. Clin. Invest.* 58: 1240–1248, 1976.
- [27440] 992.Raum, D.; Glass, D.; Carpenter, C. B.; Schur, P. H.; Alper, C.A.: Mapping for the structural gene for the second component of complement with respect to the human major histocompatibility complex. *Am. J. Hum. Genet.* 31: 35–41, 1979.
- [27441] 993.Raum, D.; Surgenor, T.; Awdeh, Z.; Marcus, D.; Blumenthal, M.; Yunis, E. J.; Alper, C. A.: An unusual 'morphologic' variant of BfS. *Am. J. Hum. Genet.* 36: 346–351, 1984.
- [27442] 994.Rittner, C.; Grosse-Wilde, H.; Albert, E. D.: Localization of the Bf locus within the HLA region: report on an informative family and critical evaluation of available data on Bf mapping. *Hum. Genet.* 35:79–82, 1976.
- [27443] 995.Rittner, C.; Grosse-Wilde, H.; Rittner, B.; Netzel, B.; Scholz, S.; Lorenz, H.; Albert, E. D.: Linkage group HL–

A-MLC-Bf (properdin factor B): the site of the Bf locus at the immunogenetic linkage group on chromosome 6. Humangenetik 27: 173-183, 1975.

[27444] 996. Teisberg, P.; Olaisen, B.; Gedde-Dahl, T., Jr.; Thorsby, E.: On the localization of the Gb locus within the MHS region of chromosome 6. Tissue Antigens 5: 257-261, 1975.

[27445] 997. WHO-IUIS Nomenclature Sub-Committee: Nomenclature for human complement factor B*2. Europ. J. Immunogenet. 20: 307-309, 1993.

[27446] 998. Wyatt, R. J.; Julian, B. A.; Galla, J. H.: Properdin deficiency with IgA nephropathy. (Letter) New Eng. J. Med. 305: 1097 only, 1981.

[27447] 999. Ziegler, J. B.; Alper, C. A.: Properdin factor B and histocompatibility loci linked in the rhesus monkey. Nature 254: 609-610, 1975.

[27448] 1000. Kondo, I.; Shin, K.; Honmura, S.; Nakajima, H.; Yamamura, E.; Satoh, H.; Terauchi, M.; Usuki, Y.; Takita, H.; Hamaguchi, H.: A case report of a patient with retinoblastoma and chromosome 13q deletion: assignment of a new gene (gene for LCP1) on human chromosome 13. Hum. Genet. 71: 263-266, 1985.

[27449] 1001. Lin, C.-S.; Aebersold, R. H.; Kent, S. B.; Varma, M.;

Leavitt,J.: Molecular cloning and characterization of plas-
tin, a human leukocyteprotein expressed in transformed
human fibroblasts. Molec. Cell.Biol. 8: 4659–4668, 1988.

[27450] 1002.Lin, C.–S.; Chang, C.–H.; Huynh, T.: The murine L–
plastin genepromoter: identification and comparison with
the human L–plastin genepromoter. DNA Cell Biol. 16:
9–16, 1997.

[27451] 1003.Lin, C.–S.; Park, T.; Chen, Z. P.; Leavitt, J.: Human
plastingenes: comparative gene structure, chromosome
location, and differentialexpression in normal and neo–
plastic cells. J. Biol. Chem. 268: 2781–2792,1993.

[27452] 1004.McConkey, E. H.; Taylor, B. J.; Phan, D.: Human het–
erozygosity:a new estimate. Proc. Nat. Acad. Sci. 76:
6500–6504, 1979.

[27453] 1005.Murayama, N.; Tanaka, Y.; Hanyu, M.; Kobayashi, K.;
Hamaguchi,H.; Kondo, I.: Assignment of I–plastin to
13q14.3. (Abstract) HumanGenome Mapping Workshop 93
27 only, 1993.

[27454] 1006.O'Farrell, P. Z.; Goodman, H. M.; O'Farrell, P. H.:
High resolutiontwo–dimensional electrophoresis of basic
as well as acidic proteins. Cell 12:1133–1142, 1977.

[27455] 1007.Walton, K. E.; Steyer, D.; Gruenstein, E. I.: Genetic
polymorphismin normal human fibroblasts as analyzed by

two-dimensional polyacrylamidegel electrophoresis. J. Biol. Chem. 254: 7951–7960, 1979.

- [27456] 1008.Zu, Y.; Kohno, M.; Kubota, I.; Nishida, E.; Hanaoka, M.; Namba,Y.: Characterization of interleukin 2 stimulated 65-kilodalton phosphoprotein in human T cells. Biochemistry 29: 1055–1062, 1990.
- [27457] 1009.Zu, Y.; Shigesada, K.; Nishida, E.; Kubota, I.; Kohno, M.; Hanaoka,M.; Namba, Y.: 65-kilodalton protein phosphorylated by interleukin2 stimulation bears two putative actin-binding sites and two calcium-bindingsites. Biochemistry 29: 8319–8324, 1990.
- [27458] 1010.Aggarwal, B. B.; Eessalu, T. E.; Hass, P. E.: Characterizationof receptors for human tumour necrosis factor and their regulationby gamma-interferon. Nature 318: 665–667, 1985.
- [27459] 1011.Evans, A. M.; Petersen, J. W.; Sekhon, G. S.; DeMars, R.: Mappingof prolactin and tumor necrosis factor-beta genes on human chromosome6p using lymphoblastoid cell deletion mutants. Somat. Cell Molec.Genet. 15: 203–213, 1989.
- [27460] 1012.Gray, P. W.; Aggarwal, B. B.; Benton, C. V.; Bringman, T. S.; Henzel,W. J.; Jarrett, J. A.; Leung, D. W.; Moffat, B.; Ng, P.; Svedersky,L. P.; Palladino, M. A.; Nedwin, G. E.:

Cloning and expression of cDNA for human lymphotoxin, a lymphokine with tumour necrosis activity. *Nature* 312:721–724, 1984.

- [27461] 1013. Jongeneel, C. V.; Briant, L.; Udalova, I. A.; Sevin, A.; Nedospasov, S. A.; Cambon-Thomsen, A.: Extensive genetic polymorphism in the human tumor necrosis factor region and relation to extended HLA haplotypes. *Proc. Nat. Acad. Sci.* 88: 9717–9721, 1991.
- [27462] 1014. Koss, K.; Satsangi, J.; Fanning, G. C.; Welsh, K. I.; Jewell, D. P.: Cytokine (TNF- α , LT- α , and IL-10) polymorphisms in inflammatory bowel diseases and normal controls: differential effects on production and allele frequencies. *Genes Immun.* 1: 185–190, 2000.
- [27463] 1015. Camara, V. M.; Harding, J. W.; Prieur, D. J.: Inherited lysozyme deficiency in rabbits: the absence of a primary isozyme of lysozyme as the cause of the condition. *Lab. Invest.* 63: 544–550, 1990.
- [27464] 1016. Canet, D.; Sunde, M.; Last, A. M.; Miranker, A.; Spencer, A.; Robinson, C. V.; Dobson, C. M.: Mechanistic studies of the folding of human lysozyme and the origin of amyloidogenic behavior in its disease-related variants. *Biochemistry* 38: 6419–6427, 1999.
- [27465] 1017. Dayhoff, M. O.: *Atlas of Protein Sequence and Struc-*

ture. Lactalbumin and Lysozyme. Washington: National Biomedical Research Foundation (pub.) 5: 1972. Pp. D133–D140.

- [27466] 1018. Fleming, A.: On a remarkable bacteriolytic element found in tissues and secretions. *Proc. Roy. Soc. Ser. B.* 93: 306–317, 1922.
- [27467] 1019. Fleming, A.; Allison, V. D.: Observations on a bacteriolytic substance ('lysozyme') found in secretions and tissues. *Brit. J. Exp. Path.* 3: 252–260, 1922.
- [27468] 1020. Greenwald, R. A.; Cantor, J. O.; Prieur, D. J.; Young, D. M.: Composition of cartilage from lysozyme-deficient rabbits. *Biochim. Biophys. Acta* 385: 435–437, 1975.
- [27469] 1021. Neufeld, E. L.: Personal Communication. Bethesda, Maryland 1972.
- [27470] 1022. Pepys, M. B.; Hawkins, P. N.; Booth, D. R.; Vigushin, D. M.; Tennent, G. A.; Soutar, A. K.; Totty, N.; Nguyen, O.; Blake, C. C. F.; Terry, C. J.; Feest, T. G.; Zalin, A. M.; Hsuan, J. J.: Human lysozyme gene mutations cause hereditary systemic amyloidosis. *Nature* 362: 553–557, 1993.
- [27471] 1023. Peters, C. W. B.; Kruse, U.; Pollwein, R.; Grzeschik, K.-H.; Sippel, A. E.: The human lysozyme gene: sequence organization and chromosomal localization. (Abstract) *Cytogenet. Cell Genet.* 51: 1059 only, 1989.

- [27472] 1024.Prieur, D. J.: Personal Communication. Pullman, Washington 5/13/1975.
- [27473] 1025.Prieur, D. J.; Olson, H. M.; Young, D. M.: Lysozyme deficiency--an inherited disorder of rabbits. *Am. J. Path.* 77: 283-296, 1974.
- [27474] 1026.Spitznagel, J. K.; Cooper, M. R.; McCall, A. E.; DeChatelet, L.R.; Welsh, I. R.: Selective deficiency of granules associated with lysozyme and lactoferrin in human polymorphs (PMN) with reduced microbicidal capacity. (Abstract) *J. Clin. Invest.* 51: 93A only, 1972.
- [27475] 1027.Ku, C. C.; Murakami, M.; Sakamoto, A.; Kappler, J.; Marrack, P.: Control of homeostasis of CD8+ memory T cells by opposing cytokines. *Science* 288:675-678, 2000.
- [27476] 1028.Leonard, W. J.; Donlon, T. A.; Lebo, R. V.; Greene, W. C.: Localization of the gene encoding the human interleukin-2 receptor on chromosome 10. *Science* 228: 1547-1549, 1985.
- [27477] 1029.Webb, G. C.; Campbell, H. D.; Lee, J. S.; Young, I. G.: Mapping the gene for murine T-cell growth factor, IL-2, to bands B-C on chromosome 3 and for the alpha chain of the IL2-receptor, IL-2ra, to bands A2-A3 on chromosome 2. *Cytogenet. Cell Genet.* 54: 164-168, 1990.
- [27478] 1030.Du, X.; Williams, D. A.: Interleukin-11: review of

molecular, cell biology, and clinical use. *Blood* 89: 3897–3908, 1997.

- [27479] 1031. Du, X. X.; Williams, D. A.: Interleukin–11: a multi-functional growth factor derived from the hematopoietic microenvironment. *Blood* 83:2023–2030, 1994.
- [27480] 1032. McKinley, D.; Wu, Q.; Yang–Feng, T.; Yang, Y.–C.: Genomic sequence and chromosomal location of human interleukin–11 gene (IL11). *Genomics* 13:814–819, 1992.
- [27481] 1033. Paul, S. R.; Bennett, F.; Calvetti, J. A.; Kelleher, K.; Wood, C. R.; O'Hara, R. M., Jr.; Leary, A. C.; Sibley, B.; Clark, S. C.; Williams, D. A.; Yang, Y.–C.: Molecular cloning of a cDNA encoding interleukin 11, a stromal cell–derived lymphopoietic and hematopoietic cytokine. *Proc. Nat. Acad. Sci.* 87: 7512–7516, 1990.
- [27482] 1034. Yang–Feng, T. L.; Gibson, L.; Yang, Y. C.: Assignment of the gene encoding human interleukin–11 to chromosome 19q13.3–q13.4. (Abstract) *Cytogenet. Cell Genet.* 58: 2027 only, 1991.
- [27483] 1035. Grunig, G.; Warnock, M.; Wakil, A. E.; Venkayya, R.; Brombacher, F.; Rennick, D. M.; Sheppard, D.; Mohrs, M.; Donaldson, D. D.; Locksley, R. M.; Corry, D. B. Requirement for IL–13 independently of IL–4 in experimental asthma. *Science* 282: 2261–2263, 1998.

- [27484] 1036.Heinzmann, H.; Mao, X.-Q.; Akaiwa, M.; Kreomer, R. T.; Gao, P.-S.;Ohshima, K.; Umeshita, R.; Abe, Y.; Braun, S.; Yamashita, T.; Roberts,M. H.; Sugimoto, R.; and 20 others: Genetic variants of IL-13 signallingand human asthma and atopy. *Hum. Molec. Genet.* 9: 549-559, 2000.
- [27485] 1037.Howard, T. D.; Koppelman, G. H.; Xu, J.; Zheng, S. L.; Postma,D. S.; Meyers, D. A.; Bleecker, E. R.: Gene-gene interaction in asthma:IL4RA and IL13 in a Dutch population with asthma. *Am. J. Hum. Genet.* 70:230-236, 2002.
- [27486] 1038.Howard, T. D.; Whittaker, P. A.; Zaiman, A. L.; Koppelman, G. H.;Xu, J.; Hanley, M. T.; Meyers, D. A.; Postma, D. S.; Bleecker, E.R.: Identification and association of polymorphisms in the interleukin-13gene with asthma and atopy in a Dutch population. *Am. J. Resp. CellMolec. Biol.* 25: 377-384, 2001.
- [27487] 1039.Kuperman, D. A.; Huang, X.; Koth, L. L.; Chang, G. H.; Dolganov,G. M.; Zhu, Z.; Elias, J. A.; Sheppard, D.; Erle, D. J.: Direct effectsof interleukin-13 on epithelial cells cause airway hyperreactivityand mucus overproduction in asthma. *Nature Med.* 8: 885-889, 2002.
- [27488] 1040.Burshtyn, D. N.; Scharenberg, A. M.; Wagtmann, N.; Rajagopalan,S.; Berrada, K.; Yi, T.; Kinet, J.-P.; Long, E. O.: Recruitment oftyrosine phosphatase HCP by the killer cell

inhibitor receptor. *Immunity* 4:77–85, 1996.

- [27489] 1041. Dean, F. B.; Lian, L.; O'Donnell, M.: cDNA cloning and gene mapping of human homologs for *Schizosaccharomyces pombe* rad17, rad1, and hus1 and cloning of homologs from mouse, *Caenorhabditis elegans*, and *Drosophila melanogaster*. *Genomics* 54: 424–436, 1998.
- [27490] 1042. Mills, G. B.; Schmandt, R.; McGill, M.; Amendola, A.; Hill, M.; Jacobs, K.; May, C.; Rodricks, A.–M.; Campbell, S.; Hogg, D.: Expression of TTK, a novel human protein kinase, is associated with cell proliferation. *J. Biol. Chem.* 267: 16000–16006, 1992.
- [27491] 1043. Macdonald, D. H. C.; Lahiri, D.; Sampath, A.; Chase, A.; Sohal, J.; Cross, N. C. P.: Cloning and characterization of RNF6, a novel RING finger gene mapping to 13q12. *Genomics* 58: 94–97, 1999.
- [27492] 1044. Moreira, E. F.; Jaworski, C. J.; Rodriguez, I. R.: Cloning of a novel member of the reticulon gene family (RTN3): gene structure and chromosomal localization to 11q13. *Genomics* 58: 73–81, 1999.
- [27493] 1045. Strobel, M. C.; Seperack, P. K.; Copeland, N. G.; Jenkins, N. A.: Molecular analysis of two mouse dilute locus deletion mutations: spontaneous dilute lethal–20J and radiation–induced dilute prenatal lethal Aa2 alleles. *Molec.*

Cell. Biol. 10: 501–509, 1990.

- [27494] 1046. Tanaka, H.; Homma, K.; Iwane, A. H.; Katayama, E.; Ikebe, R.; Saito, J.; Yanagida, T.; Ikebe, M.: The motor domain determines the large step of myosin-V. *Nature* 415: 192–195, 2002.
- [27495] 1047. Walker, M. L.; Burgess, S. A.; Sellers, J. R.; Wang, F.; Hammer, J. A., III; Trinick, J.; Knight, P. J.: Two-headed binding of a processive myosin to F-actin. *Nature* 405: 804–807, 2000.
- [27496] 1048. Yanagida, T.; Iwane, A. H.: A large step for myosin. *Proc. Nat. Acad. Sci.* 97: 9357–9359, 2000.
- [27497] 1049. Barton, P. J. R.; Buckingham, M. E.: The myosin alkali light chain proteins and their genes. *Biochem. J.* 231: 249–261, 1985.
- [27498] 1050. Cohen-Haguénauer, O.; Barton, P. J. R.; Van Cong, N.; Serero, S.; Gross, M.-S.; Jegou-Foubert, C.; de Tand, M.-F.; Robert, B.; Buckingham, M.; Frezal, J.: Assignment of the human fast skeletal muscle myosin alkali light chains gene (MLC1F/MLC3F) to 2q32.1–2qter. *Hum. Genet.* 78:65–70, 1988.
- [27499] 1051. Seidel, U.; Bober, E.; Winter, B.; Lenz, S.; Lohse, P.; Arnold, H. H.: The complete nucleotide sequences of cDNA clones coding for human myosin light chains 1 and 3. *Nu-*

cleic Acids Res. 15: 4989 only, 1987.

- [27500] 1052. Seidel, U.; Bober, E.; Winter, B.; Lenz, S.; Lohse, P.; Goedde, H. W.; Grzeschik, K. H.; Arnold, H. H.: Alkali myosin light chains in man are encoded by a multigene family that includes the adult skeletal muscle, the embryonic or atrial, and nonsarcomeric isoforms. *Gene* 66:135–146, 1988.
- [27501] 1053. Serero, S.; Barton, P.; Van Cong, N.; Cohen-Haguenauer, O.; Robert, B.; Buckingham, M.; Frezal, J.: Assignment of the human fast skeletal muscle myosin alkali light chains gene (MLC1F/MLC3F) to 2q32.1–2qter. (Abstract) *Cytogenet. Cell Genet.* 46: 690 only, 1987.
- [27502] 1054. Macera, M. J.; Szabo, P.; Wadgaonkar, R.; Siddiqui, M. A. Q.; Verma, R. S.: Localization of the gene coding for ventricular myosin regulatory light chain (MYL2) to human chromosome 12q23–q24.3. *Genomics* 13:829–831, 1992.
- [27503] 1055. Poetter, K.; Jiang, H.; Hassanzadeh, S.; Master, S. R.; Chang, A.; Dalakas, M. C.; Rayment, I.; Sellers, J. R.; Fananapazir, L.; Epstein, N. D.: Mutations in either the essential or regulatory light chains of myosin are associated with a rare myopathy in human heart and skeletal muscle. *Nature Genet.* 13: 63–69, 1996.

- [27504] 1056.Kissel, K.; Santoso, S.; Hofmann, C.; Stroncek, D.; Bux, J.: Molecularbasis of the neutrophil glycoprotein NB1 (CD177) involved in the pathogenesisof immune neutropenias and transfusion reactions. *Europ. J. Immun.* 31:1301–1309, 2001.
- [27505] 1057.Kissel, K.; Scheffler, S.; Kerowgan, M.; Bux, J.: Molecular basisof NB1 (HNA–2a, CD177) deficiency. *Blood* 99: 4231–4233, 2002.
- [27506] 1058.Lalezari, P.; Murphy, G. B.; Allan, F. H.: NB1, a new neutrophilspecific antigen involved in the pathogenesis of neonatal neutropenia. *J.Clin. Invest.* 50: 1108–1115, 1971.
- [27507] 1059.Temerinac, S.; Klippel, S.; Strunck, E.; Roder, S.; Lubbert, M.;Lange, W.; Azemar, M.; Meinhardt, G.; Schaefer, H.–E.; Pahl, H. L.: Cloning of PRV–1, a novel member of the uPAR receptor superfamily,which is overexpressed in polycythemia rubra vera. *Blood* 95: 2569–2576,2000.
- [27508] 1060.Van Cong, N.; Ray, D.; Gross, M. S.; de Tand, M. F.; Frezal, J.;Moreau–Gachelin, F.: Localization of the human oncogene SPI1 on chromosome11, region p11.22. *Hum. Genet.* 84: 542–546, 1990.
- [27509] 1061.Dionne, C. A.; Kaplan, R.; Seuanez, H.; O'Brien, S. J.; Jaye, M.: Chromosome assignment by polymerase chain

reaction techniques: assignment of the oncogene FGF-5 to human chromosome 4. *Biotechniques* 8: 190-194, 1990.

[27510] 1062. Hebert, J. M.; Rosenquist, T.; Gotz, J.; Martin, G. R.: FGF5 as a regulator of the hair growth cycle: evidence from targeted and spontaneous mutations. *Cell* 78: 1017-1025, 1994.

[27511] 1063. Nguyen, C.; Roux, D.; Mattei, M.-G.; de Lapeyriere, O.; Goldfarb, M.; Birnbaum, D.; Jordan, B. R.: The FGF-related oncogenes *hst* and *int.2*, and the *bcl.1* locus are contained within one megabase in band q13 of chromosome 11, while the *fgf.5* oncogene maps to 4q21. *Oncogene* 3:703-708, 1988.

[27512] 1064. Zhan, X.; Bates, B.; Hu, X.; Goldfarb, M.: The human FGF-5 oncogene encodes a novel protein related to fibroblast growth factors. *Molec. Cell. Biol.* 8: 3487-3495, 1988.

[27513] 1065. Befort, K.; Mattei, M.-G.; Roeckel, N.; Kieffer, B.: Chromosomal localization of the delta opioid receptor gene to human 1p34.3-p36.1 and mouse 4D bands by in situ hybridization. *Genomics* 20: 143-145, 1994.

[27514] 1066. Bzdega, T.; Chin, H.; Kim, H.; Jung, H. H.; Kozak, C. A.; Klee, W. A.: Regional expression and chromosomal localization of the delta opiate receptor gene. *Proc. Nat. Acad. Sci.* 90: 9305-9309, 1993.

- [27515] 1067. Evans, C. J.; Keith, D. E.; Morrison, H.; Magendzo, K.; Edwards, R. H.: Cloning of a delta opioid receptor by functional expression. *Science* 258:1952–1955, 1992.
- [27516] 1068. Filliol, D.; Ghazizadeh, S.; Chluba, J.; Martin, M.; Matthes, H.W. D.; Simonin, F.; Befort, K.; Gaveriaux-Ruff, C.; Dierich, A.; LeMeur, M.; Valverde, O.; Maldonado, R.; Kieffer, B. L.: Mice deficient for delta- and mu-opioid receptors exhibit opposing alterations of emotional responses. *Nature Genet.* 25: 195–200, 2000.
- [27517] 1069. Jordan, B. A.; Devi, L. A.: G-protein-coupled receptor heterodimerization modulates receptor function. *Nature* 399: 697–700, 1999.
- [27518] 1070. Kaufman, D. L.; Xia, Y.-R.; Keith, D. E., Jr.; Newman, D.; Evans, C. J.; Lusis, A. J.: Localization of the delta-opioid receptor gene to mouse chromosome 4 by linkage analysis. *Genomics* 19: 405–406, 1994.
- [27519] 1071. Mayer, P.; Tischmeyer, H.; Jayasinghe, M.; Bonnekoh, B.; Gollnick, H.; Teschemacher, H.; Holtt, V.: A delta-opioid receptor lacking the third cytoplasmic loop is generated by atypical mRNA processing in human malignomas. *FEBS Lett.* 480: 156–160, 2000.
- [27520] 1072. Whistler, J. L.; Enquist, J.; Marley, A.; Fong, J.; Glader, F.; Tsuruda, P.; Murray, S. R.; von Zastrow, M.: Modu-

lation of postendocytosorting of G protein-coupled receptors. *Science* 297: 615–620, 2002.

[27521] 1073. Xu, K.; Liu, X.; Nagarajan, S.; Gu, X.-Y.; Goldman, D.: Relationship of the delta-opioid receptor gene to heroin abuse in a large Chinese case/control sample. *Am. J. Med. Genet.* 11: 45–50, 2002.

[27522] 1074. Bartoshuk, L. M.; Duffy, V. B.; Miller, I. J.: PTC/PROP tasting: anatomy, psychophysics, and sex effects. *Physiol. Behav.* 56: 1165–1171, 1994.

[27523] 1075. Chautard-Freire-Maia, E. A.: Linkage relationships between 22 autosomal markers. *Ann. Hum. Genet.* 38: 191–198, 1974.

[27524] 1076. Conneally, P. M.; Dumont-Driscoll, M.; Huntzinger, R. S.; Nance, W. E.; Jackson, C. E.: Linkage relations of the loci for Kell and phenylthiocarbamide (PTC) taste sensitivity. *Hum. Hered.* 26: 267–271, 1976.

[27525] 1077. Spence, M. A.; Falk, C. T.; Neiswanger, K.; Field, L. L.; Marazita, M. L.; Allen, F. H., Jr.; Siervogel, R. M.; Roche, A. F.; Crandall, B. F.; Sparkes, R. S.: Estimating the recombination frequency for the PTC-Kell linkage. *Hum. Genet.* 67: 183–186, 1984.

[27526] 1078. Bhattacharyya, N.; Banerjee, S.: A novel role of XRCC1 in the functions of a DNA polymerase beta variant.

Biochemistry 40: 9005–9013,2001.

- [27527] 1079.Laborda, J.; Sausville, E. A.; Hoffman, T.; Notario, V.: dlk,a putative mammalian homeotic gene differentially expressed in smallcell lung carcinoma and neuroendocrine tumor cell line. J. Biol.Chem. 268: 3817–3820, 1993.
- [27528] 1080.Dryja, T. P.; Rapaport, J.; McGee, T. L.; Nork, T. M.; Schwartz,T. L.: Molecular etiology of low-penetrance retinoblastoma in twopedigrees. Am. J. Hum. Genet. 52: 1122–1128, 1993.
- [27529] 1081.Dryja, T. P.; Rapaport, J. M.; Joyce, J. M.; Petersen, R. A.:Molecular detection of deletions involving band q14 of chromosome13 in retinoblastomas. Proc. Nat. Acad. Sci. 83: 7391–7394, 1986.
- [27530] 1082.Dryja, T. P.; Rapaport, J. M.; Weichselbaum, R.; Bruns, G. A.P.: Chromosome 13 restriction fragment length polymorphisms. Hum.Genet. 65: 320–324, 1984.
- [27531] 1083.Duane, T. B.: Clinical Ophthalmology. Hagerstown: Harper andRow (pub.) 3: 1980. Pp. 13 only.
- [27532] 1084.Duncan, A. M. V.; Morgan, C.; Gallie, B. L.; Phillips, R. A.;Squire, J.: Re-evaluation of the sublocalization of esterase D andits relation to the retinoblastoma locus by in situ hybridization. Cytogenet.Cell Genet. 44: 153–157,

1987.

- [27533] 1085.Dunn, J. M.; Phillips, R. A.; Becker, A. J.; Gallie, B. L.: Identification of germline and somatic mutations affecting the retinoblastoma gene. *Science* 241:1797–1800, 1988.
- [27534] 1086.Dunn, J. M.; Phillips, R. A.; Zhu, X.; Becker, A.; Gallie, B.L.: Mutations in the RB1 gene and their effects on transcription. *Molec.Cell. Biol.* 9: 4596–4604, 1989.
- [27535] 1087.Ejima, Y.; Sasaki, M. S.; Kaneko, A.; Tanooka, H.: Types, rates, origin and expressivity of chromosome mutations involving 13q14 in retinoblastoma patients. *Hum. Genet.* 79: 118–123, 1988.
- [27536] 1088.Ejima, Y.; Sasaki, M. S.; Kaneko, A.; Tanooka, H.; Hara, Y.; Hida, T.; Kinoshita, Y.: Possible inactivation of part of chromosome 13 due to 13qXp translocation associated with retinoblastoma. *Clin.Genet.* 21: 357–361, 1982.
- [27537] 1089.Eldridge, R.; O'Meara, K.; Kitchin, D.: Superior intelligence in sighted retinoblastoma patients and their families. *J. Med. Genet.* 9:331–335, 1972.
- [27538] 1090.Falls, H. F.; Neel, J. V.: Genetics of retinoblastoma. *Arch.Ophthal.* 46: 367–389, 1951.
- [27539] 1091.Fitzgerald, P. H.; Stewart, J.; Suckling, R. D.: Retinoblastoma mutation rate in New Zealand and support

for the two-hit model. Hum.Genet. 64: 128–130, 1983.

- [27540] 1092.Francke, U.: Retinoblastoma and chromosome 13. Cytogenet. CellGenet. 14: 131–134, 1976.
- [27541] 1093.Francois, J.: Retinoblastoma and osteogenic sarcoma. Ophthalmologica 175:185–191, 1977.
- [27542] 1094.Francois, J.: Hereditary malignant tumor of the eye. CongenitalAnomalies of The Eye. St. Louis: C. V. Mosby Co. (pub.) 1968.Pp. 205–246.
- [27543] 1095.Francois, J.; Matton, M. T.; De Bie, S.; Tanaka, Y.; Vandenbulcke,D.: Genesis and genetics of retinoblastoma. Ophthalmologica 170:405–425, 1975.
- [27544] 1096.Friend, S. H.; Bernards, R.; Rogelj, S.; Weinberg, R. A.; Rapaport,J. M.; Albert, D. M.; Dryja, T. P.: A human DNA segment with propertiesof the gene that predisposes to retinoblastoma and osteosarcoma. Nature 323:643–646, 1986.
- [27545] 1097.Friend, S. H.; Dryja, T. P.; Weinberg, R. A.: Oncogenes and tumor-suppressinggenes. New Eng. J. Med. 318: 618–622, 1988.
- [27546] 1098.Friend, S. H.; Horowitz, J. M.; Gerber, M. R.; Wang, X.–F.; Bogenmann,E.; Li, F. P.; Weinberg, R. A.: Deletions of a DNA sequence in retinoblastomasand mesenchymal tumors: organization of the sequence and its encoded–protein. Proc. Nat. Acad. Sci. 84: 9059–9063, 1987. Note:

Correction:Proc. Nat. Acad. Sci. 85: 2234 only, 1988..

- [27547] 1099.Fukushima, Y.; Kuroki, Y.; Ito, T.; Kondo, I.; Nishigaki, I.:Familial retinoblastoma (mother and son) with 13q14 deletion. Hum.Genet. 77: 104–107, 1987.
- [27548] 1100.Fung, Y.–K. T.; Murphree, A. L.; T'Ang, A.; Qian, J.; Hinrichs,S. H.; Benedict, W. F.: Structural evidence for the authenticityof the human retinoblastoma gene. Science 236: 1657–1661, 1987.
- [27549] 1101.Gallie, B. L.: Predictive testing for retinoblastoma comes ofage. (Editorial) Am. J. Hum. Genet. 61: 279–281, 1997.
- [27550] 1102.Gallie, B. L.; Ellsworth, R. M.; Abramson, D. M.; Phillips, R.A.: Retinoma: spontaneous regression of retinoblastoma or benignmanifestation of the mutation? Brit. J. Cancer 45: 513–521, 1982.
- [27551] 1103.Gallie, B. L.; Phillips, R. A.: Multiple manifestations of theretinoblastoma gene. Birth Defects Orig. Art. Ser. 18(6): 689–701,1982.
- [27552] 1104.Garcia–Cao, M.; Gonzalo, S.; Dean, D.; Blasco, M. A.: A rolefor the Rb family of proteins in controlling telomere length. NatureGenet. 15Oct.: , 2002. Note: Advance Electronic Publication.
- [27553] 1105.Gey, W.: Dq–, multiple Missbildungen und

Retinoblastom. Humangenetik 10:362–365, 1970.

- [27554] 1106.Huang, Y. Z.; Wang, Q.; Xiong, W. C.; Mei, L.: Erbin is a protein concentrated at postsynaptic membranes that interacts with PSD-95. J.Biol. Chem. 276: 19318–19326, 2001.
- [27555] 1107.Aalto-Setälä, K.; Helve, E.; Kovanen, P. T.; Kontula, K.: Finnish type of low density lipoprotein receptor gene mutation (FH-Helsinki) deletes exons encoding the carboxy-terminal part of the receptor and creates an internalization-defective phenotype. J. Clin. Invest. 84:499–505, 1989.
- [27556] 1108.Aalto-Setälä, K.; Koivisto, U.-M.; Miettinen, T. A.; Gylling, H.; Kesäniemi, Y. A.; Savolainen, M.; Pyörälä, K.; Ebeling, T.; Mononen, I.; Turtola, H.; Viikari, J.; Kontula, K.: Prevalence and geographical distribution of major LDL receptor gene rearrangements in Finland. J.Intern. Med. 231: 227–234, 1992.
- [27557] 1109.Clark, G. J.; Cooper, B.; Fitzpatrick, S.; Green, B. J.; Hart, D. N. J.: The gene encoding the immunoregulatory signaling molecule CMRF-35A localized to human chromosome 17 in close proximity to other members of the CMRF-35 family. Tissue Antigens 57: 415–423, 2001.
- [27558] 1110.Daish, A.; Starling, G. C.; McKenzie, J. L.; Nimmo, J.

C.; Jackson, D. G.; Hart, D. N. J.: Expression of the CMRF-35 antigen, a new member of the immunoglobulin gene superfamily, is differentially regulated on leucocytes. *Immunology* 79: 55–63, 1993.

[27559] 1111. Jackson, D. G.; Hart, D. N. J.; Starling, G.; Bell, J. I.: Molecular cloning of a novel member of the immunoglobulin gene superfamily homologous to the polymeric immunoglobulin receptor. *Europ. J. Immun.* 22: 1157–1163, 1992.

[27560] 1112. Matesic, L. E.; Yip, R.; Reuss, A. E.; Swing, D. A.; O'Sullivan, T. N.; Fletcher, C. F.; Copeland, N. G.; Jenkins, N. A.: Mutations in *Mlph*, encoding a member of the Rab effector family, cause the melanosomal transport defects observed in leaden mice. *Proc. Nat. Acad. Sci.* 98: 10238–10243, 2001.

[27561] 1113. Silvers, W. K.: *The Coat Colors of Mice*. New York: Springer, 1979.

[27562] 1114. Chen, W.; Kubota, S.; Kim, K.-S.; Cheng, J.; Kuriyama, M.; Eggertsen, G.; Bjorkhem, I.; Seyama, Y.: Novel homozygous and compound heterozygous mutations of sterol 27-hydroxylase gene (*CYP27*) cause cerebrotendinous xanthomatosis in three Japanese patients from two unrelated families. *J. Lipid Res.* 38: 870–879,

1997.

- [27563] 1115.Chen, W.; Kubota, S.; Ujike, H.; Ishihara, T.; Seyama, Y.: A novelarg362ser mutation in the sterol 27-hydroxylase gene (CYP27): itseffects on pre-mRNA splicing and enzyme activity. *Biochemistry* 37:15050–15056, 1998.
- [27564] 1116.Kim, K.–K.; Kubota, S.; Kuriyama, M.; Fujiyama, J.; Bjorkhem, I.;Eggertsen, G.; Seyama, Y.: Identification of new mutations in sterol27-hydroxylase gene in Japanese patients with cerebrotendinous xanthomatosis(CTX). *J. Lipid Res.* 35: 1031–1039, 1994.
- [27565] 1117.Lamon–Fava, S.; Schaefer, E. J.; Garuti, R.; Salen, G.; Calandra,S.: Two novel mutations in the sterol 27-hydroxylase gene causingcerebrotendinous xanthomatosis. *Clin. Genet.* 61: 185–191, 2002.
- [27566] 1118.Leitersdorf, E.; Reshef, A.; Meiner, V.; Levitzki, R.; Schwartz,S. P.; Dann, E. J.; Berkman, N.; Cali, J. J.; Klapholz, L.; Berginer,V. M.: Frameshift and splice-junction mutations in the sterol 27-hydroxylasegene cause cerebrotendinous xanthomatosis in Jews of Moroccan origin. *J.Clin. Invest.* 91: 2488–2496, 1993.
- [27567] 1119.Rosen, H.; Reshef, A.; Maeda, N.; Lippoldt, A.; Shpizen, S.; Triger,L.; Eggertsen, G.; Bjorkhem, I.; Leiters–

dorf, E.: Markedly reduced bile acid synthesis but maintained levels of cholesterol and vitamin D metabolites in mice with disrupted sterol 27-hydroxylase gene. *J. Biol. Chem.* 273: 14805–14812, 1998.

- [27568] 1120. Shiga, K.; Fukuyama, R.; Kimura, S.; Nakajima, K.; Fushiki, S.: Mutation of the sterol 27-hydroxylase gene (CYP27) results in truncation of mRNA expressed in leukocytes in a Japanese family with cerebrotendinous xanthomatosis. *J. Neurol. Neurosurg. Psychiatr.* 67: 675–677, 1999.
- [27569] 1121. Toba, H.; Fukuyama, R.; Sasaki, M.; Shiga, K.; Ishibashi, S.; Fushiki, S.: A Japanese patient with cerebrotendinous xanthomatosis has different mutations within two functional domains of CYP27. (Letter) *Clin. Genet.* 61: 77–78, 2002.
- [27570] 1122. Juwana, J.-P.; Henderikx, P.; Mischo, A.; Wadle, A.; Fadle, N.; Gerlach, K.; Arends, J. W.; Hoogenboom, H.; Pfreundschuh, M.; Renner, C.: EB/RP gene family encodes tubulin binding proteins. *Int. J. Cancer* 81: 275–284, 1999.
- [27571] 1123. Renner, C.; Pfitzenmeier, J.-P.; Gerlach, K.; Held, G.; Ohnesorge, S.; Sahin, U.; Bauer, S.; Pfreundschuh, M.: RP1, a new member of the adenomatous polyposis coli-binding EB1-like gene family, is differently expressed in activated

T cells. *J. Immun.* 159: 1276–1283, 1997.

[27572] 1124.Wadle, A.; Thiel, G.; Mischo, A.; Jung, V.; Pfreundschuh, M.; Renner,C.: Chromosomal localization and promoter analysis of the adenomatouspolyposis coli binding protein RP1. *Oncogene* 20: 5920–5929, 2001.

[27573] 1125.Dunn, N. R.; Hogan, B. L. M.: How does the mouse get its trunk? *NatureGenet.* 27: 351–352, 2001.

[27574] 1126.Kalantry, S.; Manning, S.; Haub, O.; Tomihara–Newberger, C.; Lee,H.–G.; Fangman, J.; Disteché, C. M.; Manova, K.; Lacy, E.: The amnionlessgene, essential for mouse gastrulation, encodes a visceral–endoderm–specificprotein with an extracellular cysteine–rich domain. *Nature Genet.* 27:412–416, 2001.

[27575] 1127.Tomihara–Newberger, C.; Haub, O.; Lee, H.–G.; Soares, V.; Manova,K.; Lacy, E.: The amn gene product is required in extraembryonictissues for the generation of middle primitive streak derivatives. *Dev.Biol.* 204: 34–54, 1998.

[27576] 1128.Wang, X.; Bornslaeger, E. A.; Haub, O.; Tomihara–Newberger, C.;Lonberg, N.; Dinulos, M. B.; Disteché, C. M.; Copeland, N.; Gilbert,D. J.; Jenkins, N. A.; Lacy, E.: A candidate gene for the amnionlessgastrulation stage mouse mutation encodes a TRAF–related protein. *Dev.Biol.* 177:

274–290, 1996.

- [27577] 1129. Awasthi, S.; Cheng, J.; Singhal, S. S.; Saini, M. K.; Pandya, U.; Pikula, S.; Bandorowicz-Pikula, J.; Singh, S. V.; Zimniak, P.; Awasthi, Y. C.: Novel function of human RLIP76: ATP-dependent transport of glutathione conjugates and doxorubicin. *Biochemistry* 39: 9327–9334, 2000.
- [27578] 1130. Jullien-Flores, V.; Dorseuil, O.; Romero, R.; Letourneur, F.; Saragosti, S.; Berger, R.; Tavitian, A.; Gacon, G.; Camonis, J. H.: Bridging Ral GTPase to Rho pathways: RLIP76, a Ral effector with CDC42/Rac GTPase-activating protein activity. *J. Biol. Chem.* 270: 22473–22477, 1995.
- [27579] 1131. Mas, C.; Bourgeois, F.; Bulfone, A.; Levacher, B.; Mugnier, C.; Simonneau, M.: Cloning and expression analysis of a novel gene, RP42, mapping to an autism susceptibility locus on 6q16. *Genomics* 65: 70–74, 2000.
- [27580] 1132. Faulkner, G.; Pallavicini, A.; Formentin, E.; Comelli, A.; Ievolella, C.; Trevisan, S.; Bortoletto, G.; Scannapieco, P.; Salamon, M.; Mouly, V.; Valle, G.; Lanfranchi, G.: ZASP: a new Z-band alternatively spliced PDZ-motif protein. *J. Cell Biol.* 146: 465–475, 1999.
- [27581] 1133. Zhou, Q.; Ruiz-Lozano, P.; Martone, M. E.; Chen, J.: Cypher, a striated muscle-restricted PDZ and LIM domain-

containing protein, binds to alpha-actinin-2 and protein kinase C. *J. Biol. Chem.* 274:19807–19813, 1999.

[27582] 1134. Feng, S.-L. Y.; Guo, Y.; Factor, V. M.; Thorgeirsson, S. S.; Bell, D. W.; Testa, J. R.; Peifley, K. A.; Winkles, J. A.: The Fn14 immediate-early response gene is induced during liver regeneration and highly expressed in both human and murine hepatocellular carcinomas. *Am. J. Path.* 156:1253–1261, 2000.

[27583] 1135. Meighan-Mantha, R. L.; Hsu, D. K. W.; Guo, Y.; Brown, S. A. N.; Feng, S.-L. Y.; Peifley, K. A.; Alberts, G. F.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Richards, C. M.; Winkles, J. A.: The mitogen-inducible Fn14 gene encodes a type I transmembrane protein that modulates fibroblast adhesion and migration. *J. Biol. Chem.* 274: 33166–33176, 1999.

[27584] 1136. Ikeda, H.; Lethe, B.; Lehmann, F.; van Baren, N.; Baurain, J.-F.; de Smet, C.; Chambost, H.; Vitale, M.; Moretta, A.; Boon, T.; Coulie, P. G.: Characterization of an antigen that is recognized on a melanoma showing partial HLA loss by CTL expressing an NK inhibitory receptor. *Immunity* 6:199–208, 1997.

[27585] 1137. van Baren, N.; Chambost, H.; Ferrant, A.; Michaux, L.; Ikeda, H.; Millard, I.; Olive, D.; Boon, T.; Coulie, P. G.:

PRAME, a gene encoding an antigen recognized on a human melanoma by cytolytic T cells, is expressed in acute leukaemia cells. *Brit. J. Haemat.* 102: 1376–1379, 1998.

[27586] 1138. Burgener, R.; Wolf, M.; Ganz, T.; Baggiolini, M.: Purification and characterization of a major phosphatidylserine-binding phosphoprotein from human platelets.

Biochem. J. 269: 729–734, 1990.

[27587] 1139. Gustincich, S.; Schneider, C.: Serum deprivation response genes induced by serum starvation but not by contact inhibition. *Cell Growth Diff.* 4: 753–760, 1993.

[27588] 1140. Gustincich, S.; Vatta, P.; Goruppi, S.; Wolf, M.; Saccone, S.; Della Valle, G.; Baggiolini, M.; Schneider, C.: The human serum deprivation response gene (SDPR) maps to 2q32–q33 and codes for a phosphatidylserine-binding protein. *Genomics* 57: 120–129, 1999.

[27589] 1141. Eder, P. S.; Kekuda, R.; Stolc, V.; Altman, S.: Characterization of two scleroderma autoimmune antigens that copurify with human ribonuclease P. *Proc. Nat. Acad. Sci.* 94: 1101–1106, 1997.

[27590] 1142. Barton, D. E.; Spritz, R. A.; Francke, U.: RPU1 encoding the 68kDa U1 snRNP-associated protein is located on chromosome 19. (Abstract) *Cytogenet. Cell Genet.* 46: 577 only, 1987.

- [27591] 1143. Du, H.; Rosbash, M.: The U1 snRNP protein U1C recognizes the 5-primesplice site in the absence of base pairing. *Nature* 419: 86–90, 2002.
- [27592] 1144. Montzka, K. A.; Steitz, J. A.: Additional low-abundance human small nuclear ribonucleoproteins: U11, U12, etc. *Proc. Nat. Acad. Sci.* 85: 8885–8889, 1988.
- [27593] 1145. Nelissen, R. L.; Sillekens, P. T.; Beijer, R. P.; Geurts van Kessel, A. H.; van Venrooij, W. J.: Structure, chromosomal localization and evolutionary conservation of the gene encoding human U1 snRNP-specific A protein. *Gene* 102: 189–196, 1991.
- [27594] 1146. Spritz, R. A.; Strunk, K.; Surowy, C. S.; Hoch, S. O.; Barton, D. E.; Francke, U.: The human 70-kD SnRNP protein: cDNA cloning, chromosomal localization, and expression. (Abstract) *Am. J. Hum. Genet.* 41: A239 only, 1987.
- [27595] 1147. Spritz, R. A.; Strunk, K.; Surowy, C. S.; Hoch, S. O.; Barton, D. E.; Francke, U.: Human U1–70K snRNP protein: cDNA cloning, chromosomal localization, expression, alternative splicing and RNA-binding. *Nucleic Acids Res.* 15: 10373–10391, 1987.
- [27596] 1148. John, M. E.; John, M. C.; Ashley, P.; MacDonald, R. J.; Simpson, E. R.; Waterman, M. R.: Identification and characterization of cDNA clones specific for cholesterol side–

chain cleavage cytochrome P-450. Proc.Nat. Acad. Sci. 81: 5628-5632, 1984.

[27597] 1149.Morohashi, K.; Fujii-Kuriyama, Y.; Okada, Y.; Sogawa, K.; Hirose,T.; Inayama, S.; Omura, T.: Molecular cloning and nucleotide sequence of cDNA for mRNA of mitochondrial cytochrome P-450(SCC) of bovine adrenal cortex. Proc. Nat. Acad. Sci. 81: 4647-4651, 1984.

[27598] 1150.Tajima, T.; Fujieda, K.; Kouda, N.; Nakae, J.; Miller, W. L.:Heterozygous mutation in the cholesterol side chain cleavage enzyme(P450scc) gene in a patient with 46,XY sex reversal and adrenal insufficiency. J.Clin. Endocr. Metab. 86: 3820-3825, 2001.

[27599] 1151.Bain, P. A.; Meisler, M. H.; Taylor, B. A.; Payne, A. H.: The genes encoding gonadal and nongonadal forms of 3-beta-hydroxysteroid dehydrogenase/delta-5-delta-4 isomerase are closely linked on mouse chromosome 3. Genomics 16: 219-223, 1993.

[27600] 1152.Ruiz-Perez, V. L.; Ide, S. E.; Strom, T. M.; Lorenz, B.; Wilson,D.; Woods, K.; King, L.; Francomano, C.; Freisinger, P.; Spranger,S.; Marino, B.; Dallapiccola, B.; Wright, M.; Meitinger, T.; Polymeropoulos,M. H.; Goodship, J.: Mutations in a new gene in Ellis-van Creveld syndrome and Weyers acrodistal dysostosis. Nature Genet. 24:

283–286,2000. Note: Erratum: Nature Genet. 25: 125 only, 2000.

- [27601] 1153.Douhan, J., III; Hauber, I.; Eibl, M. M.; Glimcher, L. H.: Genetic evidence for a new type of major histocompatibility complex class II combined immunodeficiency characterized by a dyscoordinate regulation of HLA-D alpha and beta chains. J. Exp. Med. 183: 1063–1069, 1996.
- [27602] 1154.Ahmed, C. M. I.; Ware, D. H.; Lee, S. C.; Patten, C. D.; Ferrer-Montiel, A. V.; Schinder, A. F.; McPherson, J. D.; Wagner-McPherson, C. B.; Wasmuth, J. J.; Evans, G. A.; Montal, M.: Primary structure, chromosomal localization, and functional expression of a voltage-gated sodium-channel from human brain. Proc. Nat. Acad. Sci. 89: 8220–8224, 1992.
- [27603] 1155.Han, J.; Lu, C.-M.; Brown, G. B.; Rado, T. A.: Direct amplification of a single dissected chromosomal segment by polymerase chain reaction: a human brain sodium channel gene is on chromosome 2q22–q23. Proc. Nat. Acad. Sci. 88: 335–339, 1991.
- [27604] 1156.George, A. L.; Gellens, M. E.; Kallen, R. G.; Barchi, R. L.: Molecular cloning and chromosomal location of two human muscle voltage-gated sodium channels. Soc. Neurosci. Abst. 16: 184 only, 1990.

- [27605] 1157. George, A. L., Jr.; Knops, J. F.; Han, J.; Finley, W. H.; Knittle, T. J.; Tamkun, M. M.; Brown, G. B.: Assignment of a human voltage-dependent sodium channel alpha-subunit gene (SCN6A) to 2q21-q23. *Genomics* 19:395-397, 1994.
- [27606] 1158. Han, J.; Lu, C.-M.; Brown, G. B.; Rado, T. A.: Direct amplification of a single dissected chromosome segment by polymerase chain reaction: a human brain sodium channel gene is on chromosome 2q22-q23. *Proc. Nat. Acad. Sci.* 88: 335-339, 1991.
- [27607] 1159. Calvo, R. M.; Asuncion, M.; Telleria, D.; Sancho, J.; San Millan, J. L.; Escobar-Morreale, H. F.: Screening for mutations in the steroidogenic acute regulatory protein and steroidogenic factor-1 genes, and in CYP11A and dosage-sensitive sex reversal-adrenal hypoplasia gene on the X chromosome, gene-1 (DAX-1), in hyperandrogenic hirsute women. *J. Clin. Endocr. Metab.* 86: 1746-1749, 2001.
- [27608] 1160. Bertrand, P.; Poirier, J.; Oda, T.; Finch, C. E.; Pasinetti, G. M.: Association of apolipoprotein E genotype with brain levels of apolipoprotein E and apolipoprotein J (clusterin) in Alzheimer disease. *Molec. Brain Res.* 33: 174-178, 1995.
- [27609] 1161. Birkenmeier, E. H.; Letts, V. A.; Frankel, W. N.; Ma-

genheimer, B. S.; Calvet, J. P.: Sulfated glycoprotein-2 (Sgp-2) maps to mouse chromosome 14. *Mammalian Genome* 4: 131-132, 1993.

[27610] 1162. Danik, M.; Chabot, J.-G.; Hassan-Gonzalez, D.; Suh, M.; Quirion, R.: Localization of sulfated glycoprotein-2/clusterin mRNA in the rat brain by in situ hybridization. *J. Comp. Neurol.* 334: 209-227, 1993.

[27611] 1163. Pan, H.; Yin, C.; Dyson, N. J.; Harlow, E.; Yamasaki, L.; VanDyke, T.: Key roles for E2F1 in signaling p53-dependent apoptosis and in cell division within developing tumors. *Molec. Cell* 2: 283-292, 1998.

[27612] 1164. Phillips, A. C.; Ernst, M. K.; Bates, S.; Rice, N. R.; Vousden, K. H.: E2F-1 potentiates cell death by blocking antiapoptotic signaling pathways. *Molec. Cell* 4: 771-781, 1999.

[27613] 1165. Saenz Robles, M. T.; Symonds, H.; Chen, J.; Van Dyke, T.: Induction versus progression of brain tumor development: differential functions for the pRB- and p53-targeting domains of simian virus 40 T antigen. *Molec. Cell. Biol.* 14: 2686-2698, 1994.

[27614] 1166. Sherr, C. J.: Tumor surveillance via the ARF-p53 pathway. *Genes Dev.* 12: 2984-2991, 1998.

[27615] 1167. Tsai, K. Y.; Hu, Y.; Macleod, K. F.; Crowley, D.; Ya-

masaki, L.;Jacks, T.: Mutation of E2f-1 suppresses apoptosis and inappropriateS phase entry and extends survival of Rb-deficient mouse embryos. Molec.Cell 2: 293-304, 1998.

[27616] 1168.Weinberg, R. A.: E2F and cell proliferation: a world turned upsidedown. Cell 85: 457-459, 1996.

[27617] 1169.Wu, L.; Timmers, C.; Maiti, B.; Saavedra, H. I.; Sang, L.; Chong,G. T.; Nuckolls, F.; Giangrande, P.; Wright, F. A.; Field, S. J.;Greenberg, M. E.; Orkin, S.; Nevins, J. R.; Robinson, M. L.; Leone,G.: The E2F1-3 transcription factors are essential for cellular proliferation. Nature 414:457-462, 2001.

[27618] 1170.Yamasaki, L.; Jacks, T.; Bronson, R.; Goillot, E.; Harlow, E.;Dyson, N. J.: Tumor induction and tissue atrophy in mice lackingE2F-1. Cell 85: 537-548, 1996.

[27619] 1171.Zhang, H. S.; Postigo, A. A.; Dean, D. C.: Active transcriptionalrepression by the Rb-E2F complex mediates G1 arrest triggered by p16(INK4a),TGF-beta, and contact inhibition. Cell 97: 53-61, 1999.

[27620] 1172.Zhang, Y.; Chellappan, S. P.: Cloning and characterization ofhuman DP2, a novel dimerization partner of E2F. Oncogene 10: 2085-2093,1995.

[27621] 1173.Fantes, J. A.; Oghene, K.; Boyle, S.; Danes, S.;

Fletcher, J. M.; Bruford, E. A.; Williamson, K.; Seawright, A.; Schedl, A.; Hanson, I.; Zehetner, G.; Bhogal, R.; Lehrach, H.; Gregory, S.; Williams, J.; Little, P. F. R.; Sellar, G. C.; Hoovers, J.; Mannens, M.; Weissenbach, J.; Junien, C.; van Heyningen, V.; Bickmore, W. A.: A high-resolution integrated physical, cytogenetic, and genetic map of human chromosome 11: distal p13 to proximal p15.1. *Genomics* 25: 447–461, 1995.

[27622] 1174. Flores, O.; Lu, H.; Reinberg, D.: Factors involved in specific transcription by mammalian RNA polymerase II: identification and characterization of factor IIH. *J. Biol. Chem.* 267: 2786–2793, 1992.

[27623] 1175. Heng, H. H. Q.; Xiao, H.; Shi, X.-M.; Greenblatt, J.; Tsui, L.-C.: Genes encoding general initiation factors for RNA polymerase II transcription are dispersed in the human genome. *Hum. Molec. Genet.* 3:61–64, 1994.

[27624] 1176. Lu, H.; Zawel, L.; Fisher, L.; Egly, J.-M.; Reinberg, D.: Human general transcription factor IIH phosphorylates the C-terminal domain of RNA polymerase II. *Nature* 358: 641–645, 1992.

[27625] 1177. Marinoni, J.-C.; Roy, R.; Vermeulen, W.; Miniou, P.; Lutz, Y.; Weeda, G.; Seroz, T.; Gomez, D. M.; Hoeijmakers, J. H. J.; Egly, J.-M.: Cloning and characterization of p52,

the fifth subunit of the core of the transcription/DNA repair factor TFIIH. EMBO J. 16: 1093–1102, 1997.

- [27626] 1178. Shiekhata, R.; Mermelstein, F.; Fisher, R. P.; Drapkin, R.; Dynlacht, B.; Wessling, H. C.; Morgan, D. O.; Reinberg, D.: Cdk-activating kinase complex is a component of human transcription factor TFIIH. Nature 374:283–287, 1995.
- [27627] 1179. Cheng, S.-D.; Peng, H.-L.; Chang, H.-Y.: Localization of the human UGP2 gene encoding the muscle isoform of UDPglucose pyrophosphorylase to 2p13–p14 by fluorescence in situ hybridization. Genomics 39:414–416, 1997.
- [27628] 1180. Peng, H.-L.; Chang, H.-Y.: Cloning of a human liver UDP-glucose pyrophosphorylase cDNA by complementation of the bacterial galU mutation. FEBS Lett. 329: 153–158, 1993.
- [27629] 1181. Shows, T. B.; Brown, J. A.; Goggin, A. P.; Haley, L. L.; Byers, M. G.; Eddy, R. L.: Assignment of a molecular form of UDP glucose pyrophosphorylase (UGPP-2) to chromosome 2 in man. Cytogenet. Cell Genet. 22: 215–218, 1978.
- [27630] 1182. Balow, R.-M.; Ragnarsson, U.; Zetterqvist, O.: Tripeptidyl aminopeptidase in the extralysosomal fraction of rat liver. J. Biol. Chem. 258:11622–11628, 1983.

- [27631] 1183. Bermingham, N. A.; McKay, T.; Hoyle, J.; Hernandez, D.; Martin, J. E.; Fisher, E. M. C.: The gene encoding tripeptidyl peptidase II maps to chromosome 1 in the mouse. *Mammalian Genome* 7: 390 only, 1996.
- [27632] 1184. Geier, E.; Pfeifer, G.; Wilm, M.; Lucchiari-Hartz, M.; Baumeister, W.; Eichmann, K.; Niedermann, G.: A giant protease with potential to substitute for some functions of the proteasome. *Science* 283: 978–981, 1999.
- [27633] 1185. Martinsson, T.; Vujic, M.; Tomkinson, B.: Localization of the human tripeptidyl peptidase II gene (TPP2) to 13q32–q33 by nonradioactive in situ hybridization and somatic cell hybrids. *Genomics* 17: 493–495, 1993.
- [27634] 1186. Tomkinson, B.: Nucleotide sequence of cDNA covering the N-terminus of human tripeptidyl peptidase II. *Biomed. Biochim. Acta* 50: 727–729, 1991.
- [27635] 1187. Tomkinson, B.; Jonsson, A.-K.: Characterization of cDNA for human tripeptidyl peptidase II: the N-terminal part of the enzyme is similar to subtilisin. *Biochemistry* 30: 168–174, 1991.
- [27636] 1188. Altafaj, X.; Dierssen, M.; Baamonde, C.; Marti, E.; Visa, J.; Guimera, J.; Oset, M.; Gonzalez, J. R.; Florez, J.; Filat, C.; Estivill, X.: Neurodevelopmental delay, motor abnormalities and cognitive deficits in transgenic mice over-

expressing Dyrk1A (minibrain), a murine model of Down's syndrome. Hum. Molec. Genet. 10: 1915–1923, 2001.

- [27637] 1189. Tanaka, T.; Nakahara, K.; Kato, N.; Imai, T.; Yamazaki, T.; Tomita, H.; Shimokawa, H.; Matsushashi, H.; Sato, N.; Matsui, M.; Kihira, S.; Shimizu, A.; Sano, T.; Haneda, N.; Kino, M.; Miyakita, Y.; Matsuoka, R.; Nagai, R.; Yazaki, Y.; Nakamura, Y.: Genetic linkage analyses of Romano–Ward syndrome (RWS) in 13 Japanese families. Hum. Genet. 94:380–384, 1994.
- [27638] 1190. Towbin, J. A.; Li, H.; Taggart, T. R.; Lehmann, M. H.; Schwartz, P. J.; Satler, C. A.; Ayyagari, R.; Robinson, J. L.; Moss, A.; Hejtmancik, J. F.: Evidence of genetic heterogeneity in Romano–Ward long QT syndrome: analysis of 23 families. Circulation 90: 2635–2644, 1994.
- [27639] 1191. Tye, K.–H.; Dessler, K. B.; Benchimol, A.: Survival following spontaneous ventricular flutter–fibrillation associated with QT syndrome: documentation during ambulatory monitoring. Arch. Intern. Med. 140:255–256, 1980.
- [27640] 1192. Tyson, J.; Tranebjaerg, L.; McEntagart, M.; Larsen, L. A.; Christiansen, M.; Whiteford, M. L.; Bathen, J.; Aslaksen, B.; Sorland, S. J.; Lund, O.; Pembrey, M. E.; Malcolm, S.; Bitner–Glindzicz, M.: Mutational spectrum in the cardioauditory syndrome of Jervell and Lange–Nielsen. Hum. Genet.

107: 499–503, 2000.

- [27641] 1193. Van der Straaten, P. J. C.; Bruins, C. L. D.: A family with heritable electrocardiographic Q–T prolongation. *J. Med. Genet.* 10: 158–160, 1973.
- [27642] 1194. Vincent, G. M.: The heart rate of Romano–Ward syndrome patients. *Am. Heart J.* 112: 61–64, 1986.
- [27643] 1195. Vincent, G. M.; Timothy, K. W.; Leppert, M.; Keating, M.: The spectrum of symptoms and QT intervals in carriers of the gene for the long–QT syndrome. *New Eng. J. Med.* 327: 846–852, 1992.
- [27644] 1196. Ward, O. C.: A new familial cardiac syndrome in children. *J. Irish Med. Assoc.* 54: 103–106, 1964.
- [27645] 1197. Weitkamp, L. R.; Moss, A. J.: The long QT (Romano–Ward) syndrome locus, LQT, is probably linked to the HLA loci. (Abstract) *Cytogenet. Cell Genet.* 40: 775 only, 1985.
- [27646] 1198. Weitkamp, L. R.; Moss, A. J.; Lewis, R. A.; Hall, W. J.; MacCluer, J. W.; Schwartz, P. J.; Locati, E. H.; Tzivoni, D.; Vincent, G. M.; Robinson, J. L.; Guttormsen, S. A.: Analysis of HLA and disease susceptibility: chromosome 6 genes and sex influence long–QT phenotype. *Am. J. Hum. Genet.* 55: 1230–1241, 1994.
- [27647] 1199. Weitkamp, L. R.; Moss, A. J.; Schwartz, P. J.; Locati,

E.; Tzivoni,D.; Vincent, G. M.; Robinson, J.; Guttormsen, S.: Analysis of HLAhaplotypes in long QT syndrome: withdrawal of the preliminary assignmentof LQT to the HLA linkage group. (Abstract) Cytogenet. Cell Genet. 51:1106 only, 1989.

- [27648] 1200.Yang, W.-P.; Levesque, P. C.; Little, W. A.; Conder, M. L.; Shalaby,F. Y.; Blonar, M. A.: KvLQT1, a voltage-gated potassium channel responsiblefor human cardiac arrhythmias. Proc. Nat. Acad. Sci. 94: 4017-4021,1997.
- [27649] 1201.Cavalli, A.; Lattion, A.-L.; Hummler, E.; Nenniger, M.; Pedrazzini,T.; Aubert, J.-F.; Michel, M. C.; Yang, M.; Lembo, G.; Vecchione,C.; Mostardini, M.; Schmidt, A.; Beermann, F.; Cotecchia, S.: Decreasedblood pressure response in mice deficient of the alpha(1b)-adrenergicreceptor. Proc. Nat. Acad. Sci. 94: 11589-11594, 1997.
- [27650] 1202.Samilchuk, E. I.; Chuchalin, A. G.: Mis-sense mutation of alpha-1-antichymotrypsingene and chronic lung disease. (Letter) Lancet 342: 624, 1993.
- [27651] 1203.Hildebrand, C. E.; Gonzalez, F. J.; McBride, O. W.; Nebert, D.W.: Assignment of the human 2,3,7,8-tetrachlorodibenzo-p-dioxin-induciblecytochrome P1-450 gene to chromosome 15. Nucleic Acids Res.

13:2009–2016, 1985.

- [27652] 1204.Olives, B.; Merriman, M.; Bailly, P.; Bain, S.; Barnett, A.; Todd,J.; Cartron, J.–P.; Merriman, T.: The molecular basis of the Kiddblood group polymorphism and its lack of association with type 1 diabetessusceptibility. *Hum. Molec. Genet.* 6: 1017–1020, 1997.
- [27653] 1205.Cohn, D. V.; Zangerle, R.; Fischer–Colbrie, R. R.; Chu, L. L. H.;Elting, J. J.; Hamilton, J. W.; Winkler, H.: Similarity of secretoryprotein I from parathyroid gland to chromogranin A from the adrenalmedulla. *Proc. Nat. Acad. Sci.* 79: 6056–6059, 1982.
- [27654] 1206.Adham, I. M.; Grzeschik, K.–H.; Geurts van Kessel, A. H. M.; Engel,W.: Localization of human preproacrosin to chromosome 22q13–qterby somatic cell hybrid analysis. (Abstract) *Cytogenet. Cell Genet.* 51:948 only, 1989.
- [27655] 1207.Adham, I. M.; Grzeschik, K.–H.; Geurts van Kessel, A. H. M.; Engel,W.: The gene encoding the human pre–proacrosin (ACR) maps to the q13–qterregion on chromosome 22. *Hum. Genet.* 84: 59–62, 1989.
- [27656] 1208.Adham, I. M.; Klemm, U.; Maier, W.–M.; Engel, W.: Molecular cloningof human preproacrosin cDNA. *Hum. Genet.* 84: 125–128, 1990.
- [27657] 1209.Adham, I. M.; Klemm, U.; Maier, W.–M.; Tsaousidou,

S.; Engel, W.: Molecular cloning and expression of boar and human proacrosin cDNA.(Abstract) Meeting of Gesellschaft fuer Humangenetik, Munich 149only, 4/4/1989.

- [27658] 1210.Adham, I. M.; Spitzer, U.; Schlosser, M.; Kremling, H.; Keime,S.; Engel, W.: A reply: the human proacrosin gene. Europ. J. Biochem. 207:27–28, 1992.
- [27659] 1211.Florke–Gerloff, S.; Topfer–Petersen, E.; Muller–Esterl, W.; Schill,W.–B.; Engel, W.: Acrosin and the acrosome in human spermatogenesis.Hum. Genet. 65: 61–67, 1983.
- [27660] 1212.Keime, S.; Adham, I. M.; Engel, W.: Nucleotide sequence and exon–intronorganization of the human proacrosin gene. Europ. J. Biochem. 190:195–200, 1990.
- [27661] 1213.Klemm, U.; Muller–Esterl, W.; Engel, W.: Acrosin, the peculiarsperm–specific serine protease. Hum. Genet. 87: 635–641, 1991.
- [27662] 1214.Kremling, H.; Keime, S.; Wilhelm, K.; Adham, I. M.; Hameister,H.; Engel, W.: Mouse proacrosin gene: nucleotide sequence, diploidexpression and chromosomal localization. Genomics 11: 828–834,1991.
- [27663] 1215.Vazquez–Levin, M. H.; Reventos, J.; Gordon, J. W.: Molecularcloning, sequencing and restriction mapping of the genomic sequenceencoding human proacrosin. Europ.

J. Biochem. 207: 23–26, 1992.

- [27664] 1216. Bianchi, G.; Tripodi, G.; Casari, G.; Salardi, S.; Barber, B. R.; Garcia, R.; Leoni, P.; Torielli, L.; Cusi, D.; Ferrandi, M.; Pinna, L. A.; Baralle, F. E.; Ferrari, P.: Two point mutations within the adducin genes are involved in blood pressure variation. *Proc. Nat. Acad. Sci.* 91: 3999–4003, 1994.
- [27665] 1217. Cusi, D.; Barlassina, C.; Azzani, T.; Casari, G.; Citterio, L.; Devoto, M.; Glorioso, N.; Lanzani, C.; Manunta, P.; Righetti, M.; Rivera, R.; Stella, P.; Troffa, C.; Zagato, L.; Bianchi, G.: Polymorphism of alpha-adducin and salt sensitivity in patients with essential hypertension. *Lancet* 349:1353–1357, 1997.
- [27666] 1218. Gardner, K.; Bennett, V.: A new erythrocyte membrane-associated protein with calmodulin binding activity: identification and purification. *J. Biol. Chem.* 261: 1339–1348, 1986.
- [27667] 1219. Goldberg, Y. P.; Lin, B.-Y.; Andrew, S. E.; Nasir, J.; Graham, R.; Graves, M. L.; Hutchinson, G.; Theilmann, J.; Ginzinger, D. G.; Schappert, K.; Clarke, L.; Rommens, J. M.; Hayden, M. R.: Cloning and mapping of the alpha-adducin gene close to D4S95 and assessment of its relationship to Huntington disease. *Hum. Molec. Genet.* 1:669–675, 1992.

- [27668] 1220.Hayden, M. R.: Huntington's Chorea. New York: Springer-Verlag(pub.) 1981.
- [27669] 1221.Joshi, R.; Bennett, V.: Mapping the domain structure of human erythrocyte adducin. J. Biol. Chem. 265: 13130-13136, 1990.
- [27670] 1222.Joshi, R.; Gilligan, D. M.; Otto, E.; McLaughlin, T.; Bennett,V.: Primary structure and domain organization of human alpha and beta adducin. J. Cell Biol. 115: 665-675, 1991.
- [27671] 1223.Zuffardi, O.; Caiulo, A.; Maraschio, P.; Tupler, R.; Bianchi,E.; Amisano, P.; Beluffi, G.; Moratti, R.; Liguri, G.: Regional assignment of the loci for adenylate kinase to 9q32 and for alpha(1)-acid glycoprotein to 9q31-q32: a locus for Goltz syndrome in region 9q32-qter? Hum.Genet. 82: 17-19, 1989.
- [27672] 1224.Bruns, G. A. P.; Regina, V. M.: Adenylate kinase-2, a mitochondrial enzyme. Biochem. Genet. 15: 477-486, 1977.
- [27673] 1225.Carritt, B.; King, J.; Welch, H. M.: Gene order and localization of enzyme loci on the short arm of chromosome 1. Ann. Hum. Genet. 46:329-335, 1982.
- [27674] 1226.Goss, S. J.; Harris, H.: Gene transfer by means of cell fusion.II. The mapping of 8 loci on human chromosome 1

by statistical analysis of gene assortment in somatic cell hybrids. *J. Cell Sci.* 25: 39–57, 1977.

[27675] 1227. Van Cong, N.; Billardon, C.; Rebourcet, R.; Kaouel, C. L.-B.; Picard, J. Y.; Weil, D.; Frezal, J.: The existence of a second adenylate kinase locus linked to PGM-1 and peptidase-C. *Ann. Genet.* 15: 213–218, 1972.

[27676] 1228. Cook, P. J. L.; Buckton, K. E.; Spowart, G.: Family studies on chromosome 9. *Cytogenet. Cell Genet.* 16: 284–288, 1976.

[27677] 1229. Mohandas, T.; Sparkes, R. S.; Sparkes, M. C.; Shulkin, J. D.; Toomey, K. E.; Funderburk, S. J.: Regional localization of human gene loci on chromosome 9: studies of somatic cell hybrids containing human translocation. *Am. J. Hum. Genet.* 31: 586–600, 1979.

[27678] 1230. Pilz, A.; Woodward, K.; Povey, S.; Abbott, C.: Comparative mapping of 50 human chromosome 9 loci in the laboratory mouse. *Genomics* 25: 139–149, 1995.

[27679] 1231. Povey, S.; Slaughter, C. A.; Wilson, D. E.; Gormley, I. P.; Buckton, K. E.; Perry, P.; Bobrow, M.: Evidence for the assignment of the loci AK 1, AK 3 and ACON to chromosome 9 in man. *Ann. Hum. Genet.* 39: 413–422, 1976.

[27680] 1232. Robson, E. B.; Meera Khan, P.: Report of the committee on the genetic constitution of chromosomes 7, 8,

and 9. Cytogenet. CellGenet. 32: 144–152, 1982.

- [27681] 1233.Steinbach, P.; Benz, R.: Demonstration of gene dosage effectsfor AK3 and GALT in fibroblasts from a fetus with 9p trisomy. Hum.Genet. 63: 290–291, 1983.
- [27682] 1234.Viskochil, D.; Buchberg, A. M.; Xu, G.; Cawthon, R. M.; Stevens,J.; Wolff, R. K.; Culver, M.; Carey, J. C.; Copeland, N. G.; Jenkins,N. A.; White, R.; O'Connell, P.: Deletions and a translocation interrupta cloned gene at the neurofibromatosis type 1 locus. Cell 62: 187–192,1990.
- [27683] 1235.Wilson, D. E., Jr.; Povey, S.; Harris, H.: Adenylate kinases inman: evidence for a third locus. Ann. Hum. Genet. 39: 305–313, 1976.
- [27684] 1236.Xu, G.; O'Connell, P.; Stevens, J.; White, R.: Characterizationof human adenylate kinase 3 (AK3) cDNA and mapping of the AK3 pseudogeneto an intron of the NF1 gene. Genomics 13: 537–542, 1992.
- [27685] 1237.Adinolfi, A.; Adinolfi, M.; Hopkinson, D. A.: Immunological andbiochemical characterization of the human alcohol dehydrogenase chi-ADHisozyme. Ann. Hum. Genet. 48: 1–10, 1984.
- [27686] 1238.Beisswenger, T. B.; Holmquist, B.; Vallee, B. L.: Chi-ADH is thesole alcohol dehydrogenase isozyme of mam-

malian brains: implications and inferences. Proc. Nat. Acad. Sci. 82: 8369–8373, 1985.

- [27687] 1239. Carlock, L.; Hiroshige, S.; Wasmuth, J.; Smith, M.: Assignment of the gene coding for class III ADH to human chromosome 4: 4q21–4q25. (Abstract) Cytogenet. Cell Genet. 40: 598 only, 1985.
- [27688] 1240. Engeland, K.; Hoog, J.-O.; Holmquist, B.; Estonius, M.; Jornvall, H.; Vallee, B. L.: Mutation of arg-115 of human class III alcohol dehydrogenase: a binding site required for formaldehyde dehydrogenase activity and fatty acid activation. Proc. Nat. Acad. Sci. 90: 2491–2494, 1993.
- [27689] 1241. Giri, P.; Krug, J. F.; Kozak, C.; Moretti, T.; O'Brien, S. J.; Seuanez, H. N.; Goldman, D.: Cloning and comparative mapping of a human class III (chi) alcohol dehydrogenase cDNA. Biochem. Biophys. Res. Commun. 164: 453–460, 1989.
- [27690] 1242. Goldman, D.; Rathna Giri, P.; Moretti, T. R.; Krug, J. F.; Kozak, C.; Dean, M.; Seuanez, H. N.; O'Brien, S. J.: Class III alcohol dehydrogenase (ADH5): widespread expression and synteny with other ADHs in both mouse and man. (Abstract) Am. J. Hum. Genet. 45 (suppl.): A141 only, 1989.
- [27691] 1243. Matsuo, Y.; Yokoyama, S.: Cloning and sequencing

of a processed pseudogene derived from a human class III alcohol dehydrogenase gene. *Am. J. Hum. Genet.* 46: 85–91, 1990.

[27692] 1244. Smith, M.: Genetics of human alcohol and aldehyde dehydrogenases. *Adv. Hum. Genet.* 15: 249–290, 1986.

[27693] 1245. Ramarao, C. S.; Kincade Denker, J. M.; Perez, D. M.; Gaivin, R. J.; Riek, R. P.; Graham, R. M.: Genomic organization and expression of the human alpha-1B-adrenergic receptor. *J. Biol. Chem.* 267: 21936–21945, 1992.

[27694] 1246. Yang–Feng, T. L.; Xue, F.; Zhong, W.; Cotecchia, S.; Frielle, T.; Caron, M. G.; Lefkowitz, R. J.; Francke, U.: Chromosomal organization of adrenergic receptor genes. *Proc. Nat. Acad. Sci.* 87: 1516–1520, 1990.

[27695] 1247. Herz, J.; Hamann, U.; Rogne, S.; Myklebost, O.; Gausepohl, H.; Stanley, K. K.: Surface location and high affinity for calcium of a 500 kd liver membrane protein closely related to the LDL-receptors suggest a physiological role as lipoprotein receptor. *EMBO J.* 7: 4119–4127, 1988.

[27696] 1248. Cailleau–Thomas, A.; Coullin, P.; Candelier, J.–J.; Balanzino, L.; Mennesson, B.; Oriol, R.; Mollicone, R.: FUT4 and FUT9 genes are expressed early in human embryogenesis. *Glycobiology* 10: 789–802, 2000.

[27697] 1249. Coullin, P.; Mollicone, R.; Grisard, M. C.; Gibaud, A.;

Ravise,N.; Feingold, J.; Oriol, R.: Chromosome 11q localization of one of the three expected genes for the human alpha-3-fucosyltransferases, by somatic hybridization. Cytogenet. Cell Genet. 56: 108–111, 1991.

[27698] 1250. Geurts van Kessel, A.; Tetteroo, P.; van Agthoven, T.; Paulussen, R.; van Dongen, J.; Hagemeijer, A.; von dem Borne, A.: Localization of human myeloid-associated surface antigen detected by a panel of 20 monoclonal antibodies to the q12-qter region of chromosome 11. J. Immun. 133: 1265–1269, 1984.

[27699] 1251. Sefton, L.; Kearney, P.; Kelsey, G.; Povey, S.; Wolfe, J.: Physical linkage of the genes PI and AACT. (Abstract) Cytogenet. Cell Genet. 51:1076, 1989.

[27700] 1252. Sefton, L.; Kelsey, G.; Kearney, P.; Povey, S.; Wolfe, J.: A physical map of the human PI and AACT genes. Genomics 7: 382–388, 1990.

[27701] 1253. Tachikawa, H.; Tsuda, M.; Onoe, K.; Ueno, M.; Takagi, S.; Shinohara, Y.: Alpha-1-antichymotrypsin gene A1252G variant (ACT Isehara-1) is associated with a lacunar type of ischemic cerebrovascular disease. J. Hum. Genet. 46: 45–47, 2001.

[27702] 1254. Tsuda, M.; Sei, Y.; Matsumoto, M.; Kamiguchi, H.; Yamamoto, M.; Shinohara, Y.; Igarashi, T.; Yamamura, M.:

Alpha-1-antichymotrypsinvariant detected by PCR-single strand conformation polymorphism (PCR-SSCP)and direct sequencing. Hum. Genet. 90: 467-468, 1992.

[27703] 1255.Tsuda, M.; Sei, Y.; Yamamura, M.; Yamamoto, M.; Shinohara, Y.: Detection of a new mutant alpha-1-antichymotrypsin in patients withocclusive-cerebrovascular disease. FEBS Lett. 304: 66-68, 1992.

[27704] 1256.Wang, X.; DeKosky, S. T.; Luedecking-Zimmer, E.; Ganguli, M.;Kamboh, M. I.: Genetic variation in alpha-1-antichymotrypsin andits association with Alzheimer's disease. Hum. Genet. 110: 356-365,2002.

[27705] 1257.Yamamoto, M.; Kondo, I.; Ogawa, N.; Asanuma, M.; Yamashita, Y.;Mizuno, Y.: Genetic association between susceptibility to Parkinson'sdisease and alpha-1-antichymotrypsin polymorphism. Brain Res. 759:153-155, 1997.

[27706] 1258.Azem, A.; Kessel, M.; Goloubinoff, P.: Characterization of a functionalGroEL-14(GroES-7)-2 chaperonin hetero-oligomer. Science 265: 653-656,1994.

[27707] 1259.Cheng, M. Y.; Hartl, F.-U.; Martin, J.; Pollock, R. A.; Kalousek,F.; Neupert, W.; Hallberg, E. M.; Hallberg, R. L.; Horwich, A. L.: Mitochondrial heat-shock protein hsp60 is essential for assemblyof proteins imported into yeast mi-

tochondria. *Nature* 337: 620–625,1989.

[27708] 1260.Ellis, R. J.: The molecular chaperone concept. *Semin. Cell Biol.* 1:1–9, 1990.

[27709] 1261.Fontaine, B.; Davoine, C.–S.; Durr, A.; Paternotte, C.; Feki, I.;Weissenbach, J.; Hazan, J.; Brice, A.: A new locus for autosomaldominant pure spastic paraplegia, on chromosome 2q24–q34. *Am. J.Hum. Genet.* 66: 702–707, 2000.

[27710] 1262.Rothman, J. E.: Polypeptide chain binding proteins: catalystsof protein folding and related processes in cells. *Cell* 59: 591–601,1989.

[27711] 1263.Saibil, H.; Dong, Z.; Wood, S.; auf der Mauer, A.: Binding ofchaperonins. *Nature* 353: 25–26, 1991.

[27712] 1264.Schmidt, M.; Rutkat, K.; Rachel, R.; Pfeifer, G.; Jaenicke, R.;Viitanen, P.; Lorimer, G.; Buchner, J.: Symmetric complexes of GroEchaperonins as part of the functional cycle. *Science* 265: 656–659,1994.

[27713] 1265.Venner, T. J.; Singh, B.; Gupta, R. S.: Nucleotide sequencesand novel structural features of human and Chinese hamster hsp60 (chaperonin)gene families. *DNA Cell Biol.* 9: 545–552, 1990.

[27714] 1266.Coste, H.; Rodriguez, J. C.: Orphan nuclear hormone receptor Rev–erb–alpharegulates the human apolipopro–

tein CIII promoter. J. Biol. Chem. 277:27120–27129, 2002.

- [27715] 1267.Jaiswal, A. K.; Gonzalez, F. J.; Nebert, D. W.: Comparison of human mouse P(1)450 upstream regulatory sequences in liver- and nonliver-derived cell lines. Molec. Endocr. 1: 312–320, 1987.
- [27716] 1268.Jaiswal, A. K.; Gonzalez, F. J.; Nebert, D. W.: Human P(1)–450 gene sequence and correlation of mRNA with genetic differences in benzo(a)pyrene metabolism. Nucleic Acids Res. 13: 4503–4520, 1985.
- [27717] 1269.Jaiswal, A. K.; Gonzalez, F. J.; Nebert, D. W.: Human dioxin-inducible cytochrome P1–450: complementary DNA and amino acid sequence. Science 228:80–83, 1985.
- [27718] 1270.Jaiswal, A. K.; Nebert, D. W.: Two RFLPs associated with the human P(1)450 gene linked to the MPI locus on chromosome 15 (HGM8D15S8). Nucleic Acids Res. 14: 4376, 1986.
- [27719] 1271.Jaiswal, A. K.; Nebert, D. W.; Gonzalez, F. J.: Human P(3)450:cDNA and complete amino acid sequence. Nucleic Acids Res. 14: 6773–6774, 1986.
- [27720] 1272.Jones, S. N.; Jones, P. G.; Ibarguen, H.; Caskey, C. T.; Craigen, W. J.: Induction of the Cyp1a–1 dioxin-responsive enhancer in transgenic mice. Nucleic Acids Res. 19:

6547–6551, 1991.

- [27721] 1273.Kawajiri, K.; Eguchi, H.; Nakachi, K.; Sekiya, T.; Yamamoto, M.: Association of CYP1A1 germ line polymorphisms with mutations of the p53 gene in lung cancer. *Cancer Res.* 56: 72–76, 1996.
- [27722] 1274.Kawajiri, K.; Nakachi, K.; Imai, K.; Yoshii, A.; Shinoda, N.; Watanabe, J.: Identification of genetically high risk individuals to lung cancer by DNA polymorphisms of the cytochrome P450IA1 gene. *FEBS Lett.* 263: 131–133, 1990.
- [27723] 1275.Kawajiri, K.; Watanabe, J.; Gotoh, O.; Tagashira, Y.; Sogawa, K.; Fujii-Kuriyama, Y.: Structure and drug inducibility of the human cytochrome P-450c gene. *Europ. J. Biochem.* 159: 219–225, 1986.
- [27724] 1276.Kouri, R. E.; McKinney, C. E.; Slomiany, D. J.; Snodgrass, D.R.; Wray, N. P.; McLemore, T. L.: Positive correlation between high aryl hydrocarbon hydroxylase activity and primary lung cancer as analyzed in cryopreserved lymphocytes. *Cancer Res.* 42: 5030–5037, 1982.
- [27725] 1277.McBride, O. W.: Personal Communication. Bethesda, Md. 9/16/1985.
- [27726] 1278.Mooney, L. A.; Bell, D. A.; Santella, R. M.; Van Bennekum, A.M.; Ottman, R.; Paik, M.; Blaner, W. S.; Lucier, G. W.; Covey, L.; Young, T. L.; Cooper, T. B.; Glassman, A. H.;

Perera, F. P.: Contribution of genetic and nutritional factors to DNA damage in heavy smokers. *Carcinogenesis* 18:503–509, 1997.

[27727] 1279. Nakachi, K.; Imai, K.; Hayashi, S.; Watanabe, J.; Kawajiri, K.: Genetic susceptibility to squamous cell carcinoma of the lung in relation to cigarette smoking dose. *Cancer Res.* 51: 5177–5180, 1991.

[27728] 1280. Nebert, D. W.: Personal Communication. Bethesda, Md. 2/1/1988.

[27729] 1281. Ocraft, K. P.; Muskett, J. M.; Brown, S.: Localization of the human arylhydrocarbon hydroxylase gene to the 2q31–2pter region of chromosome 2. *Ann. Hum. Genet.* 49: 237–239, 1985.

[27730] 1282. Perera, F. P.: Environment and cancer: who are susceptible? *Science* 278:1068–1073, 1997.

[27731] 1283. Petersen, D. D.; McKinney, C. E.; Ikeya, K.; Smith, H. H.; Bale, A. E.; McBride, O. W.; Nebert, D. W.: Human CYP1A1 gene: cosegregation of the enzyme inducibility phenotype and an RFLP. *Am. J. Hum. Genet.* 48:720–725, 1991.

[27732] 1284. Quattrochi, L. C.; Okino, S. T.; Pendurthi, U. R.; Tukey, R. H.: Cloning and isolation of human cytochrome P-450 cDNAs homologous to dioxin-inducible rabbit mR-

NAs encoding P-450 4 and P-450 6. DNA 4:395-400, 1985.

[27733] 1285. Tukey, R. H.; Lalley, P. A.; Nebert, D. W.: Localization of cytochrome P1-450 and P3-450 genes to mouse chromosome 9. Proc. Nat. Acad. Sci. 81:3163-3166, 1984.

[27734] 1286. Wang, X.; Zuckerman, B.; Pearson, C.; Kaufman, G.; Chen, C.; Wang, G.; Niu, T.; Wise, P. H.; Bauchner, H.; Xu, X.: Maternal cigarette smoking, metabolic gene polymorphism, and infant birth weight. J.A.M.A. 287:195-202, 2002.

[27735] 1287. Wiebel, F. J.; Hlavica, P.; Grzeschik, K. H.: Expression of aromatic polycyclic hydrocarbon-induced monooxygenase (aryl hydrocarbon hydroxylase) in man-mouse hybrids is associated with human chromosome 2. Hum. Genet. 59: 277-280, 1981.

[27736] 1288. Xu, X.; Kelsey, K. T.; Wiencke, J. K.; Wain, J. C.; Christiani, D. C.: Cytochrome P450 CYP1A1 MspI polymorphism and lung cancer susceptibility. Cancer Epidemiol. Biomarkers Prev. 5: 687-692, 1996.

[27737] 1289. Berne, R. M.: Cardiac nucleotides in hypoxia: possible role in regulation of coronary blood flow. Am. J. Physiol. 204: 317-322, 1963.

[27738] 1290. Harper, M. E.; Dugaiczyk, A.: Linkage of the evolu-

tionarily-related serum albumin and alpha-fetoprotein genes within q11-22 of human chromosome 4. Am. J. Hum. Genet. 35: 565-572, 1983.

[27739] 1291. Livingstone, F. B.: The Duffy blood groups, vivax malaria, and malaria selection in human populations: a review. Hum. Biol. 56: 413-425, 1984.

[27740] 1292. Maisonpierre, P. C.; Le Beau, M. M.; Espinosa, R., III; Ip, N. Y.; Belluscio, L.; de la Monte, S. M.; Squinto, S.; Furth, M. E.; Yancopoulos, G. D.: Human and rat brain-derived neurotrophic factor and neurotrophin-3: gene structures, distributions and chromosomal localizations. Genomics 10: 558-568, 1991.

[27741] 1293. Lefort, A.; Passage, E.; Libert, F.; Szpirer, J.; Vassart, G.; Mattei, M.-G.: Localization of human calcyphosine gene (CAPS) to the p13.3 region of chromosome 19 by in situ hybridization. Cytogenet. Cell Genet. 54: 154-155, 1990.

[27742] 1294. Yang-Feng, T. L.; Naiman, T.; Kopatz, I.; Eli, D.; Dafni, N.; Canaani, D.: Assignment of the human casein kinase II alpha-prime subunit gene (CSNK2A1) to chromosome 16p13.2-p13.3. Genomics 19: 173 only, 1994.

[27743] 1295. Bonner, T. I.; Young, A. C.; Brann, M. R.; Buckley, N. J.: Cloning and expression of the human and rat m5 muscarinic acetylcholine genes. Neuron 1: 403-410, 1988.

- [27744] 1296. Arnaudo, E.; Hirano, M.; Seelan, R. S.; Milatovich, A.; Hsieh, C.-L.; Fabrizi, G. M.; Grossman, L. I.; Francke, U.; Schon, E. A.: Tissue-specific expression and chromosome assignment of genes specifying two isoforms of subunit VIIa of human cytochrome c oxidase. *Gene* 119:299–305, 1992.
- [27745] 1297. Aman, P.; Ron, D.; Mandahl, N.; Fioretos, T.; Heim, S.; Arheden, K.; Willen, H.; Rydholm, A.; Mitelman, F.: Rearrangement of the transcription factor gene CHOP in myxoid liposarcomas with t(12;16)(q13;p11). *Genes Chromosomes Cancer* 5: 278–285, 1992.
- [27746] 1298. Jones, M. E. E.; Thorburn, A. W.; Britt, K. L.; Hewitt, K. N.; Wreford, N. G.; Proietto, J.; Oz, O. K.; Leury, B. J.; Robertson, K. M.; Yao, S.; Simpson, E. R.: Aromatase-deficient (ArKO) mice have a phenotype of increased adiposity. *Proc. Nat. Acad. Sci.* 97: 12735–12740, 2000.
- [27747] 1299. Sakai, T.; Johnson, K. J.; Murozono, M.; Sakai, K.; Magnuson, M. A.; Wieloch, T.; Cronberg, T.; Isshiki, A.; Erickson, H. P.; Fassler, R.: Plasma fibronectin supports neuronal survival and reduces brain injury following transient focal cerebral ischemia but is not essential for skin-wound healing and hemostasis. *Nature Med.* 7: 324–330, 2001.
- [27748] 1300. Weiher, H.; Noda, T.; Gray, D. A.; Sharpe, A. H.;

Jaenisch, R.: Transgenic mouse model of kidney disease: insertional inactivation of ubiquitously expressed gene leads to nephrotic syndrome. *Cell* 62:425–434, 1990.

[27749] 1301. Dausset, J.; Ivanyi, P.; Colombani, J.; Feingold, N.; Legrand, L.: Le système HLA-1: études génétiques de population et de familles. *Nouv. Rev. Franc. Hemat.* 7: 897–899, 1967.

[27750] 1302. Geurts, J. M.; Schoenmakers, E. F.; Roijer, E.; Stenman, G.; Vande Ven, W. J. M.: Expression of reciprocal hybrid transcripts of HMGIC and FHIT in a pleomorphic adenoma of the parotid gland. *Cancer Res.* 57: 13–17, 1997.

[27751] 1303. Tan, J. C.; Indelicato, S. R.; Narula, S. K.; Zavodny, P. J.; Chou, C.-C.: Characterization of interleukin-10 receptors on human and mouse cells. *J. Biol. Chem.* 268: 21053–21059, 1993.

[27752] 1304. Baumann, P.; West, S. C.: DNA end-joining catalyzed by human cell-free extracts. *Proc. Nat. Acad. Sci.* 95: 14066–14070, 1998.

[27753] 1305. Lage, H.; Dietel, M.: Cloning of a human cDNA encoding a protein with high homology to yeast methionyl-tRNA synthetase. *Gene* 178:187–189, 1996.

[27754] 1306. O'Hara, B.; Jenkins, N. A.; Gilbert, D. J.; Copeland, N. G.; Shows, T. B.; Eddy, R. L.; Bohlen, P.; Kovesdi, I.: Chro-

mosomal assignment of the heparin-binding cytokine genes MDK and PTN in mouse and man. *Cytogenet. Cell Genet.* 69: 40–43, 1995.

- [27755] 1307. Tsukamoto, A. S.; Grosschedl, R.; Guzman, R. C.; Parslow, T.; Varmus, H. E.: Expression of the int-1 gene in transgenic mice is associated with mammary gland hyperplasia and adenocarcinomas in male and female mice. *Cell* 55: 619–625, 1988.
- [27756] 1308. Jiang, X.; Li, J.; Paskind, M.; Epstein, P. M.: Inhibition of calmodulin-dependent phosphodiesterase induces apoptosis in human leukemic cells. *Proc. Nat. Acad. Sci.* 93: 11236–11241, 1996.
- [27757] 1309. Kawagishi, J.; Kumabe, T.; Yoshimoto, T.; Yamamoto, T.: Structure, organization, and transcription units of the human alpha-platelet-derived growth factor receptor gene, PDGFRA. *Genomics* 30: 224–232, 1995.
- [27758] 1310. Gispert, S.; Twells, R.; Orozco, G.; Brice, A.; Weber, J.; Heredero, L.; Scheufler, K.; Riley, B.; Allotey, R.; Nothers, C.; Hillermann, R.; Lunkes, A.; Khati, C.; Stevanin, G.; Hernandez, A.; Magarino, C.; Klockgether, T.; Durr, A.; Chneiweiss, H.; Enczmann, J.; Farrall, M.; Beckmann, J.; Mullan, M.; Wernet, P.; Agid, Y.; Freund, H.-J.; Williamson, R.; Auburger, G.; Chamberlain, S.: Chromosomal assign-

ment of the second locus for autosomal dominant cerebellar ataxia (SCA2) to chromosome 12q23–24.1. *Nature Genet.* 4: 295–299, 1993.

- [27759] 1311. Pearson, C. A.; Pearson, D.; Shibahara, S.; Hofsteenge, J.; Chiquet-Ehrismann, R.: Tenascin: cDNA cloning and induction by TGF- β . *EMBO J.* 7:2977–2981, 1988.
- [27760] 1312. Kanai, Y.; Lee, W.-S.; You, G.; Brown, D.; Hediger, M. A.: The human kidney low affinity Na(+)/glucose cotransporter SGLT2: delineation of the major renal reabsorptive mechanism for D-glucose. *J. Clin. Invest.* 93: 397–404, 1994.
- [27761] 1313. Wells, R. G.; Mohandas, T. K.; Hediger, M. A.: Localization of the Na⁺/glucose cotransporter gene SGLT2 to human chromosome 16 close to the centromere. *Genomics* 17: 787–789, 1993.
- [27762] 1314. Wells, R. G.; Pajor, A. M.; Kanai, Y.; Turk, E.; Wright, E. M.; Hediger, M. A.: Cloning of a human kidney cDNA with similarity to the sodium-glucose cotransporter. *Am. J. Physiol.* 263: F459–F465, 1992.
- [27763] 1315. Malo, D.; Schurr, E.; Dorfman, J.; Canfield, V.; Levenson, R.; Gros, P.: Three brain sodium channel α -subunit genes are clustered on the proximal segment of mouse chromosome 2. *Genomics* 10: 666–672, 1991.

- [27764] 1316.Malo, M. S.; Srivastava, K.; Andresen, J. M.; Chen, X.-N.; Korenberg, J. R.; Ingram, V. M.: Targeted gene walking by low stringency polymerase chain reaction: assignment of a putative human brain sodium channel gene (SCN3A) to chromosome 2q24-31. *Proc. Nat. Acad. Sci.* 91: 2975-2979, 1994.
- [27765] 1317.Dickinson, D. P.; Ridall, A. L.; Levine, M. J.: Human submandibular gland statherin and basic histidine-rich peptide are encoded by highly abundant mRNA's derived from a common ancestral sequence. *Biochem. Biophys. Res. Commun.* 149: 784-790, 1987.
- [27766] 1318.Sabatini, L. M.; Carlock, L. R.; Johnson, G. W.; Azen, E. A.: cDNA cloning and chromosomal localization of a gene for statherin, a regulator of calcium in saliva. (Abstract) *Am. J. Hum. Genet.* 39:A217 only, 1986.
- [27767] 1319.Sabatini, L. M.; Carlock, L. R.; Johnson, G. W.; Azen, E. A.: cDNA cloning and chromosomal localization (4q11-13) of a gene for statherin, a regulator of calcium in saliva. *Am. J. Hum. Genet.* 41:1048-1060, 1987.
- [27768] 1320.Tunnacliffe, A.; McGuire, R. S.: A physical linkage group in human chromosome band 11q23 covering a region implicated in leukocyte neoplasia. *Genomics* 8: 447-453, 1990.

- [27769] 1321. Abbott, G. W.; Butler, M. H.; Bendahhou, S.; Dalakas, M. C.; Ptacek, L. J.; Goldstein, S. A. N.: MiRP2 forms potassium channels in skeletal muscle with Kv3.4 and is associated with periodic paralysis. *Cell* 104:217–231, 2001.
- [27770] 1322. Allore, R.; O'Hanlon, D.; Price, R.; Neilson, K.; Willard, H. F.; Cox, D. R.; Marks, A.; Dunn, R. J.: Gene encoding the beta-subunit of S100 protein is on chromosome 21: implications for Down syndrome. *Science* 239:1311–1313, 1988.
- [27771] 1323. Allore, R. J.; Friend, W. C.; O'Hanlon, D.; Neilson, K. M.; Bauman, R.; Dunn, R. J.; Marks, A.: Cloning and expression of the human S100-beta gene. *J. Biol. Chem.* 265: 15537–15543, 1990.
- [27772] 1324. Duncan, A. M. V.; Higgins, J.; Dunn, R. J.; Allore, R.; Marks, A.: Refined sublocalization of the human gene encoding the beta subunit of the S100 protein (S100B) and confirmation of a subtle t(9;21) translocation using in situ hybridization. *Cytogenet. Cell Genet.* 50: 234–235, 1989.
- [27773] 1325. Reeves, R. H.; Yao, J.; Crowley, M. R.; Buck, S.; Zhang, X.; Yarowsky, P.; Gearhart, J. D.; Hilt, D. C.: Astrocytosis and axonal proliferation in the hippocampus of S100b transgenic mice. *Proc. Nat. Acad. Sci.* 91:5359–5363, 1994.

- [27774] 1326. Cloninger, C.: The psychobiological regulation of social cooperation. *Nature Med.* 1: 623–625, 1995.
- [27775] 1327. Urata, H.; Kinoshita, A.; Perez, D. M.; Misono, K. S.; Bumpus, F. M.; Graham, R. M.; Husain, A.: Cloning of the gene and cDNA for human heart chymase. *J. Biol. Chem.* 266: 17173–17179, 1991.
- [27776] 1328. Kurlan, R.; Behr, J.; Medved, L.; Shoulson, I.; Pauls, D.; Kidd, J. R.; Kidd, K. K.: Familial Tourette's syndrome: report of a large pedigree and potential for linkage analysis. *Neurology* 36: 772–776, 1986.
- [27777] 1329. Kurihara, Y.; Kurihara, H.; Suzuki, H.; Kodama, T.; Maemura, K.; Nagai, R.; Oda, H.; Kuwaki, T.; Cao, W.-H.; Kamada, N.; Jishage, K.; Ouchi, Y.; Azuma, S.; Toyoda, Y.; Ishikawa, T.; Kumada, M.; Yazaki, Y.: Elevated blood pressure and craniofacial abnormalities in mice deficient in endothelin-1. *Nature* 368: 703–710, 1994.
- [27778] 1330. Swanson, J.; Oosterlaan, J.; Murias, M.; Schuck, S.; Flodman, P.; Spence, M. A.; Wasdell, M.; Ding, Y.; Chi, H.-C.; Smith, M.; Mann, M.; Carlson, C.; Kennedy, J. L.; Sergeant, J. A.; Leung, P.; Zhang, Y.-P.; Sadeh, A.; Chen, C.; Whalen, C. K.; Babb, K. A.; Moyzis, R.; Posner, M. I.: Attention deficit/hyperactivity disorder children with a 7-repeat allele of the dopamine receptor D4 gene have extreme be-

havior but normal performance on critical neuropsychological tests of attention. Proc. Nat. Acad. Sci. 97: 4754–4759, 2000.

- [27779] 1331. Nakae, J.; Biggs, W. H., III; Kitamura, T.; Cavenee, W. K.; Wright, C. V. E.; Arden, K. C.; Accili, D.: Regulation of insulin action and pancreatic beta-cell function by mutated alleles of the gene encoding forkhead transcription factor Foxo1. Nature Genet. 32: 245–253, 2002.
- [27780] 1332. Fischer, J. A.; Egert, F.; Werder, E.; Born, W.: An inherited mutation associated with functional deficiency of the alpha-subunit of the guanine nucleotide-binding protein Gs in pseudo- and pseudopseudohypoparathyroidism. J. Clin. Endocr. Metab. 83: 935–938, 1998.
- [27781] 1333. Zhuang, Y.; Soriano, P.; Weintraub, H.: The helix-loop-helix gene E2A is required for B cell formation. Cell 79: 875–884, 1994.
- [27782] 1334. Nance, W. E.; Jackson, C. E.; Witkop, C. J., Jr.: Amish albinism: a distinctive autosomal recessive phenotype. Am. J. Hum. Genet. 22: 579–586, 1970.
- [27783] 1335. Nance, W. E.; Witkop, C. J., Jr.; Rawls, R. F.: Genetic and biochemical evidence for two forms of oculocutaneous albinism in man. In: The Clinical Delineation of Birth Defects. Eye. Baltimore: Williams and Wilkins (pub.) VIII: 1971.

Pp. 125–128.

- [27784] 1336.Okoro, A. N.: Albinism in Nigeria: a clinical and social study. *Brit.J. Derm.* 92: 485–492, 1975.
- [27785] 1337.Ramsay, M.; Colman, M.–A.; Stevens, G.; Zwane, E.; Kromberg, J.;Farrall, M.; Jenkins, T.: The tyrosinase–positive oculocutaneousalbinism locus maps to chromosome 15q11.2–q12. *Am. J. Hum. Genet.* 51:879–884, 1992.
- [27786] 1338.Rinchik, E. M.; Bultman, S. J.; Horsthemke, B.; Lee, S.–T.; Strunk,K. M.; Spritz, R. A.; Avidano, K. M.; Jong, M. T. C.; Nicholls, R.D.: A gene for the mouse pink–eyed dilution locus and for human typell oculocutaneous albinism. *Nature* 361: 72–76, 1993.
- [27787] 1339.Robinson, W. P.; Bottani, A.; Yagang, X.; Balakrishman, J.; Binkert,F.; Machler, M.; Prader, A.; Schinzel, A.: Molecular, cytogenetic,and clinical investigations of Prader–Willi syndrome patients. *Am.J. Hum. Genet.* 49: 1219–1234, 1991.
- [27788] 1340.Rosemblat, S.; Durham–Pierre, D.; Gardner, J. M.; Nakatsu, Y.;Brilliant, M. H.; Orlow, S. J.: Identification of a melanosomal membraneprotein encoded by the pink–eyed dilution (type II oculocutaneousalbinism) gene. *Proc. Nat. Acad. Sci.* 91: 12071–12075, 1994.
- [27789] 1341.Spritz, R. A.; Fukai, K.; Holmes, S. A.; Luande, J.:

Frequent intragenic deletion of the P gene in Tanzanian patients with type II oculocutaneous albinism (OCA2). *Am. J. Hum. Genet.* 56: 1320–1323, 1995.

[27790] 1342. Stevens, G.; Ramsay, M.; Jenkins, T.: Oculocutaneous albinism (OCA2) in sub-Saharan Africa: distribution of the common 2.7-kb P gene deletion mutation. *Hum. Genet.* 99: 523–527, 1997.

[27791] 1343. Stevens, G.; van Beukering, J.; Jenkins, T.; Ramsay, M.: An intragenic deletion of the P gene is the common mutation causing tyrosinase-positive oculocutaneous albinism in Southern African Negroids. *Am. J. Hum. Genet.* 56: 586–591, 1995.

[27792] 1344. Trevor-Roper, P. D.: Marriage of two complete albinos with normally pigmented offspring. *Brit. J. Ophthalmol.* 36: 107–108, 1952.

[27793] 1345. Trevor-Roper, P. D.: Albinism. *Proc. Roy. Soc. Med.* 56: 21–24, 1963.

[27794] 1346. Waardenburg, P. J.: *Genetics and Ophthalmology*. Springfield, Ill.: Charles C Thomas (pub.) 1: 1961. Pp. 732 only.

[27795] 1347. Walpole, I. R.; Mulcahy, M. T.: Tyrosinase positive albinism with familial 46,XY,t(2;4)(q31.2;q31.22) balanced translocation. *J. Med. Genet.* 28: 482–484, 1991.

- [27796] 1348. Witkop, C. J., Jr.: Dental problems of an hereditary nature. In: Witkop, C. J.: Genetics and Dental Health. New York: McGraw-Hill (pub.) 1962.
- [27797] 1349. Witkop, C. J., Jr.: Personal Communication. Minneapolis, Minn. 1966.
- [27798] 1350. Witkop, C. J., Jr.; Niswander, J. D.; Bergsma, D. R.; Workman, P. L.; White, J. G.: Tyrosinase-positive oculocutaneous albinism among the Zuni and the Brandywine triracial isolate: biochemical and clinical characteristics and fertility. *Am. J. Phys. Anthropol.* 36:397-405, 1972.
- [27799] 1351. Witkop, C. J., Jr.; Quevedo, W. C., Jr.; Fitzpatrick, T. B.; King, R. A.: Albinism. In: Scriver, C. R.; Beaudet, A. L.; Sly, W. S.; Valle, D.: *The Metabolic Basis of Inherited Disease*. Vol. II. New York: McGraw-Hill (pub.) (6th ed.): 1989. Pp. 2905-2947.
- [27800] 1352. Witkop, C. J., Jr.; Van Scott, E. J., Jr.; Jacoby, G. A.: Evidence for two forms of autosomal recessive albinism in man. (Abstract) *Proc. Second Int. Cong. Hum. Genet.*, Rome, Sept. 6-12, 1961 2: 1064-1065, 1963.
- [27801] 1353. Woolf, C. M.: Albinism among Indians in Arizona and New Mexico. *Am. J. Hum. Genet.* 17: 23-35, 1965.
- [27802] 1354. Woolf, C. M.; Dukepoo, F. C.: Hopi Indians, inbreeding and albinism. *Science* 164:30-37, 1969.

- [27803] 1355.Dittmar, G. A. G.; Wilkinson, C. R. M.; Jedrzejewski, P. T.; Finley,D.: Role of a ubiquitin–like modification in polarized morphogenesis. *Science* 295:2442–2446, 2002.
- [27804] 1356.Friedman, J. S.; Koop, B. F.; Raymond, V.; Walter, M. A.: Isolationof a ubiquitin–like (UBL5) gene from a screen identifying highly expressedand conserved iris genes. *Genomics* 71: 252–255, 2001.
- [27805] 1357.Jarrous, N.; Eder, P. S.; Guerrier–Takada, C.; Hoog, C.; Altman,S.: Autoantigenic properties of some protein subunits of catalyticallyactive complexes of human ribonuclease P. *RNA* 4: 407–417, 1998.
- [27806] 1358.MacArthur, C. A.; Shackleford, G. M.: Npm3: A novel, widely expressedgene encoding a protein related to the molecular chaperones nucleoplasminand nucleophosmin. *Genomics* 42: 137–140, 1997.
- [27807] 1359.Shackleford, G. M.; Ganguly, A.; MacArthur, C. A.: Cloning, expressionand nuclear localization of human NPM3, a member of the nucleophosmin/nucleoplasminfamily of nuclear chaperones. *BMC Genomics* 2: 8, 2001. Note: ElectronicArticle.
- [27808] 1360.Hauf, S.; Waizenegger, I. C.; Peters, J.–M.: Cohesin cleavageby separase required for anaphase and cytokinesis in human cells. *Science* 293:1320–1323, 2001.

- [27809] 1361.Hoque, M. T.; Ishikawa, F.: Human chromatid cohesin componenthRad21 is phosphorylated in M phase and associated with metaphasecentromeres. *J. Biol. Chem.* 276: 5059–5067, 2001.
- [27810] 1362.McKay, M. J.; Troelstra, C.; van der Spek, P.; Kanaar, R.; Smit,B.; Hagemeijer, A.; Bootsma, D.; Hoeijmakers, J. H. J.: Sequenceconservation of the rad21 *Schizosaccharomyces pombe* DNA double-strandbreak repair gene in human and mouse. *Genomics* 36: 305–315, 1996.
- [27811] 1363.Sadano, H.; Sugimoto, H.; Sakai, F.; Nomura, N.; Os-umi, T.: NXP-1,a human protein related to Rad21/Scc1/Mcd1, is a component of thenuclear matrix. *Biochem. Biophys. Res. Commun.* 267: 418–422, 2000.
- [27812] 1364.Fujimoto, K.; Shen, M.; Noshiro, M.; Matsubara, K.; Shingu, S.;Honda, K.; Yoshida, E.; Suardita, K.; Matsuda, Y.; Kato, Y.: Molecularcloning and characterization of DEC2, a new member of basic helix-loop-helixproteins. *Biochem. Biophys. Res. Commun.* 280: 164–171, 2001.
- [27813] 1365.Garriga-Canut, M.; Roopra, A.; Buckley, N. J.: The basic helix-loop-helixprotein, SHARP-1, represses transcription by a histone deacetylase-dependentand histone deacetylase-independent mechanism. *J. Biol. Chem.* 276:14821–14828, 2001.

- [27814] 1366. Fukamachi, S.; Shimada, A.; Shima, A.: Mutations in the gene encoding B, a novel transporter protein, reduce melanin content in medaka. *Nature Genet.* 28: 381–385, 2001.
- [27815] 1367. Harada, M.; Li, Y. F.; El-Gamil, M.; Rosenberg, S. A.; Robbins, P. F.: Use of an in vitro immunoselected tumor line to identify shared melanoma antigens recognized by HLA-A*0201-restricted T cells. *Cancer Res.* 61: 1089–1094, 2001.
- [27816] 1368. Newton, J. M.; Cohen-Barak, O.; Hagiwara, N.; Gardner, J. M.; Davisson, M. T.; King, R. A.; Brilliant, M. H.: Mutations in the human orthologue of the mouse underwhite gene (uw) underlie a new form of oculocutaneous albinism, OCA4. *Am. J. Hum. Genet.* 69: 981–988, 2001.
- [27817] 1369. Gu, H.; Saito, K.; Klamman, L. D.; Shen, J.; Fleming, T.; Wang, Y.-P.; Pratt, J. C.; Lin, G.; Lim, B.; Kinet, J.-P.; Neel, B. G.: Essential role for Gab2 in the allergic response. *Nature* 412: 186–190, 2001.
- [27818] 1370. Appel, S.; Reichwald, K.; Zimmermann, W.; Reis, A.; Rosenthal, A.; Hennies, H. C.: Identification and localization of a new human myotubularin-related protein gene, MTMR8, on 8p22–p23. *Genomics* 75:6–8, 2001.
- [27819] 1371. Laporte, J.; Blondeau, F.; Buj-Bello, A.; Mandel, J.-L.:

The myotubularin family: from genetic disease to phosphoinositide metabolism. *Trends Genet.* 17: 221–228, 2001.

[27820] 1372. Ariizumi, K.; Shen, G.-L.; Shikano, S.; Xu, S.; Ritter, R., III; Kumamoto, T.; Edelbaum, D.; Morita, A.; Bergstresser, P. R.; Takashima, A.: Identification of a novel, dendritic cell-associated molecule, dectin-1, by subtractive cDNA cloning. *J. Biol. Chem.* 275: 20157–20167, 2000.

[27821] 1373. Brown, G. D.; Gordon, S.: A new receptor for beta-glucans. *Nature* 413:36–37, 2001.

[27822] 1374. Hernanz-Falcon, P.; Arce, I.; Roda-Navarro, P.; Fernandez-Ruiz, E.: Cloning of human dectin-1, a novel C-type lectin-like receptor gene expressed on dendritic cells. *Immunogenetics* 53: 288–295, 2001.

[27823] 1375. Yokota, K.; Takashima, A.; Bergstresser, P. R.; Ariizumi, K.: Identification of a human homologue of the dendritic cell-associated C-type lectin-1, dectin-1. *Gene* 272: 51–60, 2001.

[27824] 1376. Mackay, F.; Woodcock, S. A.; Lawton, P.; Ambrose, C.; Baetscher, M.; Schneider, P.; Tschopp, J.; Browning, J. L.: Mice transgenic for BAFF develop lymphocytic disorders along with autoimmune manifestations. *J. Exp. Med.* 190:

1697–1710, 1999.

- [27825] 1377. Thompson, J. S.; Bixler, S. A.; Qian, F.; Vora, K.; Scott, M. L.; Cachero, T. G.; Hession, C.; Schneider, P.; Sizing, I. D.; Mullen, C.; Strauch, K.; Zafari, M.; Benjamin, C. D.; Tschopp, J.; Browning, J. L.; Ambrose, C.: BAFF-R, a newly identified TNF receptor that specifically interacts with BAFF. *Science* 293: 2108–2111, 2001.
- [27826] 1378. Xu, S.; Lam, K.-P.: B-cell maturation protein, which binds the tumor necrosis factor family members BAFF and APRIL, is dispensable for humoral immune responses. *Molec. Cell. Biol.* 21: 4067–4074, 2001.
- [27827] 1379. Yan, M.; Wang, H.; Chan, B.; Roose-Girma, M.; Erickson, S.; Baker, T.; Tumas, D.; Grewal, I. S.; Dixit, V. M.: Activation and accumulation of B cells in TACI-deficient mice. *Nature Immun.* 2: 638–643, 2001.
- [27828] 1380. Hagen, G.; Muller, S.; Beato, M.; Suske, G.: Sp1-mediated transcriptional activation is repressed by Sp3. *EMBO J.* 13: 3843–3851, 1994.
- [27829] 1381. Kalff-Suske, M.; Kunz, J.; Grzeschik, K.-H.; Suske, G.: Human Sp3 transcriptional regulator gene (SP3) maps to chromosome 2q31. *Genomics* 37: 410–412, 1996.
- [27830] 1382. Kingsley, C.; Winoto, A.: Cloning of GT box-binding proteins: a novel Sp1 multigene family regulating T-cell

receptor gene expression. *Molec.Cell. Biol.* 12:
4251–44261, 1992.

- [27831] 1383.Carmeci, C.; Thompson, D. A.; Ring, H. Z.; Francke, U.; Weigel, R. J.: Identification of a gene (GPR30) with homology to the G-protein-coupled receptor superfamily associated with estrogen receptor expression in breast cancer. *Genomics* 45: 607–617, 1997.
- [27832] 1384.Feng, Y.; Gregor, P.: Cloning of a novel member of the G protein-coupled receptor family related to peptide receptors. *Biochem. Biophys. Res. Commun.* 231: 651–654, 1997.
- [27833] 1385.Kvingedal, A. M.; Smeland, E. B.: A novel putative G-protein-coupled receptor expressed in lung, heart and lymphoid tissue. *FEBS Lett.* 407:59–62, 1997.
- [27834] 1386.Owman, C.; Blay, P.; Nilsson, C.; Lolait, S. J.: Cloning of human cDNA encoding a novel heptahelix receptor expressed in Burkitt's lymphoma and widely distributed in brain and peripheral tissues. *Biochem. Biophys. Res. Commun.* 228: 285–292, 1996.
- [27835] 1387.Takada, Y.; Kato, C.; Kondo, S.; Korenaga, R.; Ando, J.: Cloning of cDNAs encoding G protein-coupled receptor expressed in human endothelial cells exposed to fluid shear stress. *Biochem. Biophys. Res. Commun.*

240;-737-741,1997.

- [27836] 1388.Inohara, N.; Koseki, T.; Chen, S.; Wu, X.; Nunez, G.: CIDE, a novel family of cell death activators with homology to the 45 kDa subunit of the DNA fragmentation factor. *EMBO J.* 17: 2526-2533,1998.
- [27837] 1389.Leek, J. P.; Carr, I. M.; Bell, S. M.; Markham, A. F.; Lench, N.J.: Assignment of the DNA fragmentation factor gene (DFFA) to human chromosome bands 1p36.3-p36.2 by in situ hybridization. *Cytogenet.Cell Genet.* 79: 212-213, 1997.
- [27838] 1390.Liu, X.; Zou, H.; Slaughter, C.; Wang, X.: DFF, a heterodimeric protein that functions downstream of caspase-3 to trigger DNA fragmentation during apoptosis. *Cell* 89: 175-184, 1997.
- [27839] 1391.Judson, H.; van Roy, N.; Strain, L.; Vandesompele, J.; Van Gele, M.; Speleman, F.; Bonthron, D. T.: Structure and mutation analysis of the gene encoding DNA fragmentation factor 40 (caspase-activated nuclease), a candidate neuroblastoma tumour suppressor gene. *Hum.Genet.* 106: 406-413, 2000.
- [27840] 1392.Liu, X.; Li, P.; Widlak, P.; Zou, H.; Luo, X.; Garrard, W. T.; Wang, X.: The 40-kDa subunit of DNA fragmentation factor induces DNA fragmentation and chromatin conden-

sation during apoptosis. Proc.Nat. Acad. Sci. 95:
8461–8466, 1998.

- [27841] 1393.Mukae, N.; Enari, M.; Sakahira, H.; Fukuda, Y.; In-
azawa, J.; Toh,H.; Nagata, S.: Molecular cloning and char-
acterization of human caspase-activatedDNase. Proc. Nat.
Acad. Sci. 95: 9123–9128, 1998.
- [27842] 1394.Garrett-Sinha, L. A.; Eberspaecher, H.; Seldin, M. F.;
de Crombrughe,B.: A gene for a novel zinc-finger protein
expressed in differentiatedepithelial cells and transiently
in certain mesenchymal cells. J.Biol. Chem. 271:
31384–31390, 1996.
- [27843] 1395.Segre, J. A.; Bauer, C.; Fuchs, E.: Klf4 is a transcrip-
tion factorrequired for establishing the barrier function of
the skin. NatureGenet. 22: 356–360, 1999.
- [27844] 1396.Shields, J. M.; Christy, R. J.; Yang, V. W.: Identifica-
tion andcharacterization of a gene encoding a gut-
enriched Kruppel-like factorexpressed during growth ar-
rest. J. Biol. Chem. 271: 20009–20017,1996.
- [27845] 1397.Yet, S.-F.; McA'Nulty, M. M.; Folta, S. C.; Yen, H.-W.;
Yoshizumi,M.; Hsieh, C.-M.; Layne, M. D.; Chin, M. T.;
Wang, H.; Perrella, M.A.; Jain, M. K.; Lee, M.-E.: Human
EZF, a Kruppel-like zinc fingerprotein, is expressed in
vascular endothelial cells and contains transcriptionalacti-

vation and repression domains. J. Biol. Chem. 273:
1026–1031,1998.

[27846] 1398.Sherman, P. M.; Sun, H.; Macke, J. P.; Williams, J.;
Smallwood,P. M.; Nathans, J.: Identification and character-
ization of a conservedfamily of protein serine/threonine
phosphatases homologous to Drosophilaretinal degenera-
tion C (rdgC). Proc. Nat. Acad. Sci. 94:
11639–11644,1997.

[27847] 1399.Ohira, M.; Ootsuyama A.; Suzuki, E.; Ichikawa, H.;
Seki, N.; Nagase,T.; Monura, N.; Ohki, M.: Identification of
a novel human gene containingthe tetratricopeptide re-
peat domain from the Down syndrome regionof chromo-
some 21. DNA Res. 3: 9–16, 1996.

[27848] 1400.Tsukahara, F.; Hattori, M.; Muraki, T.; Sakaki, Y.:
Identificationand cloning of a novel cDNA belonging to
tetratricopeptide repeatgene family from Down syn-
drome–critical region 21q22.2. J. Biochem. 120:820–827,
1996.

[27849] 1401.Albrecht, U.; Sun, Z. S.; Eichele, G.; Lee, C. C.: A dif-
ferentialresponse to two putative mammalian circadian
regulators, mper1 andmper2, to light. Cell 91:
1055–1064, 1997.

[27850] 1402.Duan, W.; Zhang, Z.; Gash, D. M.; Mattson, M. P.:

Participation of prostate apoptosis response-4 in degeneration of dopaminergic neurons in models of Parkinson's disease. *Ann. Neurol.* 46: 587–597, 1999.

[27851] 1403. Johnstone, R. W.; See, R. H.; Sells, S. F.; Wang, J.; Muthukkumar, S.; Englert, C.; Haber, D. A.; Licht, J. D.; Sugrue, S. P.; Roberts, T.; Rangnekar, V. M.; Shi, Y.: A novel repressor, par-4, modulates transcription and growth suppression functions of the Wilms' tumor suppressor WT1. *Molec. Cell. Biol.* 16: 6945–6956, 1996.

[27852] 1404. Johnstone, R. W.; Tommerup, N.; Hansen, C.; Vissing, H.; Shi, Y.: Mapping of the human PAWR (par-4) gene to chromosome 12q21. *Genomics* 53:241–243, 1998.

[27853] 1405. Bai, J.; Uehara, Y.; Montell, D. J.: Regulation of invasive cell behavior by taiman, a *Drosophila* protein related to ALB1, a steroid receptor coactivator amplified in breast cancer. *Cell* 103: 1047–1058, 2000.

[27854] 1406. Chen, H.; Lin, R. J.; Schiltz, R. L.; Chakravarti, D.; Nash, A.; Nagy, L.; Privalsky, M. L.; Nakatani, Y.; Evans, R. M.: Nuclear receptor coactivator ACTR is a novel histone acetyltransferase and forms a multimeric activation complex with P/CAF and CBP/p300. *Cell* 90:569–580, 1997.

[27855] 1407. Guan, X.-Y.; Xu, J.; Anzick, S. L.; Zhang, H.; Trent, J. M.; Meltzer, P. S.: Hybrid selection of transcribed se-

quences from microdissected DNA: isolation of genes within amplified region at 20q11–q13.2 in breast cancer. *Cancer Res.* 56: 3446–3450, 1996.

- [27856] 1408. Shirazi, S. K.; Bober, M. A.; Coetzee, G. A.: Polymorphic exonic CAG microsatellites in the gene amplified in breast cancer (AIB1 gene). *Clin. Genet.* 54: 102–103, 1998.
- [27857] 1409. Takeshita, A.; Cardona, G. R.; Koibuchi, N.; Suen, C.-S.; Chin, W. W.: TRAM-1, a novel 160-kDa thyroid hormone receptor activator molecule, exhibits distinct properties from steroid receptor coactivator-1. *J. Biol. Chem.* 272: 27629–27634, 1997.
- [27858] 1410. Stoss, O.; Schwaiger, F.-W.; Cooper, T. A.; Stamm, S.: Alternative splicing determines the intracellular localization of the novel nuclear protein Nop30 and its interaction with the splicing factor SRp30c. *J. Biol. Chem.* 274: 10951–10962, 1999.
- [27859] 1411. Arrigo, G.; Gherzi, R.; Bonaglia, M. C.; Leprini, A.; Zuffardi, O.; Zardi, L.: Assignment of the tenascin-R gene (Tnr) to mouse chromosome 4 band E2 by fluorescence in situ hybridization; refinement of the human TNR location to chromosome 1q24. *Cytogenet. Cell Genet.* 78:145–146, 1997.
- [27860] 1412. Carnemolla, B.; Leprini, A.; Borsi, L.; Querze, G.;

Urbini, S.;Zardi, L.: Human tenascin-R: complete primary structure, pre-mRNAalternative splicing and gene localization on chromosome 1q23-q24. J.Biol. Chem. 271: 8157-8160, 1996.

- [27861] 1413.Erickson, H. P.: Tenascin-C, tenascin-R and tenascin-X: a familyof talented proteins in search of functions. Curr. Opin. Cell Biol. 5:869-876, 1993.
- [27862] 1414.Leprini, A.; Gherzi, R.; Siri, A.; Querze, G.; Viti, F.; Zardi,L.: The human tenascin-R gene. J. Biol. Chem. 271: 31251-31254,1996.
- [27863] 1415.Williams, H.; Schachner, M.; Wang, B.; Kenwrick, S.: Radiationhybrid mapping of the genes for tenascin-R (TNR), phosducin (PDC),Laminin C1 (LAMC1), and TAX in 1q25-q32. Genomics 46: 165-166, 1997.
- [27864] 1416.Chou, J. J.; Li, H.; Salvesen, G. S.; Yuan, J.; Wagner, G.: Solutionstructure of BID, an intracellular amplifier of apoptotic signaling. Cell 95:615-624, 1999.
- [27865] 1417.Footz, T. K.; Birren, B.; Minoshima, S.; Asakawa, S.; Shimizu,N.; Ali Riazi, M.; McDermid, H. E.: The gene for death agonist BIDmaps to the region of human 22q11.2 duplicated in cat eye syndromechromosomes and to mouse chromosome 6. Genomics 51: 472-475, 1998.
- [27866] 1418.Li, H.; Zhu, H.; Xu, C.; Yuan, J.: Cleavage of BID by

caspase8 mediates the mitochondrial damage in the Fas pathway of apoptosis. Cell 94:491–501, 1998.

[27867] 1419.Luo, X.; Budihardjo, I.; Zou, H.; Slaughter, C.; Wang, X.: Bid,a Bcl2 interacting protein, mediates cytochrome c release from mitochondriain response to activation of cell surface death receptors. Cell 94:481–490, 1998.

[27868] 1420.McDonnell, J. M.; Fushman, D.; Milliman, C. L.; Korsmeyer, S. J.;Cowburn, D.: Solution structure of the proapoptotic molecule BID:a structural basis for apoptotic agonists and antagonists. Cell 96:625–634, 1999.

[27869] 1421.Wang, K.; Yin, X.–M.; Chao, D. T.; Milliman, C. L.; Korsmeyer,S. J.: BID: a novel BH3 domain–only death agonist. Genes Dev. 10:2859–2869, 1996.

[27870] 1422.Wang, K.; Yin, X.–M.; Copeland, N. G.; Gilbert, D. J.; Jenkins,N. A.; Keck, C. L.; Zimonjic, D. B.; Popescu, N. C.; Korsmeyer, S.J.: BID, a proapoptotic BCL–2 family member, is localized to mousechromosome 6 and human chromosome 22q11. Genomics 53: 235–238, 1998.

[27871] 1423.Yin, X.–M.; Wang, K.; Gross, A.; Zhao, Y.; Zinkel, S.; Klocke,B.; Roth, K. A.; Korsmeyer, S. J.: Bid–deficient mice are resistantto Fas–induced hepatocellular apoptosis. Nature 400: 886–891, 1999.

[27872] 1424.Zha, J.; Weiler, S.; Oh, K. J.; Wei, M. C.; Korsmeyer, S.

J.:Posttranslational N-myristoylation of BID as a molecular switch fortargeting mitochondria and apoptosis. Science 290: 1761–1765, 2000.

[27873] 1425.Giguere, V.; Yang, N.; Segui, P.; Evans, R. M.: Identificationof a new class of steroid hormone receptors. Nature 331: 91–94,1988.

[27874] 1426.Sladek, R.; Bader, J.–A.; Giguere, V.: The orphan nuclear receptorestrogen–related receptor alpha is a transcriptional regulator ofthe human medium–chain acyl coenzyme A dehydrogenase gene. Molec.Cell. Biol. 17: 5400–5409, 1997.

[27875] 1427.Sladek, R.; Beatty, B.; Squire, J.; Copeland, N. G.; Gilbert, D.J.; Jenkins, N. A.; Giguere, V.: Chromosomal mapping of the humanand murine orphan receptors ERR–alpha (ESRRA) and ERR–beta (ESRRB)and identification of a novel human ERR–alpha–related pseudogene. Genomics 45:320–326, 1997.

[27876] 1428.Bengtson, P.; Larson, C.; Lundblad, A.; Larson, G.; Pahlsson, P.: Identification of a missense mutation (G329A; arg110–to–gln) inthe human FUT7 gene. J. Biol. Chem. 276: 31575–31582, 2001.

[27877] 1429.Mahdavi, J.; Sonden, B.; Hurtig, M.; Olfat, F. O.; Forsberg, L.;Roche, N.; Angstrom, J.; Larsson, T.; Teneberg, S.;

Karlsson, K.-A.; Altraja, S.; Wadstrom, T.; and 11 others: Helicobacter pylori SabA adhesin in persistent infection and chronic inflammation. Science 297:573–578, 2002.

[27878] 1430. Natsuka, S.; Gersten, K. M.; Zenita, K.; Kannagi, R.; Lowe, J. B.: Molecular cloning of a cDNA encoding a novel human leukocyte α -1,3-fucosyltransferase capable of synthesizing the sialyl Lewis x determinant. J. Biol. Chem. 269: 16789–16794, 1994.

[27879] 1431. Sasaki, K.; Kurata, K.; Funayama, K.; Nagata, M.; Watanabe, E.; Ohta, S.; Hannai, N.; Nishi, T.: Expression cloning of a novel α -1,3-fucosyltransferase that is involved in biosynthesis of the sialyl Lewis x carbohydrate determinants in leukocytes. J. Biol. Chem. 269: 14730–14737, 1994.

[27880] 1432. Bale, T. L.; Contarino, A.; Smith, G. W.; Chan, R.; Gold, L. H.; Sawchenko, P. E.; Koob, G. F.; Vale, W. W.; Lee, K.-F.: Mice deficient for corticotropin-releasing hormone receptor-2 display anxiety-like behaviour and are hypersensitive to stress. Nature Genet. 24: 410–414, 2000.

[27881] 1433. Coste, S. C.; Kesterson, R. A.; Heldwein, K. A.; Stevens, S. L.; Heard, A. D.; Hollis, J. H.; Murray, S. E.; Hill, J. K.; Pantely, G. A.; Hohimer, A. R.; Hatton, D. C.; Phillips, T. J.; Finn, D. A.; Low, M. J.; Rittenberg, M. B.; Stenzel, P.;

Stenzel-Poore, M. P.: Abnormal adaptations to stress and impaired cardiovascular function in mice lacking corticotropin-releasing hormone receptor-2. *Nature Genet.* 24: 403–409, 2000.

[27882] 1434. Hsu, S. Y.; Hsueh, A. J. W.: Human stresscopin and stresscopin-related peptide are selective ligands for the type 2 corticotropin-releasing hormone receptor. *Nature Med.* 7: 605–611, 2001.

[27883] 1435. Kishimoto, T.; Radulovic, J.; Radulovic, M.; Lin, C. R.; Schrick, C.; Hooshmand, F.; Hermanson, O.; Rosenfeld, M. G.; Spiess, J.: Deletion of *Crhr2* reveals an anxiolytic role for corticotropin-releasing hormone receptor-2. *Nature Genet.* 24: 415–419, 2000.

[27884] 1436. Kostich, W. A.; Chen, A.; Sperle, K.; Largent, B. L.: Molecular identification and analysis of a novel human corticotropin-releasing factor (CRF) receptor: the CRF2-gamma receptor. *Molec. Endocr.* 12: 1077–1085, 1998.

[27885] 1437. Lesh, J. S.; Burrows, H. L.; Seasholtz, A. F.; Camper, S. A.: Mapping of the mouse corticotropin-releasing hormone receptor 2 gene (*Crhr2*) to chromosome 6. *Mammalian Genome* 8: 944–945, 1997.

[27886] 1438. Liaw, C. W.; Lovenberg, T. W.; Barry, G.; Oltersdorf,

T.; Grigoriadis, D. E.; De Souza, E. B.: Cloning and characterization of the human corticotropin-releasing factor-2 receptor complementary deoxyribonucleic acid. *Endocrinology* 137: 72–77, 1996.

- [27887] 1439. Meyer, A. H.; Ullmer, C.; Schmuck, K.; Morel, C.; Wishart, W.; Lubbert, H.; Engels, P.: Localization of the human CRF2 receptor to 7p21–p15 by radiation hybrid mapping and FISH analysis. *Genomics* 40:189–190, 1997.
- [27888] 1440. Bastians, H.; Krebber, H.; Hoheisel, J.; Ohl, S.; Lichter, P.; Ponstingl, H.; Joos, S.: Assignment of the human serine/threonine protein phosphatase 4 gene (PPP4C) to chromosome 16p11–p12 by fluorescence in situ hybridization. *Genomics* 42: 181–182, 1997.
- [27889] 1441. Brewis, N. D.; Cohen, P. T. W.: Protein phosphatase X has been highly conserved during mammalian evolution. *Biochim. Biophys. Acta* 1171:231–233, 1992.
- [27890] 1442. Dallery, E.; Galiegue-Zouitina, S.; Collyn-d'Hooghe, M.; Quief, S.; Denis, C.; Hildebrand, M.-P.; Lantoine, D.; Deweindt, C.; Tilly, H.; Bastard, C.; Kerckaert, J.-P.: TTF, a gene encoding a novel small G protein, fuses to the lymphoma-associated LAZ3 gene by t(3;4) chromosomal translocation. *Oncogene* 10: 2171–2178, 1995.
- [27891] 1443. Dallery-Prudhomme, E.; Roumier, C.; Denis, C.;

Preudhomme, C.;Kerckaert, J.-P.; Galiegue-Zouitina, S.:
Genomic structure and assignment of the RhoH/TTF small
GTPase gene (ARHH) to 4p13 by in situ hybridization. Ge-
nomics 43:89–94, 1997.

[27892] 1444.Ensinger, C.; Obrist, P.; Mikuz, G.; Merx, G.;
Smeets, D.; Banziger, R.; Bachmann, F.; Burger, M.: Assign-
ment of the p150 subunit of the eukaryotic initiation factor
3A gene (EIF3A) to human chromosome band 10q26 by in
situ hybridisation. Cytogenet. Cell Genet. 83: 74–75, 1998.

[27893] 1445.Johnson, K. R.; Merrick, W. C.; Zoll, W. L.; Zhu, Y.:
Identification of cDNA clones for the large subunit of eu-
karyotic translation initiation factor 3: comparison of ho-
mologues from human, *Nicotiana tabacum*, *Caenorhabditis*
elegans, and *Saccharomyces cerevisiae*. J. Biol. Chem.
272:7106–7113, 1997.

[27894] 1446.Aaltonen, J.; Björnsen, P.; Sandkuijl, L.; Perheentupa,
J.; Peltonen, L.: An autosomal locus causing autoimmune
disease: autoimmune polyglandular disease type I assigned
to chromosome 21. Nature Genet. 8: 83–87, 1994.

[27895] 1447.Aaltonen, J.; Horelli-Kuitunen, N.; Fan, J.-B.; Björnsen,
P.; Perheentupa, J.; Myers, R.; Palotie, A.; Peltonen, L.:
High-resolution physical and transcriptional mapping of
the autoimmune polyendocrinopathy–candida–

sis-ectodermaldystrophy locus on chromosome 21q22.3 by FISH. *Genome Res.* 7: 820-829,1997.

- [27896] 1448. Ahonen, P.: Autoimmune polyendocrinopathy--candidosis--ectodermaldystrophy (APECED): autosomal recessive inheritance. *Clin. Genet.* 27:535-542, 1985.
- [27897] 1449. Ahonen, P.; Myllarniemi, S.; Sipila, I.; Perheentupa, J.: Clinical variation of autoimmune polyendocrinopathy-candidiasis-ectodermaldystrophy (APECED) in a series of 68 patients. *New Eng. J. Med.* 322:1829-1836, 1990.
- [27898] 1450. Arulanantham, K.; Kwyer, J. M.; Genel, M.: Evidence for defective immunoregulation in the syndrome of familial candidiasis endocrinopathy. *New Eng. J. Med.* 300: 164-168, 1979.
- [27899] 1451. Betterle, C.; Greggio, N. A.; Volpato, M.: Autoimmune polyglandular syndrome type 1. *J. Clin. Endocr. Metab.* 83: 1049-1055, 1998.
- [27900] 1452. Bjorses, P.; Aaltonen, J.; Vikman, A.; Perheentupa, J.; Ben-Zion, G.; Chiumello, G.; Dahl, N.; Heideman, P.; Hoorweg-Nijman, J. J. G.; Mathivon, L.; Mullis, P. E.; Pohl, M.; Ritzen, M.; Romeo, G.; Shapiro, M. S.; Smith, C. S.; Solyom, J.; Zlotogora, J.; Peltonen, L.: Genetic homogeneity of autoimmune polyglandular disease type I. *Am. J. Hum. Genet.* 59: 879-886, 1996.

- [27901] 1453.Bjorses, P.; Halonen, M.; Palvimo, J. J.; Kolmer, M.; Aaltonen,J.; Ellonen, P.; Perheentupa, J.; Ulmanen, I.; Peltonen, L.: Mutationsin the AIRE gene: effects on subcellular location and transactivationfunction of the autoimmune polyendocrinopathy-candidiasis-ectodermaldystrophy protein. *Am. J. Hum. Genet.* 66: 378-392, 2000.
- [27902] 1454.Bjorses, P.; Pelto-Huikko, M.; Kaukonen, J.; Aaltonen, J.; Peltonen,L.; Ulmanen, I.: Localization of the APECED protein in distinct nuclearstructures. *Hum. Molec. Genet.* 8: 259-266, 1999.
- [27903] 1455.Blizzard, R. M.; Kyle, M. A.: Studies of the adrenal antigensand antibodies in Addison's disease. *J. Clin. Invest.* 42: 1653-1660,1963.
- [27904] 1456.Castells, S.; Fikrig, S.; Inamdar, S.; Orti, E.: Familial moniliasis,defective delayed hypersensitivity, and adrenocorticotrophic hormonedeficiency. *J. Pediat.* 79: 72-79, 1971.
- [27905] 1457.Charache, S.: Methemoglobinemia--sleuthing for a new cause. (Editorial) *NewEng. J. Med.* 314: 776-778, 1986.
- [27906] 1458.Dayhoff, M. O.: Atlas of Protein Sequence and Structure. CytochromeB group. Washington: National Biomed-

cal Research Foundation (pub.)5: 1972. Pp. D29–D33.

- [27907] 1459.Giordano, S.; Steggles, A. W.: The human reticulo-
cyte and liver cytochrome b5 mRNA's are the products of
one gene. (Series)Miami Short Reports. Advances in Gene
Technology: The Molecular Biology of Human Genetic Dis-
ease. New York: IRL Press (pub.) 1: 1991.Pp. 8 only.
- [27908] 1460.Giordano, S. J.; Kaftory, A.; Steggles, A. W.: A splic-
ing mutation in the cytochrome b5 gene from a patient
with congenital methemoglobinemia and pseudo-
hermaphroditism. Hum. Genet. 93: 568–570, 1994.
- [27909] 1461.Giordano, S. J.; Yoo, M.; Ward, D. C.; Bhatt, M.; Over-
hauser, J.; Steggles, A. W.: The human cytochrome b-5
gene and two of its pseudogenes are located on chromo-
somes 18q23, 14q31–32.1 and 20p11.2, respectively.
Hum.Genet. 92: 615–618, 1993.
- [27910] 1462.Hackett, C. S.; Strittmatter, P.: Covalent cross-link-
ing of the active sites of vesicle-bound cytochrome b5 and
NADH-cytochrome b5 reductase. J. Biol. Chem. 259:
3275–3282, 1984.
- [27911] 1463.Hegesh, E.; Hegesh, J.; Kaftory, A.: Congenital
methemoglobinemia with a deficiency of cytochrome b5.
New Eng. J. Med. 314: 757–761, 1986.
- [27912] 1464.Hultquist, D. E.; Passon, P. G.: Catalysis of methe-

moglobin reduction by erythrocyte cytochrome b5 and cytochrome b5 reductase. *Nature* N.B. 229: 252–254, 1971.

[27913] 1465. Li, X. R.; Giordano, S. J.; Yoo, M.; Steggles, A. W.:

The isolation and characterization of the human cytochrome b5 gene. *Biochem. Biophys. Res. Commun.* 209: 894–900, 1995.

[27914] 1466. Lloyd, E.; Ferrer, J. C.; Funk, W. D.; Mauk, M. R.;

Mauk, A. G.: Recombinant human erythrocyte cytochrome b5. *Biochemistry* 33:11432–11437, 1994.

[27915] 1467. Shephard, E. A.; Povey, S.; Spurr, N. K.; Phillips, I. R.:

Chromosomal localization of a cytochrome b(5) gene to human chromosome 18 and a cytochrome b(5) pseudo-gene to the X chromosome. *Genomics* 11: 302–308, 1991.

[27916] 1468. Steggles, A. W.; Kaftory, A.; Giordano, S. J.: The analysis of type IV methemoglobinemia: identification of a patient lacking cytochrome b5. (Abstract) *Am. J. Hum. Genet.* 51 (suppl.): A177 only, 1992.

[27917] 1469. Yoo, M.; Steggles, A. W.: The complete nucleotide sequence of human liver cytochrome b(5) mRNA. *Biochem. Biophys. Res. Commun.* 156:576–580, 1988.

[27918] 1470. Tsai, H.-M.: Physiologic cleavage of von Willebrand factor by a plasma protease is dependent on its conformation and requires calcium ion. *Blood* 87: 4235–4244,

1996.

- [27919] 1471.Tsai, H.-M.; Lian, E. C.-Y.: Antibodies to von Willebrand factor-cleaving protease in acute thrombotic thrombocytopenic purpura. *New Eng. J.Med.* 339: 1585-1594, 1998.
- [27920] 1472.Nagase, T.; Ishikawa, K.; Nakajima, D.; Ohira, M.; Seki, N.; Miyajima, N.; Tanaka, A.; Kotani, H.; Nomura, N.; Ohara, O.: Prediction of the coding sequences of unidentified human genes. VII. The complete sequences of 100 new cDNA clones from brain which can code for large proteins in vitro. *DNA Res.* 4: 141-150, 1997.
- [27921] 1473.Bastians, H.; Krebber, H.; Vetrie, D.; Hoheisel, J.; Lichter, P.; Ponstingl, H.; Joos, S.: Localization of the novel serine/threonine protein phosphatase 6 gene (PPP6C) to human chromosome Xq22.3. *Genomics* 41:296-297, 1997.
- [27922] 1474.Bastians, H.; Ponstingl, H.: The novel human protein serine/threonine phosphatase 6 is a functional homologue of budding yeast Sit4p and fission yeast ppe1, which are involved in cell cycle regulation. *J.Cell Sci.* 109: 2865-2874, 1996.
- [27923] 1475.Prueitt, R. L.; Ross, J. L.; Zinn, A. R.: Physical mapping of nine Xq translocation breakpoints and identifica-

tion of XPNPEP2 as a premature ovarian failure candidate gene. *Cytogenet. Cell Genet.* 89:44–50, 2000.

- [27924] 1476. Sprinkle, T. J.; Stone, A. A.; Venema, R. C.; Denslow, N. D.; Caldwell, C.; Ryan, J. W.: Assignment of the membrane-bound human aminopeptidase P gene (XPNPEP2) to chromosome Xq25. *Genomics* 50: 114–116, 1998.
- [27925] 1477. Venema, R. C.; Ju, H.; Zou, R.; Venema, V. J.; Ryan, J. W.: Cloning and tissue distribution of human membrane-bound aminopeptidase P. *Biochim. Biophys. Acta* 1354: 45–48, 1997.
- [27926] 1478. Dear, N.; Matena, K.; Vingron, M.; Boehm, T.: A new subfamily of vertebrate calpains lacking a calmodulin-like domain: implications for calpain regulation and evolution. *Genomics* 45: 175–184, 1997.
- [27927] 1479. Fong, Y. W.; Zhou, Q.: Stimulatory effect of splicing factors on transcriptional elongation. *Nature* 414: 929–933, 2001.
- [27928] 1480. Bonnert, T. P.; McKernan, R. M.; Farrar, S.; le Bourdelles, B.; Heavens, R. P.; Smith, D. W.; Hewson, L.; Rigby, M. R.; Sirinathsinghji, D. J. S.; Brown, N.; Wafford, K. A.; Whiting, P. J.: Theta, a novel gamma-aminobutyric acid type A receptor subunit. *Proc. Nat. Acad. Sci.* 96: 9891–9896, 1999.

- [27929] 1481. Whiting, P. J.; Bonnert, T. P.; McKernan, R. M.; Farrar, S.; leBourdelle, B.; Heavens, R. P.; Smith, D. W.; Hewson, L.; Rigby, M. R.; Sirinathsinghji, D. J. S.; Thompson, S. A.; Wafford, K. A.: Molecular and functional diversity of the expanding GABA-A receptor gene family. *Ann. N.Y. Acad. Sci.* 645–653, 1999.
- [27930] 1482. Board, P. G.: Identification of cDNAs encoding two human alpha class glutathione transferases (GSTA3 and GSTA4) and the heterologous expression of GSTA4-4. *Biochem. J.* 330: 827–831, 1998.
- [27931] 1483. Desmots, F.; Rauch, C.; Henry, C.; Guillouzo, A.; Morel, F.: Genomic organization, 5-prime-flanking region and chromosomal localization of the human glutathione transferase A4 gene. *Biochem. J.* 336: 437–442, 1998.
- [27932] 1484. Hubatsch, I.; Ridderstrom, M.; Mannervik, B.: Human glutathione transferase A4-4: an alpha class enzyme with high catalytic efficiency in the conjugation of 4-hydroxynonenal and other genotoxic products of lipid peroxidation. *Biochem. J.* 330: 175–179, 1998.
- [27933] 1485. Zhang, F.; Zhang, W.; Liu, L.; Fisher, C. L.; Hui, D.; Childs, S.; Dorovini-Zis, K.; Ling, V.: Characterization of ABCB9, an ATP binding cassette protein associated with lysosomes. *J. Biol. Chem.* 275:23287–23294, 2000.

- [27934] 1486. Allikmets, R.; Gerrard, B.; Glavac, D.; Ravnik-Glavac, M.; Jenkins, N. A.; Gilbert, D. J.; Copeland, N. G.; Modi, W.; Dean, M.: Characterization and mapping of three new mammalian ATP-binding transporter genes from an EST database. *Mammalian Genome* 6: 114–117, 1995.
- [27935] 1487. Zhang, F.; Hogue, D. L.; Liu, L.; Fisher, C. L.; Hui, D.; Childs, S.; Ling, V.: M-ABC2, a new human mitochondrial ATP-binding cassette membrane protein. *FEBS Lett.* 478: 89–94, 2000.
- [27936] 1488. Lee, J.; Ho, W.-H.; Maruoka, M.; Corpuz, R. T.; Baldwin, D. T.; Foster, J. S.; Goddard, A. D.; Yansura, D. G.; Vandlen, R. L.; Wood, W. L.; Gurney, A. L.: IL-17E, a novel proinflammatory ligand for the IL-17 receptor homolog IL-17Rh1. *J. Biol. Chem.* 276: 1660–1664, 2001.
- [27937] 1489. Tian, E.; Sawyer, J. R.; Largaespada, D. A.; Jenkins, N. A.; Copeland, N. G.; Shaughnessy, J. D., Jr.: Evi27 encodes a novel membrane protein with homology to the IL17 receptor. *Oncogene* 19: 2098–2109, 2000.
- [27938] 1490. Lee, M.-H.; Lu, K.; Hazard, S.; Yu, H.; Shulenin, S.; Hidaka, H.; Kojima, H.; Allikmets, R.; Sakuma, N.; Pegoraro, R.; Srivastava, A. K.; Salen, G.; Dean, M.; Patel, S. B.: Identification of a gene, ABCG5, important in the regulation of dietary cholesterol absorption. *Nature Genet.* 27: 79–83,

2001.

- [27939] 1491.Lu, K.; Lee, M.-H.; Hazard, S.; Brooks-Wilson, A.; Hidaka, H.;Kojima, H.; Ose, L.; Stalenhoef, A. F. H.; Miettinen, T.; Bjorkhem,I.; Bruckert, E.; Pandya, A.; Brewer, H. B., Jr.; Salen, G.; Dean,M.; Srivastava, A.; Patel, S. B.: Two genes that map to the STSLlocus cause sitosterolemia: genomic structure and spectrum of mutation-involving sterolin-1 and sterolin-2, encoded by ABCG5 and ABCG8, respectively. *Am.J. Hum. Genet.* 69: 278-290, 2001.
- [27940] 1492.Ueki, N.; Oda, T.; Kondo, M.; Yano, K.; Noguchi, T.; Muramatsu,M.: Selection system for genes encoding nuclear-targeted proteins. *Nat.Biotech.* 16: 1338-1342, 1998.
- [27941] 1493.Ueki, N.; Seki, N.; Yano, K.; Masuho, Y.; Saito, T.; Muramatsu,M.: Isolation and characterization of a novel human gene (HFB30)which encodes a protein with a RING finger motif. *Biochim. Biophys.Acta* 232-236, 1999.
- [27942] 1494.Oikawa, E.; Iijima, H.; Suzuki, T.; Sasano, H.; Sato, H.; Kamataki,A.; Nagura, H.; Kang, M. J.; Fujino, T.; Suzuki, H.; Yamamoto, T.T.: A novel acyl-CoA synthetase, ACS5, expressed in intestinal epithelialcells and proliferating preadipocytes. *J. Biochem.* 124: 679-685,1998.

- [27943] 1495. Yamashita, Y.; Kumabe, T.; Cho, Y.-Y.; Watanabe, M.; Kawagishi, J.; Yoshimoto, T.; Fujino, T.; Kang, M.-J.; Yamamoto, T. T. Fatty acid induced glioma cell growth is mediated by the acyl-CoA synthetase 5 gene located on chromosome 10q25.1-q25.2, a region frequently deleted in malignant gliomas. *Oncogene* 19: 5919-5925, 2000.
- [27944] 1496. de Luis, O.; Valero, M. C.; Perez Jurado, L. A.: WB-SCR14, a putative transcription factor gene deleted in Williams-Beuren syndrome: complete characterisation of the human gene and the mouse ortholog. *Europ. J. Hum. Genet.* 8: 215-222, 2000.
- [27945] 1497. Meng, X.; Lu, X.; Li, Z.; Green, E. D.; Massa, H.; Trask, B. J.; Morris, C. A.; Keating, M. T.: Complete physical map of the common deletion region in Williams syndrome and identification and characterization of three novel genes. *Hum. Genet.* 103: 590-599, 1998.
- [27946] 1498. Nakajima, H.; Cella, M.; Langen, H.; Friedlein, A.; Colonna, M.: Activating interactions in human NK cell recognition: the role of 2B4-CD48. *Europ. J. Immun.* 29: 1676-1683, 1999.
- [27947] 1499. Watzl, C.; Stebbins, C. C.; Long, E. O.: Cutting edge: NK cell inhibitory receptors prevent tyrosine phosphorylation of the activation receptor 2B4 (CD244). *J. Immun.* 165:

3545–3548, 2000.

- [27948] 1500. Bloomfield, C. D.; Garson, O. M.; Volin, L.; Knuutila, S.; de laChapelle, A.: t(1;3)(p36;q21) in acute nonlymphocytic leukemia: a new cytogenetic–clinicopathologic association. *Blood* 66: 1409–1413, 1985.
- [27949] 1501. Moir, D. J.; Jones, P. A.; Pearson, J.; Duncan, J. R.; Cook, P.; Buckle, V. J.: A new translocation, t(1;3)(p36;q21), in myelodysplastic disorders. *Blood* 64: 553–555, 1984.
- [27950] 1502. Secker–Walker, L. M.; Mehta, A.; Bain, B.: Abnormalities of 3q21 and 3q26 in myeloid malignancy: a United Kingdom Cancer Cytogenetic Group study. *Brit. J. Haemat.* 91: 490–501, 1995.
- [27951] 1503. Welborn, J. L.; Lewis, J. P.; Jenks, H.; Walling, P.: Diagnostic and prognostic significance of t(1;3)(p36;q21) in the disorders of hematopoiesis. *Cancer Genet. Cytogenet.* 28: 277–285, 1987.
- [27952] 1504. Mitchelmore, C.; Troelsen, J. T.; Sjostrom, H.; Noren, O.: The HOXC11 homeodomain protein interacts with the lactose–phlorizin hydrolase promoter and stimulates HNF1–alpha–dependent transcription. *J. Biol. Chem.* 273: 13297–13306, 1998.
- [27953] 1505. Watson, M. A.; Darrow, C.; Zimonjic, D. B.; Popescu,

N.; Fleming, T. P.: Structure and transcriptional regulation of the human mammaglobin gene, a breast cancer associated member of the uteroglobin gene family localized to chromosome 11q13. *Oncogene* 16: 817–824, 1998.

[27954] 1506. Watson, M. A.; Fleming, T. P.: Mammaglobin, a mammary-specific member of the uteroglobin gene family, is overexpressed in human breast cancer. *Cancer Res.* 56: 860–865, 1996.

[27955] 1507. Watson, M. A.; Fleming, T. P.: Isolation of differentially expressed sequence tags from human breast cancer. *Cancer Res.* 54: 4598–4602, 1994.

[27956] 1508. Heidebrecht, H. J.; Buck, F.; Steinmann, J.; Sprenger, R.; Wacker, H. H.; Parwaresch, R.: p100: a novel proliferation-associated nuclear protein specifically restricted to cell cycle phases S, G2, and M. *Blood* 90: 226–233, 1997.

[27957] 1509. Larrick, J. W.; Lee, J.; Ma, S.; Li, X.; Francke, U.; Wright, S. C.; Balint, R. F.: Structural, functional analysis and localization of the human CAP18 gene. *FEBS Lett.* 398: 74–80, 1996.

[27958] 1510. Nagaoka, I.; Hirota, S.; Niyonsaba, F.; Hirata, M.; Adachi, Y.; Tamura, H.; Heumann, D.: Cathelicidin family of antibacterial peptides CAP18 and CAP11 inhibit the expression of TNF- α by blocking the binding of LPS to

CD14+ cells. *J. Immun.* 167: 3329–3338, 2001.

- [27959] 1511.Hu, X.; Ray, P. N.; Murphy, E. G.; Thompson, M. W.; Worton, R.G.: Duplicational mutation at the Duchenne muscular dystrophy locus:its frequency, distribution, origin, and phenotype–genotype correlation. *Am.J. Hum. Genet.* 46: 682–695, 1990.
- [27960] 1512.Hu, X.; Ray, P. N.; Worton, R. G.: Mechanisms of tandem duplication in the Duchenne muscular dystrophy gene include both homologous and nonhomologous intrachromosomal recombination. *EMBO J.* 10: 2471–2477, 1991.
- [27961] 1513.Hu, X.; Worton, R. G.: Partial gene duplication as a cause of human disease. *Hum. Mutat.* 1: 3–12, 1992.
- [27962] 1514.Ingram, V. M.: Gene evolution and the haemoglobins. *Nature* 189:704–708, 1961.
- [27963] 1515.Itagaki, Y.; Saida, K.; Iwamura, K.: Regenerative capacity of mdx mouse muscles after repeated applications of myo–necrotic bupivacaine. *Acta Neuropath.* 89: 380–384, 1995.
- [27964] 1516.Kaplan, J.–C.; Kahn, A.; Chelly, J.: Illegitimate transcription:its use in the study of inherited disease. *Hum. Mutat.* 1: 357–360, 1992.
- [27965] 1517.Kavaslar, G. N.; Telatar, M.; Serdaroglu, P.; Deymeer, F.; Ozdemir, C.; Tolun, A.: Identification of a one–basepair

deletion in exon6 of the dystrophin gene. Hum. Mutat. 6: 85–86, 1995.

- [27966] 1518. Kilimann, M. W.; Pizzuti, A.; Grompe, M.; Caskey, C. T.: Pointmutations and polymorphisms in the human dystrophin gene identified in genomic DNA sequences amplified by multiplex PCR. Hum. Genet. 89:253–258, 1992.
- [27967] 1519. Kim, T.-W.; Wu, K.; Black, I. B.: Deficiency of brain synaptic dystrophin in human Duchenne muscular dystrophy. Ann. Neurol. 38:446–449, 1995.
- [27968] 1520. Kneppers, A. L. J.; Deutz–Terlouw, P. P.; van Ommen, G. J. B.; Bakker, E.: Point mutation screening for Duchenne muscular dystrophy (DMD) by SSCP–analysis of multiplex PCR products by use of the PhastSystem(TM). Am. J. Hum. Genet. Suppl. 53: Abstract–1493, 1993.
- [27969] 1521. Ostendorff, H. P.; Bossenz, M.; Mincheva, A.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Lichter, P.; Bach, I.: Functional characterization of the gene encoding RLIM, the corepressor of LIM homeodomain factors. Genomics 69: 120–130, 2000.
- [27970] 1522. Ostendorff, H. P.; Peirano, R. I.; Peters, M. A.; Schluter, A.; Bossenz, M.; Scheffner, M.; Bach, I.: Ubiquitination–dependent cofactor exchange on LIM homeodomain transcription factors. Nature 416: 99–103, 2002.

[27971] 1523.Koenig, M.: Personal Communication. Boston, Mass. 10/8/1987.100. Koenig, M.; Beggs, A. H.; Moyer, M.; Scherpf, S.; Heindrich,K.; Bettecken, T.; Meng, G.; Muller, C. R.; Lindlof, M.; Kaariainen,H.; de la Chapelle, A.; Kiuru, A.; and 24 others: The molecularbasis for Duchenne ver- sus Becker muscular dystrophy: correlation ofseverity with type of deletion. Am. J. Hum. Genet. 45: 498–506,1989.101. Koenig, M.; Bertelson, C. J.; Monaco, A. P.; Hoffman, E.; Feener,C. C.; Kunkel, L. M.: Complete cloning of the Duchenne muscular dystrophycDNA and an analysis of the entire DMD locus. (Abstract) Am. J. Hum.Genet. 41: A222, 1987.102. Koenig, M.; Hoffman, E. P.; Bertelson, C. J.; Monaco, A. P.;Feener, C.; Kunkel, L. M.: Complete cloning of the Duchenne muscular dystrophy (DMD) cDNA and preliminary genomic organization of the DMDgene in normal and affected individuals. Cell 50: 509–517, 1987.103. Koenig, M.; Monaco, A. P.; Kunkel, L. M.: The complete sequenceof dystrophin predicts a rod- shaped cytoskeletal protein. Cell 53:219–228, 1988.104. Koh, J.; Bartlett, R. J.; Pericak–Vance, M. A.; Speer, M. C.;Yamaoka, L. H.; Phillips, K.; Hung, W.–Y.; Ray, P. N.; Worton, R.G.; Gilbert, J. R.; Lee, J. E.; Siddique, T.; Kandt, R. S.; Roses,A. D.: Inherited deletion at Duchenne dystro–

phy locus in normal male.(Letter) Lancet II: 1154–1155,
 1987.105. Kunkel, L. M.: Analysis of deletions in DNA
 from patients with Becker and Duchenne muscular dystro-
 phy. Nature 322: 73–77, 1986.106. Kunkel, L. M.;
 Monaco, A. P.; Middlesworth, W.; Ochs, H. D.; Latt, S. A.:
 Specific cloning of DNA fragments absent from the DNA of
 a male patient with an X chromosome deletion. Proc. Nat.
 Acad. Sci. 82: 4778–4782, 1985.107. Laing, N. G.; Layton,
 M. G.; Johnsen, R. D.; Chandler, D. C.; Mears, M. E.; Gold-
 blatt, J.; Kakulas, B. A.: Two distinct mutations in a single
 dystrophin gene: chance occurrence or premutation? Am. J.
 Med. Genet. 42: 688–692, 1992.108. Lederfein, D.; Levy,
 Z.; Augier, N.; Mornet, D.; Morris, G.; Fuchs, O.; Yaffe, D.;
 Nudel, U.: A 71-kilodalton protein is a major product of
 the Duchenne muscular dystrophy gene in brain and oth-
 er nonmuscle tissues. Proc. Nat. Acad. Sci. 89: 5346–5350,
 1992.109. Lederfein, D.; Yaffe, D.; Nudel, U.: A house-
 keeping type promoter, located in the 3-prime region of
 the Duchenne muscular dystrophy gene, controls the ex-
 pression of Dp71, a major product of the gene.
 Hum. Molec. Genet. 2: 1883–1888, 1993.110. Lee, C. C.;
 Pearlman, J. A.; Chamberlain, J. S.; Caskey, C. T.: Expres-
 sion of recombinant dystrophin and its localization to the-

cell membrane. *Nature* 349: 334–336, 1991.111. Lee, G.-H.; Badorff, C.; Knowlton, K. U.: Dissociation of sarco-glycans and the dystrophin carboxyl terminus from the sarcolemma in enteroviral cardiomyopathy. *Circ. Res.* 87: 489–495, 2000.112. Lenk, U.; Hanke, R.; Kraft, U.; Grade, K.; Grunewald, I.; Speer, A.: Non-isotopic analysis of single strand conformation polymorphism (SSCP) in the exon 13 region of the human dystrophin gene. *J. Med. Genet.* 30: 951–954, 1993.113. Lenk, U.; Hanke, R.; Speer, A.: Carrier detection in DMD families with point mutations, using PCR-SSCP and direct sequencing. *Neuromusc. Disord.* 4: 411–418, 1994.114. Lenk, U.; Hanke, R.; Thiele, H.; Speer, A.: Point mutations at the carboxy terminus of the human dystrophin gene: implications for an association with mental retardation in DMD patients. *Hum. Molec. Genet.* 2: 1877–1881, 1993.115. Lenk, U.; Oexle, K.; Voit, T.; Ancker, U.; Hellner, K.-A.; Speer, A.; Hubner, C.: A cysteine 3340 substitution in the dystroglycan-binding domain of dystrophin associated with Duchenne muscular dystrophy, mental retardation and absence of the ERG b-wave. *Hum. Molec. Genet.* 973–975, 1996.116. Liechti-Gallati, S.; Braga, S.; Hirsiger, H.; Moser, H.: Familial deletion in Becker type muscular dystrophy within the pXJ region.

Hum.Genet. 77: 267–268, 1987.117. Lindlof, M.; Kaari-
 ainen, H.; van Ommen, G. J. B.; de la Chapelle, A.: Mi-
 crodeletions in patients with X-linked muscular dystro-
 phy:molecular-clinical correlations. Clin. Genet. 33:
 131–139, 1988.118. Lindlof, M.; Kiuru, A.; Kaariainen, H.;
 Kalimo, H.; Lang, H.; Pihko, H.; Rapola, J.; Somer, H.;
 Somer, M.; Savontaus, M.-L.; de la Chapelle, A.: Gene dele-
 tions in X-linked muscular dystrophy. Am.J. Hum. Genet.
 44: 496–503, 1989.119. Mankin, A. S.; Liebman, S. W.:
 Baby, don't stop! Nature Genet. 23:8–10, 1999.120. Mao,
 Y.; Cremer, M.: Detection of Duchenne muscular dystro-
 phy carriers by dosage analysis using the DMD cDNA clone
 8. Hum. Genet. 81:193–195, 1989.121. Matsuo, M.; Ma-
 sumura, T.; Nakajima, T.; Kitoh, Y.; Takumi, T.; Nishio, H.;
 Koga, J.; Nakamura, H.: A very small frame-shifting dele-
 tion within exon 19 of the Duchenne muscular dystrophy
 gene. Biochem.Biophys. Res. Commun. 170: 963–967,
 1990.122. Matsuo, M.; Masumura, T.; Nishio, H.; Naka-
 jima, T.; Kitoh, Y.; Takumi, T.; Koga, J.; Nakamura, H.: Exon
 skipping during splicing of dystrophin mRNA precursor
 due to an intraexon deletion in the dystrophin gene of
 Duchenne muscular dystrophy. J. Clin. Invest. 87:
 2127–2131, 1991.123. McArdle, A.; Edwards, R. H. T.;

Jackson, M. J.: Time course of changes in plasma membrane permeability in the dystrophin-deficient mdx mouse. *Muscle Nerve* 17: 1378–1384, 1994.124. McCabe, E. R. B.; Towbin, J.; Chamberlain, J.; Baumbach, L.; Witkowski, J.; van Ommen, G. J. B.; Koenig, M.; Kunkel, L. M.; Seltzer, W. K.: Complementary DNA probes for the Duchenne muscular dystrophy locus demonstrate a previously undetectable deletion in a patient with dystrophic myopathy, glycerol kinase deficiency, and congenital adrenal hypoplasia. *J. Clin. Invest.* 83: 95–99, 1989.125.

Milasin, J.; Muntoni, F.; Severini, G. M.; Bartoloni, L.; Vatta, M.; Krajcinovic, M.; Mateddu, A.; Angelini, C.; Camerini, F.; Falaschi, A.; Mestroni, L.; Giacca, M.; Heart Muscle Disease Study Group: A point mutation in the 5-prime splice site of the dystrophin gene first intron responsible for X-linked dilated cardiomyopathy. *Hum. Molec. Genet.* 5: 73–79, 1996.126.

Minetti, C.; Bonilla, E.: Mosaic expression of dystrophin in carriers of Becker's muscular dystrophy and the X-linked syndrome of myalgia and cramps. (Letter) *New Eng. J. Med.* 327: 1100, 1992.127.

Moizard, M.-P.; Toutain, A.; Fournier, D.; Berret, F.; Raynaud, M.; Billard, C.; Andres, C.; Moraine, C.: Severe cognitive impairment in DMD: obvious clinical indi-

cation for Dp71 isoform point mutations screening. *Europ. J. Hum. Genet.* 8: 552–556, 2000.128. Monaco, A. P.; Bertelson, C. J.; Liechti-Gallati, S.; Moser, H.; Kunkel, L. M.: An explanation for phenotypic differences between patients bearing partial deletions of DMD locus. *Genomics* 2: 90–95, 1988.129. Monaco, A. P.; Kunkel, L. M.: A giant locus for the Duchenne and Becker muscular dystrophy gene. *Trends Genet.* 3: 33–37, 1987.130. Monaco, A. P.; Neve, R. L.; Colletti-Feener, C.; Bertelson, C. J.; Kurnit, D. M.; Kunkel, L. M.: Isolation of candidate cDNAs for portions of the Duchenne muscular dystrophy gene. *Nature* 323: 646–650, 1986.131. Muntoni, F.; Cau, M.; Ganau, A.; Congiu, R.; Arvedi, G.; Mateddu, A.; Marrosu, M. G.; Cianchetti, C.; Realdi, G.; Cao, A.; Melis, M. A.: Deletion of the dystrophin muscle-promoter region associated with X-linked dilated cardiomyopathy. *New Eng. J. Med.* 329: 921–925, 1993.132. Muntoni, F.; Melis, M. A.; Ganau, A.; Dubowitz, V.: Transcription of the dystrophin gene in normal tissues and in skeletal muscle of a family with X-linked dilated cardiomyopathy. *Am. J. Hum. Genet.* 56:151–157, 1995.133. Muntoni, F.; Wilson, L.; Marrosu, G.; Marrosu, M. G.; Cianchetti, C.; Mestroni, L.; Ganau, A.; Dubowitz, V.; Sewry, C.: A mutation in the dystrophin gene

selectively affecting dystrophin expression in the heart. J. Clin. Invest. 96: 693–699, 1995.134. Nevin, N. C.; Hughes, A. E.; Calwell, M.; Lim, J. H. K.: Duchenne muscular dystrophy in a female with a translocation involving Xp21. J. Med. Genet. 23: 171–187, 1986.135. Nigro, V.; Politano, L.; Nigro, G.; Romano, S. C.; Molinari, A. M.; Puca, G. A.: Detection of a nonsense mutation in the dystrophin gene by multiple SSCP. Hum. Molec. Genet. 1: 517–520, 1992.136. Nobile, C.; Marchi, J.; Nigro, V.; Roberts, R. G.; Danieli, G. A.: Exon–intron organization of the human dystrophin gene. Genomics 45:421–424, 1997.137. Nobile, C.; Toffolatti, L.; Rizzi, F.; Simionati, B.; Nigro, V.; Cardazzo, B.; Patarnello, T.; Valle, G.; Danieli, G. A.: Analysis of 22 deletion breakpoints in dystrophin intron 49. Hum. Genet. 110:418–421, 2002.138. Norman, A.; Harper, P.: A survey of manifesting carriers of Duchenne and Becker muscular dystrophy in Wales. Clin. Genet. 36:31–37, 1989.139. Ohno, S.: Evolution by Gene Duplication. Berlin: Springer–Verlag(pub.) 1970.140. Ortiz–Lopez, R.; Li, H.; Su, J.; Goytia, V.; Towbin, J. A.: Evidence for a dystrophin missense mutation as a cause of X–linked dilated cardiomyopathy. Circulation 95: 2434–2440, 1997.141. Palmucci, L.; Doriguzzi, C.; Mongini, T.;

Restagno, G.; Chiado-Piat, L.; Maniscalco, M.: Unusual expression and very mild course of Xp21 muscular dystrophy (Becker type) in a 60-year-old man with 26 percent deletion of the dystrophin gene. *Neurology* 44: 541–543, 1994.142. Passos-Bueno, M. R.; Bakker, E.; Kneppers, A. L. J.; Takata, R. I.; Rapaport, D.; den Dunnen, J. T.; Zatz, M.; van Ommen, G. J. B.: Different mosaicism frequencies for proximal and distal Duchenne muscular dystrophy (DMD) mutations indicate difference in etiology and recurrence risk. *Am. J. Hum. Genet.* 51: 1150–1155, 1992.143. Paulson, K. E.; Deka, N.; Schmid, C. W.; Misra, R.; Schindler, C. W.; Rush, M. G.; Kadyk, L.; Leinwand, L.: A transposon-like element in human DNA. *Nature* 316: 359–361, 1985.144. Pernelle, J.-J.; Chafey, P.; Chelly, J.; Wahrmann, J. P.; Kaplan, J.-C.; Tome, F.; Fardeau, M.: Nebulin seen in DMD males including one patient with a large DNA deletion encompassing the DMD gene. *Hum. Genet.* 78: 285, 1988.145. Pillers, D.-A. M.; Fitzgerald, K. M.; Duncan, N. M.; Rash, S. M.; White, R. A.; Dwinnell, S. J.; Powell, B. R.; Schnur, R. E.; Ray, P. N.; Cibis, G. W.; Weleber, R. G.: Duchenne/Becker muscular dystrophy: correlation of phenotype by electroretinography with sites of dystrophin mutations. *Hum. Genet.* 105: 2–9, 1999.146. Pizzuti, A.;

Pieretti, M.; Fenwick, R. G.; Gibbs, R. A.; Caskey, C. T.: A transposon-like element in the deletion-prone region of the dystrophin gene. *Genomics* 13: 594–600, 1992.147.

Porter, J. D.; Khanna, S.; Kaminski, H. J.; Rao, J. S.; Merriam, A. P.; Richmonds, C. R.; Leahy, P.; Li, J.; Guo, W.; Andrade, F. H.: A chronic inflammatory response dominates the skeletal muscle molecular signature in dystrophin-deficient mdx mice. *Hum. Molec. Genet.* 11:263–272, 2002.148.

Prior, T. W.; Papp, A. C.; Snyder, P. J.; Burghes, A. H. M.; Bartolo, C.; Sedra, M. S.; Western, L. M.; Mendell, J. R.: A missense mutation in the dystrophin gene in a Duchenne muscular dystrophy patient. *Nature Genet.* 4: 357–360, 1993.149.

Prior, T. W.; Papp, A. C.; Snyder, P. J.; Burghes, A. H. M.; Sedra, M. S.; Western, L. M.; Bartello, C.; Mendell, J. R.: Identification of two point mutations and a one base deletion in exon 19 of the dystrophin gene by heteroduplex formation. *Hum. Molec. Genet.* 2: 311–313, 1993.150.

Prior, T. W.; Papp, A. C.; Snyder, P. J.; Burghes, A. H. M.; Sedra, M. S.; Western, L. M.; Bartolo, C.; Mendell, J. R.: Exon 44 nonsense mutation in two Duchenne muscular dystrophy brothers detected by heteroduplex analysis. *Hum. Mutat.* 2: 192–195, 1993.151.

Prior, T. W.; Papp, A. C.; Snyder, P. J.; Sedra, M. S.; Western, L. M.; Bar-

tolo, C.; Moxley, R. T.; Mendell, J. R.: Heteroduplex analysis of the dystrophin gene: application to point mutation and carrier detection. *Am. J. Med. Genet.* 50: 68–73, 1994.152. Rafael, J. A.; Sunada, Y.; Cole, N. M.; Campbell, K. P.; Faulkner, J. A.; Chamberlain, J. S.: Prevention of dystrophic pathology in mdx mice by a truncated dystrophin isoform. *Hum. Molec. Genet.* 3:1725–1733, 1994.153. Rafael, J. A.; Townsend, E. R.; Squire, S. E.; Potter, A. C.; Chamberlain, J. S.; Davies, K. E.: Dystrophin and utrophin influence fiber type composition and post-synaptic membrane structure. *Hum. Molec. Genet.* 9: 1357–1367, 2000.154. Ray, P. N.; Belfall, B.; Duff, C.; Logan, C.; Kean, V.; Thompson, M. W.; Sylvester, J. E.; Gorski, J. L.; Schmickel, R. D.; Worton, R. G.: Cloning of the breakpoint of an X;21 translocation associated with Duchenne muscular dystrophy. *Nature* 318: 672–675, 1985.155. Read, A. P.; Mountford, R. C.; Forrest, S. M.; Kenwright, S. J.; Davies, K. E.; Harris, R.: Patterns of exon deletions in Duchenne and Becker muscular dystrophy. *Hum. Genet.* 80: 152–156, 1988.156. Rininsland, F.; Hahn, A.; Niemann-Seyde, S.; Slomski, R.; Hanefeld, F.; Reiss, J.: Identification of a new DMD gene deletion by ectopic transcript analysis. *J. Med. Genet.* 29: 647–651, 1992.157. Roberts,

R. G.; Bentley, D. R.; Bobrow, M.: Infidelity in the structure of ectopic transcripts: a novel exon in lymphocyte dystrophin transcripts. *Hum. Mutat.* 2: 293–299, 1993.158.

Roberts, R. G.; Bobrow, M.; Bentley, D. R.: The spectrum of mild X-linked recessive muscular dystrophy. *Arch. Neurol.* 34: 408–416, 1992.159.

Roberts, R. G.; Bobrow, M.; Bentley, D. R.: Point mutations in the dystrophin gene. *Proc. Nat. Acad. Sci.* 89: 2331–2335, 1992.160.

Roberts, R. G.; Gardner, R. J.; Bobrow, M.: Searching for the 1 in 2,400,000: a review of dystrophin gene point mutations. *Hum. Mutat.* 4: 1–11, 1994.161.

Roberts, R. G.; Passos-Bueno, M. R.; Bobrow, M.; Vainzof, M.; Zatz, M.: Point mutation in a Becker muscular dystrophy patient. *Hum. Molec. Genet.* 2: 75–77, 1992.162.

Rowland, L. P.: Biochemistry of muscle membranes in Duchenne muscular dystrophy. *Muscle Nerve* 3: 3–20, 1980.163.

Ryder-Cook, A. S.; Sicinski, P.; Thomas, K.; Davies, K. E.; Worton, R. G.; Barnard, E. A.; Darlison, M. G.; Barnard, P. J.: Localization of the mdx mutation within the mouse dystrophin gene. *EMBO J.* 7:3017–3021, 1988.164.

Saad, F. A.; Vita, G.; Mora, M.; Morandi, L.; Vitiello, L.; Oliviero, S.; Danieli, G. A.: A novel nonsense mutation in the human dystrophin gene. *Hum. Mutat.* 2: 314–316, 1993.165.

Saad, F. A.;

Vita, G.; Toffolatti, L.; Danieli, G. A.: A possible missense mutation detected in the dystrophin gene by double strand conformation analysis (DSCA). *Neuromusc. Disord.* 4: 335–341, 1994.166. Sakamoto, M.; Yuasa, K.; Yoshimura, M.; Yokota, T.; Ikemoto, T.; Suzuki, M.; Dickson, G.; Miyagoe-Suzuki, Y.; Takeda, S.: Micro-dystrophin cDNA ameliorates dystrophic phenotypes when introduced into mdx mice as a transgene. *Biochem. Biophys. Res. Commun.* 293: 1265–1272, 2002.167. Sarig, R.; Mezger-Lallemand, V.; Gitelman, I.; Davis, C.; Fuchs, O.; Yaffe, D.; Nudel, U.: Targeted inactivation of Dp71, the major non-muscle product of the DMD gene: differential activity of the Dp71 promoter during development. *Hum. Molec. Genet.* 8: 1–10, 1999.168. Sarkar, G.; Sommer, S. S.: Access to a messenger RNA sequence or its protein product is not limited by tissue or species specificity. *Science* 244:331–334, 1989.169. Schwartz, L. S.; Tarleton, J.; Popovich, B.; Seltzer, W. K.; Hoffman, E. P.: Fluorescent multiplex linkage analysis and carrier detection for Duchenne/Becker muscular dystrophy. *Am. J. Hum. Genet.* 51:721–729, 1992.170. Scott, M. O.; Sylvester, J. E.; Heiman-Patterson, T.; Shi, Y.-J.; Fieles, W.; Stedman, H.; Burghes, A.; Ray, P.; Worton, R.; Fischbeck, K.

H.: Duchenne muscular dystrophy gene expression in normal and diseased human muscle. *Science* 239: 1418–1420, 1988.171. Sharp, N. J. H.; Kornegay, J. N.; Van Camp, S. D.; Herbstreith, M. H.; Secore, S. L.; Kettle, S.; Hung, W.-Y.; Constantinou, C. D.; Dykstra, M. J.; Roses, A. D.; Bartlett, R. J.: An error in dystrophin mRNA processing in golden retriever muscular dystrophy, an animal homologue of Duchenne muscular dystrophy. *Genomics* 13: 115–121, 1992.172. Shiga, N.; Takeshima, Y.; Sakamoto, H.; Inoue, K.; Yokota, Y.; Yokoyama, M.; Matsuo, M.: Disruption of the splicing enhancer sequence within exon 27 of the dystrophin gene by a nonsense mutation induces partial skipping of the exon and is responsible for Becker muscular dystrophy. *J. Clin. Invest.* 100: 2204–2210, 1997.173. Sicinski, P.; Geng, Y.; Ryder-Cook, A. S.; Barnard, E. A.; Darlison, M. G.; Barnard, P. J.: The molecular basis of muscular dystrophy in the mdx mouse: a point mutation. *Science* 244: 1578–1580, 1989.174. Smithies, O.; Connell, G. E.; Dixon, G. H.: Chromosomal rearrangements and the evolution of haptoglobin genes. *Nature* 196: 232–236, 1962.175. Southern, E. M.: Detection of specific sequences among DNA fragments separated by gel electrophoresis. *J. Molec. Biol.* 98: 503–517, 1975.176.

Stratford-Perricaudet, L. D.; Makeh, I.; Perricaudet, M.; Briand, P.: Widespread long-term gene transfer to mouse skeletal muscles and heart. *J. Clin. Invest.* 90: 626–630, 1992.177. Takeshima, Y.; Nishio, H.; Narita, N.; Wada, H.; Ishikawa, Y.; Ishikawa, Y.; Minami, R.; Nakamura, H.; Matsuo, M.: Amino-terminal deletion of 53% of dystrophin results in an intermediate Duchenne-Becker muscular dystrophy phenotype. *Neurology* 44: 1648–1651, 1994.178. Tennyson, C. N.; Klamut, H. J.; Worton, R. G.: The human dystrophin gene requires 16 hours to be transcribed and is cotranscriptionally spliced. *Nature Genet.* 9: 184–190, 1995.179. Tinsley, J. M.; Blake, D. J.; Davies, K. E.: Apo-dystrophin-3: a 2.2kb transcript from the DMD locus encoding the dystrophin glycoprotein binding site. *Hum. Molec. Genet.* 2: 521–524, 1993.180. Tinsley, J. M.; Potter, A. C.; Phelps, S. R.; Fisher, R.; Trickett, J. I.; Davies, K. E.: Amelioration of the dystrophic phenotype of mdx mice using a truncated utrophin transgene. *Nature* 384: 349–353, 1996.181. Todorova, A.; Danieli, G. A.: Large majority of single-nucleotide mutations along the dystrophin gene can be explained by more than one mechanism of mutagenesis. *Hum. Mutat.* 9: 537–547, 1997.182. Torelli, S.; Muntoni, F.: Alternative splicing of dys-

trophinexon 4 in normal human muscle. Hum. Genet. 97: 521–523, 1996.183. Towbin, J. A.; Hejtmancik, J. F.; Brink, P.; Gelb, B.; Zhu, X.M.; Chamberlain, J. S.; McCabe, E. R. B.; Swift, M.: X-linked dilated cardiomyopathy: molecular genetic evidence of linkage to the Duchenne muscular dystrophy (dystrophin) gene at the Xp21 locus. Circulation 87:1854–1865, 1993.184. Towbin, J. A.; Ortiz-Lopez, R.: X-linked dilated cardiomyopathy.(Letter) New Eng. J. Med. 330: 369–370, 1994.185. Towbin, J. A.; Zhu, X. M.; Gelb, B.; Bies, R.; Chamberlain, J.; Maichele, A.; Ohlendieck, K.; Campbell, K.; McCabe, E. R. B.; Swift, M.: X-linked dilated cardiomyopathy (XLCM): molecular characterization.(Abstract) Am. J. Hum. Genet. 49 (suppl.): 421, 1991.186. Tuffery, S.; Lenk, U.; Roberts, R. G.; Coubes, C.; Demaille, J.; Claustres, M.: Protein truncation test: analysis of two novel point mutations at the carboxy-terminus of the human dystrophin gene associated with mental retardation. Hum. Mutat. 6: 126–135, 1995.187. Valentine, B. A.; Winand, N. J.; Pradhan, D.; Moise, N. S.; deLahunta, A.; Kornegay, J. N.; Cooper, B. J.: Canine X-linked muscular dystrophy as an animal model of Duchenne muscular dystrophy: a review. Am. J. Med. Genet. 42: 352–356, 1992.188. Verellen-Dumoulin, C.; Freund, M.; De Meyer,

R.; Laterre, C.; Frederic, J.; Thompson, M. W.; Markovic, V. D.; Worton, R. G.: Expression of an X-linked muscular dystrophy in a female due to translocation involving Xp21 and non-random inactivation of the normal X chromosome. Hum. Genet. 67: 115–119, 1984.189. Wehling, M.; Spencer, M. J.; Tidball, J. G.: A nitric oxide synthase transgene ameliorates muscular dystrophy in mdx mice. J. Cell Biol. 155: 123–131, 2001.190. Werner, W.; Spiegler, A. W. J.: Inherited deletion of subband Xp21.13 in a male with Duchenne muscular dystrophy. J. Med. Genet. 25:377–382, 1988.191. Wilton, S. D.; Chandler, D. C.; Kakulas, B. A.; Laing, N. G.: Identification of a point mutation and germinal mosaicism in a Duchenne muscular dystrophy family. Hum. Mutat. 3: 133–140, 1994.192. Wilton, S. D.; Johnsen, R. D.; Pedretti, J. R.; Laing, N. G.: Two distinct mutations in a single dystrophin gene: identification of an altered splice-site as the primary Becker muscular dystrophy mutation. Am. J. Med. Genet. 46: 563–569, 1993.193. Winnard, A. V.; Jia-Hsu, Y.; Gibbs, R. A.; Mendell, J. R.; Burghes, A. H. M.: Identification of a 2 base pair nonsense mutation causing a cryptic splice site in a DMD patient. Hum. Molec. Genet. 1: 645–646, 1992.194. Wood, D. S.; Zeviani, M.; PELLE, A.; Bonilla, E.; Salviati, G.;

Miranda, A. F.; DiMauro, S.; Rowland, L. P.: Is nebulin the defective gene product in Duchenne muscular dystrophy? (Letter) *New Eng. J. Med.* 316: 107–108, 1987.195. Worton, R. G.: Dystrophin: the long and short of it. (Editorial) *J. Clin. Invest.* 93: 4, 1994.196. Worton, R. G.: Personal Communication. Toronto, Ontario, Canada 9/12/1987.197. Xiong, D.; Lee, G.-H.; Badorff, C.; Dorner, A.; Lee, S.; Wolf, P.; Knowlton, K. U.: Dystrophin deficiency markedly increases enterovirus-induced cardiomyopathy: a genetic predisposition to viral heart disease. *Nature Med.* 8: 872–877, 2002.198. Yang, T. P.; Patel, P. I.; Chinault, A. C.; Stout, J. T.; Jackson, L. G.; Hildebrand, B. M.; Caskey, C. T.: Molecular evidence for new mutation at the HPRT locus in Lesch–Nyhan patients. *Nature* 310:412–414, 1984.199. Yoshida, K.; Ikeda, S.; Nakamura, A.; Kagoshima, M.; Takeda, S.; Shoji, S.; Yanagisawa, N.: Molecular analysis of the Duchenne muscular dystrophy gene in patients with Becker muscular dystrophy presenting with dilated cardiomyopathy. *Muscle Nerve* 16: 1161–1166, 1993.200. Yoshida, K.; Nakamura, A.; Yazaki, M.; Ikeda, S.; Takeda, S.: Insertional mutation by transposable element, L1, in the DMD gene results in X-linked dilated cardiomyopathy. *Hum. Molec. Genet.* 7:1129–1132, 1998.201. Zubrzycka–

Gaarn, E. E.; Bulman, D. E.; Karpati, G.; Burghes, A. H. M.; Belfall, B.; Klamut, H. J.; Talbot, J.; Hodges, R. S.; Ray, P. N.; Worton, R. G.: The Duchenne muscular dystrophy gene product is localized in sarcolemma of human skeletal muscle. *Nature* 333:466–469, 1988.

[27972] 1524. Tanner, S.; Stagljar, I.; Georgiev, O.; Schaffner, W.; Bourquin, J.-P.: A novel SR-related protein specifically interacts with the carboxy-terminal domain (CTD) of RNA polymerase II through a conserved interaction domain. *Biol. Chem.* 378: 565–571, 1997.

[27973] 1525. Zhang, W.-J.; Wu, J. Y.: Sip1, a novel RS domain-containing protein essential for pre-mRNA splicing. *Molec. Cell. Biol.* 18: 676–684, 1998.

[27974] 1526. Salbaum, J. M.: Genomic structure and chromosomal localization of the mouse gene Punc. *Mammalian Genome* 10: 107–111, 1999.

[27975] 1527. Yang, W.; Li, C.; Mansour, S. L.: Impaired motor coordination in mice that lack punc. *Molec. Cell. Biol.* 21: 6031–6043, 2001.

[27976] 1528. Tsukaguchi, H.; Tokui, T.; Mackenzie, B.; Berger, U. V.; Chen, X.-Z.; Wang, Y.; Brubaker, R. F.; Hediger, M. A.: A family of mammalian Na(+)-dependent L-ascorbic acid transporters. *Nature* 399: 70–75, 1999.

- [27977] 1529.Fukuta, M.; Inazawa, J.; Torii, T.; Tsuzuki, K.; Shimada, E.; Habuchi, O.: Molecular cloning and characterization of human keratan sulfatase-6-sulfotransferase. *J. Biol. Chem.* 272: 32321–32328, 1997.
- [27978] 1530.Iida, A.; Saito, S.; Sekine, A.; Mishima, C.; Kitamura, Y.; Kondo, K.; Harigae, S.; Osawa, S.; Nakamura, Y.: Catalog of 77 single-nucleotide polymorphisms (SNPs) in the carbohydrate sulfotransferase 1 (CHST1) and carbohydrate sulfotransferase 3 (CHST3) genes. *J. Hum. Genet.* 47:14–19, 2002.
- [27979] 1531.Mazany, K. D.; Peng, T.; Watson, C. E.; Tabas, I.; Williams, K.J.: Human chondroitin 6-sulfotransferase: cloning, gene structure, and chromosomal localization. *Biochim. Biophys. Acta* 1407: 92–97, 1998.
- [27980] 1532.Ayres, J. A.; Shum, L.; Akarsu, A. N.; Dashner, R.; Takahashi, K.; Ikura, T.; Slavkin, H. C.; Nuckolls, G. H.: DACH: genomic characterization, evaluation as a candidate for postaxial polydactyly type A2, and developmental expression pattern of the mouse homologue. *Genomics* 77: 18–26, 2001.
- [27981] 1533.Davis, R. J.; Shen, W.; Sandler, Y. I.; Amoui, M.; Purcell, P.; Maas, R.; Ou, C.-N.; Vogel, H.; Beaudet, A. L.; Mardon, G.: Dach1 mutant mice bear no gross abnormalities in

eye, limb, and brain development and exhibit postnatal lethality. *Molec. Cell. Biol.* 21: 1484–1490, 2001.

[27982] 1534. Hammond, K. L.; Hanson, I. M.; Brown, A. G.; Lettice, L. A.; Hill, R. E.: Mammalian and *Drosophila* dachshund genes are related to the *Ski* proto-oncogene and are expressed in eye and limb. *Mech. Dev.* 74:121–131, 1998.

[27983] 1535. Hammond, K. L.; Lettice, L. A.; Hill, R. E.; Lee, M.; Boyle, S.; Hanson, I. M.: Human (DACH) and mouse (Dach) homologues of *Drosophila* dachshund map to chromosomes 13q22 and 14E3, respectively. *Genomics* 55:252–253, 1999.

[27984] 1536. Li, X.; Perissi, V.; Liu, F.; Rose, D. W.; Rosenfeld, M. G.: Tissue-specific regulation of retinal and pituitary precursor cell proliferation. *Science* 297:1180–1183, 2002.

[27985] 1537. Kuramochi, S.; Matsuda, Y.; Okamoto, M.; Kitamura, F.; Yonekawa, H.; Karasuyama, H.: Molecular cloning of the human gene *STK10* encoding lymphocyte-oriented kinase, and comparative chromosomal mapping of the human, mouse, and rat homologues. *Immunogenetics* 49: 369–375, 1999.

[27986] 1538. Kuramochi, S.; Moriguchi, T.; Kuida, K.; Endo, J.; Semba, K.; Nishida, E.; Karasuyama, H.: *LOK* is a novel mouse *STE20*-like protein kinase that is expressed pre-

dominantly in lymphocytes. J. Biol. Chem.

272:22679–22684, 1997.

[27987] 1539. Berg, L.-P.; Shamsheer, M. K.; El-Daher, S. S.; Kakkar, V. V.; Authi, K. S.: Expression of human TRPC genes in the megakaryocytic cell lines MEG01, DAMI and HEL. FEBS Lett. 403: 83–86, 1997.

[27988] 1540. Wes, P. D.; Chevesich, J.; Jeromin, A.; Rosenberg, C.; Stetten, G.; Montell, C.: TRPC1, a human homolog of a *Drosophila* store-operated channel. Proc. Nat. Acad. Sci. 92: 9652–9656, 1995.

[27989] 1541. Xu, X.-Z. S.; Li, H.-S.; Guggino, W. B.; Montell, C.: Coassembly of TRP and TRPL produces a distinct store-operated conductance. Cell 89: 1155–1164, 1997.

[27990] 1542. Zhu, X.; Chu, P. B.; Peyton, M.; Birnbaumer, L.: Molecular cloning of a widely expressed human homologue for the *Drosophila* trp gene. FEBS Lett. 373: 193–198, 1995.

[27991] 1543. Zhu, X.; Jiang, M.; Peyton, M.; Boulay, G.; Hurst, R.; Stefani, E.; Birnbaumer, L.: trp, a novel mammalian gene family essential for agonist-activated capacitative Ca^{2+} entry. Cell 85: 661–671, 1996.

[27992] 1544. Zitt, C.; Zobel, A.; Obukhov, A. G.; Harteneck, C.; Kalkbrenner, F.; Luckhoff, A.; Schultz, G.: Cloning and

functional expression of a human Ca^{2+} -permeable cation channel activated by calcium store depletion. *Neuron* 16: 1189–1196, 1996.

- [27993] 1545. Baudier, J.; Deloulme, J. C.; Van Dorsselaer, A.; Black, D.; Matthes, H. W. D.: Purification and characterization of a brain-specific protein kinase C substrate, neurogranin (p17): identification of a consensus amino acid sequence between neurogranin and neuromodulin (GAP43) that corresponds to the protein kinase C phosphorylation site and the calmodulin-binding domain. *J. Biol. Chem.* 266: 229–237, 1991.
- [27994] 1546. Martinez de Arrieta, C.; Morte, B.; Coloma, A.; Bernal, J.: The human RC3 gene homolog, NRGN contains a thyroid hormone-responsive element located in the first intron. *Endocrinology* 140: 335–343, 1999.
- [27995] 1547. Martinez de Arrieta, C.; Perez Jurado, L.; Bernal, J.; Coloma, A.: Structure, organization, and chromosomal mapping of the human neurogranin gene (NRGN). *Genomics* 41: 243–249, 1997.
- [27996] 1548. Gantz, I.; Konda, Y.; Yang, Y.-K.; Miller, D. E.; Dierick, H. A.; Yamada, T.: Molecular cloning of a novel receptor (CMKLR1) with homology to the chemotactic factor receptors. *Cytogenet. Cell Genet.* 74: 286–290, 1996.

- [27997] 1549.Methner, A.; Hermey, G.; Schinke, B.; Hermans-Borgmeyer, I.: A novel G protein-coupled receptor with homology to neuropeptide and chemoattractant receptors expressed during bone development. *Biochem. Biophys. Res. Commun.* 233: 336–342, 1997.
- [27998] 1550.Owman, C.; Lolait, S. J.; Santen, S.; Olde, B.: Molecular cloning and tissue distribution of cDNA encoding a novel chemoattractant-like receptor. *Biochem. Biophys. Res. Commun.* 241: 390–394, 1997.
- [27999] 1551.Blanco, G.; Irving, N. G.; Brown, S. D. M.; Miller, C. C. J.; McLoughlin, D. M.: Mapping of the human and murine X11-like genes (APBA2 and Apba2), the murine Fe65 gene (Apbb1), and the human Fe65-like gene (APBB2): genes encoding phosphotyrosine-binding domain proteins that interact with the Alzheimer's disease amyloid precursor protein. *Mammalian Genome* 9: 473–475, 1998.
- [28000] 1552.Butz, S.; Okamoto, M.; Sudhof, T. C.: A tripartite protein complex with the potential to couple synaptic vesicle exocytosis to cell adhesion in brain. *Cell* 94: 773–782, 1998.
- [28001] 1553.Chen, W.-J.; Goldstein, J. L.; Brown, M. S.: NPXY, a sequence often found in cytoplasmic tails, is required for coated pit-mediated internalization of the low density

lipoprotein receptor. J. Biol.Chem. 265: 3116–3123, 1990.

- [28002] 1554.Duclos, F.; Boschert, U.; Sirugo, G.; Mandel, J.-L.; Hen, R.; Koenig,M.: Gene in the region of the Friedreich ataxia locus encodes a putativetransmembrane protein expressed in the nervous system. Proc. Nat.Acad. Sci. 90: 109–113, 1993.
- [28003] 1555.Duclos, F.; Koenig, M.: Comparison of primary structure of a neuron–specificprotein, X11, between human and mouse. Mammalian Genome 6: 57–58,1995.
- [28004] 1556.Okamoto, M.; Sudhof, T. C.: Mints, Munc18–interacting proteinsin synaptic vesicle exocytosis. J. Biol. Chem. 272: 31459–31464,1997.
- [28005] 1557.van der Geer, P.; Pawson, T.: The PTB domain: a new protein moduleimplicated in signal transduction. Trends Biochem. Sci. 20: 277–280,1995.
- [28006] 1558.Loh, N. Y.; Ambrose, H. J.; Guay–Woodford, L. M.; DasGupta, S.;Nawrotzki, R. A.; Blake, D. J.; Davies, K. E.: Genomic organizationand refined mapping of the mouse beta–dystrobrevin gene. MammalianGenome 9: 857–862, 1998.
- [28007] 1559.Rodius, F.; Duclos, F.; Wrogemann, K.; Le Paslier, D.; Ougen, P.;Billault, A.; Belal, S.; Musenger, C.; Brice, A.; Durr, A.; Mignard,C.; Sirugo, G.; Weissenbach, J.; Cohen,

D.; Hentati, F.; Ben Hamida, M.; Mandel, J.-L.; Koenig, M.: Recombinations in individuals homozygous by descent localize the Friedreich ataxia locus in a cloned 450-kb interval. *Am. J. Hum. Genet.* 54: 1050–1059, 1994.

[28008] 1560. Peters, M. F.; O'Brien, K. F.; Sadoulet-Puccio, H. M.; Kunkel, L. M.; Adams, M. E.; Froehner, S. C.: Beta-dystrobrevin, a new member of the dystrophin family: identification, cloning, and protein associations. *J. Biol. Chem.* 272: 50:–31561–31569, 1997.

[28009] 1561. Morris, M. E.; Viswanathan, N.; Kuhlman, S.; Davis, F. C.; Weitz, C. J.: A screen for genes induced in the suprachiasmatic nucleus by light. *Science* 279: 1544–1547, 1998.

[28010] 1562. Patwardhan, S.; Gashler, A.; Siegel, M. G.; Chang, L. C.; Joseph, L. J.; Shows, T. B.; Le Beau, M. M.; Sukhatme, V. P.: EGR3, a novel member of the Egr family of genes encoding immediate-early transcription factors. *Oncogene* 6: 917–928, 1991.

[28011] 1563. Tourtellotte, W. G.; Milbrandt, J.: Sensory ataxia and muscle spindle agenesis in mice lacking the transcription factor Egr3. *Nature Genet.* 20: 87–91, 1998.

[28012] 1564. Cavailles, V.; Dauvois, S.; Horset, L. F.; Lopez, G.; Hoare, S.; Kushner, P. J.; Parker, M. G.: Nuclear factor

RIP140 modulates transcriptional activation by the estrogen receptor. *EMBO J.* 14: 3741–3751, 1995.

- [28013] 1565. Katsanis, N.; Ives, J. H.; Groet, J.; Nizetic, D.; Fisher, E. M. C.: Localisation of receptor interacting protein 140 (RIP140) within 100 kb of D21S13 on 21q11, a gene-poor region of the human genome. *Hum. Genet.* 102: 221–223, 1998.
- [28014] 1566. Bodnar, J. S.; Chatterjee, A.; Castellani, L. W.; Ross, D. A.; Ohmen, J.; Cavalcoli, J.; Wu, C.; Dains, K. M.; Catanese, J.; Chu, M.; Sheth, S. S.; Charugundla, K.; Demant, P.; West, D. B.; de Jong, P.; Lusis, A. J.: Positional cloning of the combined hyperlipidemia gene *Hyplip1*. *Nature Genet.* 30: 110–116, 2002.
- [28015] 1567. Basile, A.; Sica, A.; d'Aniello, E.; Breviario, F.; Garrido, G.; Castellano, M.; Mantovani, A.; Introna, M.: Characterization of the promoter for the human long pentraxin *PTX3*: role of NF- κ B in tumor necrosis factor- α and interleukin-1- β regulation. *J. Biol. Chem.* 272: 8172–8178, 1997.
- [28016] 1568. Brown, J. L.; Stowers, L.; Baer, M.; Trejo, J.; Coughlin, S.; Chant, J.: Human Ste20 homologue hPAK1 links GTPases to the JNK MAP kinase pathway. *Curr. Biol.* 6: 598–605, 1996.

- [28017] 1569.Knaus, U. G.; Morris, S.; Dong, H.-J.; Chernoff, J.; Bokoch, G.M.: Regulation of human leukocyte p21-activated kinases through Gprotein-coupled receptors. *Science* 269: 221-223, 1995.
- [28018] 1570.Lei, M.; Lu, W.; Meng, W.; Parrini, M.-C.; Eck, M. J.; Mayer, B.J.; Harrison, S. C.: Structure of PAK1 in an autoinhibited conformation reveals a multistage activation switch. *Cell* 102: 387-397, 2000.
- [28019] 1571.Parrini, M. C.; Lei, M.; Harrison, S. C.; Mayer, B. J.: Pak1 kinase homodimers are autoinhibited in trans and dissociated upon activation by Cdc42 and Rac1. *Molec. Cell* 9: 73-83, 2002.
- [28020] 1572.Sanders, L. C.; Matsumura, F.; Bokoch, G. M.; de Lanerolle, P.: Inhibition of myosin light chain kinase by p21-activated kinase. *Science* 283:2083-2085, 1999.
- [28021] 1573.Arany, Z.; Sellers, W. R.; Livingston, D. M.; Eckner, R.: E1A-associated p300 and CREB-associated CBP belong to a conserved family of coactivators.(Letter) *Cell* 77: 799-800, 1994.
- [28022] 1574.Gayther, S. A.; Batley, S. J.; Linger, L.; Bannister, A.; Thorpe, K.; Chin, S.-F.; Daigo, Y.; Russell, P.; Wilson, A.; Sowter, H. M.; Delhanty, J. D. A.; Ponder, B. A. J.; Kouzarides, T.; Caldas, C.: Mutations truncating the EP300

acetylase in human cancers. *Nature Genet.* 24: 300–303, 2000.

[28023] 1575. Ida, K.; Kitabayashi, I.; Taki, T.; Taniwaki, M.; Noro, K.; Yamamoto, M.; Ohki, M.; Hayashi, Y.: Adenoviral E1A-associated protein p300 is involved in acute myeloid leukemia with t(11;22)(q23;q13). *Blood* 90:4699–4704, 1997.

[28024] 1576. Muraoka, M.; Konishi, M.; Kikuchi-Yanoshita, R.; Tanaka, K.; Shitara, N.; Chong, J.-M.; Iwama, T.; Miyaki, M.: p300 gene alterations in colorectal and gastric carcinomas. *Oncogene* 12: 1565–1569, 1996.

[28025] 1577. Yao, T. P.; Oh, S. P.; Fuchs, M.; Zhou, N.-D.; Ch'ng, L.-E.; Newsome, D.; Bronson, R. T.; Li, E.; Livingston, D. M.; Eckner, R.: Gene dosage-dependent embryonic development and proliferation defects in mice lacking the transcriptional integrator p300. *Cell* 93: 361–372, 1998.

[28026] 1578. Parant, J.; Chavez-Reyes, A.; Little, N. A.; Yan, W.; Reinke, V.; Jochemsen, A. G.; Lozano, G.: Rescue of embryonic lethality in Mdm4-null mice by loss of Trp53 suggests a nonoverlapping pathway with MDM2 to regulate p53. *Nature Genet.* 29: 92–95, 2001.

[28027] 1579. Shvarts, A.; Bazuine, M.; Dekker, P.; Ramos, Y. F. M.; Steegenga, W. T.; Merckx, G.; van Ham, R. C. A.; van der

Houven van Oordt, W.;van der Eb, A. J.; Jochemsen, A. G.: Isolation and identification of the human homolog of a new p53-binding protein, Mdmx. *Genomics* 43:34–42, 1997.

- [28028] 1580.Hosaka, M.; Sudhof, T. C.: Synapsin III, a novel synapsin with an unusual regulation by Ca^{2+} . *J. Biol. Chem.* 273: 13371–13374, 1998.
- [28029] 1581.Kao, H.-T.; Porton, B.; Czernik, A. J.; Feng, J.; Yiu, G.; Haring, M.; Benfenati, F.; Greengard, P.: A third member of the synapsin gene family. *Proc. Nat. Acad. Sci.* 95: 4667–4672, 1998.
- [28030] 1582.Schizophrenia Collaborative Linkage Group (Chromosome 22): A combined analysis of D22S278 marker alleles in affected sib-pairs: support for a susceptibility locus for schizophrenia at chromosome 22q12. *Am. J. Med. Genet.* 67: 40–45, 1996.
- [28031] 1583.Tsai, M.-T.; Hung, C.-C.; Tsai, C.-Y.; Liu, M.-Y.; Su, Y.-C.; Chen, Y.-H.; Hsiao, K.-J.; Chen, C.-H.: Mutation analysis of synapsin III gene in schizophrenia. *Am. J. Med. Genet. (Neuropsychiat. Genet.)* 114:79–83, 2002.
- [28032] 1584.Dubiel, W.; Ferrell, K.; Pratt, G.; Rechsteiner, M.: Subunit 4 of the 26 S protease is a member of a novel eukaryotic ATPase family. *J. Biol. Chem.* 267: 22699–22702, 1992.

- [28033] 1585.Hoyle, J.; Fisher, E. M. C.: Genomic organization and mapping of the mouse P26s4 ATPase gene: a member of the remarkably conserved AAA gene family. *Genomics* 31: 115–118, 1996.
- [28034] 1586.Dubiel, W.; Ferrell, K.; Rechsteiner, M.: Tat-binding protein 7 is a subunit of the 26S protease. *Biol. Chem. Hoppe-Seyler* 375:237–240, 1994.
- [28035] 1587.Kull, F. J.; Sablin, E. P.; Lau, R.; Fletterick, R. J.; Vale, R.D.: Crystal structure of the kinesin motor domain reveals a structural similarity to myosin. *Nature* 380: 550–555, 1996.
- [28036] 1588.Navone, F.; Niclas, J.; Hom-Booher, N.; Sparks, L.; Bernstein, H. D.; McCaffrey, G.; Vale, R. D.: Cloning and expression of a human kinesin heavy chain gene: interaction of the COOH-terminal domain with cytoplasmic microtubules in transfected CV-1 cells. *J. Cell Biol.* 117: 1263–1275, 1992.
- [28037] 1589.Niclas, J.; Navone, F.; Hom-Booher, N.; Vale, R. D.: Cloning and localization of a conventional kinesin motor expressed exclusively in neurons. *Neuron* 12: 1059–1072, 1994.
- [28038] 1590.Tanaka, Y.; Kanai, Y.; Okada, Y.; Nonaka, S.; Takeda, S.; Harada, A.; Hirokawa, N.: Targeted disruption of mouse

conventional kinesin heavy chain, kif5B, results in abnormal perinuclear clustering of mitochondria. *Cell* 93: 1147–1158, 1998.

- [28039] 1591. Wang, H.; Huang, Z.-Q.; Xia, L.; Feng, Q.; Erdjument-Bromage, H.; Strahl, B. D.; Briggs, S. D.; Allis, C. D.; Wong, J.; Tempst, P.; Zhang, Y.: Methylation of histone H4 at arginine 3 facilitating transcriptional activation by nuclear hormone receptor. *Science* 293: 853–857, 2001.
- [28040] 1592. Sun, Z.; Unutmaz, D.; Zou, Y.-R.; Sunshine, M. J.; Pierani, A.; Brenner-Morton, S.; Mebius, R. E.; Littman, D. R.: Requirement for ROR-gamma in thymocyte survival and lymphoid organ development. *Science* 288: 2369–2373, 2000.
- [28041] 1593. Albala, J. S.; Thelen, M. P.; Prange, C.; Fan, W.; Christensen, M.; Thompson, L. H.; Lennon, G. G.: Identification of a novel human RAD51 homolog, RAD51B. *Genomics* 46: 476–479, 1997.
- [28042] 1594. Cartwright, R.; Dunn, A. M.; Simpson, P. J.; Tambini, C. E.; Thacker, J.: Isolation of novel human and mouse genes of the recA/RAD51 recombination-repair gene family. *Nucleic Acids Res.* 26: 1653–1659, 1998.
- [28043] 1595. Rice, M. C.; Smith, S. T.; Bullrich, F.; Havre, P.; Kmiec, E. B.: Isolation of human and mouse genes based

on homology to REC2, arecombinational repair gene from the fungus *Ustilago maydis*. *Proc.Nat. Acad. Sci.* 94: 7417–7422, 1997.

- [28044] 1596.Abramovich, C.; Yakobson, B.; Chebath, J.; Revel, M.: A protein–argininemethyltransferase binds to the intracytoplasmic domain of the IFNAR1chain in the type I interferon receptor. *EMBO J.* 16: 260–266, 1997.
- [28045] 1597.Lin, W.–J.; Gary, J. D.; Yang, M. C.; Clarke, S.; Herschman, H.R.: The mammalian immediate–early TIS21 protein and the leukemia–associatedBTG1 protein interact with a protein–arginine N–methyltransferase. *J.Biol. Chem.* 271: 15034–15044, 1996.
- [28046] 1598.Mowen, K. A.; Tang, J.; Zhu, W.; Schurter, B. T.; Shuai, K.; Herschman,H. R.; David, M.: Arginine methylation of STAT1 modulates IFN–alpha/beta–induced transcription. *Cell* 104: 731–741, 2001.
- [28047] 1599.Nikawa, J.; Nakano, H.; Ohi, N.: Structural and functional conservationof human and yeast HCP1 genes which can suppress the growth defectof the *Saccharomyces cerevisiae* ire15 mutant. *Gene* 171: 107–111,1996.
- [28048] 1600.Pawlak, M. R.; Scherer, C. A.; Chen, J.; Roshon, M. J.; Ruley,H. E.: Arginine N–methyltransferase 1 is required for early postimplantationmouse development, but cells defi–

cient in the enzyme are viable. *Molec.Cell. Biol.* 20: 4859–4869, 2000.

[28049] 1601.Scorilas, A.; Black, M. H.; Talieri, M.; Diamandis, E. P.: Genomic organization, physical mapping, and expression analysis of the human protein arginine methyltransferase 1 gene. *Biochem. Biophys. Res. Commun.* 278: 349–359, 2000.

[28050] 1602.Baxendale, S.; MacDonald, M. E.; Mott, R.; Francis, F.; Lin, C.; Kirby, S. F.; James, M.; Zehetner, G.; Hummerich, H.; Valdes, J.; Collins, F. S.; Deaven, L. J.; Gusella, J. F.; Lehrach, H.; Bates, G. P.: A cosmid contig and high resolution restriction map of the 2 megabase region containing the Huntington's disease gene. *Nature Genet.* 4: 181–186, 1993.

[28051] 1603.Richelda, R.; Ronchetti, D.; Baldini, L.; Cro, L.; Viggiano, L.; Marzella, R.; Rocchi, M.; Otsuki, T.; Lombardi, L.; Maiolo, A. T.; Neri, A.: A novel chromosomal translocation t(4;14)(p16.3;q32) in multiple myeloma involves the fibroblast growth-factor receptor 3 gene. *Blood* 90: 4062–4070, 1997.

[28052] 1604.Wright, T. J.; Ricke, D. O.; Denison, K.; Abmayr, S.; Cotter, P.D.; Hirschhorn, K.; Keinänen, M.; McDonald-McGinn, D.; Somer, M.; Spinner, N.; Yang-Feng, T.; Zackai,

E.; Altherr, M. R.: A transcriptmap of the newly defined 165 kb Wolf–Hirschhorn syndrome criticalregion. Hum. Molec. Genet. 6: 317–324, 1997.

[28053] 1605.Chen, H.; Rossier, C.; Lalioti, M. D.; Lynn, A.; Chakravarti, A.;Perrin, G.; Antonarakis, S. E.: Cloning of the cDNA for a human homologueof the Drosophila white gene and mapping to chromosome 21q22.3. Am.J. Hum. Genet. 59: 66–75, 1996.

[28054] 1606.Croop, J. M.; Tiller, G. E.; Fletcher, J. A.; Lux, M. L.; Raab,E.; Goldenson, D.; Son, D.; Arciniegas, S.; Wu, R. L.: Isolationand characterization of a mammalian homolog of the Drosophila whitegene. Gene 185: 77–85, 1997.

[28055] 1607.Klucken, J.; Buchler, C.; Orso, E.; Kaminski, W. E.; Porsch–Ozcurumez,M.; Liebisch, G.; Kapinsky, M.; Diederich, W.; Drobnik, W.; Dean,M.; Allikmets, R.; Schmitz, G.: ABCG1 (ABC8), the human homolog ofthe Drosophila white gene, is a regulator of macrophage cholesteroland phospholipid transport. Proc. Nat. Acad. Sci. 97: 817–822, 2000.

[28056] 1608.Langmann, T.; Porsch–Ozcurumez, M.; Unkelbach, U.; Klucken, J.;Schmitz, G.: Genomic organization and characterization of the promoterof the human ATP–binding cassette transporter–G1 (ABCG1) gene.

Biochim.Biophys. Acta 1494: 175–180, 2000.

- [28057] 1609.Lorkowski, S.; Rust, S.; Engel, T.; Jung, E.; Tegelkamp, K.; Galinski,E. A.; Assmann, G.; Cullen, P.: Genomic sequence and structure of the human ABCG1 (ABC8) gene. Biochem. Biophys. Res. Commun. 280:121–131, 2001.
- [28058] 1610.Savary, S.; Denizot, F.; Luciani, M.–F.; Mattei, M.–G.; Chimini,G.: Molecular cloning of a mammalian ABC transporter homologous to *Drosophila* white gene. Mammalian Genome 7: 673–676, 1996.
- [28059] 1611.Chen, P.; Luo, C.; Deng, Y.; Ryan, K.; Register, J.; Margosiak,S.; Tempczyk–Russell, A.; Nguyen, B.; Myers, P.; Lundgren, K.; Kan,C.–C.; O'Connor, P. M.: The 1.7–angstrom crystal structure of human cell cycle checkpoint kinase Chk1: implications for Chk1 regulation. Cell 100:681–692, 2000.
- [28060] 1612.Flaggs, G.; Plug, A. W.; Dunks, K. M.; Mundt, K. E.; Ford, J. C.;Quiggle, M. R. E.; Taylor, E. M.; Westphal, C. H.; Ashley, T.; Hoekstra,M. F.; Carr, A. M.: Atm–dependent interactions of a mammalian Chk1homolog with meiotic chromosomes. Curr. Biol. 7: 977–986, 1997.
- [28061] 1613.Liu, Q.; Guntuku, S.; Cui, X.–S.; Matsuoka, S.; Cortez, D.; Tamai,K.; Luo, G.; Carattini–Rivera, S.; DeMayo, F.;

Bradley, A.; Donehower, L. A.; Elledge, S. J.: Chk1 is an essential kinase that is regulated by Atr and required for the G2/M DNA damage checkpoint. *Genes Dev.* 14:1448–1459, 2000.

[28062] 1614. Sanchez, Y.; Wong, C.; Thoma, R. S.; Richman, R.; Wu, Z.; Piwnicka-Worms, H.; Elledge, S. J.: Conservation of the Chk1 checkpoint pathway in mammals: linkage of DNA damage to Cdk regulation through Cdc25. *Science* 277:1497–1501, 1997.

[28063] 1615. Takai, H.; Tominaga, K.; Motoyama, N.; Minamishima, Y. A.; Nagahama, H.; Tsukiyama, T.; Ikeda, K.; Nakayama, K.; Nakanishi, M.; Nakayama, K.: Aberrant cell cycle checkpoint function and early embryonic death in Chk1^{-/-} mice. *Genes Dev.* 14: 1439–1447, 2000.

[28064] 1616. Borden, L. A.; Smith, K. E.; Gustafson, E. L.; Branchek, T. A.; Weinshank, R. L.: Cloning and expression of a betaine/GABA transporter from human brain. *J. Neurochem.* 64: 977–984, 1995.

[28065] 1617. Rasola, A.; Galletta, L. J. V.; Barone, V.; Romeo, G.; Bagnasco, S.: Molecular cloning and functional characterization of a GABA/betaine transporter from human kidney. *FEBS Lett.* 373: 229–233, 1995.

[28066] 1618. Yamauchi, A.; Uchida, S.; Kwon, H. M.; Preston, A. S.;

Robey, R.B.; Garcia-Perez, A.; Burg, M. B.; Handler, J. S.: Cloning of a Na(+)and Cl(-)-dependent betaine transporter that is regulated by hypertonicity. J.Biol. Chem. 267: 649-652, 1992.

[28067] 1619.Eisses, J. F.; Kaplan, J. H.: Molecular characterization of hCTR1,the human copper uptake protein. J. Biol. Chem. 277: 29162-29171,2002.

[28068] 1620.Klomp, A. E. M.; Tops, B. B. J.; van den Berg, I. E. T.; Berger,R.; Klomp, L. W. J.: Biochemical characterization and subcellularlocalization of human copper transporter 1 (hCTR1). Biochem. J. 364:497-505, 2002.

[28069] 1621.Kuo, Y.-M.; Zhou, B.; Cosco, D.; Gitschier, J.: The copper transporterCTR1 provides an essential function in mammalian embryonic development. Proc.Nat. Acad. Sci. 98: 6836-6841, 2001.

[28070] 1622.Lee, J.; Pena, M. M. O.; Nose, Y.; Thiele, D. J.: Biochemicalcharacterization of the human copper transporter Ctr1. J. Biol. Chem. 277:4380-4387, 2002.

[28071] 1623.Lee, J.; Prohaska, J. R.; Thiele, D. J.: Essential role for mammaliancopper transporter Ctr1 in copper homeostasis and embryonic development. Proc.Nat. Acad. Sci. 98: 6842-6847, 2001.

[28072] 1624.Moller, L. B.; Petersen, C.; Lund, C.; Horn, N.: Char-

acterization of the hCTR1 gene: genomic organization, functional expression, and identification of a highly homologous processed gene. *Gene* 257:13–22, 2000.

[28073] 1625. Zhou, B.; Gitschier, J.: hCTR1: a human gene for copper uptake identified by complementation in yeast. *Proc. Nat. Acad. Sci.* 94:7481–7486, 1997.

[28074] 1626. Schwienbacher, C.; Sabbioni, S.; Campi, M.; Veronese, A.; Bernardi, G.; Menegatti, A.; Hatada, I.; Mukai, T.; Ohashi, H.; Barbanti-Brodano, G.; Croce, C. M.; Negrini, M.: Transcriptional map of 170-kb region at chromosome 11p15.5: identification and mutational analysis of the BWR1A gene reveals the presence of mutations in tumor samples. *Proc. Nat. Acad. Sci.* 95: 3873–3878, 1998.

[28075] 1627. Hirono, Y.; Fushida, S.; Yonemura, Y.; Yamamoto, H.; Watanabe, H.; Raz, A.: Expression of autocrine motility factor receptor correlates with disease progression in human gastric cancer. *Brit. J. Cancer* 74:2003–2007, 1996.

[28076] 1628. Huang, B.; Xie, Y.; Raz, A.: Identification of an upstream region that controls the transcription of the human autocrine motility factor receptor. *Biochem. Biophys. Res. Commun.* 212: 727–742, 1995.

[28077] 1629. Silletti, S.; Yao, J.; Sanford, J.; Mohammed, A. N.; Otto, T.; Wolman, S. R.; Raz, A.: Autocrine motility factor

receptor in human bladder carcinoma: gene expression, loss of cell–contact regulation and chromosomal mapping. *Int. J. Oncol.* 3: 801–807, 1993.

- [28078] 1630. Watanabe, H.; Carmi, P.; Hogan, V.; Raz, T.; Silletti, S.; Nabi, I. R.; Raz, A.: Purification of human tumor cell autocrine motility factor and molecular cloning of its receptor. *J. Biol. Chem.* 266:13442–13448, 1991.
- [28079] 1631. Apergis, G. A.; Crawford, N.; Ghosh, D.; Stepan, C. M.; Vorachek, W. R.; Wen, P.; Locker, J.: A novel nk-2-related transcription factor associated with human fetal liver and hepatocellular carcinoma. *J. Biol. Chem.* 273: 2917–2925, 1998.
- [28080] 1632. Amlal, H.; Burnham, C. E.; Soleimani, M.: Characterization of Na(+)/HCO(3-) cotransporter isoform NBC-3. *Am. J. Physiol.* 276:F903–F913, 1999.
- [28081] 1633. Burnham, C. E.; Wang, Z.; Soleimani, M.: Personal Communication. Cincinnati, Oh. 6/1/2000.
- [28082] 1634. Choi, I.; Aalkjaer, C.; Boulpaep, E. L.; Boron, W. F.: An electroneutral sodium/bicarbonate cotransporter NBCn1 and associated sodium channel. *Nature* 405:571–575, 2000.
- [28083] 1635. Ishibashi, K.; Sasaki, S.; Marumo, F.: Molecular cloning of a new sodium bicarbonate cotransporter cDNA

from human retina. *Biochem.Biophys. Res. Commun.* 246: 535–538, 1998.

- [28084] 1636.Pushkin, A.; Abuladze, N.; Lee, I.; Newman, D.; Hwang, J.; Kurtz,I.: Mapping of the human NBC3 (SLC4A7) gene to chromosome 3p22. *Genomics* 57:321–322, 1999. Note: Correction: *Genomics* 58: 216 and 321–322, 1999.
- [28085] 1637.Pushkin, A.; Abuladze, N.; Lee, I.; Newman, D.; Hwang, J.; Kurtz,I.: Cloning, tissue distribution, genomic organization, and functional characterization of NBC3, a new member of the sodium bicarbonate cotransporter–family. *J. Biol. Chem.* 274: 16569–16575, 1999.
- [28086] 1638.Brown, M. S.; Goldstein, J. L.: The SREBP pathway: regulation of cholesterol metabolism by proteolysis of a membrane–bound transcription factor. *Cell* 89: 331–340, 1997.
- [28087] 1639.Duncan, E. A.; Brown, M. S.; Goldstein, J. L.; Sakai, J.: Cleavage site for sterol–regulated protease localized to a leu–ser bond in the lumenal loop of sterol regulatory element–binding protein–2. *J.Biol. Chem.* 272: 12778–12785, 1997.
- [28088] 1640.Nagase, T.; Miyajima, N.; Tanaka, A.; Sazuka, T.; Seki, N.; Sato,S.; Tabata, S.; Ishikawa, K.; Kawarabayasi, Y.; Kotani, H.; Nomura,N.: Prediction of the coding sequences

of unidentified human genes.III. The coding sequences of 40 new genes (KIAA0081–KIAA0120) deducedby analysis of cDNA clones from human cell line KG–1. DNA Res. 2:37–43, 1995.

[28089] 1641.Nakajima, T.; Iwaki, K.; Kodama, T.; Inazawa, J.; Emi, M.: Genomicstructure and chromosomal mapping of the human site–1 protease (S1P)gene. J. Hum. Genet. 45: 212–217, 2000.

[28090] 1642.Sakai, J.; Duncan, E. A.; Rawson, R. B.; Hua, X.; Brown, M. S.;Goldstein, J. L.: Sterol–regulated release of SREBP–2 from cell membranesrequires two sequential cleavages, one within a transmembrane segment. Cell 85:1037–1046, 1996.

[28091] 1643.Sakai, J.; Rawson, R. B.; Espenshade, P. J.; Cheng, D.; Seegmiller,A. C.; Goldstein, J. L.; Brown, M. S.: Molecular identification ofthe sterol–regulated luminal protease that cleaves SREBPs and controlslipid composition of animal cells. Molec. Cell 2: 505–514, 1998.

[28092] 1644.Watt, S. M.; Buhring, H.–J.; Rappold, I.; Chan, J. Y.–H.; Lee–Prudhoe,J.; Jones, T.; Zannettino, A. C. W.; Simmons, P. J.; Doyonnas, R.;Sheer, D.; Butler, L. H.: CD164, a novel sialomucin on CD34+ anderythroid subsets, is located on human chromosome 6q21. Blood 92:849–866,

1998.

- [28093] 1645. Zannettino, A. C. W.; Buhning, H.-J.; Niutta, S.; Watt, S. M.; Benton, M. A.; Simmons, P. J.: The sialomucin CD164 (MGC-24v) is an adhesive glycoprotein expressed by human hematopoietic progenitors and bone marrow stromal cells that serves as a potent negative regulator of hematopoiesis. *Blood* 92: 2613–2628, 1998.
- [28094] 1646. Abts, H. F.; Breuhahn, K.; Michel, G.; Kohrer, K.; Esser, P.; Ruzicka, T.: Analysis of UVB modulated gene expression in human keratinocytes by mRNA differential display polymerase chain reaction. *Photochem. Photobiol.* 66: 363–367, 1997.
- [28095] 1647. Abts, H. F.; Welss, T.; Mirmohammadsadegh, A.; Kohrer, K.; Michel, G.; Ruzicka, T.: Cloning and characterization of hurpin (protease inhibitor 13): a new skin-specific, UV-repressible serine proteinase inhibitor of the ovalbumin serpin family. *J. Molec. Biol.* 293: 29–39, 1999.
- [28096] 1648. Nakashima, T.; Pak, S. C.; Silverman, G. A.; Spring, P. M.; Frederick, M. J.; Clayman, G. L.: Genomic cloning, mapping, structure and promoter analysis of HEADPIN, a serpin which is down-regulated in head and neck cancer cells. *Biochim. Biophys. Acta* 1492: 441–446, 2000.
- [28097] 1649. Spring, P.; Nakashima, T.; Frederick, M.; Henderson,

Y.; Clayman, G.: Identification and cDNA cloning of head-pin, a novel differentially expressed serpin that maps to chromosome 18q. *Biochem. Biophys. Res. Commun.* 264: 299–304, 1999.

- [28098] 1650. Alimova-Kost, M. V.; Imreh, S.; Buchman, V. L.; Ninkina, N. N.: Assignment of phosphotriesterase-related gene (PTER) to human chromosome band 10p12 by in situ hybridization. *Cytogenet. Cell Genet.* 83:16–17, 1998.
- [28099] 1651. Davies, J. A.; Buchman, V. L.; Krylova, O.; Ninkina, N. N.: Molecular cloning and expression pattern of rpr-1, a resiniferatoxin-binding, phosphotriesterase-related protein, expressed in rat kidney tubules. *FEBS Lett.* 410: 378–382, 1997.
- [28100] 1652. Jones, P. G.; Lombardi, S. J.; Cockett, M. I.: Cloning and tissue distribution of the human G protein beta-5 cDNA. *Biochim. Biophys. Acta* 1402: 288–291, 1998.
- [28101] 1653. Watson, A. J.; Aragay, A. M.; Slepak, V. Z.; Simon, M. I.: A novel form of the G protein beta subunit G-beta-5 is specifically expressed in the vertebrate retina. *J. Biol. Chem.* 271: 28154–28160, 1996.
- [28102] 1654. Watson, A. J.; Katz, A.; Simon, M. I.: A fifth member of the mammalian G-protein beta subunit family: expression in brain and activation of the beta-2 isotype of phos-

pholipase C. J. Biol. Chem. 269: 22150–22156,1994.

- [28103] 1655.Dixon, B.; Sahely, B.; Liu, L.; Pohajdak, B.: Cloning a cDNA from human NK/T cells which codes for an unusual leucine zipper containing protein. Biochim. Biophys. Acta 1216: 321–324, 1993.
- [28104] 1656.Kim, H.–S.: Assignment of the human B3–1 gene (PSCDBP) to chromosome 2 band q11.2 by radiation hybrid mapping. Cytogenet. Cell Genet. 84:95 only, 1999.
- [28105] 1657.Tang, P.; Cheng, T. P.; Agnello, D.; Wu, C.–Y.; Hissong, B. D.; Watford, W. T.; Ahn, H.–J.; Galon, J.; Moss, J.; Vaughan, M.; O'Shea, J. J.; Gadina, M.: Cybr, a cytokine–inducible protein that binds cytohesin–1 and regulates its activity. Proc. Nat. Acad. Sci. 99:2625–2629, 2002.
- [28106] 1658.Nakayama, Y.; Goebel, M.; O'Brine Greco, B.; Lemon, S.; Pingchang Chow, E.; Kirchhausen, T.: The medium chains of the mammalian clathrin–associated proteins have a homolog in yeast. Europ. J. Biochem. 202: 569–574,1991.
- [28107] 1659.Hiramoto, T.; Nakanishi, T.; Sumiyoshi, T.; Fukuda, T.; Matsuura, S.; Tauchi, H.; Komatsu, K.; Shibasaki, Y.; Inui, H.; Watatani, M.; Yasutomi, M.; Sumii, K.; Kajiyama, G.; Kamada, N.; Miyagawa, K.; Kamiya, K.: Mutations of a novel human RAD54 homologue, RAD54B, in prima–

rycancer. *Oncogene* 18: 3422–3426, 1999.

- [28108] 1660. Miyagawa, K.; Tsuruga, T.; Kinomura, A.; Usui, K.; Katsura, M.; Tashiro, S.; Mishima, H.; Tanaka, K.: A role for RAD54B in homologous recombination in human cells. *EMBO J.* 21: 175–180, 2002.
- [28109] 1661. Tanaka, K.; Hiramoto, T.; Fukuda, T.; Miyagawa, K.: A novel human Rad54 homologue, Rad54B, associates with Rad51. *J. Biol. Chem.* 275:26316–26321, 2000.
- [28110] 1662. Pillutla, R. C.; Shimamoto, A.; Furuichi, Y.; Shatkin, A. J.: Human mRNA capping enzyme (RNGTT) and cap methyltransferase (RNMT) map to 6q16 and 18p11.22–p11.23, respectively. *Genomics* 54: 351–353, 1998.
- [28111] 1663. Tsukamoto, T.; Shibagaki, Y.; Murakoshi, T.; Suzuki, M.; Nakamura, A.; Gotoh, H.; Mizumoto, K.: Cloning and characterization of two human cDNAs encoding the mRNA capping enzyme. *Biochem. Biophys. Res. Commun.* 243: 101–108, 1998.
- [28112] 1664. Yamada–Okabe, T.; Doi, R.; Shimmi, O.; Arisawa, M.; Yamada–Okabe, H.: Isolation and characterization of a human cDNA for mRNA 5–prime–capping enzyme. *Nucleic Acids Res.* 26: 1700–1706, 1998.
- [28113] 1665. Yue, Z.; Maldonado, E.; Pillutla, R.; Cho, H.; Rein–

berg, D.; Shatkin, A. J.: Mammalian capping enzyme complements mutant *Saccharomyces cerevisiae* lacking mRNA guanylyltransferase and selectively binds the elongating form of RNA polymerase II. *Proc. Nat. Acad. Sci.* 94:12898–12903, 1997.

[28114] 1666. Ishikawa, K.; Nagase, T.; Nakajima, D.; Seki, N.; Ohira, M.; Miyajima, N.; Tanaka, A.; Kotani, H.; Nomura, N.; Ohara, O.: Prediction of the coding sequences of unidentified human genes. VIII. 78 new cDNA clones from brain which code for large proteins in vitro. *DNA Res.* 4:307–313, 1997.

[28115] 1667. Pillutla, R. C.; Yue, Z.; Maldonado, E.; Shatkin, A. J.: Recombinant human mRNA cap methyltransferase binds capping enzyme/RNA polymerase complexes. *J. Biol. Chem.* 273: 21443–21446, 1998.

[28116] 1668. Tsukamoto, T.; Shibagaki, Y.; Niikura, Y.; Mizumoto, K.: Cloning and characterization of three human cDNAs encoding mRNA (guanine-7)-methyltransferase, an mRNA cap methylase. *Biochem. Biophys. Res. Commun.* 251: 27–34, 1998.

[28117] 1669. Misra-Press, A.; Cooke, N. E.; Liebhaber, S. A.: Complex alternative splicing partially inactivates the human chorionic somatomammotropin-like (hCS-L) gene. *J. Biol.*

Chem. 269: 23220–23229, 1994.

- [28118] 1670.Sawamura, T.; Kume, N.; Aoyama, T.; Moriwaki, H.; Hoshikawa, H.;Aiba, Y.; Tanaka, T.; Miwa, S.; Katsura, Y.; Kita, T.; Masaki, T.: An endothelial receptor for oxidized low-density lipoprotein. *Nature* 386:73–77, 1997.
- [28119] 1671.Yamanaka, S.; Zhang, X.–Y.; Miura, K.; Kim, S.; Iwao, H.: Thehuman gene encoding the lectin-type oxidized LDL receptor (OLR1) isa novel member of the natural killer gene complex with a unique expressionprofile. *Genomics* 54: 191–199, 1998.
- [28120] 1672.Gorboulev, V.; Ulzheimer, J. C.; Akhoundova, A.; Ulzheimer–Teuber,I.; Karbach, U.; Quester, S.; Baumann, C.; Lang, F.; Busch, A. E.;Koepsell, H.: Cloning and characterization of two human polyspecificorganic cation transporters. *DNA Cell Biol.* 16: 871–881, 1997.
- [28121] 1673.Grundemann, D.; Gorboulev, V.; Gambaryan, S.; Veyhl, M.; Koepsell,H.: Drug excretion mediated by a new prototype of polyspecific transporter. *Nature* 372:549–552, 1994.
- [28122] 1674.Hayer, M.; Bonisch, H.; Bruss, M.: Molecular cloning, functionalcharacterization and genomic organization of four alternatively splicedisoforms of the human organic cation transporter 1 (hOCT1/SLC22A1). *Ann.Hum. Genet.*

63: 473–482, 1999.

- [28123] 1675.Koehler, M. R.; Wissinger, B.; Gorboulev, V.; Koepsell, H.; Schmid,M.: The two human organic cation transporter genes SLC22A1 and SLC22A2are located on chromosome 6q26. *Cytogenet. Cell Genet.* 79: 198–200,1997.
- [28124] 1676.Zhang, L.; Dresser, M. J.; Gray, A. T.; Yost, S. C.; Terashita,S.; Giacomini, K. M.: Cloning and functional expression of a humanliver organic cation transporter. *Molec. Pharm.* 51: 913–921, 1997.
- [28125] 1677.Grundemann, D.; Schomig, E.: Gene structures of the human non-neuronalmonoamine transporters EMT and OCT2. *Hum. Genet.* 106: 627–635, 2000.
- [28126] 1678.Mooslehner, K. A.; Allen, N. D.: Cloning of the mouse organiccation transporter 2 gene, Slc22a2, from an enhancer–trap transgeneintegration locus. *Mammalian Genome* 10: 218–224, 1999.
- [28127] 1679.Fujiwara, T.; Watanabe, T. K.; Tanaka, K.; Slaughter, C. A.; DeMartino,G. N.: cDNA cloning of p42, a shared subunit of two proteasome regulatoryproteins, reveals a novel member of the AAA protein family. *FEBSLett.* 387: 184–188, 1996.
- [28128] 1680.Hu, Q.; Cool, B. H.; Wang, B.; Hearn, M. G.; Martin,

G. M.: A candidate molecular mechanism for the association of an intronic polymorphism of FE65 with resistance to very late onset dementia of the Alzheimer type. *Hum. Molec. Genet.* 11: 465–475, 2002.

[28129] 1681. Hu, Q.; Kukull, W. A.; Bressler, S. L.; Gray, M. D.; Cam, J. A.; Larson, E. B.; Martin, G. M.; Deeb, S. S.: The human FE65 gene: genomic structure and an intronic biallelic polymorphism associated with sporadic dementia of the Alzheimer type. *Hum. Genet.* 103: 295–303, 1998.

[28130] 1682. McLoughlin, D. M.; Miller, C. C. J.: The intracellular cytoplasmic domain of the Alzheimer's disease amyloid precursor protein interacts with phosphotyrosine-binding domain proteins in the yeast two-hybrid system. *FEBS Lett.* 397: 197–200, 1996.

[28131] 1683. Guenette, S. Y.; Chen, J.; Jondro, P. D.; Tanzi, R. E.: Association of a novel human FE65-like protein with the cytoplasmic domain of the beta-amyloid precursor protein. *Proc. Nat. Acad. Sci.* 93: 10832–10837, 1996.

[28132] 1684. Bressler, S. L.; Gray, M. D.; Sopher, B. L.; Hu, Q.; Hearn, M. G.; Pham, D. G.; Dinulos, M. B.; Fukuchi, K.-I.; Sisodia, S. S.; Miller, M. A.; Distèche, C. M.; Martin, G. M.: cDNA cloning and chromosomal mapping of the human Fe65 gene: interaction of the conserved cytoplasmic do-

mains of the human beta-amyloid precursor protein and its homologues with the mouse Fe65 protein. *Hum. Molec. Genet.* 5: 1589–1598, 1996.

- [28133] 1685. Duilio, A.; Faraonio, R.; Minopoli, G.; Zambrano, N.; Russo, T.: Fe65L2: a new member of the Fe65 protein family interacting with the intracellular domain of the Alzheimer's beta-amyloid precursor protein. *Biochem. J.* 330: 513–519, 1998.
- [28134] 1686. Tanahashi, H.; Tabira, T.: Genome structure and chromosomal mapping of the gene for Fe65L2 interacting with Alzheimer's beta-amyloid precursor protein. *Biochem. Biophys. Res. Commun.* 258: 385–389, 1999.
- [28135] 1687. Tanahashi, H.; Tabira, T.: Molecular cloning of human Fe65L2 and its interaction with the Alzheimer's beta-amyloid precursor protein. *Neurosci. Lett.* 261: 143–146, 1999.
- [28136] 1688. Howard, L.; Nelson, K. K.; Maciewicz, R. A.; Blobel, C. P.: Interaction of the metalloprotease disintegrins MDC9 and MDC15 with two SH3 domain-containing proteins, endophilin I and SH3PX1. *J. Biol. Chem.* 274: 31693–31699, 1999.
- [28137] 1689. Galliano, M.-F.; Huet, C.; Frygeliuss, J.; Polgren, A.; Wewer, U.M.; Engvall, E.: Binding of ADAM12, a marker of

skeletal muscle regeneration, to the muscle-specific actin-binding protein, alpha-actinin-2, is required for myoblast fusion. *J. Biol. Chem.* 275: 13933–13939, 2000.

- [28138] 1690. Gilpin, B. J.; Loechel, F.; Mattei, M.-G.; Engvall, E.; Albrechtsen, R.; Wewer, U. M.: A novel, secreted form of human ADAM 12 (meltrin alpha) provokes myogenesis in vivo. *J. Biol. Chem.* 273: 157–166, 1998.
- [28139] 1691. Yagami-Hiromasa, T.; Sato, T.; Kurisaki, T.; Kamijo, K.; Nabeshima, Y.; Fujisawa-Sehara, A.: A metalloprotease-disintegrin participating in myoblast fusion. *Nature* 377: 652–656, 1995.
- [28140] 1692. Brancolini, C.; Bottega, S.; Schneider, C.: Gas2, a growth arrest-specific protein, is a component of the microfilament network system. *J. Cell Biol.* 117: 1251–1261, 1992.
- [28141] 1693. Collavin, L.; Buzzai, M.; Saccone, S.; Bernard, L.; Federico, C.; Della Valle, G.; Brancolini, C.; Schneider, C.: cDNA characterization and chromosome mapping of the human GAS2 gene. *Genomics* 48: 265–269, 1998.
- [28142] 1694. Chantry, D.; Vojtek, A.; Kashishian, A.; Holtzman, D. A.; Wood, C.; Gray, P. W.; Cooper, J. A.; Hoekstra, M. F.: p110-delta, a novel phosphatidylinositol 3-kinase catalytic subunit that associates with p85 and is expressed pre-

dominantly in leukocytes. J. Biol. Chem.

272:19236–19241, 1997.

[28143] 1695.Okkenhaug, K.; Bilancio, A.; Farjot, G.; Priddle, H.; Sancho, S.;Peskett, E.; Pearce, W.; Meek, S. E.; Salpekar, A.; Waterfield, M.D.; Smith, A. J. H.; Vanhaesebroeck, B.: Impaired B and T cell antigenreceptor signaling in p110–delta PI 3–kinase mutant mice. Science 297:1031–1034, 2002.

[28144] 1696.Seki, N.; Nimura, Y.; Ohira, M.; Saito, T.; Ichimiya, S.; Nomura,N.; Nakagawara, A.: Identification and chromosome assignment of a human gene encoding a novel phosphatidylinositol–3 kinase. DNA Res. 4:355–358, 1997.

[28145] 1697.Vanhaesebroeck, B.; Welham, M. J.; Kotani, K.; Stein, R.; Warne,P. H.; Zvelebil, M. J.; Higashi, K.; Volinia, S.; Downward, J.; Waterfield,M. D.: p110–delta, a novel phosphoinositide 3–kinase in leukocytes. Proc.Nat. Acad. Sci. 94: 4330–4335, 1997.

[28146] 1698.Bowman, M. R.; Crimmins, M. A. V.; Yetz–Aldape, J.; Kriz, R.; Kelleher,K.; Herrmann, S.: The cloning of CD70 and its identification as the ligand for CD27. J. Immun. 152: 1756–1761, 1994.

[28147] 1699.Berger, R.; Mezey, E.; Clancy, K. P.; Harta, G.; Wright,

R. M.; Repine, J. E.; Brown, R. H.; Brownstein, M.; Patterson, D.: Analysis of aldehyde oxidase and xanthine dehydrogenase/oxidase as possible candidate genes for autosomal recessive familial amyotrophic lateral sclerosis. *Somat. Cell Molec. Genet.* 21: 121–131, 1995.

[28148] 1700. Turner, N. A.; Doyle, W. A.; Ventom, A. M.; Bray, R. C.: Properties of rabbit liver aldehyde oxidase and the relationship of the enzyme to xanthine oxidase and dehydrogenase. *Europ. J. Biochem.* 232: 646–657, 1995.

[28149] 1701. Agarwal, A. K.; White, P. C.: Structure of the VPATPD gene encoding subunit D of the human vacuolar proton ATPase. *Biochem. Biophys. Res. Commun.* 279: 543–547, 2000.

[28150] 1702. Mune, T.; Rogerson, F. M.; Nikkila, H.; Agarwal, A. K.; White, P. C.: Human hypertension caused by mutations in the kidney isozyme of 11- β -hydroxysteroid dehydrogenase. *Nature Genet.* 10: 394–399, 1995.

[28151] 1703. Nikkila, H.; Tannin, G. M.; New, M. I.; Taylor, N. F.; Kalaitzoglou, G.; Monder, C.; White, P. C.: Defects in the HSD11 gene encoding 11- β -hydroxysteroid dehydrogenase are not found in patients with apparent mineralocorticoid excess or 11-oxoreductase deficiency. *J. Clin. Endocr. Metab.* 77: 687–691, 1993.

- [28152] 1704.Cheng, G.; Ye, Z.-S.; Baltimore, D.: Binding of Bruton's tyrosinekinase to Fyn, Lyn, or Hck through a Src homology 3 domain-mediatedinteraction. *Proc. Nat. Acad. Sci.* 91: 8152–8155, 1994.
- [28153] 1705.Gibson, S.; Leung, B.; Squire, J. A.; Hill, M.; Arima, N.; Goss,P.; Hogg, D.; Mills, G. B.: Identification, cloning, and characterizationof a novel human T-cell-specific tyrosine kinase located at the hematopoietincomplex on chromosome 5q. *Blood* 82: 1561–1572, 1993.
- [28154] 1706.Janis, E. M.; Siliciano, J. D.; Isaac, D. D.; Griffin, C. A.; Hawkins,A. L.; Kozak, C. A.; Desiderio, S.: Mapping of the gene for the tyrosinekinase Itk to a region of conserved synteny between mouse chromosome11 and human chromosome 5q. *Genomics* 23: 269–271, 1994.
- [28155] 1707.Schaeffer, E. M.; Debnath, J.; Yap, G.; McVicar, D.; Liao, X. C.;Littman, D. R.; Sher, A.; Varmus, H. E.; Lenardo, M. J.; Schwartzberg,P. L.: Requirement for Tec kinases Rlk and Itk in T cell receptorsignaling and immunity. *Science* 284: 638–641, 1999.
- [28156] 1708.Woods, M. L.; Kivens, W. J.; Adelsman, M. A.; Qiu, Y.; August,A.; Shimizu, Y.: A novel function for the Tec family tyrosine kinaseltk in activation of beta-1 integrins by the T-cell receptor. *EMBOJ.* 20: 1232–1244, 2001.

- [28157] 1709. Blanche, H.; Massart, C.; Dausset, J.; Cann, H.: TCP1 is not linked to HLA, GLO1, PGK1P2 and other markers in a 45cM map of the short arm of chromosome 6 (6p). (Abstract) Cytogenet. Cell Genet. 46:581–582, 1987.
- [28158] 1710. Blanche, H.; Wright, L. G.; Vergnaud, G.; de Gouyon, B.; Lauthier, V.; Silver, L. M.; Dausset, J.; Cann, H. M.; Spielman, R. S.: Genetic mapping of three human homologues of murine t-complex genes localizes TCP10 to 6q27, 15 cM distal to TCP1 and PLG. Genomics 12: 826–828, 1992.
- [28159] 1711. Fonatsch, C.; Gradl, G.; Ragousis, J.; Ziegler, A.: Assignment of the TCP1 locus to the long arm of human chromosome 6 by in situ hybridization. Cytogenet. Cell Genet. 45: 109–112, 1987.
- [28160] 1712. Willison, K.; Dudley, K.; Spurr, N.; Goodfellow, P.: Chromosomal assignment of TCP-1, the human homologue of a mouse t-complex locus. (Abstract) Cytogenet. Cell Genet. 40: 779–780, 1985.
- [28161] 1713. Parker, N. J.; Begley, C. G.; Fox, R. M.: Human R1 subunit of ribonucleotide reductase (RRM1): 5-prime flanking region of the gene. Genomics 19:91–96, 1994.
- [28162] 1714. Parker, N. J.; Begley, C. G.; Fox, R. M.: Human gene for the large subunit of ribonucleotide reductase (RRM1):

functional analysis of the promoter. *Genomics* 27:
280–285, 1995.

[28163] 1715. Bisbal, C.; Martinand, C.; Silhol, M.; Lebleu, B.; Salehzada, T.: Cloning and characterization of a RNase L inhibitor: a new component of the interferon-regulated 2–5A pathway. *J. Biol. Chem.* 270: 13308–13317, 1995.

[28164] 1716. Elliott, R. W.; Samuelson, L. C.; Lambert, M. S.; Meisler, M. H.: Assignment of pancreatic ribonuclease gene to mouse chromosome 14. *Cytogenet. Cell Genet.* 42: 110–112, 1986.

[28165] 1717. Piccoli, R.; Di Gaetano, S.; De Lorenzo, C.; Grauso, M.; Monaco, C.; Spalletti-Cernia, D.; Laccetti, P.; Cinatl, J.; Matousek, J.; D'Alessio, G.: A dimeric mutant of human pancreatic ribonuclease with selective cytotoxicity toward malignant cells. *Proc. Nat. Acad. Sci.* 96: 7768–7773, 1999.

[28166] 1718. Zhang, J.; Zhang, Y.; Rosenberg, H. F.: Adaptive evolution of a duplicated pancreatic ribonuclease gene in a leaf-eating monkey. *Nature Genet.* 30: 416–420, 2002.

[28167] 1719. Shen, S.; Battersby, S.; Weaver, M.; Clark, E.; Stephens, K.; Harmar, A. J.: Refined mapping of the human serotonin transporter (SLC6A4) gene within 17q11 adjacent to the CPD and NF1 genes. *Europ. J. Hum. Genet.* 8:

75–78, 2000.

[28168] 1720.Sukonick, D. L.; Pollock, B. G.; Sweet, R. A.; Mulsant, B. H.;Rosen, J.; Klunk, W. E.; Kastango, K. B.; DeKosky, S. T.; Ferrell,R. E.: The 5-HTTPR*S/*L polymorphism and aggressive behavior in Alzheimerdisease. Arch. Neurol. 58: 1425–1428, 2001.

[28169] 1721.Lilja, H.; Abrahamsson, P.–A.; Lundwall, A.: Semenogelin, thepredominant protein in human semen: primary structure and identificationof closely related proteins in the male accessory sex glands and onthe spermatozoa. J. Biol. Chem. 264: 1894–1900, 1989.

[28170] 1722.Loeffler, C.; Rao, V. V. N. G.; Schnittger, S.; Pfau, H. P.; Lundwall,A.; Schaefer, R.; Stolz, F. M.; Hansmann, I.: Personal Communication. 8/18/1991.

[28171] 1723.Ulvsback, M.; Lazure, C.; Lilja, H.; Spurr, N. K.; Rao, V. V.;Loffler, C.; Hansmann, I.; Lundwall, A.: Gene structure of semenogelinI and II: the predominant proteins in human semen are encoded by twohomologous genes on chromosome 20. J. Biol. Chem. 267: 18080–18084,1992.

[28172] 1724.Byrne, P. C.; Shipley, J. M.; Chave, K. J.; Sanders, P. G.; Snell,K.: Characterisation of a human serine hydroxymethyltransferase pseudogeneand its localisation of 1p32.3–33. Hum. Genet. 97: 340–344, 1996.

- [28173] 1725.Elsea, S. H.; Juyal, R. C.; Jiralerspong, S.; Finucane, B. M.; Pandolfo, M.; Greenberg, F.; Baldini, A.; Stover, P.; Patel, P. I.: Haploinsufficiency of cytosolic serine hydroxymethyltransferase in the Smith–Magenis syndrome. *Am. J. Hum. Genet.* 57: 1342–1350, 1995.
- [28174] 1726.Baecher, C. M.; Dorfman, K. S.; Mattei, M.–G.; Frelinger, J. G.: cDNA cloning and localization of the mouse leukosialin gene (Ly48) to chromosome 7. *Immunogenetics* 31: 307–314, 1990.
- [28175] 1727.Bazil, V.; Strominger, J. L.: CD43, the major sialoglycoprotein of human leukocytes, is proteolytically cleaved from the surface of stimulated lymphocytes and granulocytes. *Proc. Nat. Acad. Sci.* 90:3792–3796, 1993.
- [28176] 1728.Delon, J.; Kaibuchi, K.; Germain, R. N.: Exclusion of CD43 from the immunological synapse is mediated by phosphorylation–regulated relocation of the cytoskeletal adaptor moesin. *Immunity* 15: 691–701, 2001.
- [28177] 1729.Pallant, A.; Eskenazi, A.; Mattei, M.–G.; Fournier, R. E. K.; Carlsson, S. R.; Fukuda, M.; Frelinger, J. G.: Characterization of cDNAs encoding human leukosialin and localization of the leukosialin gene to chromosome 16. *Proc. Nat. Acad. Sci.* 86: 1328–1332, 1989.
- [28178] 1730.Park, J. K.; Rosenstein, Y. J.; Remold–O'Donnell, E.;

Bierer, B.E.; Rosen, F. S.; Burakoff, S. J.: Enhancement of T-cell activation by the CD43 molecule whose expression is defective in Wiskott–Aldrich syndrome. *Nature* 350: 706–709, 1991.

[28179] 1731. Rosenstein, Y.; Park, J. K.; Hahn, W. C.; Rosen, F. S.; Bierer, B. E.; Burakoff, S. J.: CD43, a molecule defective in Wiskott–Aldrich syndrome, binds ICAM–1. *Nature* 354: 233–235, 1991.

[28180] 1732. Schmid, K.; Hediger, M. A.; Brossmer, R.; Collins, J. H.; Haupt, H.; Marti, T.; Offner, G. D.; Schaller, J.; Takagaki, K.; Walsh, M. T.; Schwick, H. G.; Rosen, F. S.; Remold–O'Donnell, E.: Amino acid sequence of human plasma galactoglycoprotein: identity with the extracellular region of CD43 (sialophorin). *Proc. Nat. Acad. Sci.* 89: 663–667, 1992.

[28181] 1733. Shelley, C. S.; Carroll, M. C.; Davis, A. E., III; Bruns, G. A. P.; Whitehead, A. S.; Finberg, R.; Remold–O'Donnell, E.; Rosen, F. S.: Human sialophorin: cDNA cloning and gene location. (Abstract) *FASEBJ.* 2: A1659 only, 1988.

[28182] 1734. Shelley, C. S.; Remold–O'Donnell, E.; Davis, A. E., III; Bruns, G. A. P.; Rosen, F. S.; Carroll, M. C.; Whitehead, A. S.: Molecular characterization of sialophorin (CD43), the lymphocyte surface sialoglycoprotein defective in Wiskott–

Aldrich syndrome. *Proc. Nat. Acad. Sci.* 86:2819–2823, 1989.

[28183] 1735.Shelley, C. S.; Remold–O'Donnell, E.; Rosen, F. S.; Whitehead,A. S.: Structure of the human sialophorin (CD43) gene: identificationof features atypical of genes encoding integral membrane proteins. *Biochem.J.* 270: 569–576, 1990.

[28184] 1736.van den Berg, T. K.; Nath, D.; Ziltener, H. J.; Vestweber, D.;Fukuda, M.; van Die, I.; Crocker, P. R.: Cutting edge: CD43 functionsas a T cell counterreceptor for the macrophage adhesion receptor sialoadhesin(Siglec–1). *J. Immun.* 166: 3637–3640, 2001.

[28185] 1737.Crackower, M. A.; Scherer, S. W.; Rommens, J. M.; Hui, C.–C.; Poorkaj,P.; Soder, S.; Cobben, J. M.; Hudgins, L.; Evans, J. P.; Tsui, L.–C.: Characterization of the split hand/split foot malformation locusSHFM1 at 7q21.3–q22.1 and analysis of a candidate gene for its expressionduring limb development. *Hum. Molec. Genet.* 5: 571–579, 1996.

[28186] 1738.Tong, Q.; Xing, S.; Jhiang, S. M.: Leucine zipper-mediated dimerizationis essential for the PTC1 oncogenic activity. *J. Biol. Chem.* 272:9043–9047, 1997.

[28187] 1739.Vellucci, V. F.; Reiss, M.: Cloning and genomic orga–

nization of the human transforming growth factor- β type I receptor gene. *Genomics* 46:278–283, 1997.

- [28188] 1740. Allen, R. C.; Webster, A. R.; Sui, R.; Brown, J.; Taylor, C. M.; Stone, E. M.: Molecular characterization and ophthalmic investigation of a large family with type 2A von Hippel–Lindau disease. *Arch. Ophthalmol.* 119:1659–1665, 2001.
- [28189] 1741. Beebe, S. J.; Oyen, O.; Sandberg, M.; Froysa, A.; Hansson, V.; Jahnsen, T.: Molecular cloning of a tissue-specific protein kinase (C γ) from human testis—representing a third isoform for the catalytic subunit of cAMP-dependent protein kinase. *Molec. Endocr.* 4:465–475, 1990.
- [28190] 1742. Foss, K. B.; Berube, D.; Simard, J.; Beebe, S. J.; Sandberg, M.; Grzeschik, K.-H.; Gagne, R.; Hansson, V.; Jahnsen, T.: Localization of the catalytic subunit C- γ of cAMP-dependent protein kinase on human chromosome 9q13. (Abstract) *Cytogenet. Cell Genet.* 58:1937–1938, 1991.
- [28191] 1743. Foss, K. B.; Simard, J.; Berube, D.; Beebe, S. J.; Sandberg, M.; Grzeschik, K.-H.; Gagne, R.; Hansson, V.; Jahnsen, T.: Localization of the catalytic subunit C- γ of the cAMP-dependent protein kinase gene (PRKACG) to

human chromosome region 9q13. *Cytogenet. Cell Genet.* 60:22–25, 1992.

- [28192] 1744.Reinton, N.; Haugen, T. B.; Orstavik, S.; Skalhegg, B. S.; Hansson,V.; Jahnsen, T.; Tasken, K.: The gene encoding the C gamma catalyticsubunit of cAMP–dependent protein kinase is a transcribed retroposon. *Genomics*49: 290–297, 1998.
- [28193] 1745.Orstavik, S.; Natarajan, V.; Tasken, K.; Jahnsen, T.; Sandberg,M.: Characterization of the human gene encoding the type I–alphaand type I–beta cGMP–dependent protein kinase (PRKG1). *Genomics* 42:311–318, 1997.
- [28194] 1746.Orstavik, S.; Sandberg, M.; Berube, D.; Natarajan, V.; Simard,J.; Walter, U.; Gagne, R.; Hansson, V.; Jahnsen, T.: Localizationof the human gene for type I cyclic GMP–dependent protein kinase to chromosome 10. *Cytogenet. Cell Genet.* 59: 270–273, 1992.
- [28195] 1747.Osborne, K. A; Robichon, A.; Burgess, E.; Butland, S.; Shaw, R.A.; Coulthard, A.; Pereira, H. S.; Greenspan, R. J.; Sokolowski, M.B.: Natural behavior polymorphism due to a cGMP–dependent proteinkinase of *Drosophila*. *Science* 277: 834–836, 1997.
- [28196] 1748.Pfeifer, A.; Klatt, P.; Massberg, S.; Ny, L.; Sausbier, M.; Hirneill,C.; Wang, G.–X.; Korth, M.; Aszodi, A.; Anders–

son, K.-E.; Krombach, F.; Mayerhofer, A.; Ruth, P.; Fassler, R.; Hofmann, F.: Defective smooth muscle regulation in cGMP kinase I-deficient mice. *EMBO J.* 17:3045–3051, 1998.

[28197] 1749. Sandberg, M.; Natarajan, V.; Ronander, I.; Kalderon, D.; Walter, U.; Lohmann, S. M.; Jahnsen, T.: Molecular cloning and predicted full-length amino acid sequence of the type I beta isozyme of cGMP-dependent protein kinase from human placenta: tissue distribution and developmental changes in rat. *FEBS Lett.* 255: 321–329, 1989.

[28198] 1750. Tamura, N.; Itoh, H.; Ogawa, Y.; Nakagawa, O.; Harada, M.; Chun, T.-H.; Suga, S.; Yoshimasa, T.; Nakao, K.: cDNA cloning and gene expression of human type I- α cGMP-dependent protein kinase. *Hypertension* 27:552–557, 1996.

[28199] 1751. Oyen, O.; Myklebust, F.; Scott, J. D.; Hansson, V.; Jahnsen, T.: Human testis cDNA for the regulatory subunit RII α of cAMP-dependent protein kinase encodes an alternate amino-terminal region. *FEBS Lett.* 246:57–64, 1989.

[28200] 1752. Tasken, K.; Naylor, S. L.; Solberg, R.; Jahnsen, T.: Mapping of the gene encoding the regulatory subunit RII- α of cAMP-dependent protein kinase (locus PRKAR2A)

to human chromosome region 3p21.3–p21.2. *Genomics* 50:378–381, 1998.

[28201] 1753.Solberg, R.; Sistonen, P.; Traskelin, A.–L.; Berube, D.; Simard,J.; Krajci, P.; Jahnsen, T.; de la Chapelle, A.: Mapping of the regulatory subunits RI–beta and RII–beta of cAMP–dependent protein kinase genes on human chromosome 7. *Genomics* 14: 63–69, 1992.

[28202] 1754.Cummings, D. E.; Brandon, E. P.; Planas, J. V.; Motamed, K.; Idzerda,R. L.; McKnight, G. S.: Genetically lean mice result from targeted disruption of the RII–beta subunit of protein kinase A. *Nature* 382::622–626, 1996.

[28203] 1755.Scambler, P.; Oyen, O.; Wainwright, B.; Farrall, M.; Law, H.–Y.;Estivill, X.; Sandberg, M.; Williamson, R.; Jahnsen, T.: Exclusion of catalytic and regulatory subunits of cAMP–dependent protein kinase as candidate genes for the defect causing cystic fibrosis. *Am. J.Hum. Genet.* 41: 925–932, 1987.

[28204] 1756.Wainwright, B.; Lench, N.; Davies, K.; Scambler, P.; Kruyer, H.;Williamson, R.; Jahnsen, T.; Farrall, M.: A human regulatory subunit of type II cAMP–dependent protein kinase localized by its linkage relationship to several cloned chromosome 7q markers. *Cytogenet.Cell Genet.* 45: 237–239, 1987.

- [28205] 1757.Norman, S. A.; Mott, D. M.: Molecular cloning and chromosomal localization of a human skeletal muscle PP-1-gamma-1 cDNA. *MammalianGenome* 5: 41–45, 1994.
- [28206] 1758.McGrogan, M.; Kennedy, J.; Li, M. P.; Hsu, C.; Scott, R. W.; Simonsen, C. C.; Baker, J. B.: Molecular cloning and expression of two forms of human protease nexin I. *Bio/Technology* 6: 172–177, 1988.
- [28207] 1759.Sommer, J.; Gloor, S.; Rovelli, G. F.; Hofsteenge, J.; Nick, H.; Meier, R.; Monard, D.: cDNA sequence coding for a rat glia-derived nexin and its homology to members of the serpin family. *Biochemistry* 26:6407–6410, 1987.
- [28208] 1760.Sugawara, S.; Uehara, A.; Nochi, T.; Yamaguchi, T.; Ueda, H.; Sugiyama, A.; Hanzawa, K.; Kumagai, K.; Okamura, H.; Takada, H.: Neutrophil proteinase 3-mediated induction of bioactive IL-18 secretion by human oral epithelial cells. *J. Immun.* 167: 6568–6575, 2001.
- [28209] 1761.Akiyama, K.; Yokota, K.; Kagawa, S.; Shimbara, N.; Tamura, T.; Akioka, H.; Nothwang, H. G.; Noda, C.; Tanaka, K.; Ichihara, A.: cDNA cloning and interferon gamma down-regulation of proteasomal subunits X and Y. *Science* 265: 1231–1234, 1994.
- [28210] 1762.Driscoll, J.; Brown, M. G.; Finley, D.; Monaco, J. J.: MHC-linked LMP gene products specifically alter peptidase

activities of the proteasome. *Nature* 365:262–264, 1993.

[28211] 1763.Gaczynska, M.; Rock, K. L.; Goldberg, A. L.: Gamma-interferon and expression of MHC genes regulate peptide hydrolysis by proteasomes. *Nature* 365:264–267, 1993.

[28212] 1764.Glynne, R.; Powis, S. H.; Beck, S.; Kelly, A.; Kerr, L. A.; Trowsdale, J.: A proteasome-related gene between the two ABC transporter loci in the class II region of the human MHC. *Nature* 353: 357–360, 1991.

[28213] 1765.Milatovich, A.; Song, K.; Heller, R. A.; Francke, U.: Tumor necrosis factor receptor genes, TNFR1 and TNFR2, on human chromosomes 12 and 1. *Somat. Cell Molec. Genet.* 17: 519–523, 1991.

[28214] 1766.Schall, T. J.; Lewis, M.; Koller, K. J.; Lee, A.; Rice, G. C.; Wong, G. H. W.; Gatanaga, T.; Granger, G. A.; Lentz, R.; Raab, H.; Kohr, W. J.; Goeddel, D. V.: Molecular cloning and expression of a receptor for human tumor necrosis factor. *Cell* 61: 361–370, 1990.

[28215] 1767.Schievella, A. R.; Chen, J. H.; Graham, J. R.; Lin, L.-L.: MADD, a novel death domain protein that interacts with the type 1 tumor necrosis factor receptor and activates mitogen-activated protein kinase. *J. Biol. Chem.* 272: 12069–12075, 1997.

[28216] 1768.Beltinger, C. P.; White, P. S.; Maris, J. M.; Sulman, E.

P.; Jensen, S. J.; LePaslier, D.; Stallard, B. J.; Goeddel, D. V.; de Sauvage, F. J.; Brodeur, G. M.: Physical mapping and genomic structure of the human TNFR2 gene. *Genomics* 35: 94–100, 1996.

[28217] 1769. Kaufman, B. A.; White, P. S.; Steinbrueck, T.; Donis-Keller, H.; Brodeur, G. M.: Linkage mapping of the tumor necrosis factor receptor 2 (TNFR2) gene to 1p36.2 using the single-strand conformation polymorphism technique. *Hum. Genet.* 94: 418–422, 1994.

[28218] 1770. Kemper, O.; Derre, J.; Cherif, D.; Engelmann, H.; Wallach, D.; Berger, R.: The gene for the type II (p75) tumor necrosis factor receptor (TNF-RII) is localized on band 1p36.2–p36.3. *Hum. Genet.* 87: 623–624, 1991.

[28219] 1771. Li, X.; Yang, Y.; Ashwell, J. D.: TNF-RII and c-IAP1 mediate ubiquitination and degradation of TRAF2. *Nature* 416: 345–349, 2002.

[28220] 1772. Santee, S. M.; Owen-Schaub, L. B.: Human tumor necrosis factor receptor p75/80 (CD120b) gene structure and promoter characterization. *J. Biol. Chem.* 271: 21151–21159, 1996.

[28221] 1773. Mochizuki, N.; Yamashita, S.; Kurokawa, K.; Ohba, Y.; Nagai, T.; Miyawaki, A.; Matsuda, M.: Spatio-temporal images of growth-factor-induced activation of Ras and

Rap1. Nature 411: 1065–1068, 2001.

[28222] 1774.Hon, W.-C.; Wilson, M. I.; Harlos, K.; Claridge, T. D. W.; Schofield, C. J.; Pugh, C. W.; Maxwell, P. H.; Ratcliffe, P. J.; Stuart, D. I.; Jones, E. Y.: Structural basis for the recognition of hydroxyproline in HIF-1- α by pVHL. Nature 417: 975–978, 2002.

[28223] 1775.Lin, K.; Thomas, J. T.; McBride, O. W.; Luyten, F. P.: Assignment of a new TGF- β superfamily member, human cartilage-derived morphogenetic protein-1, to chromosome 20q11.2. Genomics 34: 150–151, 1996.

[28224] 1776.Williams, J. M.; Chen, G.-C.; Zhu, L.; Rest, R. F.: Using the yeast two-hybrid system to identify human epithelial cell proteins that bind gonococcal Opa proteins: intracellular gonococci bind pyruvate kinase via their Opa proteins and require host pyruvate for growth. Molec. Microbiol. 27: 171–186, 1998.

[28225] 1777.Ho, L.; Javed, A. A.; Pepin, R. A.; Thekkumkara, T. J.; Raefsky, C.; Mole, J. E.; Caliendo, A. M.; Kwon, M. S.; Kerr, D. S.; Patel, M. S.: Identification of a cDNA clone for the β -subunit of the pyruvate dehydrogenase component of human pyruvate dehydrogenase complex. Biochem. Biophys. Res. Commun. 150: 904–908, 1988.

[28226] 1778.Koike, K.; Ohta, S.; Urata, Y.; Kagawa, Y.; Koike, M.:

Cloning and sequencing of cDNAs encoding alpha and beta subunits of human pyruvate dehydrogenase. *Proc. Nat. Acad. Sci.* 85: 41–45, 1988.

[28227] 1779. Koike, K.; Urata, Y.; Koike, M.: Molecular cloning and characterization of human pyruvate dehydrogenase beta subunit gene. *Proc. Nat. Acad. Sci.* 87: 5594–5597, 1990.

[28228] 1780. Olson, S.; Song, B. J.; Huh, T.–L.; Chi, Y.–T.; Veech, R. L.; McBride, O. W.: Three genes for enzymes of the pyruvate dehydrogenase complex map to human chromosomes 3, 7, and X. *Am. J. Hum. Genet.* 46: 340–349, 1990.

[28229] 1781. Koken, M. H. M.; Smit, E. M. E.; Jaspers–Dekker, I.; Oostra, B. A.; Hagemeijer, A.; Bootsma, D.; Hoeijmakers, J. H. J.: Localization of two human homologs, HHR6A and HHR6B, of the yeast DNA repair gene RAD6 to chromosomes Xq24–q25 and 5q23–q31. *Genomics* 12: 447–453, 1992.

[28230] 1782. Lench, N. J.; Thompson, J.; Markham, A. F.; Robinson, P. A.: (CGG) trinucleotide repeat polymorphism in the 5–prime region of the HHR6B gene: the human homolog of the yeast DNA repair gene RAD6. *Hum. Genet.* 96: 369–370, 1995.

[28231] 1783. Roest, H. P.; van Klaveren, J.; de Wit, J.; van Gurp, C. G.; Koken, M. H. M.; Vermey, M.; van Roijen, J. H.; Hooger–

brugge, J. W.; Vreeburg, J. T. M.; Baarends, W. M.; Bootsma, D.; Grootegoed, J. A.; Hoeijmakers, J. H. J.: Inactivation of the HR6B ubiquitin-conjugating DNA repair enzyme in mice causes male sterility associated with chromatin modification. *Cell* 86:799–810, 1996.

[28232] 1784. Roller, M. L.; Lossie, A. C.; Koken, M. H. M.; Smit, E. M. E.; Hagemeijer, A.; Camper, S. A.: Localization of sequences related to the human RAD6 DNA repair gene on mouse chromosomes 11 and 13. *Mammalian Genome* 6: 305–306, 1995.

[28233] 1785. Scozzari, R.; Iodice, C.; Sellitto, D.; Brdicka, R.; Mura, G.; Santachiara-Benerecetti, A. S.: Population studies on human phosphoglucomutase-1 thermostability polymorphism. *Hum. Genet.* 68: 314–317, 1984.

[28234] 1786. Shinoda, T.; Matsunaga, E.: Polymorphism of red cell phosphoglucomutase among Japanese. *Jpn. J. Hum. Genet.* 14: 316–323, 1970.

[28235] 1787. Spencer, N.; Hopkinson, D. A.; Harris, H.: Phosphoglucomutase polymorphism in man. *Nature* 204: 742–745, 1964.

[28236] 1788. Takahashi, N.; Neel, J. V.: Intragenic recombination at the human phosphoglucomutase 1 locus: predictions fulfilled. *Proc. Nat. Acad. Sci.* 90: 10725–10729, 1993.

- [28237] 1789.Takahashi, N.; Neel, J. V.; Satoh, C.; Nishizaki, J.; Masunari,N.: A phylogeny for the principal alleles of the human phosphoglucomutase-1locus. *Proc. Nat. Acad. Sci.* 79: 6636–6640, 1982.
- [28238] 1790.Tchen, P.; Seger, J.; Bois, E.; Neel, J. V.: Is there a PGM(1)4allele specific to Amerindian populations? *Hum. Genet.* 53: 229–231,1980.
- [28239] 1791.Welch, S. G.; Swindlehurst, C. A.; McGregor, I. A.; Williams,K.: Isoelectric focusing of human red cell phosphoglucomutase: thedistribution of variant phenotypes in a village population from TheGambia, West Africa. *Hum. Genet.* 43: 307–313, 1978.
- [28240] 1792.Westerveld, A.; Bootsma, D.: Personal Communication. 1971.
- [28241] 1793.Whitehouse, D. B.; Putt, W.; Lovegrove, J. U.; Morrison, K.; Hollyoake,M.; Fox, M. F.; Hopkinson, D. A.; Edwards, Y. H.: Phosphoglucomutase1: complete human and rabbit mRNA sequences and direct mapping ofthis highly polymorphic marker on human chromosome 1. *Proc. Nat.Acad. Sci.* 89: 411–415, 1992.
- [28242] 1794.Yip, S. P.; Lovegrove, J. U.; Rana, N. A.; Hopkinson, D. A.; Whitehouse,D. B.: Mapping recombination hotspots in human phosphoglucomutase(PGM1). *Hum. Molec.*

Genet. 8: 1699–1706, 1999.

[28243] 1795.Baur, M. P.; Rittner, C.: Application of a computer program for the mapping of a gene locus to the disputed PGM-3 localization on human chromosome 6. Tissue Antigens 12: 341–349, 1978.

[28244] 1796.Burgess, R. M.; Sutton, J. G.: An improved method of typing hair sheath cells using the PGM-3 locus following starch gel electrophoresis. Hum.Genet. 56: 391–393, 1981.

[28245] 1797.Hopkinson, D. A.; Harris, H.: A third phosphoglucose mutase locus in man. Ann. Hum. Genet. 31: 359–368, 1968.

[28246] 1798.Jahannsmann, R.; Schwinger, E.; Grzeschik, K. H.: Assignment of the gene locus for human phosphoglucose mutase 3 to chromosome 6q12–qter. Ann.Genet. 23: 12–14, 1980.

[28247] 1799.Jongsma, A.; Van Someren, H.; Westerveld, A.; Hagemeijer, A.; Pearson, P. L.: Localization of genes on human chromosomes by studies of human–Chinese hamster somatic cell hybrids. Assignment of PGM to chromosome C6 and regional mapping of the PGD, PGM, and pep-C genes on chromosome A1. Humangenetik 20:195–202, 1973.

- [28248] 1800.Kompf, J.; Bissbort, S.; Gohler, F.; Schunter, F.; Wernet, P.:Mapping of the linkage group GLO-Bf-HLA-B, C, A-PGM. I. Recombinationfrequencies. Hum. Genet. 44: 313-319, 1978.
- [28249] 1801.Lamm, L. U.; Jorgensen, F.; Kissmeyer-Nielsen, F.: On the mappingof PGM-3 in relation to HLA. Tissue Antigens 17: 245-246, 1981.
- [28250] 1802.Lamm, L. U.; Kissmeyer-Nielsen, F.; Henningsen, K.: Linkage andassociation studies of two phosphoglucomutase loci (PGM-1 and PGM-3)to eighteen other markers. Hum. Hered. 20: 305-318, 1970.
- [28251] 1803.Li, C.; Rodriguez, M.; Banerjee, D.: Cloning and characterizationof complementary DNA encoding human N-acetylglucosamine-phosphatmutase protein. Gene 242: 97-103, 2000.
- [28252] 1804.McCaw, B. K.; Hecht, F.; Linder, D.; Lovrien, E. W.; Wyandt, H.;Bacon, D.; Clark, B.; Lea, N.: Ovarian teratomas: cytologic data. Cytogenet.Cell Genet. 16: 391-395, 1976.
- [28253] 1805.Mohandas, T.; Sparkes, R. S.; Shulkin, J. D.; Sparkes, M. C.;Moedjono, S.: Assignment of PGM3 to the long arm of human chromosome6: studies using Chinese hamster-human cell hybrids containing a human6-15 transloca-

tion. Cytogenet. Cell Genet. 28: 116–120, 1980.

- [28254] 1806.Nadeau, J. H.; Kompf, J.; Siebert, G.; Taylor, B. A.: Linkage of Pgm-3 in the house mouse and homologies of three phosphoglucomutase loci in mouse and man. Biochem. Genet. 19: 465–474, 1981.
- [28255] 1807.Pang, H.; Koda, Y.; Soejima, M.; Kimura, H.: Identification of human phosphoglucomutase 3 (PGM3) as N-acetylglucosamine-phosphate mutase (AGM1). Ann. Hum. Genet. 66: 139–144, 2002.
- [28256] 1808.Parrington, J.; West, L.; Povey, S.: Gene mapping from ovarian teratomas. (Abstract) Cytogenet. Cell Genet. 25: 196 only, 1979.
- [28257] 1809.Van Someren, H.; Van Henegouwen, H. B.; Los, W.; Wurzer-Figurelli, E.; Doppert, B.; Vervloet, M.; Meera Khan, P.: Enzyme electrophoresis on cellulose acetate gel: zymogram patterns in man–Chinese hamster somatic cell hybrids. Humangenetik 25: 189–201, 1974.
- [28258] 1810.Cmarik, J. L.; Hegamyer, G.; Gerrard, B.; Dean, M.; Colburn, N.H.: cDNA cloning and mapping of mouse pleckstrin (Plek), a gene upregulated in transformation-resistant cells. Genomics 66: 204–212, 2000.
- [28259] 1811.Tyers, M.; Haslam, R. J.; Rachubinski, R. A.; Harley, C. B.: Molecular analysis of pleckstrin: the major protein

kinase C substrate of platelets. *J.Cell. Biochem.* 40: 133–145, 1989.

[28260] 1812.Tyers, M.; Rachubinski, R. A.; Stewart, M. I.; Varri-
chio, A. M.;Shorr, R. G. L.; Haslam, R. J.; Harley, C. B.:
Molecular cloningand expression of the major protein ki-
nase C substrate of platelets. *Nature* 333:470–473, 1988.

[28261] 1813.Tobe, T.; Minoshima, S.; Yamase, S.; Choi, N.-H.;
Tomita, M.;Shimizu, N.: Assignment of a human serum
glycoprotein SP-40,40 gene(CLI) to chromosome 8. *Cyto-
genet. Cell Genet.* 57: 193–195, 1991.

[28262] 1814.Wong, P.; Pineault, J.; Lakins, J.; Taillefer, D.; Leger,
J.;Wang, C.; Tenniswood, M.: Genomic organization and
expression ofthe rat TRPM-2 (clusterin) gene, a gene im-
plicated in apoptosis. *J.Biol. Chem.* 268: 5021–5031,
1993.

[28263] 1815.Wong, P.; Taillefer, D.; Lakins, J.; Pineault, J.; Chader,
G.;Tenniswood, M.: Molecular characterization of human
TRPM-2/clusterin,a gene associated with sperm matura-
tion, apoptosis and neurodegeneration. *Europ.J. Biochem.*
221: 917–925, 1994.

[28264] 1816.Astuti, D.; Latif, F.; Dallol, A.; Dahia, P. L. M.; Dou-
glas, F.;George, E.; Skoldberg, F.; Husebye, E. S.; Eng, C.;
Maher, E. R.:Gene mutations in the succinate dehydroge-

nase subunit SDHB cause susceptibility to familial pheochromocytoma and to familial paraganglioma. *Am. J. Hum. Genet.* 69: 49–54, 2001.

- [28265] 1817. Au, H. C.; Ream–Robinson, D.; Bellew, L. A.; Broomfield, P. L. E.; Saghbini, M.; Scheffler, I. E.: Structural organization of the gene encoding the human iron–sulfur subunit of succinate dehydrogenase. *Gene* 159:249–253, 1995.
- [28266] 1818. Kita, K.; Oya, H.; Gennis, R. B.; Ackrell, B. A. C.; Kasahara, M.: Human complex II (succinate–ubiquinone oxidoreductase): cDNA cloning of iron sulphur (Ip) subunit of liver mitochondria. *Biochem. Biophys. Res. Commun.* 166: 101–108, 1990.
- [28267] 1819. Leckschat, S.; Ream–Robinson, D.; Scheffler, I. E.: The gene for the iron sulfur protein of succinate dehydrogenase (SDH–IP) maps to human chromosome 1p35–36.1. *Somat. Cell Molec. Genet.* 19: 505–511, 1993.
- [28268] 1820. Mascarello, J. T.; Soderberg, K.; Scheffler, I. E.: Assignment of a gene for succinate dehydrogenase to human chromosome 1 by somatic cell hybridization. *Cytogenet. Cell Genet.* 28: 121–135, 1980.
- [28269] 1821. Oostveen, F. G.; Au, H. C.; Meijer, P.–J.; Scheffler, I. E.: A Chinese hamster mutant cell line with a defect in the

integral membraneprotein C-II-3 of complex II of the mitochondrial electron transportchain. J. Biol. Chem. 270: 26104-26108, 1995.

[28270] 1822.Seilhamer, J. J.; Randall, T. L.; Johnson, L. K.; Heinzmann, C.;Klisak, I.; Sparkes, R. S.; Lusi, A. J.: Novel gene exon homologousto pancreatic phospholipase A(2): sequence and chromosomal mappingof both human genes. J. Cell. Biochem. 39: 327-337, 1989.

[28271] 1823.Seilhamer, J. J.; Randall, T. L.; Johnson, L. K.; Lusi, A.; Sparkes,R. S.; Heinzman, C.: Chromosomal mapping of human pancreatic PLA2and a homologous PLA2 exon. (Abstract) J. Cell. Biochem. Suppl.12E: 55 only, 1988.

[28272] 1824.Seilhamer, J. J.; Randall, T. L.; Yamanaka, M.; Johnson, L. K.: Pancreatic phospholipase A(2): isolation of the human gene and cDNAsfrom porcine pancreas and human lung. DNA 5: 519-527, 1986.

[28273] 1825.Sparkes, R. S.; Mohandas, T.; Heinzmann, C.; Seilhamer, J. J.;Lusi, A. J.; Johnson, L. K.: Assignment of the gene for pancreaticphospholipase A2 to human chromosome 12. (Abstract) Cytogenet. CellGenet. 46: 697, 1987.

[28274] 1826.van Kuijk, F. J. G. M.; Sevanian, A.; Handelman, G. J.; Dratz,E. A.: A new role for phospholipase A(2): protection of membranesfrom lipid peroxidation damage. Trends

Biochem. Sci. 12: 31–34,1987.

- [28275] 1827.Cormier, R. T.; Hong, K. H.; Halberg, R. B.; Hawkins, T. L.; Richardson,P.; Mulherkar, R.; Dove, W. F.; Lander, E. S.: Secretory phospholipasePla2g2a confers resistance to intestinal tumorigenesis. Nature Genet. 17:88–91, 1997.
- [28276] 1828.Dennis, E. A.: Diversity of group types, regulation, and functionof phospholipase A2. J. Biol. Chem. 269: 13057–13060, 1994.
- [28277] 1829.Dietrich, W. F.; Lander, E. S.; Smith, J. S.; Moser, A. R.; Gould,K. A.; Luongo, C.; Borenstein, N.; Dove, W.: Genetic identificationof Mom–1, a major modifier locus affecting Min–induced intestinalneoplasia in the mouse. Cell 75: 631–639, 1993.
- [28278] 1830.Dobbie, Z.; Heinimann, K.; Bishop, D. T.; Muller, H.; Scott, R.J.: Identification of a modifier gene locus on chromosome 1p35–36in familial adenomatous polyposis. Hum. Genet. 99: 653–657, 1997.
- [28279] 1831.Dragani, T. A.; Manenti, G.: Mom1 leads the pack. Nature Genet. 17:7–8, 1997.
- [28280] 1832.Haapamaki, M. M.; Gronroos, J. M.; Nurmi, H.; Alanen, K.; Kallajoki,M.; Nevalainen, T. J.: Gene expression of group II phospholipaseA2 in intestine in ulcerative colitis. Gut 40: 95–101, 1997.

- [28281] 1833.Johnson, L. K.; Frank, S.; Vades, P.; Pruzanski, W.; Lusi, A.J.; Seilhamer, J. J.: Localization and evolution of two human phospholipaseA2 genes and two related genetic elements.In: Wong, P. Y.-K.; Dennis,E. A.: Phospholipase A2. New York: Plenum Press (pub.) 1990.Pp. 17-34.
- [28282] 1834.Johnson, L. K.; Seilhamer, J. J.; Frank, S.; Lusi, A.; Vadas,P.; Pruzanski, W.: Synovial fluid phospholipase A(2): chromosomalco-localization with homologous genes may provide disease-relatedPLA(2) diversity. (Abstract) Arthritis Rheum. 33 (suppl.): S79,1990.
- [28283] 1835.MacPhee, M.; Chepenik, K. P.; Liddell, R. A.; Nelson, K. K.; Siracusa,L. D.; Buchberg, A. M.: The secretory phospholipase A2 gene is acandidate for the Mom1 locus, a major modifier of Apc(Min)-inducedintestinal neoplasia. Cell 81: 957-966, 1995.
- [28284] 1836.Masharani, U.; Coleman, R. T.; Johnson, L. K.; Seilhamer, J. J.: EcoRI and NsiI RFLPs at a human PLA2 gene on chromosome 1. NucleicAcids Res. 16: 9073, 1988.
- [28285] 1837.Moser, A. R.; Dove, W. F.; Roth, K. A.; Gordon, J. I.: The Min(multiple intestinal neoplasia) mutation: its effect on gut epithelialcell differentiation and interaction with a modifier system. J. CellBiol. 116: 1517-1526, 1992.
- [28286] 1838.Moser, A. R.; Mattes, E. M.; Dove, W. F.; Lindstrom,

M. J.; Haag, J. D.; Gould, M. N.: Apc(Min), a mutation in the murine Apc gene, predisposes to mammary carcinomas and focal alveolar hyperplasias. Proc. Nat. Acad. Sci. 90: 8977–8981, 1993.

[28287] 1839. Moser, A. R.; Pitot, H. C.; Dove, W. F.: A dominant mutation that predisposes to multiple intestinal neoplasia in the mouse. Science 247:322–324, 1990.

[28288] 1840. Nadeau, J. H.: Modifier genes in mice and humans. Nature Rev. 2:165–174, 2001.

[28289] 1841. Nimmrich, I.; Friedl, W.; Kruse, R.; Pietsch, S.; Hentsch, S.; Deuter, R.; Winde, G.; Muller, O.: Loss of the PLA2G2A gene in a sporadic colorectal tumor of a patient with a PLA2G2A germline mutation and absence of PLA2G2A germline alterations in patients with FAP. Hum. Genet. 100: 345–349, 1997.

[28290] 1842. Seilhamer, J. J.; Plant, S.; Pruzanski, W.; Schilling, J.; Stefanski, E.; Vadas, P.; Johnson, L. K.: Multiple forms of phospholipase A(2) in arthritic synovial fluid. J. Biochem. 106: 38–42, 1989.

[28291] 1843. Seilhamer, J. J.; Pruzanski, W.; Vadas, P.; Plant, S.; Miller, J. A.; Kloss, J.; Johnson, L. K.: Cloning and recombinant expression of phospholipase A(2) present in rheumatoid arthritic synovial fluid. J. Biol. Chem. 264: 5335–5338,

1989.

- [28292] 1844.Seilhamer, J. J.; Randall, T. L.; Johnson, L. K.; Lusi, A.; Sparkes, R. S.; Heinzman, C.: Chromosomal mapping of human pancreatic PLA₂ and a homologous PLA₂ exon. (Abstract) J. Cell. Biochem. (Suppl.12E): 55 only, 1988.
- [28293] 1845.Su, L.-K.; Kinzler, K. W.; Vogelstein, B.; Preisinger, A. C.; Moser, A. R.; Luongo, C.; Gould, K. A.; Dove, W. F.: Multiple intestinal neoplasia caused by a mutation in the murine homolog of the APC gene. Science 256:668–670, 1992.
- [28294] 1846.Bristol, A.; Hall, S. M.; Kriz, R. W.; Stahl, M. L.; Fan, Y. S.; Byers, M. G.; Eddy, R. L.; Shows, T. B.; Knopf, J. L.: Phospholipase C-148: chromosomal location and deletion mapping of functional domains. ColdSpring Harbor Symp. Quant. Biol. 53: 915–920, 1988.
- [28295] 1847.Nelson, K. K.; Knopf, J. L.; Siracusa, L. D.: Localization of phospholipase C-γ 1 to mouse chromosome 2. Mammalian Genome 3:597–600, 1992.
- [28296] 1848.Rothschild, C. B.; Akots, G.; Fajans, S. S.; Bowden, D. W.: Amicrosatellite polymorphism associated with the PLC1 (phospholipase C) locus: identification, mapping, and linkage to the MODY locus on chromosome 20. Genomics 13: 560–564, 1992.

- [28297] 1849.Stahl, M. L.; Ferenz, C. R.; Kelleher, K. L.; Kriz, R. W.; Knopf,J. L.: Sequence similarity of phospholipase C with the non-catalyticregion of src. *Nature* 332: 269–272, 1988.
- [28298] 1850.Phillips, S. A.; Barr, V. A.; Haft, D. H.; Taylor, S. I.; Haft,C. R.: Identification and characterization of SNX15, a novel sortingnexin involved in protein trafficking. *J. Biol. Chem.* 276: 5074–5084,2001.
- [28299] 1851.Roberts, W. M.; Look, A. T.; Ruossel, M. F.; Sherr, C. J.: Tandemlinkage of human CSF–1 receptor (c-fms) and PDGF receptor genes. *Cell* 55:655–661, 1988.
- [28300] 1852.Smith, E. A.; Seldin, M. F.; Martinez, L.; Watson, M. L.; GhoshChoudhury, G.; Lalley, P. A.; Pierce, J.; Aaronson, S.; Barker, J.;Naylor, S. L.; Sakaguchi, A. Y.: Mouse platelet–derived growth factorreceptor alpha gene is deleted in W–19H and patch mutations on chromosome5. *Proc. Nat. Acad. Sci.* 88: 4811–4815, 1991.
- [28301] 1853.Stenman, G.; Eriksson, A.; Claesson–Welsh, L.: Human PDGFA receptorgene maps to the same region on chromosome 4 as the KIT oncogene. *GenesChromosomes Cancer* 1: 155–158, 1989.
- [28302] 1854.Stephenson, D. A.; Mercola, M.; Anderson, E.; Wang, C.; Stiles,C. D.; Bowen–Pope, D. F.; Chapman, V. M.:

Platelet-derived growthfactor receptor alpha-subunit gene (Pdgfra) is deleted in the mousepatch (Ph) mutation. Proc. Nat. Acad. Sci. 88: 6–10, 1991.

- [28303] 1855.Xie, J.; Aszterbaum, M.; Zhang, X.; Bonifas, J. M.; Zachary, C.;Epstein, E.; McCormick, F.: A role of PDGFR-alpha in basal cell carcinomaproliferation. Proc. Nat. Acad. Sci. 98: 9255–9259, 2001.
- [28304] 1856.Berube, D.; Luu The, V.; Lachance, Y.; Gagne, R.; Labrie, F.:Assignment of the human 3 beta-hydroxysteroid dehydrogenase gene (HSDB3)to the p13 band of chromosome 1. Cytogenet. Cell Genet. 52: 199–200,1989.
- [28305] 1857.Nekrep, N.; Jabrane-Ferrat, N.; Wolf, H. M.; Eibl, M. M.; Geyer,M.; Peterlin, B. M.: Mutation in a winged-helix DNA-binding motifcauses atypical bare lymphocyte syndrome. Nature Immun. 30Sept,2002. Note: Advance Electronic Publication.
- [28306] 1858.Spencer, S.; Dowbenko, D.; Cheng, J.; Li, W.; Brush, J.; Utzig,S.; Simanis, V.; Lasky, L. A.: PSTPIP: a tyrosine phosphorylatedcleavage furrow-associated protein that is a substrate for PEST tyrosinephosphatase. J. Cell Biol. 138: 845–860, 1997.
- [28307] 1859.Yokota, J.; Tsunetsugu-Yokota, Y.; Battifora, H.; Le

Fevre, C.;Cline, M. J.: Alterations of myc, myb, and ras(Ha) proto-oncogenes in cancers are frequent and show clinical correlation. Science 231:261–265, 1986.

[28308] 1860.Ahmed, Z. M.; Riazuddin, S.; Bernstein, S. L.; Ahmed, Z.; Khan,S.; Griffith, A. J.; Morell, R. J.; Friedman, T. B.; Riazuddin, S.;Wilcox, E. R.: Mutations of the protocadherin gene PCDH15 cause Ushersyndrome type 1F. Am. J. Hum. Genet. 69: 25–34, 2001.

[28309] 1861.Falkenberg, M.; Tom, C.; DeYoung, M. B.; Wen, S.; Linnemann, R.;Dichek, D. A.: Increased expression of urokinase during atherosclerotic lesion development causes arterial constriction and lumen loss, and accelerates lesion growth. Proc. Nat. Acad. Sci. 99: 10665–10670,2002.

[28310] 1862.Lijnen, H. R.; Van Hoef, B.; Nelles, L.; Holmes, W. E.; Collen,D.: Enzymatic properties of single-chain and two-chain forms of alys(158)–to–glu(158) mutant of urokinase-type plasminogen activator. Europ.J. Biochem. 172: 185–188, 1988.

[28311] 1863.Nagai, M.; Hiramatsu, R.; Kaneda, T.; Hayasuke, N.; Arimura, H.;Nishida, M.; Suyama, T.: Molecular cloning of cDNA coding for humanpreprourokinase. Gene 36: 183–188, 1985.

- [28312] 1864.Nelles, L.; Lijnen, H. R.; Collen, D.; Holmes, W. E.: Characterization of recombinant human single chain urokinase-type plasminogen activator mutants produced by site-specific mutagenesis of lysine 158. *J. Biol.Chem.* 262: 5682–5689, 1987.
- [28313] 1865.Riccio, A.; Grimaldi, G.; Verde, P.; Sebastio, G.; Boast, S.; Blasi, F.: The human urokinase-plasminogen activator gene and its promoter. *NucleicAcids Res.* 13: 1759–2771, 1985.
- [28314] 1866.Salerno, G.; Verde, P.; Nolli, M. L.; Corti, A.; Szots, H.; Meo, T.; Johnson, J.; Bullock, S.; Cassani, G.; Blasi, F.: Monoclonal antibodies to human urokinase identify the single-chain pro-urokinase precursor. *Proc. Nat. Acad. Sci.* 81: 110–114, 1984.
- [28315] 1867.Tripputi, P.; Blasi, F.; Verde, P.; Cannizzaro, L. A.; Emanuel, B. S.; Croce, C. M.: Human urokinase gene is located on the long arm of chromosome 10. *Proc. Nat. Acad. Sci.* 82: 4448–4452, 1985.
- [28316] 1868.Deng, Z.; Johnson, K.; Engleward, B. P.; Lane, S.; Callen, D. F.; Samson, L. D.; Davisson, M. T.; Siciliano, M. J.: New regions of conserved synteny and linkage between human chromosome 16p12–p13 and mouse chromosomes 16 and 11. (Abstract) *Cytogenet. Cell Genet.* 68:180 only,

1995.

- [28317] 1869.Fukuoka, S.-I.; Matsuda, Y.: Assignment of the Tamm-Horsfall protein/uromodulingene (Umod) to mouse chromosome bands 7F1-F2 and rat chromosome bands 1q36-q37 by in situ hybridization. *Cytogenet. Cell Genet.* 79: 241-242, 1997.
- [28318] 1870.Jeanpierre, C.; Whitmore, S. A.; Austruy, E.; Cohen-Salmon, M.; Callen, D. F.; Junien, C.: Chromosomal assignment of the uromodulingene (UMOD) to 16p13.11. *Cytogenet. Cell Genet.* 62: 185-187, 1993.
- [28319] 1871.Muchmore, A. V.; Decker, J. M.: Uromodulin: a unique 85-kilodalton immunosuppressive glycoprotein isolated from urine of pregnant women. *Science* 229:479-481, 1985.
- [28320] 1872.Pennica, D.; Kohr, W. J.; Kuang, W.-J.; Glaister, D.; Aggarwal, B. B.; Chen, E. Y.; Goeddel, D. V.: Identification of human uromodulin as the Tamm-Horsfall urinary glycoprotein. *Science* 236: 83-88, 1987.
- [28321] 1873.Pook, M. A.; Jeremiah, S.; Scheinman, S. J.; Povey, S.; Thakker, R. V.: Localization of the Tamm-Horsfall glycoprotein (uromodulin) gene to chromosome 16p12.3-16p13.11. *Ann. Hum. Genet.* 57: 285-290, 1993.
- [28322] 1874.Tamm, I.; Horsfall, F. L., Jr.: A mucoprotein derived

from human urine which reacts with influenza, mumps, and Newcastle disease viruses. *J. Exp. Med.* 95: 71–97, 1952.

[28323] 1875. Hoffman, H. M.; Mueller, J. L.; Broide, D. H.; Wanderer, A. A.; Kolodner, R. D.: Mutation of a new gene encoding a putative pyrin-like protein causes familial cold autoinflammatory syndrome and Muckle-Wells syndrome. *Nature Genet.* 29: 301–305, 2001.

[28324] 1876. Leibiger, B.; Leibiger, I. B.; Moede, T.; Kemper, S.; Kulkarni, R. N.; Kahn, C. R.; de Vargas, L. M.; Berggren, P.-O.: Selective insulin signaling through A and B insulin receptors regulates transcription of insulin and glucokinase genes in pancreatic beta cells. *Molec. Cell* 7: 559–570, 2001.

[28325] 1877. Gervais, F. G.; Xu, D.; Robertson, G. S.; Vaillancourt, J. P.; Zhu, Y.; Huang, J.; LeBlanc, A.; Smith, D.; Rigby, M.; Shearman, M. S.; Clarke, E. E.; Zheng, H.; Van Der Ploeg, L. H. T.; Ruffolo, S. C.; Thornberry, N. A.; Xanthoudakis, S.; Zamboni, R. J.; Roy, S.; Nicholson, D. W.: Involvement of caspases in proteolytic cleavage of Alzheimer's amyloid-beta precursor protein and amyloidogenic A-beta peptide formation. *Cell* 97: 395–406, 1999.

[28326] 1878. Gotz, J.; Chen, F.; van Dorpe, J.; Nitsch, R. M.: For-

mation of neurofibrillary tangles in P301L tau transgenic mice induced by A-beta42 fibrils. *Science* 293: 1491–1495, 2001.

- [28327] 1879. Abonia, J. P.; Abel, K. J.; Eddy, R. L.; Elliott, R. W.; Chapman, V. M.; Shows, T. B.; Gross, K. W.: Linkage of Agt and Actsk-1 to distal mouse chromosome 8 loci: a new conserved linkage. *Mammalian Genome* 4: 25–32, 1993.
- [28328] 1880. Borrow, J.; Shearman, A. M.; Stanton, V. P., Jr.; Becher, R.; Collins, T.; Williams, A. J.; Dube, I.; Katz, F.; Kwong, Y. L.; Morris, C.; Ohyashiki, K.; Toyama, K.; Rowley, J.; Housman, D. E.: The t(7;11)(p15;p15) translocation in acute myeloid leukaemia fuses the genes for nucleoporin-NUP98 and class I homeoprotein HOXA9. *Nature Genet.* 12: 159–167, 1996.
- [28329] 1881. Berry, R.; Stevens, T. J.; Walter, N. A. R.; Wilcox, A. S.; Rubano, T.; Hopkins, J. A.; Weber, J.; Goold, R.; Soares, M. B.; Sikela, J. M.: Gene-based sequence-tagged-sites (STSs) as the basis for a human gene map. *Nature Genet.* 10: 415–423, 1995.
- [28330] 1882. Renouf, S.; Beullens, M.; Wera, S.; Van Eynde, A.; Sikela, J.; Stalmans, W.; Bollen, M.: Molecular cloning of a human polypeptide related to yeast sds22, a regulator of protein phosphatase-1. *FEBS Lett.* 375: 75–78, 1995.

- [28331] 1883.Hatzfeld, M.; Kristjansson, G. I.; Plessmann, U.; Weber, K.: Band6 protein, a major constituent of desmosomes from stratified epithelia, is a novel member of the armadillo multigene family. *J. Cell Sci.* 107:2259–2270, 1994.
- [28332] 1884.Heid, H. W.; Schmidt, A.; Zimbelmann, R.; Schafer, S.; Winter-Simanowski, S.; Stumpp, S.; Keith, M.; Figge, U.; Schnolzer, M.; Franke, W. W.: Cell type-specific desmosomal plaque proteins of the plakoglobin family: plakophilin 1 (band 6 protein). *Differentiation* 58: 113–131, 1994.
- [28333] 1885.McGrath, J. A.; McMillan, J. R.; Shemanko, C. S.; Runswick, S.K.; Leigh, I. M.; Lane, E. B.; Garrod, D. R.; Eady, R. A. J.: Mutations in the plakophilin 1 gene result in ectodermal dysplasia/skin fragility syndrome. *Nature Genet.* 17: 240–244, 1997.
- [28334] 1886.Schmidt, A.; Langbein, L.; Pratzel, S.; Rode, M.; Rackwitz, H.-R.; Franke, W. W.: Plakophilin 3—a novel cell-type-specific desmosomal plaque protein. *Differentiation* 64: 291–306, 1999.
- [28335] 1887.Schmidt, A.; Langbein, L.; Rode, M.; Pratzel, S.; Zimbelmann, R.; Franke, W. W.: Plakophilins 1a and 1b: widespread nuclear proteins recruited in specific epithelial cells as desmosomal plaque components. *Cell Tissue Res.*

290: 481–499, 1997.

- [28336] 1888. Whittock, N. V.; Haftek, M.; Angoulvant, N.; Wolf, F.; Perrot, H.; Eady, R. A. J.; McGrath, J. A.: Genomic amplification of the human plakophilin 1 gene and detection of a new mutation in ectodermal dysplasia/skin fragility syndrome. *J. Invest. Derm.* 115: 368–374, 2000.
- [28337] 1889. Aburatani, H.; Hippo, Y.; Ishida, T.; Takashima, R.; Matsuba, C.; Kodama, T.; Takao, M.; Yasui, A.; Yamamoto, K.; Asano, M.; Fukasawa, K.; Yoshinari, T.; Inoue, H.; Ohtsuka, E.; Nishimura, S.: Cloning and characterization of mammalian 8-hydroxyguanine-specific DNA glycosylase/apurinic, apyrimidinic lyase, a functional mutM homologue. *Cancer Res.* 57: 2151–2156, 1997.
- [28338] 1890. Arai, K.; Morishita, K.; Shinmura, K.; Kohno, T.; Kim, S.-R.; Nohmi, T.; Taniwaki, M.; Ohwada, S.; Yokota, J.: Cloning of a human homolog of the yeast OGG1 gene that is involved in the repair of oxidative DNA damage. *Oncogene* 14: 2857–2861, 1997.
- [28339] 1891. Audebert, M.; Chevillard, S.; Levalois, C.; Gyapay, G.; Vieillefond, A.; Klijanienko, J.; Vielh, P.; El Naggar, A. K.; Oudard, S.; Boiteux, S.; Radicella, J. P.: Alterations of the DNA repair gene OGG1 in human clear cell carcinomas of the kidney. *Cancer Res.* 60: 4740–4744, 2000.

- [28340] 1892.Bjoras, M.; Luna, L.; Johnsen, B.; Hoff, E.; Haug, T.; Rognes,T.; Seeberg, E.: Opposite base-dependent reactions of a human baseexcision repair enzyme on DNA containing 7,8-dihydro-8-oxoguanineand abasic sites. EMBO J. 16: 6314-6322, 1997.
- [28341] 1893.Ishida, T.; Hippo, Y.; Nakahori, Y.; Matsushita, I.; Kodama, T.;Nishimura, S.; Aburatani, H.: Structure and chromosome location ofhuman OGG1. Cytogenet. Cell Genet. 85: 232-236, 1999.
- [28342] 1894.Kohno, T.; Shinmura, K.; Tosaka, M.; Tani, M.; Kim, S.-R.; Sugimura,H.; Nohmi, T.; Kasai, H.; Yokota, J.: Genetic polymorphisms and alternativesplicing of the hOGG1 gene, that is involved in the repair of 8-hydroxyguaninein damaged DNA. Oncogene 16: 3219-3225, 1998.
- [28343] 1895.Kuo, F. C.; Sklar, J.: Augmented expression of a human gene for8-oxoguanine DNA glycosylase (MutM) in B lymphocytes of the dark zonein lymph node germinal centers. J. Exp. Med. 186: 1547-1556, 1997.
- [28344] 1896.Lu, R.; Nash, H. M.; Verdine, G. L.: A mammalian DNA repair enzymethat excises oxidatively damaged guanines maps to a locus frequentlylost in lung cancer. Curr. Biol. 7: 397-407, 1997.
- [28345] 1897.Radicella, J. P.; Dherin, C.; Desmaze, C.; Fox, M. S.;

Boiteux,S.: Cloning and characterization of hOGG1, a human homolog of theOGG1 gene of *Saccharomyces cerevisiae*. *Proc. Nat. Acad. Sci.* 94:8010–8015, 1997.

[28346] 1898.Guo, S.; Yamaguchi, Y.; Schilbach, S.; Wada, T.; Lee, J.; Goddard,A.; French, D.; Handa, H.; Rosenthal, A.: A regulator of transcriptionalelongation controls vertebrate neuronal development. *Nature* 408:366–369, 2000.

[28347] 1899.DiLeone, R. J.; King, J. A.; Storm, E. E.; Copeland, N. G.; Jenkins,N. A.; Kingsley, D. M.: The Bmp8 gene is expressed in developingskeletal tissue and maps near the achondroplasia locus on mouse chromosome4. *Genomics* 40: 196–198, 1997.

[28348] 1900.Burtis, K. C.; Baker, B. S.: *Drosophila* doublesex gene controlssomatic sexual differentiation by producing alternatively splicedmRNAs encoding related sex-specific polypeptides. *Cell* 56: 997–1010,1989.

[28349] 1901.Arai, Y.; Hosoda, F.; Nakayama, K.; Ohki, M.: A yeast artificialchromosome contig and NotI restriction map that spans the tumor suppressorgene(s) locus, 11q.22.2–q23.3. *Genomics* 35: 196–206, 1996.

[28350] 1902.Hemmer, S.; Wasenius, V.–M.; Haglund, C.; Zhu, Y.; Knuutila, S.;Franssila, K.; Joensuu, H.: Alterations in the suppressor gene PPP2R1Bin parathyroid hyperplasias and

adenomas. *Cancer Genet. Cytogenet.* 134:13–17, 2002.

[28351] 1903. Walter, G.; Mumby, M.: Protein serine/threonine phosphatases and cell transformation. *Biochim. Biophys. Acta* 1155: 207–226, 1993.

[28352] 1904. Wang, S. S.; Esplin, E. D.; Li, J. L.; Huang, L.; Gazdar, A.; Minna, J.; Evans, G. A.: Alterations of the PPP2R1B gene in human lung and colon cancer. *Science* 282: 284–287, 1998.

[28353] 1905. Moog-Lutz, C.; Bouillet, P.; Regnier, C. H.; Tomasetto, C.; Mattei, M. G.; Chenard, M. P.; Anglard, P.; Rio, M. C.; Basset, P.: Comparative expression of the psoriasin (S100A7) and S100C genes in breast carcinoma and co-localization to human chromosome 1q21–q22. *Int. J. Cancer* 63:297–303, 1995.

[28354] 1906. Tanaka, M.; Adzuma, K.; Iwami, M.; Yoshimoto, K.; Monden, Y.; Itakura, M.: Human calgizzarin: one colorectal cancer-related gene selected by a large scale random cDNA sequencing and northern blot analysis. *Cancer Lett.* 89: 195–200, 1995.

[28355] 1907. Todoroki, H.; Kobayashi, R.; Watanabe, M.; Minami, H.; Hidaka, H.: Purification, characterization, and partial sequence analysis of a newly identified EF-hand type 13-kDa Ca(2+)-binding protein from smooth muscle and

non-muscle tissues. J. Biol. Chem. 266:
18668–18673, 1991.

- [28356] 1908. Watanabe, M.; Ando, Y.; Todoroki, H.; Minami, H.; Hidaka, H.: Molecular cloning and sequencing of a cDNA clone encoding a new calcium-binding protein, named calgizzarin, from rabbit lung. Biochem. Biophys. Res. Commun. 181: 644–649, 1991.
- [28357] 1909. Abdelhaleem, M. M.; Hameed, S.; Klassen, D.; Greenberg, A. H.: Leukophysin: an RNA helicase A-related molecule identified in cytotoxic T cell granules and vesicles. J. Immun. 156: 2026–2035, 1996.
- [28358] 1910. Lee, C.-G.; Eki, T.; Okumura, K.; da Costa Soares, V.; Hurwitz, J.: Molecular analysis of the cDNA and genomic DNA encoding mouse RNA helicase A. Genomics 47: 365–371, 1998.
- [28359] 1911. Lee, C.-G.; Hurwitz, J.: Human RNA helicase A is homologous to the maleless protein of Drosophila. J. Biol. Chem. 268: 16822–16830, 1993.
- [28360] 1912. Lee, C. G.; Hurwitz, J.: A new RNA helicase isolated from HeLa cells that catalytically translocates in the 3-prime to 5-prime direction. J. Biol. Chem. 267: 4398–4407, 1992.
- [28361] 1913. Zhang, S.; Grosse, F.: Domain structure of human

nuclear DNA helicase II (RNA helicase A). J. Biol. Chem. 272: 11487–11494, 1997.

[28362] 1914. Zhang, S.; Maacke, H.; Grosse, F.: Molecular cloning of the gene encoding nuclear DNA helicase II: a bovine homologue of human RNA helicase A and Drosophila Mle protein. J. Biol. Chem. 270: 16422–16427, 1995.

[28363] 1915. Thomson, R. B.; Igarashi, P.; Biemesderfer, D.; Kim, R.; Abu-Alfa, A.; Soleimani, M.; Aronson, P. S.: Isolation and cDNA cloning of Ksp-cadherin, a novel kidney-specific member of the cadherin multigene family. J. Biol. Chem. 270: 17594–17601, 1995.

[28364] 1916. Thomson, R. B.; Ward, D. C.; Quaggin, S. E.; Igarashi, P.; Muckler, Z. E.; Aronson, P. S.: cDNA cloning and chromosomal localization of the human and mouse isoforms of Ksp-cadherin. Genomics 51: 445–451, 1998.

[28365] 1917. Ambruso, D. R.; Knall, C.; Abell, A. N.; Panepinto, J.; Kurkchubasche, A.; Thurman, G.; Gonzalez-Aller, C.; Hieste, A.; deBoer, M.; Harbeck, R. J.; Oyer, R.; Johnson, G. L.; Roos, D.: Human neutrophil immunodeficiency syndrome is associated with an inhibitory Rac2 mutation. Proc. Nat. Acad. Sci. 97: 4654–4659, 2000.

[28366] 1918. Courjal, F.; Chuchana, P.; Theillet, C.; Fort, P.: Structure and chromosomal assignment to 22q12 and 17qter of

the ras-related Rac2 and Rac3 human genes. *Genomics* 44: 242–246, 1997.

[28367] 1919. Diebold, B. A.; Bokoch, G. M.: Molecular basis for Rac2 regulation of phagocyte NADPH oxidase. *Nature Immun.* 2: 211–215, 2001.

[28368] 1920. Li, B.; Yu, H.; Zheng, W.; Voll, R.; Na, S.; Roberts, A. W.; Williams, D. A.; Davis, R. J.; Ghosh, S.; Flavell, R. A.: Role of the guanosine triphosphatase Rac2 in T helper 1 cell differentiation. *Science* 288:2219–2222, 2000.

[28369] 1921. Williams, D. A.; Tao, W.; Yang, F.; Kim, C.; Gu, Y.; Mansfield, P.; Levine, J. E.; Petryniak, B.; Derrow, C. W.; Harris, C.; Jia, B.; Zheng, Y.; Ambruso, D. R.; Lowe, J. B.; Atkinson, S. J.; Dinanuer, M. C.; Boxer, L.: Dominant negative mutation of the hematopoietic-specific Rho GTPase, Rac 2, is associated with a human phagocyte immunodeficiency. *Blood* 96:1646–1654, 2000.

[28370] 1922. Haataja, L.; Groffen, J.; Heisterkamp, N.: Characterization of RAC3, a novel member of the Rho family. *J. Biol. Chem.* 272: 20384–20388, 1997.

[28371] 1923. Morris, C. M.; Haataja, L.; McDonald, M.; Gough, S.; Markie, D.; Groffen, J.; Heisterkamp, N.: The small GTPase RAC3 gene is located within chromosome band 17q25.3 outside and telomeric of a region commonly deleted in

breast and ovarian tumours. *Cytogenet. Cell Genet.* 89:18–23, 2000.

- [28372] 1924.El Rouby, S.; Newcomb, E. W.: Identification of Bcd, a novel proto-oncogene expressed in B-cells. *Oncogene* 13: 2623–2630, 1996.
- [28373] 1925.El Rouby, S.; Rao, P. H.; Newcomb, E. W.: Assignment of the human B-cell-derived (BCD1) proto-oncogene to 10p14–p15. *Genomics* 43:395–397, 1997.
- [28374] 1926.Koritschoner, N. P.; Bocco, J. L.; Panzetta–Dutari, G. M.; Dumur, C. I.; Flury, A.; Patrino, L. C.: A novel human zinc finger protein that interacts with the core promoter element of a TATA box-less gene. *J. Biol. Chem.* 272: 9573–9580, 1997.
- [28375] 1927.Narla, G.; Heath, K. E.; Reeves, H. L.; Li, D.; Giono, L. E.; Kimmelman, A. C.; Glucksman, M. J.; Narla, J.; Eng, F. J.; Chan, A. M.; Ferrari, A. C.; Martignetti, J. A.; Friedman, S. L.: KLF6, a candidate tumor suppressor gene mutated in prostate cancer. *Science* 294: 2563–2566, 2001.
- [28376] 1928.Onyango, P.; Koritschoner, N. P.; Patrino, L. C.; Zenke, M.; Weith, A.: Assignment of the gene encoding the core promoter element binding protein (COPEB) to human chromosome 10p15 by somatic hybrid analysis and fluorescence in situ hybridization. *Genomics* 48: 143–144,

1998.

- [28377] 1929.Patrino, L. C.; Bocco, J. L.: Personal Communication. Cordoba,Argentina 6/17/1998.
- [28378] 1930.Ratzliff, V.; Lalazar, A.; Wong, L.; Dang, Q.; Collins, C.; Shaulian,E.; Jensen, S.; Friedman, S. L.: Zf9, a Kruppel-like transcriptionfactor up-regulated in vivo during early hepatic fibrosis. Proc.Nat. Acad. Sci. 95: 9500–9505, 1998.
- [28379] 1931.Chieffo, C.; Garvey, N.; Gong, W.; Roe, B.; Zhang, G.; Silver,L.; Emanuel, B. S.; Budarf, M. L.: Isolation and characterizationof a gene from the DiGeorge chromosomal region homologous to the mouseTbx1 gene. Genomics 43: 267–277, 1997.
- [28380] 1932.Stachora, A. A.; Schafer, R. E.; Pohlmeier, M.; Maier, G.; Ponstingl,H.: Human Supt5h protein, a putative modulator of chromatin structure,is reversibly phosphorylated in mitosis. FEBS Lett. 409: 74–78,1997.
- [28381] 1933.Williams, J. B.; Lanahan, A. A.: A mammalian delayed-early responsegene encodes HNP36, a novel, conserved nucleolar protein. Biochem.Biophys. Res. Commun. 213: 325–333, 1995.
- [28382] 1934.Williams, J. B.; Rexer, B.; Sirripurapu, S.; John, S.; Goldstein,R.; Phillips, J. A., III; Haley, L. L.; Sait, S. N. J.;

Shows, T.B.; Smith, C. M.; Gerhard, D. S.: The human HNP36 gene is localized to chromosome 11q13 and produces alternative transcripts that are not mutated in multiple endocrine neoplasia, type 1 (MEN I) syndrome. *Genomics* 42:325–330, 1997.

[28383] 1935. Kohler, M.; Hirschberg, B.; Bond, C. T.; Kinzie, J. M.; Marrion, N. V.; Maylie, J.; Adelman, J. P.: Small-conductance, calcium-activated potassium channels from mammalian brain. *Science* 273: 1709–1714, 1996.

[28384] 1936. Austin, C. P.; Holder, D. J.; Ma, L.; Mixson, L. A.; Caskey, C. T.: Mapping of hKCa3 to chromosome 1q21 and investigation of linkage of CAG repeat polymorphism to schizophrenia. *Molec. Psychiat.* 4:261–266, 1999.

[28385] 1937. Bond, C. T.; Sprengel, R.; Bissonnette, J. M.; Kaufmann, W. A.; Pribnow, D.; Neelands, T.; Storck, T.; Baetscher, M.; Jerecic, J.; Maylie, J.; Knaus, H.-G.; Seeburg, P. H.; Adelman, J. P.: Respiration and parturition affected by conditional overexpression of the Ca(2+)-activated K(+) channel subunit, SK3. *Science* 289: 1942–1946, 2000.

[28386] 1938. Chandy, K. G.; Fantino, E.; Wittekindt, O.; Kalman, K.; Tong, L.-L.; Ho, T.-H.; Gutman, G. A.; Crocq, M.-A.; Ganguli, R.; Nimgaonkar, V.; Morris-Rosendahl, D. J.; Gargus, J. J.: Isolation of a novel potassium channel gene

hSKCa3 containing a polymorphic CAG repeat: a candidate for schizophrenia and bipolar disorder? *Molec. Psychiat.* 3: 32–37, 1998.

[28387] 1939. Frebourg, T.; Bonnet-Brilhault, F.; Laurent, C.; Campion, D.; Thibaut, F.; Deleuze, J. F.; Petit, M.; Mallet, J.: No evidence for the involvement of the hSKCa3 potassium channel gene in familial and sporadic cases of schizophrenia. (Abstract) *Am. J. Hum. Genet.* (suppl.) 63: A326 only, 1998.

[28388] 1940. Sun, G.; Tomita, H.; Shakkottai, V. G.; Gargus, J. J.: Genomic organization and promoter analysis of human KCNN3 gene. *J. Hum. Genet.* 46: 463–470, 2001.

[28389] 1941. Loeffen, J.; van den Heuvel, L.; Smeets, R.; Triepels, R.; Sengers, R.; Trijbels, F.; Smeitink, J.: cDNA sequence and chromosomal localization of the remaining three human nuclear encoded iron sulphur protein (IP) subunits of complex I: the human IP fraction is completed. *Biochem. Biophys. Res. Commun.* 247: 751–758, 1998.

[28390] 1942. Navon, R.; Shamir, E.; Dror, V.; Ghanshani, S.; Litmanovitch, T.; Kimchi, R.; Swartz, M.; Barak, Y.; Fantino, E.; Kalman, K.; Jones, E. G.; Avivi, L.; Chandy, K. G.; Gargus, J. J.; Gutman, G. A.: Strong association between schizophrenia and long CAG repeats in the hKCa3/KCNN3 gene,

mapped to 1q21, among Israeli Jews. (Abstract) Am. J. Hum.Genet. 63 (suppl.): A337 only, 1998.

- [28391] 1943.Wittekindt, O.; Jauch, A.; Burgert, E.; Scharer, L.; Holtgreve-Grez,H.; Yvert, G.; Imbert, G.; Zimmer, J.; Hoehe, M. R.; Macher, J.-P.;Chiaroni, P.; van Calker, D.; Crocq, M.-A.; Morris-Rosendahl, D. J.: The human small conductance calcium-regulated potassium channelgene (hSKCa3) contains two CAG repeats in exon 1, is on chromosome1q21.3, and shows a possible association with schizophrenia. Neurogenetics 1:259-265, 1998.
- [28392] 1944.Schenker, T.; Lach, C.; Kessler, B.; Calderara, S.; Trueb, B.:A novel GTP-binding protein which is selectively repressed in SV40transformed fibroblasts. J. Biol. Chem. 269: 25447-25453, 1994.
- [28393] 1945.Daigo, Y.; Suzuki, K.; Maruyama, O.; Miyoshi, Y.; Yasuda, T.; Kabuto,T.; Imaoka, S.; Fujiwara, T.; Takahashi, E.; Fujino, M. A.; Nakamura,Y.: Isolation, mapping, and mutation analysis of a human cDNA homologousto the doc-1 gene of the Chinese hamster, a candidate tumor suppressorfor oral cancer. Genes Chromosomes Cancer 20: 204-207, 1997.
- [28394] 1946.Todd, R.; McBride, J.; Tsuji, T.; Donoff, R. B.; Nagai, M.; Chou,M. Y.; Chiang, T.; Wong, D. T. W.: Deleted in oral

cancer-1 (doc-1), a novel oral tumor suppressor gene.

FASEB J. 1362-1370, 1995.

- [28395] 1947. Yamagata, T.; Tsuru, T.; Momoi, M. Y.; Suwa, K.; Nozaki, Y.; Mukasa, T.; Ohashi, H.; Fukushima, Y.; Momoi, T.: Genome organization of human 48-kDa oligosaccharyl-transferase (DDOST). *Genomics* 45: 535-540, 1997.
- [28396] 1948. Caplen, N. J.; Taylor, J. P.; Statham, V. S.; Tanaka, F.; Fire, A.; Morgan, R. A.: Rescue of polyglutamine-mediated cytotoxicity by double-stranded RNA-mediated RNA interference. *Hum. Molec. Genet.* 11:175-184, 2002.
- [28397] 1949. La Spada, A. R.; Wilson, E. M.; Lubahn, D. B.; Harding, A. E.; Fischback, K. H.: Androgen receptor gene mutations in X-linked spinal and bulbar muscular atrophy. *Nature* 352: 77-79, 1991.
- [28398] 1950. Iizasa, T.; Taira, M.; Shimada, H.; Ishijima, S.; Tatibana, M.: Molecular cloning and sequencing of human cDNA for phosphoribosylpyrophosphate synthetase subunit II. *FEBS Lett.* 244: 47-50, 1989.
- [28399] 1951. Taira, M.; Ishijima, S.; Kita, K.; Yamada, K.; Iizasa, T.; Tatibana, M.: Nucleotide and deduced amino acid sequences of two distinct cDNAs for rat phosphoribosylpyrophosphate synthetase. *J. Biol. Chem.* 262:14867-14870, 1987.

- [28400] 1952.Taira, M.; Kudoh, J.; Minoshima, S.; Iizasa, T.; Shimada, H.; Shimizu,Y.; Tatibana, M.; Shimizu, N.: Localization of human phosphoribosylpyrophosphatesynthetase subunit I and II genes (PRPS1 and PRPS2) to different regions of the X chromosome and assignment of two PRPS1-related genes to autosomes. *Somat.Cell Molec. Genet.* 15: 29–37, 1989.
- [28401] 1953.Wang, J. C.; Passage, M. B.; Ellison, J.; Becker, M. A.; Yen, P.H.; Shapiro, L. J.; Mohandas, T. K.: Physical mapping of loci in the distal half of the short arm of the human X chromosome: implications for the spreading of X-chromosome inactivation. *Somat. Cell Molec.Genet.* 18: 195–200, 1992.
- [28402] 1954.Sullivan, J. L.; Byron, K. S.; Brewster, F. E.; Baker, S. M.; Ochs, H. D.: X-linked lymphoproliferative syndrome: natural history of the immunodeficiency. *J. Clin. Invest.* 71: 1765–1778, 1983.
- [28403] 1955.Sullivan, J. L.; Byron, K. S.; Brewster, F. E.; Purtilo, D. T.: Deficient natural killer cell activity in X-linked lymphoproliferative syndrome. *Science* 210: 543–545, 1980.
- [28404] 1956.Sumazaki, R.; Kanegane, H.; Osaki, M.; Fukushima, T.; Tsuchida,M.; Matsukura, H.; Shinozaki, K.; Kimura, H.; Matsui, A.; Miyawaki,T.: SH2D1A mutations in Japanese

males with severe Epstein–Barr virus–associated illnesses.
Blood 98: 1268–1270, 2001.

- [28405] 1957.Sumegi, J.; Gross, T. G.; Seemayer, T. A.: The molecular genetics of X–linked lymphoproliferative (Duncan's) disease. Cancer J. Sci.Am. 5: 57–62, 1999.
- [28406] 1958.Sumegi, J.; Huang, D.; Lanyi, A.; Davis, J. D.; Seemayer, T. A.; Maeda, A.; Klein, G.; Seri, M.; Wakiguchi, H.; Purtilo, D. T.; Gross, T. G.: Correlation of mutations of the SH2D1A gene and Epstein–Barr virus infection with clinical phenotype and outcome in X–linked lymphoproliferative disease. Blood 96: 3118–3125, 2000.
- [28407] 1959.Xiang, X.; Benson, K. F.; Chada, K.: Mini–mouse: disruption of the pygmy locus in a transgenic insertional mutant. Science 247:967–969, 1990.
- [28408] 1960.Scriber, C. R.: Vitamin B6 deficiency and dependency in man. Am.J. Dis. Child. 113: 109–114, 1967.
- [28409] 1961.Scriber, C. R.; Hutchison, J. H.: The vitamin B6 deficiency syndrome in human infancy: biochemical and clinical observations. Pediatrics 31:240–250, 1963.
- [28410] 1962.Wolpert, S. M.; Barnes, P. D.: MRI in Pediatric Neuro–radiology. Mosby Year Book, St. Louis , 1992.
- [28411] 1963.Uchida, T.; Fujimori, F.; Tradler, T.; Fischer, G.; Rahfeld, J.–U.: Identification and characterization of a 14 kDa

human protein as a novel parvulin-like peptidyl prolyl cis/trans isomerase. FEBS Lett. 446:278–282, 1999.

- [28412] 1964. Aagaard, L.; Laible, G.; Selenko, P.; Schmid, M.; Dorn, R.; Schotta, G.; Kuhfittig, S.; Wolf, A.; Lebersorger, A.; Singh, P. B.; Reuter, G.; Jenuwein, T.: Functional mammalian homologues of the *Drosophila* PEV-modifier Su(var)3–9 encode centromere-associated proteins which complex with the heterochromatin component M31. EMBO J. 18: 1923–1938, 1999.
- [28413] 1965. Bannister, A. J.; Zegerman, P.; Partridge, J. F.; Miska, E. A.; Thomas, J. O.; Allshire, R. C.; Kouzarides, T.: Selective recognition of methylated lysine 9 on histone H3 by the HP1 chromo domain. Nature 410:120–124, 2001.
- [28414] 1966. Fabrizi, G. M.; Rizzuto, R.; Nakase, H.; Mita, S.; Kadenbach, B.; Schon, E. A.: Sequence of a cDNA specifying subunit VIa of human cytochrome c oxidase. Nucleic Acids Res. 17: 6409 only, 1989.
- [28415] 1967. Hey, Y.; Hoggard, N.; Burt, E.; James, L. A.; Varley, J. M.: Assignment of COX6A1 to 6p21 and a pseudogene (COX6A1P) to 1p31.1 by in situ hybridization and somatic cell hybrids. Cytogenet. Cell Genet. 77:167–168, 1997.
- [28416] 1968. Kissil, J. L.; Deiss, L. P.; Bayewitch, M.; Raveh, T.; Khaspekov, G.; Kimchi, A.: Isolation of DAP3, a novel me–

diator of interferon- γ -induced cell death. *J. Biol. Chem.* 270: 27932–27936, 1995.

- [28417] 1969. Kissil, J. L.; Kimchi, A.: Assignment of death associated protein 3 (DAP3) to human chromosome 1q21 by in situ hybridization. *Cytogenet. Cell Genet.* 77: 252 only, 1997.
- [28418] 1970. Connors, T. D.; Van Raay, T. J.; Petry, L. R.; Klinger, K. W.; Landes, G. M.; Burn, T. C.: The cloning of a human ABC gene (ABC3) mapping to chromosome 16p13.3. *Genomics* 39: 231–234, 1997.
- [28419] 1971. Klugbauer, N.; Hofmann, F.: Primary structure of a novel ABC transporter with a chromosomal localization on the band encoding the multidrug resistance-associated protein. *FEBS Lett.* 391: 61–65, 1996.
- [28420] 1972. Wu, Y.-C.; Horvitz, H. R.: The *C. elegans* cell corpse engulfment gene *ced-7* encodes a protein similar to ABC transporters. *Cell* 93: 951–960, 1998.
- [28421] 1973. Azuma, T.; Seki, N.; Yoshikawa, T.; Saito, T.; Masuho, Y.; Muramatsu, M.: cDNA cloning, tissue expression, and chromosome mapping of human homolog of SOX18. *J. Hum. Genet.* 45: 192–195, 2000.
- [28422] 1974. Greenfield, A.; Dunn, T.; Muscat, G.; Koopman, P.: The Sry-related gene Sox18 maps to distal mouse chro-

mosome 2. Genomics 36: 558–559,1996.

- [28423] 1975.Pennisi, D.; Gardner, J.; Chambers, D.; Hosking, B.; Peters, J.;Muscat, G.; Abbott, C.; Koopman, P.: Mutations in Sox18 underliecardiovascular and hair follicle defects in ragged mice. Nature Genet. 24:434–437, 2000.
- [28424] 1976.Pennisi, D. J.; James, K. M.; Hosking, B.; Muscat, G. E. O.; Koopman,P.: Structure, mapping, and expression of human SOX18. MammalianGenome 11: 1147–1149, 2000.
- [28425] 1977.Logan, M.; Tabin, C. J.: Role of Pitx1 upstream of Tbx4 in specificationof hindlimb identity. Science 283: 1736–1739, 1999.
- [28426] 1978.McCormick, M. B.; Tamimi, R. M.; Snider, L.; Asakura, A.; Bergstrom,D.; Tapscott, S. J.: neuroD2 and neuroD3: distinct expression patternsand transcriptional activation potentials within the neuroD gene family. Molec.Cell. Biol. 16: 5792–5800, 1996.
- [28427] 1979.Tamimi, R. M.; Steingrimsson, E.; Montgomery–Dyer, K.; Copeland,N. G.; Jenkins, N. A.; Tapscott, S. J.: NEU–ROD2 and NEUROD3 genesmap to human chromosomes 17q12 and 5q23–q31 and mouse chromosomes11 and 13, respectively. Genomics 40: 355–357, 1997.
- [28428] 1980.Engelen, J. J. M.; Esterling, L. E.; Albrechts, J. C. M.; Detera–Wadleigh,S. D.; van Eys, G. J. J. M.: Assignment of

the human gene for smoothelin(SMTN) to chromosome 22q12 by fluorescence in situ hybridization and radiation hybrid mapping. *Genomics* 43: 245–247, 1997.

- [28429] 1981. Rensen, S.; Merks, G.; Doevendans, P.; Geurts van Kessel, A.; van Eys, G.: Structure and chromosome location of Smtn, the mouse smoothelin gene. *Cytogenet. Cell Genet.* 89: 225–229, 2000.
- [28430] 1982. van der Loop, F. T. L.; Schaart, G.; Timmer, E. D. J.; Ramaekers, F. C. S.; van Eys, G. J. J. M.: Smoothelin, a novel cytoskeletal protein specific for smooth muscle cells. *J. Cell Biol.* 134: 401–411, 1996.
- [28431] 1983. Toshima, J.; Ohashi, K.; Okano, I.; Nunoue, K.; Kishioka, M.; Kuma, K.; Miyata, T.; Hirai, M.; Baba, T.; Mizuno, K.: Identification and characterization of a novel protein kinase, TESK1, specifically expressed in testicular germ cells. *J. Biol. Chem.* 270: 31331–31337, 1995.
- [28432] 1984. Toshima, J.; Toshima, J. Y.; Suzuki, M.; Noda, T.; Mizuno, K.: Cell-type-specific expression of a TESK1 promoter-linked lacZ gene in transgenic mice. *Biochem. Biophys. Res. Commun.* 286: 566–573, 2001.
- [28433] 1985. Garcia-Anoveros, J.; Derfler, B.; Neville-Golden, J.; Hyman, B. T.; Corey, D. P.: BNaC1 and BNaC2 constitute a new family of human neuronal sodium channels related to

degenerins and epithelial sodium channels. *Proc. Nat. Acad. Sci.* 94: 1459–1464, 1997.

- [28434] 1986.Price, M. P.; Lewin, G. R.; McIlwrath, S. L.; Cheng, C.; Xie, J.;Heppenstall, P. A.; Stucky, C. L.; Mannsfeldt, A. G.; Brennan, T.J.; Drummond, H. A.; Qiao, J.; Benson, C. J.; Tarr, D. E.; Hrstka,R. F.; Yang, B.; Williamson, R. A.; Welsh, M. J.: The mammalian sodium channel BNC1 is required for normal touch sensation. *Nature* 407:1007–1011, 2000.
- [28435] 1987.Price, M. P.; Snyder, P. M.; Welsh, M. J.: Cloning and expression of a novel human brain Na⁺ channel. *J. Biol. Chem.* 271: 7879–7882,1996.
- [28436] 1988.Waldmann, R.; Champigny, G.; Voilley, N.; Lauritzen, I.; Lazdunski,M.: The mammalian degenerin MDEG, an amiloride–sensitive cation channel activated by mutations causing neurodegeneration in *Caenorhabditis elegans*. *J. Biol. Chem.* 271: 10433–10436, 1996.
- [28437] 1989.Waldmann, R.; Voilley, N.; Mattei, M.–G.; Lazdunski, M.: The human degenerin MDEG, an amiloride–sensitive neuronal cation channel, is localized on chromosome 17q11.2–17q12 close to the microsatellite D17S798. *Genomics* 37: 269–270, 1996.
- [28438] 1990.Darlington, T. K.; Wager–Smith, K.; Ceriani, M. F.; Staknis, D.;Gekakis, N.; Steeves, T. D. L.; Weitz, C. J.;

Takahashi, J. S.; Kay, S. A.: Closing the circadian loop: CLOCK-induced transcription of its own inhibitors per and tim. Science 280: 1599–1603, 1998.

[28439] 1991. Gekakis, N.; Staknis, D.; Nguyen, H. B.; Davis, F. C.; Wilsbacher, L. D.; King, D. P.; Takahashi, J. S.; Weitz, C. J.: Role of the CLOCK protein in the mammalian circadian mechanism. Science 280: 1564–1569, 1998.

[28440] 1992. McNamara, P.; Seo, S.; Rudic, R. D.; Sehgal, A.; Chakravarti, D.; FitzGerald, G. A.: Regulation of CLOCK and MOP4 by nuclear hormone receptors in the vasculature: a humoral mechanism to reset a peripheral clock. Cell 105: 877–889, 2001.

[28441] 1993. Rutter, J.; Reick, M.; Wu, L. C.; McKnight, S. L.: Regulation of Clock and NPAS2 DNA binding by the redox state of NAD cofactors. Science 293: 510–514, 2001.

[28442] 1994. Shearman, L. P.; Sriram, S.; Weaver, D. R.; Maywood, E. S.; Chaves, I.; Zheng, B.; Kume, K.; Lee, C. C.; van der Horst, G. T. J.; Hastings, M. H.; Reppert, S. M.: Interacting molecular loops in the mammalian circadian clock. Science 288: 1013–1019, 2000.

[28443] 1995. Bowden, P. E.; Hailey, S.; Parker, G.; Hodgins, M. B.: Sequence and expression of human hair keratin genes. J. Derm. Sci. 7 (suppl.): S152–S163, 1994.

- [28444] 1996.Bowden, P. E.; Hainey, S. D.; Parker, G.; Jones, D. O.; Zimonjic,D.; Popescu, N.; Hodgins, M. B.: Characterization and chromosomallocalization of human hair-specific keratin genes and comparativeexpression during the hair growth cycle. *J. Invest. Derm.* 110: 158–164,1998.
- [28445] 1997.Fink, P.; Rogers, M. A.; Korge, B.; Winter, H.; Schweizer, J.:A cDNA encoding the human type I hair keratin hHa1. *Biochim. Biophys.Acta* 1264: 12–14, 1995.
- [28446] 1998.Heid, H. W.; Werner, E.; Franke, W. W.: The complement of nativealpha-keratin polypeptides of hair-forming cells: a subset of eightpolypeptides that differ from epithelial cytokeratins. *Differentiation* 32:101–119, 1986.
- [28447] 1999.Langbein, L.; Rogers, M. A.; Winter, H.; Silke, P.; Beckhaus, U.;Rackwitz, H.–R.; Schweizer, J.: The catalog of human hair keratins.I. Expression of the nine type I members in the hair follicle. *J.Biol. Chem.* 274: 19874–19884, 1999.
- [28448] 2000.Rogers, M. A.; Langbein, L.; Praetzel, S.; Moll, I.; Krieg, T.;Winter, H.; Schweizer, J.: Sequences and differential expressionof three novel human type-II hair keratins. *Differentiation* 61:187–194, 1997.
- [28449] 2001.Rogers, M. A.; Nischt, R.; Korge, B.; Krieg, T.; Fink, T. M.; Lichter,P.; Winter, H.; Schweizer, J.: Sequence data

and chromosomal localization of human type I and type II hair keratin genes. *Exp. Cell Res.* 220:357–362, 1995.

- [28450] 2002. Rogers, M. A.; Winter, H.; Wolf, C.; Heck, M.; Schweizer, J.: Characterization of a 190-kilobase pair domain of human type I hair keratin genes. *J. Biol. Chem.* 273: 26683–26691, 1998.
- [28451] 2003. Winter, H.; Hofmann, I.; Langbein, L.; Rogers, M. A.; Schweizer, J.: A splice site mutation in the gene of the human type I hair keratin hHa1 results in the expression of a tailless keratin isoform. *J. Biol. Chem.* 272: 32345–32352, 1997.
- [28452] 2004. Winter, H.; Langbein, L.; Krawczak, M.; Cooper, D. N.; Jave-Suarez, L. F.; Rogers, M. A.; Praetzel, S.; Heidt, P. J.; Schweizer, J.: Human type I hair keratin pseudogene phi-hHaA has functional orthologs in the chimpanzee and gorilla: evidence for recent inactivation of the human gene after the Pan-Homo divergence. *Hum. Genet.* 108: 37–42, 2001.
- [28453] 2005. Yu, J.; Yu, D.; Checkla, D. M.; Freedberg, I. M.; Bertolino, A. P.: Human hair keratins. *J. Invest. Derm.* 101 (suppl. 1): 56S–59S, 1993.
- [28454] 2006. Broccardo, C.; Troffer-Charlier, N.; Savary, S.; Mandel, J. L.; Chimini, G.: Exon organisation of the mouse gene

encoding the adrenoleukodystrophy-related protein (ALDRP). *Europ. J. Hum. Genet.* 6: 638–641, 1998.

- [28455] 2007. Lombard-Platet, G.; Savary, S.; Sarde, C.-O.; Mandel, J.-L.; Chimini, G.: A close relative of the adrenoleukodystrophy (ALD) gene codes for a peroxisomal protein with a specific expression pattern. *Proc. Nat. Acad. Sci.* 93: 1265–1269, 1996.
- [28456] 2008. Savary, S.; Troffer-Charlier, N.; Gyapay, G.; Mattei, M.-G.; Chimini, G.: Chromosomal localization of the adrenoleukodystrophy-related gene in man and mice. *Europ. J. Hum. Genet.* 5: 99–101, 1997.
- [28457] 2009. Kaiser, P.; Seufert, W.; Hofferer, L.; Kofler, B.; Sachsenmaier, C.; Herzog, H.; Jentsch, S.; Schweiger, M.; Schneider, R.: Human ubiquitin-conjugating enzyme homologous to yeast UBC8. *J. Biol. Chem.* 269: 8797–8802, 1994.
- [28458] 2010. Gray, G. E.; Mann, R. S.; Mitsiadis, E.; Henrique, D.; Carcangiu, M.-L.; Banks, A.; Leiman, J.; Ward, D.; Ish-Horowitz, D.; Artavanis-Tsakonas, S.: Human ligands of the Notch receptor. *Am. J. Path.* 154: 785–794, 1999.
- [28459] 2011. Wishart, M. J.; Taylor, G. S.; Slama, J. T.; Dixon, J. E.: PTEN and myotubularin phosphoinositide phosphatases: bringing bioinformatics to the lab bench. *Cell Biol.* 13:

172–181, 2001.

- [28460] 2012.Jacquemin, P.; Depetris, D.; Mattei, M.–G.; Martial, J. A.; Davidson,I.: Localization of human transcription factor TEF–4 and TEF–5 (TEAD2,TEAD3) genes to chromosomes 19q13.3 and 6p21.2 using fluorescencein situ hybridization and radiation hybrid analysis. *Genomics* 55:127–129, 1999.
- [28461] 2013.Kai, M.; Sakane, F.; Imai, S.; Wada, I.; Kanoh, H.: Molecularcloning of a diacylglycerol kinase isozyme predominantly expressedin human retina with a truncated and inactive enzyme expression inmost other human cells. *J. Biol. Chem.* 269: 18492–18498, 1994.
- [28462] 2014.Masai, I.; Okazaki, A.; Hosoya, T.; Hotta, Y.: *Drosophila* retinaldegeneration A gene encodes an eye-specific diacylglycerol kinasewith cysteine–rich zinc–finger motifs and ankyrin repeats. *Proc.Nat. Acad. Sci.* 90: 11157–11161, 1993.
- [28463] 2015.Stohr, H.; Klein, J.; Gehrig, A.; Koehler, M. R.; Jurk–lies, B.;Kellner, U.; Leo–Kottler, B.; Schmid, M.; Weber, B. H. F.: Mappingand genomic characterization of the gene encoding diacylglycerol kinasegamma (DAGK3): assessment of its role in dominant optic atrophy (OPA1). *Hum.Genet.* 104: 99–105, 1999.

- [28464] 2016.Valve, R.; Sivenius, K.; Miettinen, R.; Pihlajamaki, J.; Rissanen,A.; Deeb, S. S.; Auwerx, J.; Uusitupa, M.; Laakso, M.: Two polymorphisms in the peroxisome proliferator-activated receptor-gamma gene are associated with severe overweight among obese women. *J. Clin. Endocr. Metab.* 84:3708–3712, 1999.
- [28465] 2017.Wang, X. L.; Oosterhof, J.; Duarte, N.: Peroxisome proliferator-activated receptor gamma C161-T polymorphism and coronary artery disease. *Cardiovasc.Res.* 44: 588–594, 1999.
- [28466] 2018.Yen, C.-J.; Beamer, B. A.; Negri, C.; Silver, K.; Brown, K. A.;Yarnall, D. P.; Burns, D. K.; Roth, J.; Shuldiner, A. R.: Molecular scanning of the human peroxisome proliferator activated receptor gamma(hPPAR-gamma) gene in diabetic Caucasians: identification of a pro12alaPPAR-gamma-2 missense mutation. *Biochem. Biophys. Res. Commun.* 241:270–274, 1997.
- [28467] 2019.Chan, J. Y.; Han, X.-L.; Kan, Y. W.: Isolation of cDNA encoding the human NF-E2 protein. *Proc. Nat. Acad. Sci.* 90: 11366–11370,1993.
- [28468] 2020.Peters, L. L.; Andrews, N. C.; Eicher, E. M.; Davidson, M. B.;Orkin, S. H.; Lux, S. E.: Mouse microcytic anaemia caused by a defect in the gene encoding the globin en-

hancer-binding protein NF-E2. Nature 362:768–770, 1993.

[28469] 2021.Peters, L. L.; Bishop, T. R.; Andrews, N. C.: Globin-enhancerbinding factor NF-E2 is implicated in the regulation of heme biosynthesisand iron uptake in mk/mk mice. (Abstract) Blood 82 (suppl. 1): 179a,1993.

[28470] 2022.Shivdasani, R. A.; Orkin, S. H.: Erythropoiesis and globin geneexpression in mice lacking the transcription factor NF-E2. Proc.Nat. Acad. Sci. 92: 8690–8694, 1995.

[28471] 2023.Shivdasani, R. A.; Rosenblatt, M. F.; Zucker-Franklin, D.; Jackson,C. W.; Hunt, P.; Saris, C. J. M.; Orkin, S. H.: Transcription factorNF-E2 is required for platelet formation independent of the actionsof thrombopoietin/MGDF in megakaryocyte development. Cell 81: 695–704,1995.

[28472] 2024.Weremowicz, S.; Andrews, N. C.; Orkin, S. H.; Morton, C. C.: Mappingthe p45 subunit of human NFE2 to 12q13. (Abstract) Human Genome MappingWorkshop 93 25, 1993.

[28473] 2025.Natowicz, M. R.; Short, M. P.; Wang, Y.; Dickersin, G. R.; Gebhardt,M. C.; Rosenthal, D. I.; Sims, K. B.; Rosenberg, A. E.: Clinicaland biochemical manifestations of hyaluronidase deficiency. New Eng.J. Med. 335: 1029–1033, 1996.

- [28474] 2026.Triggs–Raine, B.; Salo, T. J.; Zhang, H.; Wicklow, B. A.; Natowicz,M. R.: Mutations in HYAL1, a member of a tandemly distributed multigenefamily encoding disparate hyaluronidase activities, cause a newlydescribed lysosomal disorder, mucopolysaccharidosis IX. Proc. Nat.Acad. Sci. 96: 6296–6300, 1999.
- [28475] 2027.Fogli, A.; Giglio, S.; Arrigo, G.; Lo Nigro, C.; Zollo, M.; Viggiano,L.; Rocchi, M.; Archidiacono, N.; Zuffardi, O.; Carrozzo, R.: Identificationof two paralogous regions mapping to the short and long arms of humanchromosome 2 comprising LIS1 pseudogenes. Cytogenet. Cell Genet. 86:225–232, 1999.
- [28476] 2028.Dubrovskaya, V.; Lavigne, A.–C.; Davidson, I.; Acker, J.; Staub,A.; Tora, L.: Distinct domains of hTAFII100 are required for functionalinteraction with transcription factor TFIIF–beta (RAP30) and incorporationinto the TFIID complex. EMBO J. 15: 3702–3712, 1996.
- [28477] 2029.Dubrovskaya, V.; Mattei, M.–G.; Tora, L.: Localization of thegene (TAF2D) encoding the 100–kDa subunit (hTAFII100) of the humanTFIID complex to chromosome 10 band q24–q25.2. Genomics 36: 556–557,1996.
- [28478] 2030.Tanese, N.; Saluja, D.; Vassallo, M. F.; Chen, J.–L.; Admon, A.: Molecular cloning and analysis of two subunits

of the human TFIIDcomplex: hTAFII130 and hTAFII100.

Proc. Nat. Acad. Sci. 93: 13611–13616,1996.

- [28479] 2031.Tao, Y.; Guermah, M.; Martinez, E.; Oelgeschlager, T.; Hasegawa,S.; Takada, R.; Yamamoto, T.; Horikoshi, M.; Roeder, R. G.: Specificinteractions and potential functions of human TAF(II)100. J. Biol.Chem. 272: 6714–6721, 1997.
- [28480] 2032.Charlier, C.; Coppieters, W.; Farnir, F.; Grobet, L.; Leroy, P.L.; Michaux, C.; Mni, M.; Schwers, A.; Vanman-shoven, P.; Hanset, R.;Georges, M.: The mh gene causing double-muscling in cattle maps tobovine chromosome 2. Mammalian Genome 6: 788–792, 1995.
- [28481] 2033.Ferrell, R. E.; Conte, V.; Lawrence, E. C.; Roth, S. M.; Hagberg,J. M.; Hurley, B. F.: Frequent sequence variation in the human myostatin(GDF8) gene as a marker for anal-ysis of muscle-related phenotypes. Genomics 62:203–207, 1999.
- [28482] 2034.Gonzalez–Cadavid, N. F.; Taylor, W. E.; Yarasheski, K.; Sinha–Hikim,I.; Ma, K.; Ezzat, S.; Shen, R.; Lalani, R.; Asa, S.; Mamita, M.;Nair, G.; Arver, S.; Bhasin, S.: Organi-zation of the human myostatingene and expression in healthy men and HIV–infected men with musclewasting. Proc. Nat. Acad. Sci. 95: 14938–14943, 1998.

- [28483] 2035.Grobet, L.; Martin, L. J. R.; Poncelet, D.; Pirottin, D.; Brouwers,B.; Riquet, J.; Schoeberlein, A.; Dunner, S.; Menissier, F.; Massabanda,J.; Fries, R.; Hanset, R.; Georges, M.: A deletion in the bovinemyostatin gene causes the double-muscled phenotype in cattle. *Nature-Genet.* 17: 71-74, 1997.
- [28484] 2036.Orstavik, S.; Solberg, R.; Tasken, K.; Nordahl, M.; Altherr, M.R.; Hansson, V.; Jahnsen, T.; Sandberg, M.: Molecular cloning, cDNAstructure, and chromosomal localization of the human type II cGMP-dependentprotein kinase. *Biochem. Biophys. Res. Commun.* 220: 759-765, 1996.
- [28485] 2037.Pfeifer, A.; Aszodi, A.; Seidler, U.; Ruth, P.; Hofmann, F.; Fassler,R.: Intestinal secretory defects and dwarfism in mice lacking cGMP-dependentprotein kinase II. *Science* 274: 2082-2084, 1996.
- [28486] 2038.Irminger-Finger, I.; Leung, W.-C.; Li, J.; Dubois-Dauphin, M.;Harb, J.; Feki, A.; Jefford, C. E.; Soriano, J. V.; Jaconi, M.; Montesano,R.; Krause, K.-H.: Identification of BARD1 as mediator between proapoptoticstress and p53-dependent apoptosis. *Molec. Cell* 8: 1255-1266, 2001.
- [28487] 2039.Wu, L. C.; Wang, Z. W.; Tsan, J. T.; Spillman, M. A.;

Phung, A.; Xu, X. L.; Yang, M.-C. W.; Hwang, L.-Y.; Bowcock, A. M.; Baer, R.: Identification of a RING protein that can interact in vivo with the BRCA1 gene product. *Nature Genet.* 14: 430–440, 1996.

[28488] 2040. Berge-Lefranc, J.-L.; Jay, P.; Massacrier, A.; Cau, P.; Mattei, M. G.; Bauer, S.; Marsollier, C.; Berta, P.; Fontes, M.: Characterization of the human jumonji gene. *Hum. Molec. Genet.* 5: 1637–1641, 1996.

[28489] 2041. Toyoda, M.; Kojima, M.; Takeuchi, T.: Jumonji is a nuclear protein that participates in the negative regulation of cell growth. *Biochem. Biophys. Res. Commun.* 274: 332–336, 2000.

[28490] 2042. Hoodless, P. A.; Haerry, T.; Abdollah, S.; Stapleton, M.; O'Connor, M. B.; Attisano, L.; Wrana, J. L.: MADR1, a MAD-related protein that functions in BMP2 signaling pathways. *Cell* 85: 489–500, 1996.

[28491] 2043. Lechleider, R. J.; de Caestecker, M. P.; Dehejia, A.; Polymeropoulos, M. H.; Roberts, A. B.: Serine phosphorylation, chromosomal localization, and transforming growth factor-beta signal transduction by human bsp-1. *J. Biol. Chem.* 271: 17617–17620, 1996.

[28492] 2044. Liu, F.; Hata, A.; Baker, J. C.; Doody, J.; Carcamo, J.; Harland, R. M.; Massague, J.: A human Mad protein acting

as a BMP-regulated transcriptional activator. *Nature* 381: 620–623, 1996.

[28493] 2045. Qin, B. Y.; Chacko, B. M.; Lam, S. S.; de Caestecker, M. P.; Correia, J. J.; Lin, K.: Structural basis of Smad1 activation by receptor kinase phosphorylation. *Molec. Cell* 8: 1303–1312, 2001.

[28494] 2046. Chikuba, K.; Yubisui, T.; Shirabe, K.; Takeshita, M.: Cloning and nucleotide sequence of a cDNA of the human erythrocyte NADPH-flavin reductase. *Biochem. Biophys. Res. Commun.* 198: 1170–1176, 1994.

[28495] 2047. Saito, F.; Yamaguchi, T.; Komuro, A.; Tobe, T.; Ikeuchi, T.; Tomita, M.; Nakajima, H.: Mapping of the newly identified biliverdin-IX beta-reductase gene (BLVRB) to human chromosome 19q13.13–q13.2 by fluorescence in situ hybridization. *Cytogenet. Cell Genet.* 71: 179–181, 1995.

[28496] 2048. Yamaguchi, T.; Komuro, A.; Nakano, Y.; Tomita, M.; Nakajima, H.: Complete amino acid sequence of biliverdin-IX beta reductase from human liver. *Biochem. Biophys. Res. Commun.* 197: 1518–1523, 1993.

[28497] 2049. Clarke, E. P.; Sanwal, B. D.: Cloning of a human collagen-binding protein, and its homology with rat gp46, chick hsp47 and mouse J6 proteins. *Biochim. Biophys. Acta* 1129: 246–248, 1992.

- [28498] 2050.Ikegawa, S.; Nakamura, Y.: Structure of the gene encoding humancolligin-2 (CBP2). *Gene* 194: 301-303, 1997.
- [28499] 2051.Ikegawa, S.; Sudo, K.; Okui, K.; Nakamura, Y.: Isolation, characterization and chromosomal assignment of human colligin-2 gene (CBP2). *Cytogenet.Cell Genet.* 71: 182-186, 1995.
- [28500] 2052.Weiskirchen, R.; Moser, M.; Weiskirchen, S.; Erdel, M.; Dahmen, S.; Buettner, R.; Gressner, A. M.: LIM-domain protein cysteine- and glycine-rich protein 2 (CRP2) is a novel marker of hepatic stellate cells and binding partner of the protein inhibitor of activated STAT1. *Biochem.J.* 359: 485-496, 2001.
- [28501] 2053.Furukawa, K.; Soejima, H.; Niikawa, N.; Shiku, H.; Furukawa, K.: Genomic organization and chromosomal assignment of the human beta-1,4-N-acetylgalactosaminyltransferase gene. *J. Biol. Chem.* 271: 20836-20844, 1996.
- [28502] 2054.Hamlin, P. J.; Jones, P. F.; Leek, J. P.; Bransfield, K.; Lench, N. J.; Aldersley, M. A.; Howdle, P. D.; Markham, A. F.; Robinson, P. A.: Assignment of GALGT encoding beta-1,4N-acetylgalactosaminyl-transferase (GalNAc-T) and KIF5A encoding neuronal kinesin (D12S1889) to hu-

manchromosome band 12q13 by assignment to ICI YAC 26EG10 and in situ hybridization. *Cytogenet. Cell Genet.* 82: 267–268, 1998.

- [28503] 2055. Nagata, Y.; Yamashiro, S.; Yodoi, J.; Lloyd, K. O.; Shiku, H.; Furukawa, K.: Expression cloning of beta-1,4-N-acetylgalactosaminyltransferase cDNAs that determine the expression of G(M2) and G(D2) gangliosides. *J. Biol. Chem.* 267: 12082–12089, 1992.
- [28504] 2056. Kosaki, K.; Bassi, M. T.; Kosaki, R.; Lewin, M.; Belmont, J.; Schauer, G.; Casey, B.: Characterization and mutation analysis of human LEFTYA and LEFTY B, homologues of murine genes implicated in left-right axis development. *Am. J. Hum. Genet.* 64: 712–721, 1999.
- [28505] 2057. Kothapalli, R.; Buyuksal, I.; Wu, S.-Q.; Chegini, N.; Tabibzadeh, S.: Detection of eba1, a novel human gene of the transforming growth factor beta superfamily: association of gene expression with endometrial bleeding. *J. Clin. Invest.* 99: 2342–2350, 1997.
- [28506] 2058. Meno, C.; Itoh, Y.; Saijoh, Y.; Matsuda, Y.; Tashiro, K.; Kuhara, S.; Hamada, H.: Two closely-related left-right asymmetrically expressed genes, lefty-1 and lefty-2: their distinct expression domains, chromosomal linkage and direct neuralizing activity in *Xenopus* embryos. *Genes Cells*

2: 513–524, 1997.

- [28507] 2059.Meno, C.; Saijoh, Y.; Fujii, H.; Ikeda, M.; Yokoyama, T.; Yokoyama,M.; Toyoda, Y.; Hamada, H.: Left–right asymmetric expression of theTGF–beta–family member lefty in mouse embryos. *Nature* 381: 151–155,1996.
- [28508] 2060.Meno, C.; Shimono, A.; Saijoh, Y.; Yashiro, K.; Mochida, K.; Ohishi,S.; Noji, S.; Kondoh, H.; Hamada, H.: Lefty–1 is required for left–rightdetermination as a regulator of lefty–2 and nodal. *Cell* 94: 287–297,1998.
- [28509] 2061.Tabibzadeh, S.; Mason, J. M.; Shea, W.; Cai, Y.; Murray, M. J.;Lessey, B.: Dysregulated expression of ebaf, a novel molecular defectin the endometria of patients with infertility. *J. Clin. Endocr.Metab.* 85: 2526–2536, 2000.
- [28510] 2062.Caterina, M. J.; Leffler, A.; Malmberg, A. B.; Martin, W. J.; Trafton,J.; Petersen–Zeltz, K. R.; Koltzenburg, M.; Basbaum, A. I.; Julius,D.: Impaired nociception and pain sensation in mice lacking the capsaicinreceptor. *Science* 288: 306–313, 2000.
- [28511] 2063.Caterina, M. J.; Schumacher, M. A.; Tominaga, M.; Rosen, T. A.;Levine, J. D.; Julius, D.: The capsaicin receptor: a heat–activatedion channel in the pain pathway. *Nature* 389: 816–824, 1997.
- [28512] 2064.Jordt, S.–E.; Julius, D.: Molecular basis for species–

specific sensitivity to 'hot' chili peppers. *Cell* 108: 421–430, 2002.

- [28513] 2065. Liedtke, W.; Choe, Y.; Marti-Renom, M. A.; Bell, A. M.; Denis, C. S.; Sali, A.; Hudspeth, A. J.; Friedman, J. M.; Heller, S.: Vanilloid receptor-related osmotically activated channel (VR-OAC), a candidate vertebrate osmoreceptor. *Cell* 103: 525–535, 2000.
- [28514] 2066. Liu, L.; Simon, S. A.: Similarities and differences in the currents activated by capsaicin, piperine, and zingerone in rat trigeminal ganglion cells. *J. Neurophysiol.* 76: 1858–1869, 1996.
- [28515] 2067. Prescott, J.; Stevenson, R. J.: Psychophysical responses to single and multiple presentations of the oral irritant zingerone: relationship to frequency of chili consumption. *Physiol. Behav.* 60: 617–624, 1996.
- [28516] 2068. Stevenson, R. J.; Prescott, J.: The effects of prior experience with capsaicin on ratings of its burn. *Chem. Senses* 19: 651–656, 1994.
- [28517] 2069. Stevenson, R. J.; Yeomans, M. R.: Differences in ratings of intensity and pleasantness for the capsaicin burn between chili likers and non-likers: implications for liking development. *Chem. Senses* 18: 471–482, 1993.
- [28518] 2070. Trevisani, M.; Smart, D.; Gunthorpe, M. J.; Tognetto,

M.; Barbieri, M.; Campi, B.; Amadesi, S.; Gray, J.; Jerman, J. C.; Brough, S. J.; Owen, D.; Smith, G. D.; Randall, A. D.; Harrison, S.; Bianchi, A.; Davis, J. B.; Geppetti, P.: Ethanol elicits and potentiates nociceptor responses via the vanilloid receptor-1. *Nature Neurosci.* 5: 546–551, 2002.

[28519] 2071. Nakano, M.; Yamada, K.; Fain, J.; Sener, E. C.; Sell-eck, C. J.; Awad, A. H.; Zwaan, J.; Mullaney, P. B.; Bosley, T. M.; Engle, E. C.: Homozygous mutations in *ARIX* (*PHOX2A*) result in congenital fibrosis of the extraocular muscles type 2. *Nature Genet.* 29: 315–320, 2001.

[28520] 2072. Wang, Y.; Macke, J. P.; Abella, B. S.; Andreasson, K.; Worley, P.; Gilbert, D. J.; Copeland, N. G.; Jenkins, N. A.; Nathans, J.: A large family of putative transmembrane receptors homologous to the product of the *Drosophila* tissue polarity gene *frizzled*. *J. Biol. Chem.* 271: 4468–4476, 1996.

[28521] 2073. Zhao, Z. Y.; Lee, C. C.; Baldini, A.; Caskey, C. T.: A human homologue of the *Drosophila* polarity gene *frizzled* has been identified and mapped to 17q21.1. *Genomics* 27: 370–373, 1995.

[28522] 2074. Bell, S. P.; Learned, R. M.; Jantzen, H.-M.; Tjian, R.: Functional cooperativity between transcription factors UBF1 and SL1 mediates human ribosomal RNA synthesis.

Science 241: 1192–1197, 1988.

- [28523] 2075.Chan, E. K. L.; Imai, H.; Hamel, J. C.; Tan, E. M.: Human autoantibody to RNA polymerase I transcription factor hUBF: molecular identity of nucleolus organizer region autoantigen NOR-90 and ribosomal RNA transcription upstream binding factor. J. Exp. Med. 174: 1239–1244, 1991.
- [28524] 2076.Hisatake, K.; Nishimura, T.; Maeda, Y.; Hanada, K.; Song, C.-Z.; Muramatsu, M.: Cloning and structural analysis of cDNA and the gene for mouse transcription factor UBF. Nuc. Acids Res. 19: 4631–4637, 1991.
- [28525] 2077.Jantzen, H.-M.; Admon, A.; Bell, S. P.; Tjian, R.: Nucleolar transcription factor hUBF contains a DNA-binding motif with homology to HMG proteins. Nature 344:830–836, 1990.
- [28526] 2078.Jones, K. A.; Black, D. M.; Griffiths, B. L.; Solomon, E.: Localization of the human RNA polymerase I transcription factor gene (UBTF) to the S17S183 locus on chromosome 17q21 and construction of a long-range restriction map of the region. Genomics 30: 602–604, 1995.
- [28527] 2079.Matera, A. G.; Wu, W.; Imai, H.; O'Keefe, C. L.; Chan, E. K. L.: Molecular cloning of the RNA polymerase I transcription factor hUBF/NOR-90(UBTF) gene and localization to 17q21.3 by fluorescence in situ hybridization and radia-

tion hybrid mapping. *Genomics* 41: 135–138, 1997.

[28528] 2080.O'Mahony, D. J.; Rothblum, L. I.: Identification of two forms of the RNA polymerase I transcription factor UBF. *Proc. Nat. Acad.Sci.* 88: 3180–3184, 1991.

[28529] 2081.Konishi, H.; Tsutsui, H.; Murakami, T.; Yumikura-Futatsugi, S.;Yamanaka, K.; Tanaka, M.; Iwakura, Y.; Suzuki, N.; Takeda, K.; Akira,S.; Nakanishi, K.; Mizutani, H.: IL-18 contributes to the spontaneous development of atopic dermatitis-like inflammatory skin lesion independently of IgE/stat6 under specific pathogen-free conditions. *Proc. Nat.Acad. Sci.* 99: 11340–11345, 2002.

[28530] 2082.Nolan, K. F.; Greaves, D. R.; Waldmann, H.: The human interleukin18 gene IL18 maps to 11q22.2–q22.3, closely linked to the DRD2 gene locus and distinct from mapped IDDM loci. *Genomics* 51: 161–163,1998.

[28531] 2083.Okamoto, I.; Kohno, K.; Tanimoto, T.; Iwaki, K.; Ishihara, T.;Akamatsu, S.; Ikegami, H.; Kurimoto, M.: IL-18 prevents the development of chronic graft-versus-host disease in mice. *J. Immun.* 164: 6067–6074,2000.

[28532] 2084.Okamura, H.; Tsutsui, H.; Komatsu, T.; Yutsudo, M.; Hakura, A.;Tanimoto, T.; Torigoe, K.; Okura, T.; Nukada, Y.; Hattori, K.; Akita,K.; Namba, M.; Tanabe, F.; Konishi, K.; Fukuda, S.; Kurimoto, M.:Cloning of a new cytokine

that induces IFN-gamma production by T cells. *Nature* 378:88–91, 1995.

[28533] 2085.Reddy, P.; Teshima, T.; Kukuruga, M.; Ordemann, R.; Liu, C.; Lowler,K.; Ferrara, J. L. M.: Interleukin-18 regulates acute graft-versus-hostdisease by enhancing Fas-mediated donor T cell apoptosis. *J. Exp.Med.* 194: 1433–1440, 2001.

[28534] 2086.Rothe, H.; Jenkins, N. A.; Copeland, N. G.; Kolb, H.: Active stageof autoimmune diabetes is associated with the expression of a novelcytokine, IGIF, which is located near Idd2. *J. Clin. Invest.* 99:469–474, 1997.

[28535] 2087.Sarvetnick, N.: IFN-gamma, IGIF, and IDDM. (Editorial) *J. Clin.Invest.* 99: 371–372, 1997.

[28536] 2088.Shida, K.; Shiratori, I.; Matsumoto, M.; Fukumori, Y.; Matsuhisa,A.; Kikkawa, S.; Tsuji, S.; Okamura, H.; Toyoshima, K.; Seya, T.:An alternative form of IL-18 in human blood plasma: complex formationwith IgM defined by monoclonal antibodies. *J. Immun.* 166: 6671–6679,2001.

[28537] 2089.Mishina, Y.; Rey, R.; Finegold, M. J.; Matzuk, M. M.; Josso, N.;Cate, R. L.; Behringer, R. R.: Genetic analysis of the Mullerian-inhibiting substance signal transduction pathway in mammalian sexual differentiation. *GenesDev.*

10: 2577–2587, 1996.

- [28538] 2090. Arango, N. A.; Lovell–Badge, R.; Behringer, R. R.: Targeted mutagenesis of the endogenous mouse *Mis* gene promoter: in vivo definition of genetic pathways of vertebrate sexual development. *Cell* 99: 409–419, 1999.
- [28539] 2091. Carre–Eusebe, D.; Imbeaud, S.; Harbison, M.; New, M. I.; Josso, N.; Picard, J.–Y.: Variants of the anti–Mullerian hormone gene in a compound heterozygote with the persistent Mullerian duct syndrome and his family. *Hum. Genet.* 90: 389–394, 1992.
- [28540] 2092. Zhao, J.; Dynlacht, B.; Imai, T.; Hori, T.; Harlow, E.: Expression of NPAT, a novel substrate of cyclin E–CDK2, promotes S–phase entry. *Genes Dev.* 12: 456–461, 1998.
- [28541] 2093. Margolis, R. L.; Stine, O. C.; McInnis, M. G.; Ranen, N. G.; Rubinsztein, D. C.; Leggo, J.; Brando, L. V. J.; Kidwai, A. S.; Loev, S. J.; Breschel, T. S.; Callahan, C.; Simpson, S. G.; and 12 others: cDNA cloning of a human homologue of the *Caenorhabditis elegans* cell fate–determining gene *mab–21*: expression, chromosomal localization and analysis of a highly polymorphic (CAG)_n trinucleotide repeat. *Hum. Molec. Genet.* 5:607–616, 1996.
- [28542] 2094. Margolis, R. L.; Stine, O. C.; Ward, C. M.; Franz, M. L.; Rosenblatt, A.; Callahan, C.; Sherr, M.; Ross, C. A.; Pot–

ter, N. T.: Unstable expansion of the CAG trinucleotide repeat in MAB21L1: report of a second pedigree and effect on protein expression. *J. Med. Genet.* 36:62–64, 1999.

[28543] 2095. Mariani, M.; Baldessari, D.; Francisconi, S.; Viggiano, L.; Rocchi, M.; Zappavigna, V.; Malgaretti, N.; Consalez, G. G.: Two murine and human homologs of mab-21, a cell fate determination gene involved in *Caenorhabditis elegans* neural development. *Hum. Molec. Genet.* 8:2397–2406, 1999.

[28544] 2096. Potter, N. T.: Meiotic instability associated with the CAGR1 trinucleotide repeat at 13q13. *J. Med. Genet.* 34: 411–413, 1997.

[28545] 2097. Altshuler, D.; Daly, M.; Kruglyak, L.: Guilt by association. *Nature Genet.* 26: 135–137, 2000.

[28546] 2098. Cox, N. J.: Challenges in identifying genetic variation affecting susceptibility to type 2 diabetes: examples from studies of the calpain-10 gene. *Hum. Molec. Genet.* 10: 2301–2305, 2001.

[28547] 2099. Horikawa, Y.; Oda, N.; Cox, N. J.; Li, X.; Orholm, M.; Hara, M.; Hinokio, Y.; Lindner, T. H.; Mashima, H.; Schwarz, P. E. H.; del Bosque-Plata, L.; Horikawa, Y.; and 14 others: Genetic variation in the gene encoding calpain-10 is associated with type 2 diabetes–

mellitus. Nature Genet. 26: 163–175, 2000.

[28548] 2100. Berg, J. N.; Gallione, C. J.; Stenzel, T. T.; Johnson, D. W.; Allen, W. P.; Schwartz, C. E.; Jackson, C. E.; Porteous, M. E. M.; Marchuk, D. A.: The activin receptor–like kinase 1 gene: genomic structure and mutations in hereditary hemorrhagic telangiectasia type 2. Am. J. Hum. Genet. 61: 60–67, 1997.

[28549] 2101. D'Abronzio, F. H.; Swearingen, B.; Klibanski, A.; Alexander, J. M.: Mutational analysis of activin/transforming growth factor– β type I and type II receptor kinases in human pituitary tumors. J. Clin. Endocr. Metab. 84: 1716–1721, 1999.

[28550] 2102. Futreal, P. A.; Cochran, C.; Rosenthal, J.; Miki, Y.; Swenson, J.; Hobbs, M.; Bennett, L. M.; Haugen–Strano, A.; Marks, J.; Barrett, J. C.; Tavtigian, S. V.; Shattuck–Eidens, D.; Kamb, A.; Skolnick, M.; Wiseman, R. W.: Isolation of a diverged homeobox gene, MOX1, from the BRCA1 region on 17q21 by solution hybrid capture. Hum. Molec. Genet. 3: 1359–1364, 1994.

[28551] 2103. Jones, K. A.; Black, D. M.; Brown, M. A.; Griffiths, B. L.; Nicolai, H. M.; Chambers, J. A.; Bonjardim, M.; Xu, C.–F.; Boyd, M.; McFarlane, R.; Korn, B.; Poustka, A.; North, M. A.; Schalkwyk, L.; Lehrach, H.; Solomon, E.: The detailed char–

acterisation of a 400 kb cosmid walkin the BRCA1 region:
identification and localisation of 10 genes including a
dual-specificity phosphatase. Hum. Molec. Genet. 3:
1927–1934, 1994.

[28552] 2104. Swaroop, A.; Yang-Feng, T. L.; Liu, W.; Gieser, L.;
Barrow, L.L.; Chen, K.-C.; Agarwal, N.; Meisler, M. H.;
Smith, D. I.: Molecular characterization of a novel human
gene, SEC13R, related to the yeast secretory pathway gene
SEC13, and mapping to a conserved linkage group on hu-
man chromosome 3p24–p25 and mouse chromosome 6.
Hum. Molec. Genet. 3: 1281–1286, 1994.

[28553] 2105. Kamitani, T.; Chang, H.-M.; Rollins, C.; Waneck, G.
L.; Yeh, E.T. H.: Correction of the class A defect in glyco-
sylphosphatidylinositol anchor biosynthesis in Ltk-cells by
human cDNA clone. J. Biol. Chem. 268:20733–20736,
1993.

[28554] 2106. Horton, Y. M.; Sullivan, M.; Houslay, M. D.: Molecular
cloning of a novel splice variant of human type IVA
(PDE-IVA) cyclic AMP phosphodiesterase and localization
of the gene to the p13.2–q12 region of human chromo-
some 19. Biochem. J. 308: 683–691, 1995.

[28555] 2107. Huston, E.; Pooley, L.; Julien, P.; Scotland, G.;
McPhee, I.; Sullivan, M.; Bolger, G.; Houslay, M. D.: The hu-

man cyclic AMP-specific phosphodiesterase PDE-46 (HSPDE4A4B) expressed in transfected COS7 cells occurs as both particulate and cytosolic species that exhibit distinct kinetics of inhibition by the antidepressant rolipram. J. Biol. Chem. 271: 31334–31344, 1996.

[28556] 2108. Livi, G. P.; Kmetz, P.; McHale, M. M.; Cieslinski, L. B.; Sathe, G. M.; Taylor, D. P.; Davis, R. L.; Torphy, T. J.; Balcarek, J. M.: Cloning and expression of cDNA for a human low-K(m), rolipram-sensitive cyclic AMP phosphodiesterase. Molec. Cell. Biol. 10: 2678–2686, 1990.

[28557] 2109. Obernolte, R.; Bhakta, S.; Alvarez, R.; Bach, C.; Zuppan, P.; Mulkins, M.; Jarnagin, K.; Shelton, E. R.: The cDNA of a human lymphocyte cyclic-AMP phosphodiesterase (PDE IV) reveals a multigene family. Gene 129:239–247, 1993.

[28558] 2110. Sullivan, M.; Egerton, M.; Shakur, Y.; Marquardsen, A.; Houslay, M. D.: Molecular cloning and expression, in both COS-1 cells and *S. cerevisiae*, of a human cytosolic type-IVA, cyclic AMP specific phosphodiesterase (hPDE-IVA-h6.1). Cell. Signal. 6: 793–812, 1994.

- [28559] 2111.Sullivan, M.; Rena, G.; Begg, F.; Gordon, L.; Olsen, A. S.; Houslay,M. D.: Identification and characterization of the human homologueof the short PDE4A cAMP-specific phosphodiesterase RD1 (PDE4A1) byanalysis of the human HSPDE4A gene locus located at chromosome 19p13.2. *Biochem.J.* 333: 693–703, 1998.
- [28560] 2112.Wilson, M.; Sullivan, M.; Brown, N.; Houslay, M. D.: Purification,characterization and analysis of rolipram inhibition of a human–typeIVA cyclic AMP–specific phosphodiesterase expressed in yeast. *Biochem.J.* 304: 407–415, 1994.
- [28561] 2113.Huston, E.; Lumb, S.; Russell, A.; Catterall, C.; Ross, A. H.;Steele, M. R.; Bolger, G. B.; Perry, M. J.; Owens, R. J.; Houslay,M. D.: Molecular cloning and transient expression in COS7 cells ofa novel human PDE4B cAMP–specific phosphodiesterase, HSPDE4B3. *Biochem.J.* 328: 549–558, 1997.
- [28562] 2114.Szpirer, C.; Szpirer, J.; Riviere, M.; Swinnen, J.; Vicini, E.;Conti, M.: Chromosomal localization of the human and rat genes (PDE4Dand PDE4B) encoding the cAMP–specific phosphodiesterases 3 and 4. *Cytogenet.Cell Genet.* 69: 11–14, 1995.
- [28563] 2115.Xu, R. X.; Hassell, A. M.; Vanderwall, D.; Lambert, M.

H.; Holmes, W. D.; Luther, M. A.; Rocque, W. J.; Milburn, M. V.; Zhao, Y.; Ke, H.; Nolte, R. T.: Atomic structure of PDE4: insights into phosphodiesterase mechanism and specificity. *Science* 288: 1822–1825, 2000.

[28564] 2116. Bolger, G. B.; Erdogan, S.; Jones, R. E.; Loughney, K.; Scotland, G.; Hoffmann, R.; Wilkinson, I.; Farrell, C.; Houslay, M. D.: Characterization of five different proteins produced by alternatively spliced mRNAs from the human cAMP-specific phosphodiesterase PDE4D gene. *Biochem. J.* 328: 539–548, 1997.

[28565] 2117. Dudai, Y.; Jan, Y.-N.; Byers, D.; Quinn, W. G.; Benzer, S.: *dunce*, a mutant of *Drosophila* deficient in learning. *Proc. Nat. Acad. Sci.* 73: 1684–1688, 1976.

[28566] 2118. Hansen, G.; Jin, S.-L. C.; Umetsu, D. T.; Conti, M.: Absence of muscarinic cholinergic airway responses in mice deficient in the cyclic nucleotide phosphodiesterase PDE4D. *Proc. Nat. Acad. Sci.* 97: 6751–6756, 2000.

[28567] 2119. Jin, S.-L. C.; Richard, F. J.; Kuo, W.-P.; D'Ercole, A. J.; Conti, M.: Impaired growth and fertility of cAMP-specific phosphodiesterase PDE4D-deficient mice. *Proc. Nat. Acad. Sci.* 96: 11998–12003, 1999.

[28568] 2120. Partanen, J.; Armstrong, E.; Makela, T. P.; Korhonen, J.; Sandberg, M.; Renkonen, R.; Knuutila, S.; Huebner, K.;

Alitalo, K.: A novel endothelial cell surface receptor tyrosine kinase with extracellular epidermal growth factor homology domains. *Molec. Cell. Biol.* 12:1698–1707, 1992.

[28569] 2121. Zhao, J.; Kennedy, B. K.; Lawrence, B. D.; Barbie, D. A.; Matera, A. G.; Fletcher, J. A.; Harlow, E.: NPAT links cyclin E–Cdk2 to the regulation of replication-dependent histone gene transcription. *Genes Dev.* 14: 2283–2297, 2000.

[28570] 2122. Kalaydjieva, L.; Gresham, D.; Gooding, R.; Heather, L.; Baas, F.; de Jonge, R.; Blechschmidt, K.; Angelicheva, D.; Chandler, D.; Worsley, P.; Rosenthal, A.; King, R. H. M.; Thomas, P. K.: N-myc downstream-regulated gene 1 is mutated in hereditary motor and sensory neuropathy–Lom. *Am. J. Hum. Genet.* 67: 47–58, 2000.

[28571] 2123. Kalaydjieva, L.; Hallmayer, J.; Chandler, D.; Savov, A.; Nikolova, A.; Angelicheva, D.; King, R. H. H.; Ishpekova, B.; Honeyman, K.; Calafell, F.; Shmarov, A.; Petrova, J.; Turnev, I.; Hristova, A.; Moskov, M.; Stancheva, S.; Petkova, I.; Bittles, A. H.; Georgieva, V.; Middleton, L.; Thomas, P. K.: Gene mapping in Gypsies identifies a novel demyelinating neuropathy on chromosome 8q24. *Nature Genet.* 14:214–217, 1996.

[28572] 2124. Tsujikawa, M.; Kurahashi, H.; Tanaka, T.; Okada, M.;

Yamamoto,S.; Maeda, N.; Watanabe, H.; Inoue, Y.; Kiri-
doshi, A.; Matsumoto,K.; Ohashi, Y.; Kinoshita, S.; Shimo-
mura, Y.; Nakamura, Y.; Tano,Y.: Homozygosity mapping
of a gene responsible for gelatinous drop-like corneal dys-
trophy to chromosome 1p. Am. J. Hum. Genet. 63:
1073–1077,1998.

[28573] 2125.Berkvens, T. M.; van Ormondt, H.; Gerritsen, E. J. A.;
Meera Khan,P.; van der Eb, A. J.: Identical 3250-bp dele-
tion between two Alu repeats in the ADA genes of unre-
lated ADA-SCID patients. Genomics 7:486–490, 1990.

[28574] 2126.Sanford, J. P.; Eddy, R. L.; Doyle, D.; Shows, T. B.:
Assignment of human asialoglycoprotein receptor gene
(ASGR1) to chromosome 17p11–13. Genomics
11:779–781, 1991.

[28575] 2127.Wulczyn, F. G.; Naumann, M.; Scheidereit, C.: Candi-
date proto-oncogene bcl-3 encodes a subunit-specific in-
hibitor of transcription factor NF- κ B. Nature 358:
597–599, 1992.

[28576] 2128.Falk, C. T.: New family data supporting the MN/GC
linkage. (Abstract) Cytogenet.Cell Genet. 37: 466, 1984.

[28577] 2129.Fagerlund, T.; Islander, G.; Ranklev, E.; Harbitz, I.;
Hauge,J. G.; Mogleby, E.; Berg, K.: Genetic recombination
between malignant hyperthermia and calcium release

channel in skeletal muscle. Clin.Genet. 41: 270–272, 1992.

[28578] 2130.Levitt, R. C.; Nouri, N.; Jedlicka, A. E.; McKusick, V. A.; Marks,A. R.; Shutack, J. G.; Fletcher, J. E.; Rosenberg, H.; Meyers, D.A.: Evidence for genetic heterogeneity in malignant hyperthermiasusceptibility. Genomics 11: 543–547, 1991.

[28579] 2131.MacLennan, D. H.; Duff, C.; Zorzato, F.; Fujii, J.; Phillips,M.; Korneluk, R. G.; Frodis, W.; Britt, A.; Worton, R. G.: Ryanodinereceptor gene is a candidate for predisposition to malignant hyperthermia. Nature 343:559–561, 1990.

[28580] 2132.Sanford, J. P.; Elliott, R. W.; Doyle, D.: Asialoglycoproteinreceptor genes are linked on chromosome 11 in the mouse. DNA 7:721–728, 1988.

[28581] 2133.Spiess, M.; Schwartz, A. L.; Lodish, H. F.: Sequence of humanasialoglycoprotein receptor cDNA: an internal signal sequence formembrane insertion. J. Biol. Chem. 260: 1979–1982, 1985.

[28582] 2134.Cirullo, R. E.; Arredondo–Vega, F. X.; Smith, M.; Wasmuth, J. J.: Isolation and characterization of interspecific heat–resistant hybridsbetween a temperature–sensitive Chinese hamster cell asparaginy–tRNA synthetase

mutant and normal human leukocytes: assignment of human *SN5* gene to chromosome 18. *Somat. Cell Genet.* 9: 215–233, 1983.

[28583] 2135. Shows, T. B.: Personal Communication. Buffalo, N. Y. 1/11/1983.

[28584] 2136. Akasaka, T.; Miura, I.; Takahashi, N.; Akasaka, H.; Yonetani, N.; Ohno, H.; Fukuhara, S.; Okuma, M.: A recurring translocation, $t(3;6)(q27;p21)$, in non-Hodgkin's lymphoma results in replacement of the 5'-prime regulatory region of *BCL6* with a novel H4 histone gene. *Cancer Res.* 57: 7–12, 1997.

[28585] 2137. Baron, B. W.; Nucifora, G.; McCabe, N.; Espinosa, R., III; Le Beau, M. M.; McKeithan, T. W.: Identification of the gene associated with the recurring chromosomal translocations $t(3;14)(q27;q32)$ and $t(3;22)(q27;q11)$ in B-cell lymphomas. *Proc. Nat. Acad. Sci.* 90: 5262–5266, 1993.

[28586] 2138. Capello, D.; Carbone, A.; Pastore, C.; Gloghini, A.; Saglio, G.; Gaidano, G.: Point mutations of the *BCL-6* gene in Burkitt's syndrome. *Brit. J. Haemat.* 99: 168–170, 1997.

[28587] 2139. Cattoretti, G.; Chang, C.-C.; Cechova, K.; Zhang, J.; Ye, B. H.; Falini, B.; Louie, D. C.; Offit, K.; Chaganti, R. S. K.; Dalla-Favera, R.: *BCL-6* protein is expressed in germinal-center B cells. *Blood* 86:45–53, 1995.

- [28588] 2140.Cesarman, E.; Chadburn, A.; Liu, Y.-F.; Migliazza, A.; Dalla-Favera, R.; Knowles, D. M.: BCL-6 gene mutations in posttransplantation lymphoproliferative disorders predict response to therapy and clinical outcome. *Blood* 92:2294-2302, 1998.
- [28589] 2141.Chaganti, S. R.; Chen, W.; Parsa, N.; Offit, K.; Louie, D. C.; Dalla-Favera, R.; Chaganti, R. S. K.: Involvement of BCL6 in chromosomal aberrations affecting band 3q27 in B-cell non-Hodgkin lymphoma. *Genes Chromosomes Cancer* 23: 323-327, 1998.
- [28590] 2142.Chaganti, S. R.; Rao, P. H.; Chen, W.; Dyomin, V.; Jhanwar, S.C.; Parsa, N. Z.; Dalla-Favera, R.; Chaganti, R. S. K.: Deregulation of BCL6 in non-Hodgkin lymphoma by insertion of IGH sequences in complex translocations involving band 3q27. *Genes Chromosomes Cancer* 23:328-336, 1998.
- [28591] 2143.Chang, C.-C.; Ye, B. H.; Chaganti, R. S. K.; Dalla-Favera, R.: BCL-6, a POZ/zinc-finger protein, is a sequence-specific transcriptional repressor. *Proc. Nat. Acad. Sci.* 93: 6947-6952, 1996.
- [28592] 2144.Dhordain, P.; Albagli, O.; Honore, N.; Guidez, F.; Lantoin, D.; Schmid, M.; De The, H.; Zelent, A.; Koken, M. H. M.: Colocalization and heteromerization between the

two human oncogene POZ/zinc fingerproteins, LAZ3 (BCL6) and PLZF. *Oncogene* 19: 6240–6250, 2000.

- [28593] 2145. Gaidano, G.; Capello, D.; Gloghini, A.; Fassone, L.; Vivenza, D.; Ariatti, C.; Migliazza, A.; Saglio, G.; Carbone, A.: Frequent mutation of bcl-6 proto-oncogene in high grade, but not low grade, MALT lymphomas of the gastrointestinal tract. *Haematologica* 84:582–588, 1999.
- [28594] 2146. Hamblin, T. J.; Davis, Z.; Gardiner, A.; Oscier, D. G.; Stevenson, F. K.: Unmutated Ig V(H) genes are associated with a more aggressive form of chronic lymphocytic leukemia. *Blood* 94: 1848–1854, 1999.
- [28595] 2147. Hosokawa, Y.; Maeda, Y.; Ichinohasama, R.; Miura, I.; Taniwaki, M.; Seto, M.: The Ikaros gene, a central regulator of lymphoid differentiation, fuses to the BCL6 gene as a result of t(3;7)(q27;p12) translocation in a patient with diffuse large B-cell lymphoma. *Blood* 95: 2719–2721, 2000.
- [28596] 2148. Ichii, H.; Sakamoto, A.; Hatano, M.; Okada, S.; Toyama, H.; Taki, S.; Arima, M.; Kuroda, Y.; Tokuhisa, T.: Role of Bcl-6 in the generation and maintenance of memory CD8⁺ T cells. *Nature Immun.* 3: 558–563, 2002.
- [28597] 2149. Ichinohasama, R.; Miura, I.; Funato, T.; Sato, I.; Suzuki, C.; Saito, Y.; Decoteau, J. F.; Myers, J. B.; Kadin, M. E.; Sawai, T.; Ooya, K.: A recurrent nonrandom transloca-

tion (3;7)(q27;p12) associated with BCL-6 gene rearrangement in B-cell diffuse large cell lymphoma. *Cancer Genet. Cytogenet.* 104: 19-27, 1998.

[28598] 2150. Kerckaert, J.-P.; Deweindt, C.; Tilly, H.; Quief, S.; Lecocq, G.; Bastard, C.: LAZ3, a novel zinc-finger encoding gene, is disrupted by recurring chromosome 3q27 translocations in human lymphomas. *Nature Genet.* 5: 66-70, 1993.

[28599] 2151. Liao, X.; Gilbert, D. J.; Dent, A.; Staudt, L. M.; Jenkins, N.A.; Copeland, N. G.: Mapping of the mouse Bcl6 gene to chromosome 16. *Mammalian Genome* 7: 621-622, 1996.

[28600] 2152. Migliazza, A.; Martinotti, S.; Chen, W.; Fusco, C.; Ye, B. H.; Knowles, D. M.; Offit, K.; Changanti, R. S. K.; Dalla-Favera, R.: Frequent somatic hypermutation of the 5-prime noncoding region of the BCL6 gene in B-cell lymphoma. *Proc. Nat. Acad. Sci.* 92: 12520-12524, 1995.

[28601] 2153. Miki, T.; Kawamata, N.; Hirosawa, S.; Aoki, N.: Gene involved in the 3q27 translocation associated with B-cell lymphoma, BCL5, encodes a Kruppel-like zinc-finger protein. *Blood* 83: 26-32, 1994.

[28602] 2154. Sahota, S. S.; Davis, Z.; Hamblin, T. J.; Stevenson, F. K.: Somatic mutation of bcl-6 genes can occur in the ab-

sence of V-H mutations in chronic lymphocytic leukemia. Blood 95: 3534–3540, 2000.

- [28603] 2155. Shaffer, A. L.; Yu, X.; He, Y.; Boldrick, J.; Chan, E. P.; Staudt, L. M.: BCL-6 represses genes that function in lymphocyte differentiation, inflammation, and cell cycle control. Immunity 13: 199–212, 2000.
- [28604] 2156. Shen, H. M.; Peters, A.; Baron, B.; Zhu, X.; Storb, U.: Mutation of BCL-6 gene in normal B cells by the process of somatic hypermutation of Ig genes. Science 280: 1750–1752, 1998.
- [28605] 2157. Ueda, C.; Akasaka, T.; Kurata, M.; Maesako, Y.; Nishikori, M.; Ichinohasama, R.; Imada, K.; Uchiyama, T.; Ohno, H.: The gene for interleukin-21 receptor is the partner of BCL6 in t(13;16)(q27;p11), which is recurrently observed in diffuse large B-cell lymphoma. Oncogene 21:368–376, 2002.
- [28606] 2158. Ye, B. H.; Cattoretti, G.; Shen, Q.; Zhang, J.; Hawe, N.; de Waard, R.; Leung, C.; Nouri-Shirazi, M.; Orazi, A.; Chaganti, R. S. K.; Rothman, P.; Stall, A. M.; Pandolfi, P.-P.; Dalla-Favera, R.: The BCL-6 proto-oncogene controls germinal-centre formation and Th2-type inflammation. Nature Genet. 16: 161–170, 1997.
- [28607] 2159. Ye, B. H.; Lista, F.; Lo Coco, F.; Knowles, D. M.; Offit,

K.;Chaganti, R. S. K.; Dalla-Favera, R.: Alterations of a zinc finger-encoding gene, BCL-6, in diffuse large-cell lymphoma. *Science* 262: 747-750,1993.

[28608] 2160.Ye, B. H.; Rao, P. H.; Chaganti, R. S. K.; Dalla-Favera, R.:Cloning of bcl-6, the locus involved in chromosome translocations affecting band 3q27 in B-cell lymphoma. *Cancer Res.* 53: 2732-2735,1993.

[28609] 2161.Rimokh, R.; Rouault, J. P.; Wahbi, K.; Gadoux, M.; Lafage, M.;Archimbaud, E.; Charrin, C.; Gentilhomme, O.; Germain, D.; Samarut,J.; Magaud, J. P.: A chromosome 12 coding region is juxtaposed to the MYC protooncogene locus in a t(8;12)(q24;q22) translocation in a case of B-cell chronic lymphocytic leukemia. *Genes Chromosomes-Cancer* 3: 24-36, 1991.

[28610] 2162.Gejman, P. V.; Weinstein, L. S.; Martinez, M.; Spiegel, A. M.;Cao, Q.; Hsieh, W.-T.; Hoehe, M. R.; Gershon, E. S.: Genetic mapping of the Gs-alpha subunit gene (GNAS1) to the distal long arm of chromosome 20 using a polymorphism detected by denaturing gradient gel electrophoresis. *Genomics* 9:782-783, 1991.

[28611] 2163.Fragoso, M. C. B. V.; Latronico, A. C.; Carvalho, F. M.; Zerbini,M. C. N.; Marcondes, J. A. M.; Araujo, L. M. B.; Lando, V. S.; Frazzatto,E. T.; Mendonca, B. B.; Villares, S.

M. F.: Activating mutation of the stimulatory G protein (gsp) as a putative cause of ovarian and testicular human stromal Leydig cell tumors. *J. Clin. Endocr. Metab.* 83:2074–2078, 1998.

- [28612] 2164. Gopal Rao, V. V. N.; Schnittger, S.; Hansmann, I.: G protein Gs- α (GNAS1), the probable candidate gene for Albright hereditary osteodystrophy, is assigned to human chromosome 20q12–q13.2. *Genomics* 10:257–261, 1991.
- [28613] 2165. Gorelov, V. N.; Dumon, K.; Barteneva, N. S.; Palm, D.; Roher, H.-D.; Goretzki, P. E.: Overexpression of Gs- α subunit in thyroid tumors bearing a mutated Gs- α gene. *J. Cancer Res. Clin. Oncol.* 121:219–224, 1995.
- [28614] 2166. Happle, R.: The McCune–Albright syndrome: a lethal gene surviving by mosaicism. *Clin. Genet.* 29: 321–324, 1986.
- [28615] 2167. Harris, B. A.; Robishaw, J. D.; Mumby, S. M.; Gilman, A. G.: Molecular cloning of complementary DNA for the α -subunit of the G protein that stimulates adenylate cyclase. *Science* 229: 1274–1277, 1985.
- [28616] 2168. Hayward, B.; Kamiya, M.; Takada, S.; Moran, V.; Strain, L.; Hayashizaki, Y.; Bonthron, D. T.: XL α s is a paternally derived protein product of the human GNAS1 gene. (Abstract) *Europ. J. Hum. Genet.* 6 (suppl.1): 36

only, 1998.

- [28617] 2169. Hayward, B. E.; Bonthron, D. T.: An imprinted anti-sense transcript at the human GNAS1 locus. *Hum. Molec. Genet.* 9: 835–841, 2000.
- [28618] 2170. Hayward, B. E.; Kamiya, M.; Strain, L.; Moran, V.; Campbell, R.; Hayashizaki, Y.; Bonthron, D. T.: The human GNAS1 gene is imprinted and encodes distinct paternally and biallelically expressed G proteins. *Proc. Nat. Acad. Sci.* 95: 10038–10043, 1998.
- [28619] 2171. Hayward, B. E.; Moran, V.; Strain, L.; Bonthron, D. T.: Bidirectional imprinting of a single gene: GNAS1 encodes maternally, paternally, and biallelically derived proteins. *Proc. Nat. Acad. Sci.* 95: 15475–15480, 1998.
- [28620] 2172. Hurowitz, E. H.; Melnyk, J. M.; Chen, Y.-J.; Kouros-Mehr, H.; Simon, M. I.; Shizuya, H.: Genomic characterization of the human heterotrimeric G protein alpha, beta, and gamma subunit genes. *DNAREs.* 7: 111–120, 2000.
- [28621] 2173. Iiri, T.; Farfel, Z.; Bourne, H. R.: Conditional activation defect of a human Gs-alpha mutant. *Proc. Nat. Acad. Sci.* 94: 5656–5661, 1997.
- [28622] 2174. Iiri, T.; Herzmark, P.; Nakamoto, J. M.; Van Dop, C.; Bourne, H. R.: Rapid GDP release from Gs-alpha in patients with gain and loss of endocrine function. *Nature* 371:

164–168, 1994.

- [28623] 2175. Ishikawa, Y.; Tajima, T.; Nakae, J.; Nagashima, T.; Satoh, K.; Okuhara, K.; Fujieda, K.: Two mutations of the Gs-alpha gene in two Japanese patients with sporadic pseudohypoparathyroidism type Ia. *J. Hum. Genet.* 46: 426–430, 2001.
- [28624] 2176. Jia, H.; Hingorani, A. D.; Sharma, P.; Hopper, R.; Dickerson, C.; Trutwein, D.; Lloyd, D. D.; Brown, M. J.: Association of the G(s)-alpha gene with essential hypertension and response to beta-blockade. *Hypertension* 34: 8–14, 1999.
- [28625] 2177. Kehlenbach, R. H.; Matthey, J.; Huttner, W. B.: XL alpha S is a new type of G protein. *Nature* 372: 804–809, 1994.
- [28626] 2178. Kikyo, N.; Williamson, C. M.; John, R. M.; Barton, S. C.; Beechey, C. V.; Ball, S. T.; Cattanach, B. M.; Surani, M. A.; Peters, J.: Genetic and functional analysis of neuronatin in mice with maternal or paternal duplication of distal chr. 2. *Dev. Biol.* 190: 66–77, 1997.
- [28627] 2179. Kinard, R. E.; Walton, J. E.; Buckwalter, J. A.: Pseudohypoparathyroidism. *Arch. Intern. Med.* 139: 204–207, 1979.
- [28628] 2180. Kozasa, T.; Itoh, H.; Tsukamoto, T.; Kaziro, Y.: Isola-

tion and characterization of the human Gs- α gene.

Proc. Nat. Acad. Sci. 85:2081–2085, 1988.

[28629] 2181. Juppner, H.; Schipani, E.; Bastepe, M.; Cole, D. E. C.; Lawson, M. L.; Mannstadt, M.; Hendy, G. N.; Plotkin, H.; Koshiyama, H.; Koh, T.; Crawford, J. D.; Olsen, B. R.; Vikkula, M.: The gene responsible for pseudohypoparathyroidism type Ib is paternally imprinted and maps in four unrelated kindreds to chromosome 20q13.3. Proc. Nat. Acad. Sci. 95: 11798–11803, 1998.

[28630] 2182. Landis, C. A.; Masters, S. B.; Spada, A.; Pace, A. M.; Bourne, H. R.; Vallar, L.: GTPase inhibiting mutations activate the α chain of Gs and stimulate adenylyl cyclase in human pituitary tumours. Nature 340:692–696, 1989.

[28631] 2183. Levine, M. A.; Deily, J. R.: Identification of multiple mutations in the gene encoding the α subunit of Gs in patients with pseudohypoparathyroidism type IA. (Abstract) J. Bone Miner. Res. 5: S142 only, 1990.

[28632] 2184. Levine, M. A.; Modi, W. S.; O'Brien, S. J.: Mapping of the gene encoding the α subunit of the stimulatory G protein of adenylyl cyclase (GNAS1) to 20q13.2–q13.3 in human by in situ hybridization. Genomics 11:478–479, 1991.

[28633] 2185. Levine, M. A.; Vechio, J. D.: Personal Communica–

tion. Baltimore, Md. 8/1/1990.

- [28634] 2186. Lin, C. K.; Hakakha, M. J.; Nakamoto, J. M.; Englund, A. T.; Brickman, A. S.; Scott, M. L.; Van Dop, C.: Prevalence of three mutations in the Gs- α gene among 24 families with pseudohypoparathyroidism type Ia. *Biochem. Biophys. Res. Commun.* 189: 343–349, 1992.
- [28635] 2187. Linglart, A.; Carel, J. C.; Garabedian, M.; Le, T.; Mallet, E.; Kottler, M. L.: GNAS1 lesions in pseudohypoparathyroidism Ia and Ic: genotype phenotype relationship and evidence of the maternal transmission of the hormonal resistance. *J. Clin. Endocr. Metab.* 87: 189–197, 2002.
- [28636] 2188. Malchoff, C. D.; Reardon, G.; MacGillivray, D. C.; Yamase, H.; Rogol, A. D.; Malchoff, D. M.: An unusual presentation of McCune–Albright syndrome confirmed by an activating mutation of the Gs α -subunit from a bone lesion. *J. Clin. Endocr. Metab.* 78: 803–806, 1994.
- [28637] 2189. Mantovani, G.; Romoli, R.; Weber, G.; Brunelli, V.; De Menis, E.; Beccio, S.; Beck-Peccoz, P.; Spada, A.: Mutational analysis of GNAS1 in patients with pseudohypoparathyroidism: identification of two novel mutations. *J. Clin. Endocr. Metab.* 85: 4243–4248, 2000.
- [28638] 2190. Falk, C. T.; Martin, M. D.; Walker, M. E.; Chen, T.;

Rubinstein,P.; Allen, F. H., Jr.: Family data suggesting a linkage between MN and Gc. (Abstract) Cytogenet. Cell Genet. 25: 152, 1979.

[28639] 2191.Furthmayr, H.; Metaxas, M. N.; Metaxas-Buhler, M.: M(g) and M(c):mutations within the amino-terminal region of glycophorin A. Proc.Nat. Acad. Sci. 78: 631-635, 1981.

[28640] 2192.Gedde-Dahl, T., Jr.; Olaisen, B.: MN:Ss--GC more likely than Ss:MN--GC? (Abstract) Cytogenet. Cell Genet. 32: 277-278, 1981.

[28641] 2193.German, J.; Chaganti, R. S. K.: Mapping human autosomes: assignment of the MN locus to a specific segment in the long arm of chromosome 2. Science 182: 1261-1262, 1973.

[28642] 2194.German, J.; Metaxas, M. N.; Metaxas-Buhler, M.; Louie, E.; Chaganti,R. S. K.: Further evaluation of a child with the M(k) phenotype and a translocation affecting the long arms of chromosomes 2 and 4. (Abstract) Cytogenet.Cell Genet. 25: 160, 1979.

[28643] 2195.German, J.; Walker, M. E.; Steifel, F. H.; Allen, F. H., Jr.:Autoradiographic studies of human chromosomes. II. Data concerning the position of the MN locus. Vox Sang. 16: 130-145, 1969.

[28644] 2196.German, J.; Walker, M. E.; Stiefel, F. H.; Allen, F. H.,

Jr.:MN blood-group locus: data concerning the possible chromosomal location. *Science* 162:1014–1015, 1968.

[28645] 2197.Grant, S. G.; Bigbee, W. L.: Bone marrow somatic mutation after genotoxic cancer therapy. (Letter) *Lancet* 343: 1507–1508, 1994.

[28646] 2198.Heiberg, A.; Berg, K.: Linkage data on the MNSs blood group–redcell acid phosphatase relationship. *Hum. Hered.* 25: 93–94, 1975.

[28647] 2199.Huang, C.–H.; Chen, Y.; Blumenfeld, O. O.: A novel St(a) glycophorin produced via gene conversion of pseudoexon III from glycophorin E to glycophorin A gene. *Hum. Mutat.* 15: 533–540, 2000.

[28648] 2200.Huang, C.–H.; Guizzo, M. L.; Kikuchi, M.; Blumenfeld, O. O.:Molecular genetic analysis of a hybrid gene encoding St(a) glycophorin of the human erythrocyte membrane. *Blood* 74: 836–843, 1989.

[28649] 2201.Huang, C.–H.; Puglia, K. V.; Bigbee, W. L.; Guizzo, M. L.; Hoffman, M.; Blumenfeld, O. O.: A family study of multiple mutations of alpha and delta glycophorins (glycophorins A and B). *Hum. Genet.* 81: 26–30, 1988.

[28650] 2202.Huang, C.–H.; Reid, M.; Daniels, G.; Blumenfeld, O. O.: Alteration of splice site selection by an exon mutation in the human glycophorin A gene. *J. Biol. Chem.* 268:

25902–25908, 1993.

- [28651] 2203.Huang, C.-H.; Spruell, P.; Moulds, J. J.; Blumenfeld, O. O.:Molecular basis for the human erythrocyte gly-cophorin specifying theMiltenberger class I (Mil) pheno-type. *Blood* 80: 257–263, 1992.
- [28652] 2204.Kudo, S.; Chagnovich, D.; Rearden, A.; Mattei, M. G.; Fukuda,M.: Molecular analysis of a hybrid gene encoding human glycophorinvariant Miltenberger V-like molecule. *J. Biol. Chem.* 265: 13825–13829,1990.
- [28653] 2205.Langlois, R. G.; Bigbee, W. L.; Jensen, R. H.: Mea-surements ofthe frequency of human erythrocytes with gene expression loss phenotypesat the glycophorin A lo-cus. *Hum. Genet.* 74: 353–362, 1986.
- [28654] 2206.Langlois, R. G.; Bigbee, W. L.; Kyoizumi, S.; Naka-mura, N.; Bean,M. A.; Akiyama, M.; Jensen, R. H.: Evidence for increased somaticcell mutations at the glycophorin A locus in atomic bomb survivors. *Science* 236:445–448, 1987.
- [28655] 2207.Mattei, M. G.; London, J.; Rahuel, C.; d'Auriol, L.; Colin, Y.;Le Van Kim, C.; Mattei, J. F.; Galibert, F.; Cartron, J. P.: Chromosomelocalization by in situ hybridization of the gene for human erythrocyteglycophorin to region 4q28–q31. (Abstract) *Cytogenet. Cell Genet.* 46:658,

1987.

- [28656] 2208.Mawby, W. J.; Anstee, D. J.; Tanner, M. J. A.: Immunochemical evidence for hybrid sialoglycoproteins of human erythrocytes. *Nature* 291:161–162, 1981.
- [28657] 2209.Okubo, Y.; Daniels, G. L.; Parsons, S. F.; Anstee, D. J.; Yamaguchi, H.; Tomita, T.; Seno, T.: A Japanese family with two sisters apparently homozygous for M(k). *Vox Sang.* 54: 107–111, 1988.
- [28658] 2210.Onda, M.; Fukuda, M.: Detailed physical mapping of the genes encoding glycoporphins A, B, and E, as revealed by P1 plasmids containing human genomic DNA. *Gene* 159: 225–230, 1995.
- [28659] 2211.Pasvol, G.; Wainscoat, J. S.; Weatherall, D. J.: Erythrocytes deficient in glycoporphin resist invasion by the malarial parasite *Plasmodium falciparum*. *Nature* 297: 64–66, 1982.
- [28660] 2212.Prohaska, R.; Koerner, T. A. W., Jr.; Armitage, I. M.; Furthmayr, H.: Chemical and carbon-13 nuclear magnetic resonance studies of the blood group M and N active sialoglycopeptides from human glycoporphin A. *J. Biol. Chem.* 256: 5781–5791, 1986.
- [28661] 2213.Rahuel, C.; London, J.; d'Auriol, L.; Mattei, M.-G.; Tournamille, C.; Skrzynia, C.; Lebouc, Y.; Galibert, F.;

Cartron, J.-P.: Characterization of cDNA clones for human glycophorin A: use for gene localization and for analysis of normal of glycophorin-A-deficient (Finnish type) genomic DNA. *Europ. J. Biochem.* 172: 147–153, 1988.

[28662] 2214. Rahuel, C.; London, J.; Vignal, A.; Cherif-Zahar, B.; Colin, Y.; Siebert, P.; Fukuda, M.; Cartron, J.-P.: Alteration of the genes for glycophorin A and B in glycophorin-A-deficient individuals. *Europ. J. Biochem.* 177: 605–614, 1988.

[28663] 2215. Rothman, N.; Haas, R.; Hayes, R. B.; Li, G.-L.; Wiemels, J.; Campleman, S.; Quintana, P. J. E.; Xi, L.-J.; Dosemeci, M.; Titenko-Holland, N.; Meyer, K. B.; Lu, W.; Zhang, L. P.; Bechtold, W.; Wang, Y.-Z.; Kolachana, P.; Yin, S.-N.; Blot, W.; Smith, M. T.: Benzene induces gene-duplicating but not gene-inactivating mutations at the glycophorin A locus in exposed humans. *Proc. Nat. Acad. Sci.* 92: 4069–4073, 1995.

[28664] 2216. Siebert, P. D.; Fukuda, M.: Isolation and characterization of human glycophorin A cDNA clones by a synthetic oligonucleotide approach: nucleotide sequence and mRNA structure. *Proc. Nat. Acad. Sci.* 83: 1665–1669, 1986.

[28665] 2217. Spence, M. A.; Field, L. L.; Marazita, M. L.; Joseph, J.; Sparkes, M.; Crist, M.; Crandall, B. F.; Anderson, C. E.;

Bateman, J. B.; Rotter, J. I.; Kidd, K. K.; Hodge, S. E.; Sparkes, R. S.: Estimating the recombination frequency for the MN and the Ss loci. *Hum. Hered.* 34:343–347, 1984.

[28666] 2218. Springer, G. F.; Tegtmeyer, H.: Further evidence that carbohydrates are the immunodeterminant structures of blood group M and N specificities. *Immun. Commun.* 10: 157–171, 1981.

[28667] 2219. Egan, M. F.; Goldberg, T. E.; Kolachana, B. S.; Callicott, J. H.; Mazzanti, C. M.; Straub, R. E.; Goldman, D.; Weinberger, D. R.: Effect of COMT val(108/158)met genotype on frontal lobe function and risk for schizophrenia. *Proc. Nat. Acad. Sci.* 98: 6917–6922, 2001.

[28668] 2220. Floderus, Y.; Iselius, L.; Lindsten, J.; Wetterberg, L.: Evidence for a major locus as well as a multifactorial component in the regulation of human red blood cell catechol-O-methyl-transferase activity. *Hum. Hered.* 32: 76–79, 1982.

[28669] 2221. Floderus, Y.; Wetterberg, L.: The inheritance of human erythrocyte catechol-O-methyltransferase activity. *Clin. Genet.* 19: 392–395, 1981.

[28670] 2222. Gogos, J. A.; Morgan, M.; Luine, V.; Santha, M.; Ogawa, S.; Pfaff, D.; Karayiorgou, M.: Catechol-O-methyltransferase-deficient mice exhibit sexually di-

morphic changes in catecholamine levels and behavior.

Proc.Nat. Acad. Sci. 95: 9991–9996, 1998.

[28671] 2223.Graf, W. D.; Unis, A. S.; Yates, C. M.; Sulzbacher, S.; Dinulos, M. B.; Jack, R. M.; Dugaw, K. A.; Paddock, M. N.; Parson, W. W.: Catecholamines in patients with 22q11.2 deletion syndrome and the low-activity COMT polymorphism. Neurology 57: 410–416, 2001.

[28672] 2224.Grossman, M. H.; Emanuel, B. S.; Budarf, M. L.: Chromosomal mapping of the human catechol-O-methyltransferase gene to 22q11.1–q11.2. Genomics 12:822–825, 1992.

[28673] 2225.Grossman, M. H.; Littrell, J.; Weinstein, R.; Punnett, H. H.; Emanuel, B. S.; Budarf, M.: The gene for human catechol-O-methyltransferase (COMT) maps to 22pter–22q11.1. (Abstract) Cytogenet. Cell Genet. 58:2048 only, 1991.

[28674] 2226.Gustavson, K. H.; Floderus, Y.; Jagell, S.; Wetterberg, L.; Ross, S. B.: Catechol-O-methyltransferase activity in erythrocytes in Down's syndrome: family studies. Clin. Genet. 22: 22–24, 1982.

[28675] 2227.Gustavson, K. H.; Wetterberg, L.; Backstrom, M.; Ross, S. B.: Catechol-O-methyltransferase activity in erythrocytes in Down's syndrome. Clin. Genet. 4: 279–280,

1973.

- [28676] 2228.Hoda, F.; Nicholl, D.; Bennett, P.; Arranz, M.; Aitchison, K.J.; Al-Chalabi, A.; Kunugi, H.; Vallada, H.; Leigh, P. N.; Chaudhuri, K. R.; Collier, D. A.: No association between Parkinson's disease and low-activity alleles of catechol O-methyltransferase. *Biochem. Biophys. Res. Commun.* 228: 780-784, 1996.
- [28677] 2229.Karayiorgou, M.; Altemus, M.; Galke, B. L.; Goldman, D. Murphy, D. L.; Ott, J.; Gogos, J. A.: Genotype determining low catechol-O-methyltransferase activity as a risk factor for obsessive-compulsive disorder. *Proc. Nat. Acad. Sci.* 94: 4572-4575, 1997.
- [28678] 2230.Lachman, H. M.; Morrow, B.; Shprintzen, R.; Veit, S.; Parsia, S. S.; Faedda, G.; Goldberg, R.; Kucherlapati, R.; Papoulos, D. F.: Association of codon 108/158 catechol-O-methyltransferase gene polymorphism with the psychiatric manifestations of velo-cardio-facial syndrome. *Am. J. Med. Genet.* 67: 468-472, 1996.
- [28679] 2231.Levitt, M.; Baron, M.: Human erythrocyte catechol-O-transferase: variation in thermal lability. (Abstract) Sixth Int. Cong. Hum. Genet., Jerusalem 21 only, 1981.
- [28680] 2232.Lundstrom, K.; Salminen, M.; Jalanko, A.; Savolainen, R.; Ulmanen, I.: Cloning and characterization of human

placental catechol-O-methyltransferase cDNA. DNA Cell Biol. 10: 181–189, 1991.

[28681] 2233. Scanlon, P. D.; Raymond, F. A.; Weinshilboum, R. M.: Catechol-O-methyltransferase: thermolabile enzyme in erythrocytes of subjects homozygous for allele for low activity. Science 203: 63–65, 1979.

[28682] 2234. Siervogel, R. M.; Weinshilboum, R.; Wilson, A. F.; Elston, R. C.: Major gene model for the inheritance of catechol-O-methyltransferase activity in five large families. Am. J. Med. Genet. 19: 315–323, 1984.

[28683] 2235. Spielman, R. S.; Weinshilboum, R. M.: Genetics of red cell COMT activity: analysis of thermal stability and family data. Am. J. Med. Genet. 10: 279–290, 1981.

[28684] 2236. Syvanen, A.-C.; Tilgmann, C.; Rinne, J.; Ulmanen, I.: Genetic polymorphism of catechol-O-methyltransferase (COMT): correlation of genotype with individual variation of S-COMT activity and comparison of the allele frequencies in the normal population and parkinsonian patients in Finland. Pharmacogenetics 7: 65–71, 1997.

[28685] 2237. Weinshilboum, R.; Dunnette, J.: Thermal stability and the biochemical genetics of erythrocyte catechol-O-methyltransferase and plasma dopamine-beta-hydroxylase. Clin. Genet. 19: 426–437, 1981.

- [28686] 2238.Weinshilboum, R. M.; Raymond, F. A.: Inheritance of low erythrocyte catechol-O-methyltransferase activity in man. *Am. J. Hum. Genet.* 29:125–135, 1977.
- [28687] 2239.Rouault, J.-P.; Rimokh, R.; Tessa, C.; Paranhos, G.; Ffrench, M.; Duret, L.; Garoccio, M.; Germain, D.; Samarut, J.; Magaud, J.-P.: BTG1, a member of a new family of anti-proliferative genes. *EMBO J.* 11:1663–1670, 1992.
- [28688] 2240.Couch, F. J.; Abel, K. J.; Brody, L. C.; Boehnke, M.; Collins, F. S.; Weber, B. L.: Localization of the gene for ATP citrate lyase (ACLY) distal to gastrin (GAS) and proximal to D17S856 on chromosome 17q12–q21. *Genomics* 21: 444–446, 1994.
- [28689] 2241.Elshourbagy, N. A.; Near, J. C.; Kmetz, P. J.; Sathe, G. M.; Southan, C.; Strickler, J. E.; Gross, M.; Young, J. F.; Wells, T. N. C.; Groot, P. H. E.: Rat ATP citrate-lyase: molecular cloning and sequence analysis of a full-length cDNA and mRNA abundance as a function of diet, organ, and age. *J. Biol. Chem.* 265: 1430–1435, 1990.
- [28690] 2242.Elshourbagy, N. A.; Near, J. C.; Kmetz, P. J.; Wells, T. N. C.; Groot, P. H. E.; Saxty, B. A.; Hughes, S. A.; Franklin, M.; Gloger, I. S.: Cloning and expression of a human ATP-citrate lyase cDNA. *Europ. J. Biochem.* 204: 491–499, 1992.

- [28691] 2243.Chang, Y. J.; McCabe, R. T.; Rennert, H.; Budarf, M. L.; Sayegh,R.; Emanuel, B. S.; Skolnick, P.; Strauss, J. F., III: The human 'peripheral-type' benzodiazepine receptor: regional mapping of the gene and characterization of the receptor expressed from cDNA. *DNACell Biol.* 11: 471–480, 1992.
- [28692] 2244.Lin, D.; Chang, Y. J.; Strauss, J. F., III; Miller, W. L.: The human peripheral benzodiazepine receptor gene: cloning and characterization of alternative splicing in normal tissues and in a patient with congenital lipoid adrenal hyperplasia. *Genomics* 18: 643–650, 1993.
- [28693] 2245.Riond, J.; Mattei, M. G.; Kaghad, M.; Dumont, X.; Guillemot, J.C.; Le Fur, G.; Caput, D.; Ferrara, P.: Molecular cloning and chromosomal localization of a human peripheral-type benzodiazepine receptor. *Europ.J. Biochem.* 195: 305–311, 1991.
- [28694] 2246.Bachman, E. S.; Dhillon, H.; Zhang, C.–Y.; Cinti, S.; Bianco, A.C.; Kobilka, B. K.; Lowell, B. B.: Beta-AR signaling required for diet-induced thermogenesis and obesity resistance. *Science* 297:843–845, 2002.
- [28695] 2247.Frielle, T.; Collins, S.; Daniel, K. W.; Caron, M. G.; Lefkowitz, R. J.; Kobilka, B. K.: Cloning of the cDNA for the human beta-1-adrenergic receptor. *Proc. Nat. Acad. Sci.*

84: 7920–7924, 1987.

- [28696] 2248.Frielle, T.; Kobilka, B.; Lefkowitz, R. J.; Caron, M. G.: Humanbeta-1- and beta-2-adrenergic receptors: structurally and functionallyrelated receptors derived from distinct genes. *Trends Neurosci.* 11:321–324, 1988.
- [28697] 2249.Kannel, W. B.; Kannel, C.; Paffenbarger, R. S., Jr.; Cupples, L.A.: Heart rate and cardiovascular mortality: the Framingham Study. *Am.Heart J.* 113: 1489–1494, 1987.
- [28698] 2250.Kristal-Boneh, E.; Silber, H.; Harari, G.; Froom, P.: The associationof resting heart rate with cardiovascular, cancer and all-cause mortality:eight year follow-up of 3527 male Israeli employees (the CORDIS study). *Europ.Heart J.* 21: 116–124, 2000.
- [28699] 2251.Magnusson, Y.; Marullo, S.; Hoyer, S.; Waagstein, F.; Andersson,B.; Vahlne, A.; Guillet, J.-G.; Strosberg, A. D.; Hjalmarson, A.;Hoebeke, J.: Mapping of a functional autoimmune epitope on the beta(1)-adrenergicreceptor in patients with idiopathic dilated cardiomyopathy. *J. Clin.Invest.* 86: 1658–1663, 1990.
- [28700] 2252.Mason, D. A.; Moore, J. D.; Green, S. A.; Liggett, S. B.: A gain-of-functionpolymorphism in a G-protein coupling domain of the human beta-1-adrenergicreceptor. *J. Biol. Chem.* 274: 12670–12674, 1999.

- [28701] 2253.Ranade, K.; Jorgenson, E.; Sheu, W. H.-H.; Pei, D.; Hsiung, C.A.; Chiang, F.; Chen, Y. I.; Pratt, R.; Olshen, R. A.; Curb, D.; Cox,D. R.; Botstein, D.; Risch, N.: A polymorphism in the beta-1 adrenergicreceptor is associated with resting heart rate. *Am. J. Hum. Genet.* 70:935-942, 2002.
- [28702] 2254.Small, K. M.; Wagoner, L. E.; Levin, A. M.; Kardia, S. L. R.;Liggett, S. B.: Synergistic polymorphisms of beta-1- and alpha-2C-adrenergicreceptors and the risk of congestive heart failure. *New Eng. J. Med.* 347:1135-1142, 2002.
- [28703] 2255.Wilk, J. B.; Myers, R. H.; Zhang, Y.; Lewis, C. E.; Atwood, L.;Hopkins, P. N.; Ellison, R. C.: Evidence for a gene influencing heartrate on chromosome 4 among hypertensives. *Hum. Genet.* 111: 207-213,2002.
- [28704] 2256.Yang-Feng, T. L.; Xue, F.; Zhong, W.; Cotecchia, S.; Frielle,T.; Caron, M. G.; Lefkowitz, R. J.; Francke, U.: Chromosomal organizationof adrenergic receptor genes. *Proc. Nat. Acad. Sci.* 87: 1516-1520,1990.
- [28705] 2257.Benovic, J. L.; Stone, W. C.; Huebner, K.; Croce, C.; Caron, M.G.; Lefkowitz, R. J.: cDNA cloning and chromosomal localization ofthe human beta-adrenergic receptor kinase. *FEBS Lett.* 283: 122-126,1991.
- [28706] 2258.Harding, V. B.; Jones, L. R.; Lefkowitz, R. J.; Koch, W.

J.; Rockman, H. A.: Cardiac beta-ARK1 inhibition prolongs survival and augments beta blocker therapy in a mouse model of severe heart failure. *Proc. Nat. Acad. Sci.* 98: 5809–5814, 2001.

[28707] 2259. Penn, R. B.; Benovic, J. L.: Structure of the human gene encoding the beta-adrenergic receptor kinase. *J. Biol. Chem.* 269: 14924–14930, 1994.

[28708] 2260. Rockman, H. A.; Chien, K. R.; Choi, D.-J.; Iaccarino, G.; Hunter, J. J.; Ross, J., Jr.; Lefkowitz, R. J.; Koch, W. J.: Expression of a beta-adrenergic receptor kinase 1 inhibitor prevents the development of myocardial failure in gene-targeted mice. *Proc. Nat. Acad. Sci.* 95: 7000–7005, 1998.

[28709] 2261. Spurney, R. F.; Flannery, P. J.; Garner, S. C.; Athirakul, K.; Liu, S.; Guilak, F.; Quarles, L. D.: Anabolic effects of a G protein-coupled receptor kinase inhibitor expressed in osteoblasts. *J. Clin. Invest.* 109: 1361–1371, 2002.

[28710] 2262. Benovic, J. L.; Onorato, J. J.; Arriza, J. L.; Stone, W. C.; Lohse, M.; Jenkins, N. A.; Gilbert, D. J.; Copeland, N. G.; Caron, M. G.; Lefkowitz, R. J.: Cloning, expression, and chromosomal localization of beta-adrenergic receptor kinase 2: a new member of the receptor kinase family. *J. Biol. Chem.* 266: 14939–14946, 1991.

[28711] 2263. Jin, Y.; Xu, X. L.; Yang, M.-C. W.; Wei, F.; Ayi, T.-C.;

Bowcock,A. M.; Baer, R.: Cell cycle–dependent colocalization of BARD1 andBRCA1 proteins in discrete nuclear domains. Proc. Nat. Acad. Sci. 94:12075–12080, 1997.

- [28712] 2264.Lee, J.–S.; Collins, K. M.; Brown, A. L.; Lee, C.–H.; Chung, J.H.: hCds1–mediated phosphorylation of BRCA1 regulates the DNA damageresponse. Nature 404: 201–204, 2000.
- [28713] 2265.Hess, S. D.; Daggett, L. P.; Crona, J.; Deal, C.; Lu, C.–C.; Urrutia,A.; Chavez–Noriega, L.; Ellis, S. B.; Johnson, E. C.; Velicelebi,G.: Cloning and functional characterization of human heteromericN–methyl–D–aspartate receptors. J. Pharm. Exp. Ther. 278: 808–816,1996.
- [28714] 2266.Setou, M.; Nakagawa, T.; Seog, D.–H.; Hirokawa, N.: Kinesin superfamilymotor protein KIF17 and mLin–10 in NMDA receptor–containing vesicletransport. Science 288: 1796–1802, 2000.
- [28715] 2267.Sprengel, R.; Suchanek, B.; Amico, C.; Brusa, R.; Burnashev, N.;Rozov, A.; Hvalby, O.; Jensen, V.; Paulsen, O.; Andersen, P.; Kim,J. J.; Thompson, R. F.; Sun, W.; Webster, L. C.; Grant, S. G. N.;Eilers, J.; Konnerth, A.; Li, J.; McNameara, J. O.; Seeburg, P. H.: Importance of the intracellular domain of NR2 subunits for NMDAreceptor function in vivo. Cell 92: 279–289, 1998.

- [28716] 2268.Ishii, T.; Moriyoshi, K.; Sugihara, H.; Sakurada, K.; Kadotani,H.; Yokoi, M.; Akazawa, C.; Shigemoto, R.; Mizuno, N.; Masu, M.; Nakanishi,S.: Molecular characterization of the family of the N-methyl-D-aspartatereceptor subunits. *J. Biol. Chem.* 268: 2836–2843, 1993.
- [28717] 2269.Kalsi, G.; Whiting, P.; Le Bourdelles, B.; Callen, D.; Barnard,E. A.; Gurling, H.: Localization of the human NM-DAR2D receptor subunitgene (GRIN2D) to 19q13.1–qter, the NMDAR2A subunit gene to 16p13.2(GRIN2A), and the NMDAR2C subunit gene (GRIN2C) to 17q24–q25 usingso-matic cell hybrid and radiation hybrid mapping panels. *Genomics* 47:423–425, 1998.
- [28718] 2270.Monyer, H.; Sprengel, R.; Schoepfer, R.; Herb, A.; Higuchi, M.;Lomeli, H.; Burnashev, N.; Sakmann, B.; Seeburg, P. H.: HeteromericNMDA receptors: molecular and functional distinction of subtypes. *Science* 256:1217–1221, 1992.
- [28719] 2271.Sakimura, K.; Kutsuwada, T.; Ito, I.; Manabe, T.; Takayama, C.;Kushiya, E.; Yagi, T.; Aizawa, S.; Inoue, Y.; Sugiyama, H.; Mishina,M.: Reduced hippocampal LTP and spatial learning in mice lackingNMDA receptor epsilon 1 subunit. *Nature* 373: 151–155, 1995.
- [28720] 2272.DeChiara, T. M.; Vejsada, R.; Poueymirou, W. T.;

Acheson, A.; Suri,C.; Conover, J. C. Friedman, B.; McClain, J.; Pan, L.; Stahl, N.;Ip, N. Y.; Kato, A.; Yancopoulos, G. D.: Mice lacking the CNTF receptor,unlike mice lacking CNTF, exhibit profound motor neuron deficits atbirth. Cell 83: 313–322, 1995.

[28721] 2273.Davis, S.; Aldrich, T. H.; Valenzuela, D. M.; Wong, V.; Furth,M. E.; Squinto, S. P.; Yancopoulos, G. D.: The receptor for ciliaryneurotrophic factor. Science 253: 59–63, 1991.

[28722] 2274.Donaldson, D. H.; Britt, D. E.; Jones, C.; Jackson, C. L.; Patterson,D.: Localization of the gene for the ciliary neurotrophic factorreceptor (CNTFR) to human chromosome 9. Genomics 17: 782–784, 1993.

[28723] 2275.Valenzuela, D. M.; Rojas, E.; Le Beau, M. M.; Espinosa, R., III;Brannan, C. I.; McClain, J.; Masiakowski, P.; Ip, N. Y.; Copeland,N. G.; Jenkins, N. A.; Yancopoulos, G. D.: Genomic organization andchromosomal localization of the human and mouse genes encoding thealpha receptor component for ciliary neurotrophic factor. Genomics 25:157–163, 1995.

[28724] 2276.Wakui, K.; Nishida, T.; Masuda, J.; Itoh, T.; Katsumata, D.; Ohno,T.; Fukushima, Y.: De novo interstitial deletion of 4q[46,XX,del(4)(q27q28.2)]with intact blood

group-MN locus, confining its locus to 4q28.2-4q31.1.

Jpn.J. Hum. Genet. 36: 149-153, 1991.

[28725] 2277.Wilson, A. F.; Elston, R. C.; Siervogel, R. M.; Weinshilboum, R.; Ward, L. J.: Linkage relationships between a major gene for catechol-O-methyltransferase activity and 25 polymorphic marker systems. Am. J. Med. Genet.

19:525-532, 1984.

[28726] 2278.Mattera, R.; Graziano, M. P.; Yatani, A.; Zhou, Z.; Graf, R.; Codina, J.; Birnbaumer, L.; Gilman, A. G.; Brown, A. M.: Splice variants of the alpha subunit of the G protein G(8) activate both adenylyl cyclase and calcium channels.

Science 243: 804-807, 1989.

[28727] 2279.Mehlmann, L. M.; Jones, T. L. Z.; Jaffe, L. A.: Meiotic arrest in the mouse follicle maintained by a GS protein in the oocyte. Science 297:1343-1345, 2002.

[28728] 2280.Nerlich, A.; Peschel, O.; Lohrs, U.; Parsche, F.; Betz, P.: Juvenile gigantism plus polyostotic fibrous dysplasia in the Tegernsee giant.(Letter) Lancet 338: 886-887, 1991.

[28729] 2281.Patten, J. L.; Johns, D. R.; Valle, D.; Eil, C.; Gruppuso, P.A.; Steele, G.; Smallwood, P. M.; Levine, M. A.: Mutation in the gene encoding the stimulatory G protein of adenylylate cyclase in Albright's hereditary osteodystrophy. New Eng. J. Med. 322: 1412-1419, 1990.

- [28730] 2282.Persani, L.; Borgato, S.; Lania, A.; Filopanti, M.; Mantovani,G.; Conti, M.; Spada, A.: Relevant cAMP-specific phosphodiesteraseisoforms in human pituitary: effect of Gs-alpha mutations. J. Clin.Endocr. Metab. 86: 3795-3800, 2001.
- [28731] 2283.Peters, J.; Beechey, C. V.; Ball, S. T.; Evans, E. P.: Mappingstudies of the distal imprinting region of mouse chromosome 2. Genet.Res. 63: 169-174, 1994.
- [28732] 2284.Premawardhana, L. D. K. E.; Vora, J. P.; Mills, R.; Scanlon, M.F.: Acromegaly and its treatment in the McCune-Albright syndrome. Clin.Endocr. 36: 605-608, 1992.
- [28733] 2285.Riminucci, M.; Fisher, L. W.; Majolagbe, A.; Corsi, A.; Lala,R.; De Sanctis, C.; Robey, P. G.; Bianco, P.: A novel GNAS1 mutation,R201G, in McCune-Albright syndrome. J. Bone Miner. Res. 14: 1987-1989,1999.
- [28734] 2286.Schwindinger, W. F.; Francomano, C. A.; Levine, M. A.: Identificationof a mutation in the gene encoding the alpha subunit of the stimulatoryG-protein of adenylyl cyclase in McCune-Albright syndrome. Proc.Nat. Acad. Sci. 89: 5152-5156, 1992.
- [28735] 2287.Schwindinger, W. F.; Francomano, C. A.; Levine, M. A.; McKusick,V. A.: DNA light on the Tegernsee giant.

(Letter) Lancet 338: 1454–1455,1991.

- [28736] 2288.Shapira, H.; Mouallem, M.; Shapiro, M. S.; Weisman, Y.; Farfel,Z.: Pseudohypoparathyroidism type Ia: two new heterozygous frameshiftmutations in exons 5 and 10 of the Gs alpha gene. Human Genet. 97:73–75, 1995.
- [28737] 2289.Shenker, A.; Chanson, P.; Weinstein, L. S.; Chi, P.; Spiegel,A. M.; Lomri, A.; Marie, P. J.: Osteoblastic cells derived from isolatedlesions of fibrous dysplasia contain activating somatic mutationsof the G(S)–alpha gene. Hum. Molec. Genet. 4: 1675–1676, 1995.
- [28738] 2290.Shenker, A.; Weinstein, L. S.; Moran, A.; Pescovitz, O. H.; Charest,N. J.; Boney, C. M.; Van Wyk, J. J.; Merino, M. J.; Feuillan, P. P.;Spiegel, A. M.: Severe endocrine and nonendocrine manifestationsof the McCune–Albright syndrome associated with activating mutationsof stimulatory G protein Gs. J. Pediat. 123: 509–518, 1993.
- [28739] 2291.Shore, E. M.; Ahn, J.; Jan de Beur, S.; Li, M.; Xu, M.; Gardiner,R. J. M.; Zasloff, M. A.; Whyte, M. P.; Levine, M. A.; Kaplan, F.S.: Paternally inherited inactivating mutations of the GNAS1 genein progressive osseous heteroplasia. New Eng. J. Med. 346: 99–106,2002.
- [28740] 2292.Shore, E. M.; Kaplan, F. S.; Levine, M. A.: GNAS1 mutations andprogressive osseous heteroplasia. (Letter) New

Eng. J. Med. 346:1670–1671, 2002.

- [28741] 2293. Sparkes, R. S.; Cohn, V. H.; Mohandas, T.; Zollman, S.; Cire-Eversole, P.; Amatruda, T. T.; Reed, R. R.; Lochrie, M. A.; Simon, M. I.: Mapping of genes encoding the subunits of guanine nucleotide-binding protein (G-proteins) in humans. (Abstract) Cytogenet. Cell Genet. 46: 696 only, 1987.
- [28742] 2294. Tinschert, S.; Gerl, H.; Gewies, A.; Jung, H.-P.; Nurnberg, P.: McCune-Albright syndrome: clinical and molecular evidence of mosaicism in an unusual giant patient. Am. J. Med. Genet. 83: 100–108, 1999.
- [28743] 2295. Vallar, L.; Spada, A.; Giannattasio, G.: Altered Gs and adenylate cyclase activity in human GH-secreting pituitary adenomas. Nature 330: 566–568, 1987.
- [28744] 2296. Warner, D. R.; Gejman, P. V.; Collins, R. M.; Weinstein, L. S.: A novel mutation adjacent to the switch III domain of Gs-alpha in a patient with pseudohypoparathyroidism. Molec. Endocr. 11: 1718–1727, 1997.
- [28745] 2297. Warner, D. R.; Weinstein, L. S.: A mutation in the heterotrimeric stimulatory guanine nucleotide binding protein alpha-subunit with impaired receptor-mediated activation because of elevated GTPase activity. Proc. Nat. Acad. Sci. 96: 4268–4272, 1999.

- [28746] 2298. Warner, D. R.; Weng, G.; Yu, S.; Matalon, R.; Weinstein, L. S.: A novel mutation in the switch 3 region of Gs-alpha in a patient with Albright hereditary osteodystrophy impairs GDP binding and receptor activation. *J. Biol. Chem.* 273: 23976–23983, 1998.
- [28747] 2299. Weinstein, L. S.; Gejman, P. V.; de Mazancourt, P.; American, N.; Spiegel, A. M.: A heterozygous 4-bp deletion mutation in the Gs-alpha gene (GNAS1) in a patient with Albright hereditary osteodystrophy. *Genomics* 13: 1319–1321, 1992.
- [28748] 2300. Weinstein, L. S.; Gejman, P. V.; Friedman, E.; Kadowaki, T.; Collins, R. M.; Gershon, E. S.; Spiegel, A. M.: Mutations of the Gs alpha-subunit gene in Albright hereditary osteodystrophy detected by denaturing gradient gel electrophoresis. *Proc. Nat. Acad. Sci.* 87: 8287–8290, 1990.
- [28749] 2301. Weinstein, L. S.; Shenker, A.; Gejman, P. V.; Merino, M. J.; Friedman, E.; Spiegel, A. M.: Activating mutations of the stimulatory G protein in the McCune–Albright syndrome. *New Eng. J. Med.* 325: 1688–1695, 1991.
- [28750] 2302. Weinstein, L. S.; Yu, S.: The role of genomic imprinting of GS-alpha in the pathogenesis of Albright hereditary osteodystrophy. *Trends Endocr. Metab.* 10: 81–85, 1999.
- [28751] 2303. Walker, M. E.; Rubinstein, P.; Allen, F. H., Jr.: Bio-

chemicalgenetics of MN. Vox Sang. 32: 111–120, 1977.

[28752] 2304.Weitkamp, L. R.; Adams, M. S.; Rowley, P. T.: Linkage between the MN and Hb beta loci. Hum. Hered. 22: 566–572, 1972.

[28753] 2305.Weitkamp, L. R.; Lovrien, E. W.; Olaisen, B.; Fenger, K.; Gedde–Dahl, T., Jr.; Sorensen, S. A.; Conneally, P. M.; Bias, W. B.; Ott, J.: Linkage relations of the loci for the MN blood group and red cell phosphate. Birth Defects Orig. Art. Ser. XI(3): 276–280, 1975. Note: Alternate: Cytogenet. Cell Genet. 14: 446–450, 1975.....

[28754] 2306.Neer, E. J.; Schmidt, C. J.; Nambudripad, R.; Smith, T. F.: The ancient regulatory–protein family of WD–repeat proteins. Nature 371:297–300, 1994.

[28755] 2307.Byrd, P. J.; McConville, C. M.; Cooper, P.; Parkhill, J.; Stankovic, T.; McGuire, G. M.; Thick, J. A.; Taylor, A. M. R.: Mutations revealed by sequencing the 5–prime half of the gene for ataxia telangiectasia. Hum. Molec. Genet. 5: 145–149, 1996.

[28756] 2308.Chong, M. J.; Murray, M. R.; Gosink, E. C.; Russell, H. R. C.; Srinivasan, A.; Kapsetaki, M.; Korsmeyer, S. J.; McKinnon, P. J.: Atm and Bax cooperate in ionizing radiation–induced apoptosis in the central nervous system. Proc. Nat. Acad. Sci. 97: 889–894, 2000.

- [28757] 2309. Falck, J.; Mailand, N.; Syljuasen, R. G.; Bartek, J.; Lukas, J.: The ATM–Chk2–Cdc25A checkpoint pathway guards against radioresistant DNA synthesis. *Nature* 410: 842–847, 2001.
- [28758] 2310. Falck, J.; Petrini, J. H. J.; Williams, B. R.; Lukas, J.; Bartek, J.: The DNA damage–dependent intra–S phase checkpoint is regulated by parallel pathways. *Nature Genet.* 30: 290–294, 2002.
- [28759] 2311. Kanters, J. K.; Larsen, L. A.; Orholm, M.; Agner, E.; Anderson, P. S.; Vuust, J.; Christiansen, M.: Novel donor splice site mutation in the KVLQT1 gene is associated with the long QT syndrome. *J. Cardiovasc. Electrophysiol.* 9: 620–624, 1998.
- [28760] 2312. Itoh, T.; Kikuchi, K.; Odagawa, Y.; Takata, S.; Yano, K.; Okada, S.; Haneda, N.; Ogawa, S.; Nakano, O.; Kawahara, Y.; Kasai, H.; Nakayama, T.; Fukutomi, T.; Sakurada, H.; Shimizu, A.; Yazaki, Y.; Nagai, R.; Nakamura, Y.; Tanaka, T.: Correlation of genetic etiology with response to beta–adrenergic blockade among symptomatic patients with familial long–QT syndrome. *J. Hum. Genet.* 46: 38–40, 2001.
- [28761] 2313. Keating, M.: Response to Benhorin et al. (1993). *Science* 260:1962 only, 1993.

- [28762] 2314.Keating, M.; Atkinson, D.; Dunn, C.; Timothy, K.; Vincent, G.M.; Leppert, M.: Linkage of a cardiac arrhythmia, the long QT syndrome, and the Harvey RAS-1 gene. Science 252: 704-706, 1991.
- [28763] 2315.Keating, M.; Atkinson, D.; Dunn, C.; Timothy, K.; Vincent, G.M.; Leppert, M.: Long QT syndrome is closely linked to the HarveyRAS-1 gene on chromosome 11. (Abstract) Clin. Res. 39: 317A only, 1991.
- [28764] 2316.Keating, M.; Dunn, C.; Atkinson, D.; Timothy, K.; Vincent, G.M.; Leppert, M.: Consistent linkage of the long-QT syndrome to theHarvey Ras-1 locus on chromosome 11. Am. J. Hum. Genet. 49: 1335-1339, 1991.
- [28765] 2317.Kerem, B.; Benhorin, J.; Kalman, Y. M.; Medina, A.; Dyer, T. D.;Blangero, J.; MacCluer, J. W.: Evidence for genetic heterogeneity in the long QT syndrome. (Abstract) Am. J. Hum. Genet. 51 (suppl.):A192 only, 1992.
- [28766] 2318.Klein, H. O.; Levi, A.; Kaplinsky, E.; Di Segni, E.; David, D.: Congenital long-QT syndrome: deleterious effect of long-term high-rateventricular pacing and definitive treatment by cardiac transplantation. Am.Heart J. 132: 1079-1081, 1996.
- [28767] 2319.Ko, Y.-L.; Chen, S.-A.; Tang, T. K.; Lin, J.-L.; Chiang, C.-E.;Chen, J.-J.; Teng, M.-S.; Chang, M.-S.; Lien, W.-P.;

Wu, C.-W.: Noevidence for linkage of long QT syndrome and chromosome 11p15.5 markers in a Chinese family: evidence for genetic heterogeneity. *Hum. Genet.* 94:364–366, 1994.

[28768] 2320. Kukolich, M. K.; Telsey, A.; Ott, J.; Motulsky, A. G.: Sudden infant death syndrome: normal QT intervals on ECGs of relatives. *Pediatrics* 60:51–54, 1977.

[28769] 2321. Larsen, L. A.; Fosdal, I.; Andersen, P. S.; Kanter, J. K.; Vuust, J.; Wettrell, G.; Christiansen, M.: Recessive Romano-Ward syndrome associated with compound heterozygosity for two mutations in the KVLQT1 gene. *Europ. J. Hum. Genet.* 7: 724–728, 1999.

[28770] 2322. Lee, M. P.; et al.; et al.: Loss of imprinting of a paternally expressed transcript, with antisense orientation to KVLQT1, occurs frequently in Beckwith-Wiedemann syndrome and is independent of insulin-like growth factor II imprinting. *Proc. Nat. Acad. Sci.* 96: 5203–5208, 1999.

[28771] 2323. Lee, M. P.; Hu, R.-J.; Johnson, L. A.; Feinberg, A. P.: Human KVLQT1 gene shows tissue-specific imprinting and encompasses Beckwith-Wiedemann syndrome chromosomal rearrangements. *Nature Genet.* 15: 181–185, 1997.

[28772] 2324. Li, H.; Chen, Q.; Moss, A. J.; Robinson, J.; Goytia, V.; Perry, J. C.; Vincent, G. M.; Priori, S. G.; Lehmann, M. H.;

Denfield, S.W.; Duff, D.; Kaine, S.; Shimizu, W.; Schwartz, P. J.; Wang, Q.; Towbin, J. A.: New mutations in the KVLQT1 potassium channel that cause longQT syndrome. *Circulation* 97: 1264–1269, 1998.

[28773] 2325. Locati, E. H.; Zareba, W.; Moss, A. J.; Schwartz, P. J.; Vincent, G. M.; Lehmann, M. H.; Towbin, J. A.; Priori, S. G.; Napolitano, C.; Robinson, J. L.; Andrews, M.; Timothy, K.; Hall, W. J.: Age- and sex-related differences in clinical manifestations in patients with congenital long-QT syndrome. *Circulation* 97: 2237–2244, 1998.

[28774] 2326. Mannens, M.; Wilde, A.: KVLQT1, the rhythm of imprinting. *Nature Genet.* 15: 113–115, 1997.

[28775] 2327. Martini, B.: Personal Communication. Thiene, Italy 11/9/1998.

[28776] 2328. Melki, J.; Kaplan, J.; Lucet, V.; Halley, L.; Clemenceau, S.; Kaplan, C.; Baule, M. S.; Frezal, J.: Long QT (Romano–Ward) syndrome: further definition of the linkage to the HLA loci. (Abstract) *Cytogenet. Cell Genet.* 46: 661 only, 1987.

[28777] 2329. Milne, J. R.; Ward, D. E.; Spurrell, R. A. J.; Camm, A. J.: The long QT syndrome: effects of drugs and left stellate ganglion block. *Am. Heart J.* 104: 194–198, 1982.

[28778] 2330. Mitsutake, A.; Takeshita, A.; Kuroiwa, A.; Nakamura,

M.: Usefulness of the Valsalva maneuver in management of the long QT syndrome. *Circulation* 63:1029–1035, 1981.

[28779] 2331. Moss, A. J.; McDonald, J.: Unilateral cervico–thoracic sympathetic ganglionectomy for the treatment of long Q–T interval syndrome. *New Eng. J. Med.* 285: 903–904, 1971.

[28780] 2332. Moss, A. J.; Schwartz, P. J.: Sudden death and the idiopathic long Q–T syndrome. (Editorial) *Am. J. Med.* 66: 6–7, 1979.

[28781] 2333. Moss, A. J.; Schwartz, P. J.; Crampton, R. S.; Tzivoni, D.; Locati, E. H.; MacCluer, J.; Hall, W. J.; Weitkamp, L.; Vincent, G. M.; Garson, A., Jr.; Robinson, J. L.; Benhorin, J.; Choi, S.: The long QT syndrome: prospective longitudinal study of 328 families. *Circulation* 84:1136–1144, 1991.

[28782] 2334. Murray, A.; Donger, C.; Fenske, C.; Spillman, I.; Richard, P.; Dong, Y. B.; Neyroud, N.; Chevalier, P.; Denjoy, I.; Carter, N.; Syrris, P.; Afzal, A. P.; Patton, M. A.; Guicheney, P.; Jeffery, S.: Splicing mutations in KCNQ1: a mutation hot spot at codon 344 that produces in frame transcripts. *Circulation* 100: 1077–1084, 1999.

[28783] 2335. Neyroud, N.; Denjoy, I.; Donger, C.; Gary, F.; Villain, E.; Leenhardt, A.; Benali, K.; Schwartz, K.; Coumel, P.; Guicheney, P.: Heterozygous mutation in the pore of potassium channel gene KvLQT1 causes an apparently–

normal phenotype in long QT syndrome. *Europ. J. Hum. Genet.* 6: 129–133,1998.

[28784] 2336.Neyroud, N.; Richard, P.; Vignier, N.; Donger, C.; Denjoy, I.;Demay, L.; Shkolnikova, M.; Pesce, R.; Chevalier, P.; Hainque, B.;Coumel, P.; Schwartz, K.; Guicheney, P.: Genomic organization ofthe KCNQ1 K⁺ channel gene and identification of C-terminal mutationsin the long-QT syndrome. *Circ. Res.* 84: 290–297, 1999.

[28785] 2337.Heald, R.; McLoughlin, M.; McKeon, F.: Human wee1 maintains mitotictiming by protecting the nucleus from cytoplasmically activated cdc2kinase. *Cell* 74: 463–474, 1993.

[28786] 2338.Igarashi, M.; Nagata, A.; Jinno, S.; Suto, K.; Okayama, H.: Wee1(+)-likegene in human cells. *Nature* 353: 80–83, 1991.

[28787] 2339.Annunen, P.; Helaakoski, T.; Myllyharju, J.; Veijola, J.; Pihlajaniemi,T.; Kivirikko, K. I.: Cloning of the human prolyl 4-hydroxylase alphasubunit isoform alpha(II) and characterization of the type II enzymetetramer: the alpha(I) and alpha(II) subunits do not form a mixedalpha(I)alpha(II)beta2 tetramer. *J. Biol. Chem.* 272: 17342–17348,1997.

[28788] 2340.Friedman, L.; Higgin, J. J.; Moulder, G.; Barstead, R.;

Raines, R. T.; Kimble, J.: Prolyl 4-hydroxylase is required for viability and morphogenesis in *Caenorhabditis elegans*. *Proc. Nat. Acad. Sci.* 97:4736–4741, 2000.

[28789] 2341. Helaakoski, T.; Annunen, P.; Vuori, K.; MacNeil, I. A.; Pihlajaniemi, T.; Kivirikko, K. I.: Cloning, baculovirus expression, and characterization of a second mouse prolyl 4-hydroxylase alpha-subunit isoform: formation of an alpha(2)beta(2) tetramer with the protein disulfide-isomerase/beta subunit. *Proc. Nat. Acad. Sci.* 92: 4427–4431, 1995.

[28790] 2342. Helaakoski, T.; Vuori, K.; Myllyla, R.; Kivirikko, K. I.; Pihlajaniemi, T.: Molecular cloning of the alpha-subunit of human prolyl 4-hydroxylase: the complete cDNA-derived amino acid sequence and evidence for alternative splicing of RNA transcripts. *Proc. Nat. Acad. Sci.* 86: 4392–4396, 1989.

[28791] 2343. Pajunen, L.; Jones, T. A.; Helaakoski, T.; Pihlajaniemi, T.; Solomon, E.; Sheer, D.; Kivirikko, K. I.: Assignment of the gene coding for the alpha-subunit of prolyl 4-hydroxylase to human chromosome region 10q21.3–23.1. *Am. J. Hum. Genet.* 45: 829–834, 1989.

[28792] 2344. Helaakoski, T.; Veijola, J.; Vuori, K.; Rehn, M.; Chow,

L. T.;Taillon–Miller, P.; Kivirikko, K. I.; Pihlajaniemi, T.:
Structureand expression of the human gene for the alpha
subunit of prolyl 4–hydroxylase:the two alternatively
spliced types of mRNA correspond to two homologousex–
ons the sequences of which are expressed in a variety of
tissues. J.Biol. Chem. 269: 27847–27854, 1994.

[28793] 2345.Zelinski, T.; Coghlan, G.; Myal, Y.; Shiu, R. P. C.;
Philipps,S.; White, L.; Lewis, M.: Genetic linkage between
the Kell bloodgroup system and prolactin–inducible pro–
tein loci: provisional assignmentof KEL to chromosome 7.
Ann. Hum. Genet. 55: 137–140, 1991.

[28794] 2346.Bhalla, U. S.; Ram, P. T.; Iyengar, R.: MAP kinase
phosphataseas a locus of flexibility in a mitogen–activated
protein kinase signalingnetwork. Science 297:
1018–1023, 2002.

[28795] 2347.Boulton, T. G.; Nye, S. H.; Robbins, D. J.; Ip, N. Y.;
Radziejewska,E.; Morgenbesser, S. D.; DePinho, R. A.;
Panayotatos, N.; Cobb, M.H.; Yancopoulos, G. D.: ERKs: a
family of protein–serine/threoninekinases that are acti–
vated and tyrosine phosphorylated in responseto insulin
and NGF. Cell 65: 663–675, 1991.

[28796] 2348.Cobb, M. H.; Boulton, T. G.; Robbins, D. J.: Extracel–
lular signal–regulatedkinases: ERKs in progress. Cell

Regul. 2: 965–978, 1991.

[28797] 2349. Di Cristo, G.; Berardi, N.; Cancedda, L.; Pizzorusso, T.; Putignano, E.; Ratto, G. M.; Maffei, L.: Requirement of ERK activation for visual cortical plasticity. *Science* 292: 2337–2340, 2001.

[28798] 2350. Forcet, C.; Stein, E.; Pays, L.; Corset, V.; Llambi, F.; Tessier-Lavigne, M.; Mehlen, P.: Netrin-1-mediated axon outgrowth requires deleted in colorectal cancer-dependent MAPK activation. *Nature* 417: 443–447, 2002.

[28799] 2351. Khokhlatchev, A. V.; Canagarajah, B.; Wilsbacher, J.; Robinson, M.; Atkinson, M.; Goldsmith, E.; Cobb, M. H.: Phosphorylation of the MAP kinase ERK2 promotes its homodimerization and nuclear translocation. *Cell* 93: 605–615, 1998.

[28800] 2352. Li, L.; Wisk, M.; Gonzalez, F. A.; Davis, R. J.: Genomic loci of human mitogen-activated protein kinases. *Oncogene* 9: 647–649, 1994.

[28801] 2353. Owaki, H.; Makar, R.; Boulton, T. G.; Cobb, M. H.; Geppert, T. D.: Extracellular signal-regulated kinases in T cells: characterization of human ERK1 and ERK2 cDNAs. *Biochem. Biophys. Res. Commun.* 182: 1416–1422, 1992.

[28802] 2354. Saba-El-Leil, M. K.; Malo, D.; Meloche, S.: Chromosomal localization of the mouse genes encoding the ERK1

and ERK2 isoforms of MAP kinases. *MammalianGenome* 8: 141–142, 1997.

[28803] 2355.Stefanovsky, V. Y.; Pelletier, G.; Hannan, R.; Gagnon-Kugler,T.; Rothblum, L. I.; Moss, T.: An immediate response of ribosomaltranscription to growth factor stimulation in mammals is mediatedby ERK phosphorylation of UBF. *Molec. Cell* 8: 1063–1073, 2001.

[28804] 2356.Thomas, G.: MAP kinase by any other name smells just as sweet. *Cell* 68:3–6, 1992.

[28805] 2357.Garcia, J. I.; Zalba, G.; Detera-Wadleigh, S. D.; de Miguel, C.: Isolation of a cDNA encoding the rat MAP-kinase homolog of humanp63mapk. *Mammalian Genome* 7: 810–814, 1996.

[28806] 2358.Gonzalez, F. A.; Raden, D. L.; Rigby, M. R.; Davis, R. J.: Heterogeneousexpression of four MAP kinase isoforms in human tissues. *FEBS Lett.* 304:170–178, 1992.

[28807] 2359.Zhu, A. X.; Zhao, Y.; Moller, D. E.; Flier, J. S.: Cloning andcharacterization of p97(MAPK), a novel human homolog of rat ERK-3. *Molec.Cell. Biol.* 14: 8202–8211, 1994.

[28808] 2360.Alvaro, V.; Levy, L.; Dubray, C.; Roche, A.; Peillon, F.; Querat,B.; Joubert, D.: Invasive human pituitary tumors express a point-mutatedalpha-protein kinase-C. *J. Clin.*

Endocr. 77: 1125–1129, 1993.

- [28809] 2361. Coussens, L.; Parker, P. J.; Rhee, L.; Yang–Feng, T. L.; Chen, E.; Waterfield, M. D.; Francke, U.; Ullrich, A.: Multiple, distinct forms of bovine and human protein kinase C suggest diversity in cellular signaling pathways. *Science* 233: 859–866, 1986.
- [28810] 2362. Finkenzeller, G.; Marme, D.; Hug, H.: Sequence of human protein kinase C alpha. *Nucleic Acids Res.* 18: 2183 only, 1990.
- [28811] 2363. Jones, K. W.; Shaper, M. H.; Chevrette, M.; Fournier, R. E. K.: Subtractive hybridization cloning of a tissue-specific extinguisher: TSE1 encodes a regulatory subunit of protein kinase A. *Cell* 66: 861–872, 1991.
- [28812] 2364. Latos–Bielenska, A.; Klett, C.; Just, W.; Hameister, H.: Refinement of localization of the human genes for myeloperoxidase (MPO), protein kinase C, alpha polypeptide, PRKCA, and the DNA fragment D17S21 on chromosome 17q. *Hereditas* 115: 69–72, 1991.
- [28813] 2365. Linnenbach, A. J.; Huebner, K.; Reddy, E. P.; Herlyn, M.; Parmiter, A. H.; Nowell, P. C.; Koprowski, H.: Structural alteration in the MYB protooncogene and deletion within the gene encoding alpha-type protein kinase C in human melanoma cell lines. *Proc. Nat. Acad. Sci.* 85: 74–78, 1988.

- [28814] 2366.Parker, P. J.; Coussens, L.; Totty, N.; Rhee, L.; Young, S.; Chen,E.; Stabel, S.; Waterfield, M. D.; Ullrich, A.: The complete primarystructure of protein kinase C--the major phorbol ester receptor. *Science* 233:853–859, 1986.
- [28815] 2367.Summar, M. L.; Phillips, J. A., III; Krishnamani, M. R. S.; Keefer,J.; Trofatter, J.; Schwartz, R. C.; Tsipouras, P.; Willard, H.; Ullrich,A.: Protein kinase C: a new linkage marker for growth hormone andfor COL1A1. *Genomics* 5: 163–165, 1989.
- [28816] 2368.Bayes, M.; Valverde, D.; Balcells, S.; Grinberg, D.; Vi-lageliu,L.; Benitez, J.; Ayuso, C.; Beneyto, M.; Baiget, M.; Gonzalez–Duarte,R.: Evidence against involvement of re-coverin in autosomal recessiveretinitis pigmentosa in 42 Spanish families. *Hum. Genet.* 96: 89–94,1995.
- [28817] 2369.Dizhoor, A. M.; Ray, S.; Kumar, S.; Niemi, G.; Spencer, M.; Brolley,D.; Walsh, K. A.; Philipov, P. P.; Hurley, J. B.; Stryer, L.: Recoverin:a calcium sensitive activator of retinal rod guanylate cyclase. *Science* 251:915–918, 1991.
- [28818] 2370.Premkumar, L. S.; Ahern, G. P.: Induction of vanilloid receptorchannel activity by protein kinase C. *Nature* 408: 985–990, 2000.
- [28819] 2371.Al–Maghtheh, M.; Vithana, E. N.; Inglehearn, C. F.; Moore, T.;Bird, A. C.; Bhattacharya, S. S.: Segregation of a

PRKCG mutation in two RP11 families. (Letter) Am. J. Hum. Genet. 62: 1248–1252, 1998.

- [28820] 2372. Dryja, T. P.; McEvoy, J.; McGee, T. L.; Berson, E. L.: No mutations in the coding region of the PRKCG gene in three families with retinitis pigmentosa linked to the RP11 locus on chromosome 19q. (Letter) Am. J. Hum. Genet. 65: 926–928, 1999.
- [28821] 2373. Johnson, K. J.; Jones, P. J.; Spurr, N.; Nimmo, E.; Davies, J.; Creed, H.; Weiss, M.; Williamson, R.: Linkage relationships of the protein kinase C gamma gene which exclude it as a candidate for myotonic dystrophy. Cytogenet. Cell Genet. 48: 13–15, 1988.
- [28822] 2374. McGee, T. L.; Devoto, M.; Ott, J.; Berson, E. L.; Dryja, T. P.: Evidence that the penetrance of mutations at the RP11 locus causing dominant retinitis pigmentosa is influenced by a gene linked to the homologous RP11 allele. Am. J. Hum. Genet. 61: 1059–1066, 1997.
- [28823] 2375. Saunders, A. M.; Seldin, M. F.: The syntenic relationship of proximal mouse chromosome 7 and the myotonic dystrophy gene region on human chromosome 19q. Genomics 6: 324–332, 1990.
- [28824] 2376. Vithana, E. N.; Abu-Safieh, L.; Allen, M. J.; Carey, A.; Papaioannou, M.; Chakarova, C.; Al-Maghtheh, M.;

Ebenezer, N. D.; Willis, C.; Moore, A. T.; Bird, A. C.; Hunt, D. M.; Bhattacharya, S. S.: A human homolog of yeast pre-mRNA splicing gene, PRP31, underlies autosomal dominant retinitis pigmentosa on chromosome 19q13.4 (RP11). *Molec. Cell* 8:375–381, 2001.

[28825] 2377. Gong, J.; Xu, J.; Bezanilla, M.; van Huizen, R.; Derin, R.; Li, M.: Differential stimulation of PKC phosphorylation of potassium channels by ZIP1 and ZIP2. *Science* 285: 1565–1569, 1999.

[28826] 2378. Hayashi, N.; Yokoyama, N.; Seki, T.; Azuma, Y.; Ohba, T.; Nishimoto, T.: RanBP1, a Ras-like nuclear G protein binding to Ran/TC4, inhibits RCC1 via Ran/TC4. *Molec. Gen. Genet.* 247: 661–669, 1995.

[28827] 2379. Spritz, R. A.; Strunk, K.; Surowy, C. S.; Mohrenweiser, H. W.: Human U1-70K ribonucleoprotein antigen gene: organization, nucleotide sequence, and mapping to locus 19q13.3. *Genomics* 8: 371–379, 1990.

[28828] 2380. Epstein, A. C. R.; Gleadle, J. M.; McNeill, L. A.; Hewitson, K. S.; O'Rourke, J.; Mole, D. R.; Mukherji, M.; Metzen, E.; Wilson, M. I.; Dhanda, A.; Tian, Y.-M.; Masson, N.; Hamilton, D. L.; Jaakkola, P.; Barstead, R.; Hodgkin, J.; Maxwell, P. H.; Pugh, C. W.; Schofield, C. J.; Ratcliffe, P. J.: *C. elegans* EGL-9 and mammalian homologs define a fam-

ily of dioxygenases that regulate HIF by prolyl hydroxylation. Cell 107: 43–54, 2001.

- [28829] 2381. Gatei, M.; Young, D.; Cerosaletti, K. M.; Desai-Mehta, A.; Spring, K.; Kozlov, S.; Lavin, M. F.; Gatti, R. A.; Concannon, P.; Khanna, K.: ATM-dependent phosphorylation of p53 in response to radiation exposure. Nature Genet. 25: 115–119, 2000.
- [28830] 2382. Kai, R.; Ohtsubo, M.; Sekiguchi, M.; Nishimoto, T.: Molecular cloning of a human gene that regulates chromosome condensation and is essential for cell proliferation. Molec. Cell. Biol. 6: 2027–2032, 1986.
- [28831] 2383. Nishimoto, T.; Seino, H.; Seki, N.; Hori, T.-A.: The human CHC1 gene encoding RCC1 (regulator of chromosome condensation) (CHC1) is localized to human chromosome 1p36.1. Genomics 23: 719–721, 1994.
- [28832] 2384. Ohba, T.; Nakamura, M.; Nishitani, H.; Nishimoto, T.: Self-organization of microtubule asters induced in Xenopus egg extracts by GTP-bound Ran. Science 284: 1356–1358, 1999.
- [28833] 2385. Ohtsubo, M.; Kai, R.; Furuno, N.; Sekiguchi, T.; Sekiguchi, M.; Hayashida, H.; Kuma, K.; Miyata, T.; Fukushige, S.; Murotsu, T.; Matsubara, K.; Nishimoto, T.: Isolation and characterization of the active cDNA of the

human cell cycle gene (RCC1) involved in the regulation of onset of chromosome condensation. *Genes Dev.* 1: 585–593, 1987.

[28834] 2386. Naggert, J. K.; Mu, J.-L.: The mouse very low density lipoprotein receptor (Vldlr) gene maps to chromosome 19. *Mammalian Genome* 5:453–455, 1994.

[28835] 2387. Meloni, A. M.; Dobbs, R. M.; Pontes, J. E.; Sandberg, A. A.: Translocation(X;1) in papillary renal cell carcinoma: a new cytogenetic subtype. *Cancer Genet. Cytogenet.* 65: 1–6, 1993.

[28836] 2388. Zbar, B.; Tory, K.; Merino, M.; Schmidt, L.; Glenn, G.; Choyke, P.; Walther, M. M.; Lerman, M.; Linehan, W. M.: Hereditary papillary renal cell carcinoma. *J. Urol.* 151: 561–566, 1994.

[28837] 2389. Adachi, H.; Tawaragi, Y.; Inuzuka, C.; Kubota, I.; Tsujimoto, M.; Nishihara, T.; Nakazato, H.: Primary structure of human microsomal dipeptidase deduced from molecular cloning. *J. Biol. Chem.* 265:3992–3995, 1990.

[28838] 2390. Austruy, E.; Jeanpierre, C.; Antignac, C.; Whitmore, S. A.; Van Cong, N.; Bernheim, A.; Callen, D. F.; Junien, C.: Physical and genetic mapping of the dipeptidase gene DPEP1 to 16q24.3. *Genomics* 15: 684–687, 1993.

[28839] 2391. Campbell, B. J.; Forrester, L. J.; Zahler, W. L.; Burks,

M.: Beta-lactamase activity of purified and partially characterized human renal dipeptidase. *J. Biol. Chem.* 259: 14586–14590, 1984.

[28840] 2392. Kozak, E. M.; Tate, S. S.: Glutathione-degrading enzymes of microvillus membranes. *J. Biol. Chem.* 257: 6322–6327, 1982.

[28841] 2393. Nakagawa, H.; Inazawa, J.; Inoue, K.; Misawa, S.; Kashima, K.; Adachi, H.; Nakazato, H.; Abe, T.: Assignment of the human renal dipeptidase gene (DPEP1) to band q24 of chromosome 16. (Abstract) *Cytogenet. Cell Genet.* 58: 2002 only, 1991.

[28842] 2394. Adachi, T.; Ohta, H.; Yamada, H.; Futenma, A.; Kato, K.; Hirano, K.: Quantitative analysis of extracellular-superoxide dismutase in serum and urine by ELISA with monoclonal antibody. *Clin. Chim. Acta* 212:89–102, 1992.

[28843] 2395. Folz, R. J.; Crapo, J. D.: Extracellular superoxide dismutase (SOD3): tissue-specific expression, genomic characterization, and computer-assisted sequence analysis of the human EC SOD gene. *Genomics* 22:162–171, 1994.

[28844] 2396. Hendrickson, D. J.; Fisher, J. H.; Jones, C.; Ho, Y.-S.: Regional localization of human extracellular superoxide dismutase gene to 4pter–q21. *Genomics* 8: 736–738, 1990.

- [28845] 2397.Hjalmarsson, K.; Marklund, S. L.; Engstrom, A.; Edlund, T.: Isolationand sequence of complementary DNA encoding human extracellular superoxidedismutase. *Proc. Nat. Acad. Sci.* 84: 6340–6344, 1987.
- [28846] 2398.Marklund, S. L.: Extracellular superoxide dismutase in human tissuesand human cell lines. *J. Clin. Invest.* 74: 1398–1403, 1984.
- [28847] 2399.Sandstrom, J.; Nilsson, P.; Karlsson, K.; Marklund, S. L.: 10-foldincrease in human plasma extracellular superoxide dismutase contentcaused by a mutation in heparin-binding domain. *J. Biol. Chem.* 269:19163–19166, 1994.
- [28848] 2400.Yamada, H.; Yamada, Y.; Adachi, T.; Goto, H.; Ogasawara, N.; Futenma,A.; Kitano, M.; Hirano, K.; Kato, K.: Molecular analysis of extracellular–superoxidedismutase gene associated with high level in serum. *Jpn. J. Hum.Genet.* 40: 177–184, 1995.
- [28849] 2401.McNew, J. A.; Parlati, F.; Fukuda, R.; Johnston, R. J.; Paz, K.;Paumet, F.; Sollner, T. H.; Rothman, J. E.: Compartmental specificityof cellular membrane fusion encoded in SNARE proteins. *Nature* 407:153–159, 2000.
- [28850] 2402.Weber, T.; Zemelman, B. V.; McNew, J. A.; Westermann, B.; Gmachi,M.; Parlati, F.; Sollner, T. H.; Rothman, J. E.: SNAREpins: minimalmachinery for membrane fusion.

Cell 92: 759–772, 1998.

- [28851] 2403. Rheaume, E.; Simard, J.; Morel, Y.; Mebarki, F.; Zachmann, M.; Forest, M. G.; New, M. I.; Labrie, F.: Congenital adrenal hyperplasia due to point mutations in the type II 3-beta-hydroxysteroid dehydrogenase gene. *Nature Genet.* 1: 239–245, 1992.
- [28852] 2404. Amor, M.; Parker, K. L.; Globberman, H.; New, M. I.; White, P. C.: Mutation in the CYP21B gene (ile172-to-asn) causes steroid 21-hydroxylase deficiency. *Proc. Nat. Acad. Sci.* 85: 1600–1604, 1988.
- [28853] 2405. Araujo, M.; Sanches, M. R.; Suzuki, L. A.; Guerra, G., Jr.; Farah, S. B.; De Mello, M. P.: Molecular analysis of CYP21 and C4 genes in Brazilian families with the classical form of steroid 21-hydroxylase deficiency. *Braz. J. Med. Biol. Res.* 29: 1–13, 1996.
- [28854] 2406. Aston, C. E.; Sherman, S. L.; Morton, N. E.; Speiser, P. W.; New, M. I.: Genetic mapping of the 21-hydroxylase locus: estimation of small recombination frequencies. *Am. J. Hum. Genet.* 43: 304–310, 1988.
- [28855] 2407. Bachega, T. A. S. S.; Billerbeck, A. E. C.; Madureira, G.; Marcondes, J. A. M.; Longui, C. A.; Leite, M. V.; Arnhold, I. J. P.; Mendonca, B. B.: Molecular genotyping in Brazilian patients with the classical and nonclassical forms of

21-hydroxylase deficiency. J. Clin. Endocr.Metab. 83: 4416–4419, 1998.

[28856] 2408.Aguiar–Oliveira, M. H.; Gill, M. S.; de A. Barretto, E. S.; Alcantara,M. R. S.; Miraki–Moud, F.; Menezes, C. A.; Souza, A. H. O.; Martinelli,C. E.; Pereira, F. A.; Salvatori, R.; Levine, M. A.; Shalet, S. M.;Camacho–Hubner, C.; Clayton, P. E.: Effect of severe growth hormone(GH) deficiency due to a mutation in the GH–releasing hormone receptor on insulin–like growth factors (IGFs), IGF–binding proteins, and ternarycomplex formation throughout life. J. Clin. Endocr. Metab. 84: 4118–4126,1999.

[28857] 2409.DeAlmeida, V. I.; Mayo, K. E.: Identification and binding domains of the growth hormone–releasing hormone receptor by analysis of mutant and chimeric receptor proteins. Molec. Endocr. 12: 750–765, 1998.

[28858] 2410.Gaylinn, B. D.; Harrison, J. K.; Zysk, J. R.; Lyons, C. E.; Lynch,K. R.; Thorner, M. O.: Molecular cloning and expression of a human anterior pituitary receptor for growth hormone–releasing hormone. Molec.Endocr. 7: 77–84, 1993.

[28859] 2411.Gaylinn, B. D.; von Kap–Herr, C.; Golden, W. L.; Thorner, M. O.: Assignment of the human growth hormone–releasing hormone receptorgene (GHRHR) to 7p14

by in situ hybridization. *Genomics* 19: 193–195,1994.

- [28860] 2412. Godfrey, P.; Rahal, J. O.; Beamer, W. G.; Copeland, N. G.; Jenkins, N. A.; Mayo, K. E.: GHRH receptor of little mice contains a missense mutation in the extracellular domain that disrupts receptor function. *Nature Genet.* 4: 227–232, 1993.
- [28861] 2413. Gondo, R. G.; Aguiar-Oliveira, M. H.; Hayashida, C. Y.; Toledo, S. P. A.; Abelin, N.; Levine, M. A.; Bowers, C. Y.; Souza, A. H. O.; Pereira, R. M. C.; Santos, N. L.; Salvatori, R.: Growth hormone-releasing peptide-2 stimulates GH secretion in GH-deficient patients with mutated GH-releasing hormone receptor. *J. Clin. Endocr. Metab.* 86: 3279–3283, 2001.
- [28862] 2414. Lin, S.-C.; Lin, C. R.; Gukovsky, I.; Lysis, A. J.; Sawchenko, P. E.; Rosenfeld, M. G.: Molecular basis of the little mouse phenotype and implications for cell type-specific growth. *Nature* 364: 208–213, 1993.
- [28863] 2415. Maheshwari, H. G.; Silverman, B. L.; Dupuis, J.; Baumann, G.: Phenotype and genetic analysis of a syndrome caused by an inactivating mutation in the growth hormone-releasing hormone receptor: dwarfism of Sindh. *J. Clin. Endocr. Metab.* 83: 4065–4074, 1998.
- [28864] 2416. Netchine, I.; Talon, P.; Dastot, F.; Vitaux, F.;

Goossens, M.;Amselem, S.: Extensive phenotypic analysis of a family with growthhormone (GH) deficiency caused by a mutation in the GH–releasing hormonereceptor gene. J. Clin. Endocr. Metab. 83: 432–436, 1998.

[28865] 2417.Petersenn, S.; Rasch, A. C.; Heyens, M.; Schulte, H. M.: Structureand regulation of the human growth hormone–releasing hormone receptorgene. Molec. Endocr. 12: 233–247, 1998.

[28866] 2418.Roelfsema, F.; Biermasz, N. R.; Veldman, R. G.; Veldhuis, J. D.;Frolich, M.; Stovkis–Brantsma, W. H.; Wit, J.–M.: Growth hormone(GH) secretion in patients with an inactivating defect of the GH–releasinghormone (GHRH) receptor is pulsatile: evidence for a role for non–GHRHinputs into the generation of GH pulses. J. Clin. Endocr. Metab. 86:2459–2464, 2001.

[28867] 2419.Salvatori, R.; Fan, X.; Mullis, P. E.; Haile, A.; Levine, M. A.: Decreased expression of the GHRH receptor gene due to a mutationin a Pit–1 binding site. Molec. Endocr. 16: 450–458, 2002.

[28868] 2420.Salvatori, R.; Fan, X.; Phillips, J. A., III; Espigares–Martin,R.; de Lara, I. M.; Freeman, K. L.; Plotnick, L.; Al–Ashwal, A.; Levine,M. A.: Three new mutations in the gene for the growth hormone (GH)–releasinghormone receptor

in familial isolated GH deficiency type IB. J. Clin. Endocr. Metab. 86: 273–279, 2001.

- [28869] 2421. Salvatori, R.; Hayashida, C. Y.; Aguiar-Oliveira, M. H.; Phillips, J. A., III; Souza, A. H. O.; Gondo, R. G.; Toledo, S. P. A.; Conceicao, M. M.; Prince, M.; Maheshwari, H. G.; Baumann, G.; Levine, M. A.: Familial dwarfism due to a novel mutation of the growth hormone–releasing hormone receptor gene. J. Clin. Endocr. Metab. 84: 917–923, 1999.
- [28870] 2422. Vamvakopoulos, N. C.; Kunz, J.; Olberding, U.; Scherer, S. W.; Sioutopoulou, T. O.; Schneider, V.; Durkin, A. S.; Nierman, W. C.: Mapping the human growth hormone–releasing hormone receptor (GHRHR) gene to the short arm of chromosome 7 (7p13–p21) near the epidermal growth factor receptor (EGFR) gene. Genomics 20: 338–340, 1994.
- [28871] 2423. Wajnrajch, M. P.; Gertner, J. M.; Harbison, M. D.; Chua, S. C., Jr.; Leibel, R. L.: Nonsense mutation in the human growth hormone–releasing hormone receptor causes growth failure analogous to the little (lit) mouse. Nature Genet. 12: 88–90, 1996.
- [28872] 2424. Borsani, G.; Bassi, M. T.; Sperandio, M. P.; De Grandi, A.; Buoninconti, A.; Riboni, M.; Manzoni, M.; Incerti, B.; Pepe, A.; Andria, G.; Ballabio, A.; Sebastio, G.:

SLC7A7, encoding a putative permease-related protein, is mutated in patients with lysinuric protein intolerance. *Nature Genet.* 21: 297–301, 1999.

[28873] 2425. Ghizzoni, L.; Mastorakos, G.; Street, M. E.; Vottero, A.; Mazzardo, G.; Vanelli, M.; Chrousos, G. P.; Bernasconi, S.: Spontaneous thyrotropin and cortisol secretion interactions in patients with nonclassical 21-hydroxylase deficiency and control children. *J. Clin. Endocr. Metab.* 82: 3677–3683, 1997.

[28874] 2426. Globerman, H.; Amor, H.; Parker, K. L.; New, M. I.; White, P. C.: A nonsense mutation causing steroid 21-hydroxylase deficiency. *J. Clin. Invest.* 82: 139–144, 1988.

[28875] 2427. Gordon, M. T.; Conway, D. I.; Anderson, D. C.; Harris, R.: Genetics and biochemical variability of variants of 21 hydroxylase deficiency. *J. Med. Genet.* 22: 354–360, 1985.

[28876] 2428. Gotoh, H.; Sagai, T.; Hata, J.-I.; Shiroishi, T.; Moriwaki, K.: Steroid 21-hydroxylase deficiency in mice. *Endocrinology* 123:1923–1927, 1988.

[28877] 2429. Grosse-Wilde, H.; Weil, J.; Albert, E.; Scholz, S.; Bidlingmaier, F.; Sippel, W. G.; Knorr, D.: Genetic linkage studies between congenital adrenal hyperplasia and the

HLA blood group system. Immunogenetics 8:41–49, 1979.

- [28878] 2430. Gueux, B.; Fiet, J.; Couillin, P.; Raux-Demay, M.-C.; Mornet, E.; Galons, H.; Villette, J.-M.; Boue, J.; Dreux, C.: Prenatal diagnosis of 21-hydroxylase deficiency congenital adrenal hyperplasia by simultaneous radioimmunoassay of 21-deoxycortisol and 17-hydroxyprogesterone in amniotic fluid. J. Clin. Endocr. Metab. 66: 534–537, 1988.
- [28879] 2431. Gutai, J. P.; Lee, P. A.; Johnsonbaugh, R. E.; Gareis, F.; Urban, M. D.; Migeon, C. J.: Detection of the heterozygous state in siblings of patients with congenital adrenal hyperplasia due to 21-hydroxylase deficiency. J. Pediatr. 94: 770–772, 1979.
- [28880] 2432. Haglund-Stengler, B.; Ritzen, E. M.; Gustafsson, J.; Luthman, H.: Haplotypes of the steroid 21-hydroxylase gene region encoding mild steroid 21-hydroxylase deficiency. Proc. Nat. Acad. Sci. 88:8352–8356, 1991.
- [28881] 2433. Harada, F.; Kimura, A.; Iwanaga, T.; Shimozawa, K.; Yata, J.; Sasazuki, T.: Gene conversion-like events cause steroid 21-hydroxylase deficiency in congenital adrenal hyperplasia. Proc. Nat. Acad. Sci. 84:8091–8094, 1987.
- [28882] 2434. Helmberg, A.; Tusie-Luna, M. T.; Tabarelli, M.; Kofler, R.; White, P. C.: R339H and P453S: CYP21 mutations associated with nonclassic steroid 21-hydroxylase

deficiency that are not apparent gene conversions.

Molec.Endocr. 6: 1318–1322, 1992.

[28883] 2435.Higashi, Y.; Tanae, A.; Inoue, H.; Fujii–Kuriyama, Y.: Evidencefor frequent gene conversion in the steroid 21–hydroxylase P–450(C21)gene: implications for steroid 21–hydroxylase deficiency. Am. J.Hum. Genet. 42: 17–25, 1988.

[28884] 2436.Higashi, Y.; Tanae, A.; Inoue, H.; Hiromasa, T.; Fujii–Kuriyama,Y.: Aberrant splicing and missense mutations cause steroid 21–hydroxylase(P–450[C21]) deficiency in humans: possible gene conversion products. Proc.Nat. Acad. Sci. 85: 7486–7490, 1988.

[28885] 2437.Higashi, Y.; Yoshioka, H.; Yamane, M.; Gotoh, O.; Fujii–Kuriyama,Y.: Complete nucleotide sequence of two steroid 21–hydroxylase genestandemly arranged in human chromosome: a pseudogene and a genuinegene. Proc. Nat. Acad. Sci. 83: 2841–2845, 1986.

[28886] 2438.Hirschfeld, A. J.; Fleshman, J. K.: An unusually high incidenceof salt–losing congenital adrenal hyperplasia in the Alaskan Eskimo. J.Pediat. 75: 492–494, 1969.

[28887] 2439.Holler, W.; Scholz, S.; Knorr, D.; Bidlingmaier, F.; Keller, E.;Albert, E. D.: Genetic differences between the salt–wasting, simplevirilizing, and nonclassical types of

congenital adrenal hyperplasia. J.Clin. Endocr. Metab. 60: 757–763, 1985.

[28888] 2440. Hughes, I. A.; Dyas, J.; Riad-Fahmy, D.; Laurence, K. M.: Prenatal diagnosis of congenital adrenal hyperplasia: reliability of amniotic fluid steroid analysis. J. Med. Genet. 24: 344–347, 1987.

[28889] 2441. Jaaskelainen, J.; Levo, A.; Voutilainen, R.; Partanen, J.: Population-wide evaluation of disease manifestation in relation to molecular genotype in steroid 21-hydroxylase (CYP21) deficiency: good correlation in a well defined population. J. Clin. Endocr. Metab. 82: 3293–3297, 1997.

[28890] 2442. Jaresch, S.; Kornely, E.; Kley, H.-K.; Schlaghecke, R.: Adrenal incidentaloma and patients with homozygous or heterozygous congenital adrenal hyperplasia. J. Clin. Endocr. Metab. 74: 685–689, 1992.

[28891] 2443. Jones, H. W., Jr.: Personal Communication. Baltimore, Md. 2/10/1978.

[28892] 2444. Jospe, N.; Donohoue, P. A.; Van Dop, C.; McLean, R. H.; Bias, W. B.; Migeon, C. J.: Prevalence of polymorphic 21-hydroxylase gene (CA21HB) mutations in salt-losing congenital adrenal hyperplasia. Biochem. Biophys. Res. Commun. 142: 798–804, 1987.

[28893] 2445. Kadair, R. G.; Block, M. B.; Katz, F. H.; Hofeldt, F. D.:

'Masked' 21-hydroxylase deficiency of the adrenal presenting with gynecomastia and bilateral testicular masses. Am. J. Med. 62: 278–282, 1977.

- [28894] 2446. Kawaguchi, H.; O'Huigin, C.; Klein, J.: Evolutionary origin of mutations in the primate cytochrome P450c21 gene. Am. J. Hum. Genet. 50: 766–780, 1992.
- [28895] 2447. Kirby-Keyser, L.; Porter, C. C.; Donohue, P. A.: E380D: a novel point mutation of CYP21 in an HLA-homozygous patient with salt-losing congenital adrenal hyperplasia due to 21-hydroxylase deficiency. Hum. Mutat. 9: 181–182, 1997.
- [28896] 2448. Kirkland, R. T.; Kirkland, J. L.; Keenan, B. S.; Bongiovanni, A. M.; Rosenberg, H. S.; Clayton, G. W.: Bilateral testicular tumors in congenital adrenal hyperplasia. J. Clin. Endocr. Metab. 44: 369–378, 1977.
- [28897] 2449. Klouda, P. T.; Harris, R.; Price, D. A.: Linkage and association between HLA and 21-hydroxylase deficiency. J. Med. Genet. 17: 337–341, 1980.
- [28898] 2450. Knochelhauer, E. S.; Cortet-Rudelli, C.; Cunningham, R. D.; Conway-Myers, B. A.; Dewailly, D.; Azziz, R.: Carriers of 21-hydroxylase deficiency are not at increased risk for hyperandrogenism. J. Clin. Endocr. Metab. 82: 479–485, 1997.

- [28899] 2451.Krone, N.; Braun, A.; Roscher, A. A.; Knorr, D.; Schwarz, H. P.: Predicting phenotype in steroid 21-hydroxylase deficiency? Comprehensive genotyping in 155 unrelated, well defined patients from Southern Germany. *J.Clin. Endocr. Metab.* 85: 1059–1065, 2000.
- [28900] 2452.Itoh, H.; Tomita, M.; Uchino, H.; Kobayashi, T.; Kataoka, H.; Sekiya, R.; Nawa, Y.: cDNA cloning of rat pS2 peptide and expression of trefoil peptides in acetic acid-induced colitis. *Biochem. J.* 318: 939–944, 1996.
- [28901] 2453.May, F. E. B.; Westley, B. R.: Close physical linkage of the genes encoding the pNR-2/pS2 protein and human spasmodic protein (hSP). *Hum.Genet.* 99: 303–307, 1997.
- [28902] 2454.Seib, T.; Blin, N.; Hilgert, K.; Seifert, M.; Theisinger, B.; Engel, M.; Dooley, S.; Zang, K.-D.; Welter, C.: The three human trefoil genes TFF1, TFF2, and TFF3 are located within a region of 55 kb on chromosome 21q22.3. *Genomics* 40: 200–202, 1997.
- [28903] 2455.Tomasetto, C.; Rio, M.-C.; Gautier, C.; Wolf, C.; Hareuveni, M.; Chambon, P.; Lathe, R.: hSP, the domain-duplicated homolog of pS2 protein, is co-expressed with pS2 in stomach but not in breast carcinoma. *EMBOJ.* 9: 407–414, 1990.

- [28904] 2456. Brose, N.; Petrenko, A. G.; Sudhof, T. C.; Jahn, R.: Synaptotagmin: a calcium sensor on the synaptic vesicle surface. *Science* 256: 1021–1025, 1992.
- [28905] 2457. Fernandez-Chacon, R.; Konigstorfer, A.; Gerber, S. H.; Garcia, J.; Matos, M. F.; Stevens, C. F.; Brose, N.; Rizo, J.; Rosenmund, C.; Sudhof, T. C.: Synaptotagmin I functions as a calcium regulator of release probability. *Nature* 410: 41–49, 2001.
- [28906] 2458. Geppert, M.; Archer, B. T., III; Sudhof, T. C.: Synaptotagmin II: a novel differentially distributed form of synaptotagmin. *J. Biol. Chem.* 266: 13548–13552, 1991.
- [28907] 2459. Hilbush, B. S.; Morgan, J. I.: A third synaptotagmin gene, Syt3, in the mouse. *Proc. Nat. Acad. Sci.* 91: 8195–8199, 1994.
- [28908] 2460. Jones, J. M.; Popma, S. J.; Mizuta, M.; Seino, S.; Meisler, M. H.: Synaptotagmin genes on mouse chromosomes 1, 7, and 10 and human chromosome 19. *Mammalian Genome* 6: 212–213, 1995.
- [28909] 2461. Kwon, O.-J.; Adamson, M. C.; Chin, H.; Kozak, C. A.: Genetic mapping of five mouse genes encoding synaptotagmins. *Mammalian Genome* 6: 880–881, 1995.
- [28910] 2462. Mackler, J. M.; Drummond, J. A.; Loewen, C. A.; Robinson, I. M.; Reist, N. E.: The C2B Ca^{2+} -binding motif

of synaptotagmin is required for synaptic transmission in vivo. *Nature* 418: 340–344, 2002.

[28911] 2463. Perin, M. S.; Fried, V. A.; Mignery, G. A.; Jahn, R.; Sudhof, T. C.: Phospholipid binding by a synaptic vesicle protein homologous to the regulatory region of protein kinase C. *Nature* 345: 260–263, 1990.

[28912] 2464. Perin, M. S.; Johnston, P. A.; Ozcelik, T.; Jahn, R.; Francke, U.; Sudhof, T. C.: Structural and functional conservation of synaptotagmin(p65) in *Drosophila* and humans. *J. Biol. Chem.* 266: 615–622, 1991.

[28913] 2465. Robinson, I. M.; Ranjan, R.; Schwarz, T. L.: Synaptotagmins I and IV promote transmitter release independently of Ca^{2+} binding in the C2A domain. *Nature* 418: 336–340, 2002.

[28914] 2466. Shin, O.-H.; Rizo, J.; Sudhof, T. C.: Synaptotagmin function in dense core vesicle exocytosis studied in cracked PC12 cells. *Nature Neurosci.* 5: 649–656, 2002.

[28915] 2467. Wang, C.-T.; Grishanin, R.; Earles, C. A.; Chang, P. Y.; Martin, T. F. J.; Chapman, E. R.; Jackson, M. B.: Synaptotagmin modulation of fusion pore kinetics in regulated exocytosis of dense-core vesicles. *Science* 294: 1111–1115, 2001.

[28916] 2468. Begley, C. G.; Aplan, P. D.; Davey, M. P.; Nakahara,

K.; Tchorz,K.; Kurtzberg, J.; Hershfield, M. S.; Haynes, B. F.; Cohen, D. I.;Waldmann, T. A.; Kirsch, I. R.: Chromosomal translocation in a humanleukemic stem-cell line disrupts the T-cell antigen receptor delta-chain diversity region and results in a previously unreported fusion transcript. Proc.Nat. Acad. Sci. 86: 2031–2035, 1989.

[28917] 2469.Kikuchi, M.; Tayama, T.; Hayakawa, H.; Takahashi, I.; Hoshino,H.; Ohsaka, A.: Familial thrombocytosis. Brit. J. Haemat. 89: 900–902,1995.

[28918] 2470.Wiestner, A.; Schlemper, R. J.; van der Maas, A. P. C.; Skoda,R. C.: An activating splice donor mutation in the thrombopoietingene causes hereditary thrombocythaemia. Nature Genet. 18: 49–52,1998.

[28919] 2471.Hruban, R. H.; Goggins, M.; Parsons, J.; Kern, S. E. :Clin.Cancer Res. 6: 2969–2972, 2000.

[28920] 2472.Hruban, R. H.; van Mansfield, A. D. M.; Offerhaus, G. J. A.; vanWeering, D. H. J.; Allison, D. C.; Goodman, S. N., Kensler, T. W.;Bose, K. K.; Cameron, J. L.; Bos, J. L. :Am. J. Path. 143: 545–554,1993.

[28921] 2473.Hruban, R. H.; Wilentz, R. E.; Kern, S. E. :Am. J. Pathol. 156:1821–1825, 2000.

[28922] 2474.Ruivenkamp, C. A. L.; van Wezel, T.; Zanon, C.; Stassen, A. P.M.; Vlcek, C.; Csikos, T.; Klous, A. M.;

Tripodis, N.; Perrakis, A.; Boerrigter, L.; Groot, P. C.; Lindeman, J.; Mooi, W. J.; Meijjer, G. A.; Scholten, G.; Dauwerse, H.; Paces, V.; van Zandwijk, N.; van Ommen, G. J. B.; Demant, P.: Ptp^{prj} is a candidate for the mouse colon-cancer susceptibility locus Scc1 and is frequently deleted in human cancers. *Nature Genet.* 31: 295–300, 2002.

[28923] 2475. Watanabe, T.; Mukoyama, Y.; Rhodes, M.; Thomas, M.; Kume, T.; Oishi, M.: Chromosomal location of murine protein tyrosine phosphatase (Ptp^{prj} and Ptp^{pre}) genes. *Genomics* 29: 793–795, 1995.

[28924] 2476. Elson, A.; Kozak, C. A.; Morton, C. C.; Weremowicz, S.; Leder, P.: The protein tyrosine phosphatase epsilon gene maps to mouse chromosome 7 and human chromosome 10q26. *Genomics* 31: 373–375, 1996.

[28925] 2477. Krueger, N. X.; Streuli, M.; Saito, H.: Structural diversity and evolution of human receptor-like protein tyrosine phosphatases. *EMBO J.* 9: 3241–3252, 1990.

[28926] 2478. Melhado, I. G.; Anderson, L. L.; Duncan, A. M. V.; Jirik, F. R.: The gene encoding protein-tyrosine phosphatase RPTP-epsilon (PTP^{RE}) is assigned to human chromosome 10q26. *Cytogenet. Cell Genet.* 73:168–170, 1996.

[28927] 2479. Banerjee, P.; Kleyn, P. W.; Knowles, J. A.; Lewis, C. A.;

Ross, B. M.; Parano, E.; Kovats, S. G.; Lee, J. J.; Pen-
chaszadeh, G. K.; Ott, J.; Jacobson, S. G.; Gilliam, T. C.:
TULP1 mutation in two extended Dominican kindreds with
autosomal recessive retinitis pigmentosa. *Nature Genet.*
18: 177–179, 1998.

[28928] 2480. Hagstrom, S. A.; North, M. A.; Nishina, P. M.; Berson,
E. L.; Dryja, T. P.: Recessive mutations in the gene encod-
ing the tubby-like protein TULP1 in patients with retinitis
pigmentosa. *Nature Genet.* 18: 174–176, 1998.

[28929] 2481. Knowles, J. A.; Shugart, Y.; Banerjee, P.; Gilliam, T.
C.; Lewis, C. A.; Jacobson, S. G.; Ott, J.: Identification of a
locus, distinct from RDS-peripherin, for autosomal reces-
sive retinitis pigmentosa on chromosome 6p. *Hum. Molec.*
Genet. 3: 1401–1403, 1994.

[28930] 2482. Iivanainen, A.; Sainio, K.; Sariola, H.; Tryggvason, K.:
Primary structure and expression of a novel human laminin
alpha-4 chain. *FEBS Lett.* 365: 183–188, 1995.

[28931] 2483. Richards, A. J.; Al-Imara, L.; Carter, N. P.; Lloyd, J.
C.; Leversha, M. A.; Pope, F. M.: Localization of the gene
(LAMA4) to chromosome 6q21 and isolation of a partial
cDNA encoding a variant laminin A chain. *Genomics* 22:
237–239, 1994.

[28932] 2484. Richards, A. J.; Al-Imara, L.; Carter, N. P.; Lloyd, J.

C.; Pope, F. M.: A laminin A variant gene (LAMA3) is present on chromosome 6q21 (Abstract) *J. Med. Genet.* 31: 164, 1994.

[28933] 2485. Dorow, D. S.; Devereux, L.; Dietzsch, E.; De Kretser, T.: Identification of a new family of human epithelial protein kinases containing two leucine/isoleucine-zipper domains. *Europ. J. Biochem.* 213: 701–710, 1993.

[28934] 2486. Hanks, S. K.: Eukaryotic protein kinases. *Curr. Opin. Struct. Biol.* 1: 369–383, 1991.

[28935] 2487. Balczon, R.; Bao, L.; Zimmer, W. E.: PCM-1, a 228-kD centrosome autoantigen with a distinct cell cycle distribution. *J. Cell Biol.* 124: 783–793, 1994.

[28936] 2488. Graham, M.; Shutter, J. R.; Sarmiento, U.; Sarosi, I.; Stark, K. L.: Overexpression of Agt leads to obesity in transgenic mice. (Letter) *Nature Genet.* 17: 273–274, 1997.

[28937] 2489. Kanetsky, P. A.; Swoyer, J.; Panossian, S.; Holmes, R.; Guerry, D.; Rebbeck, T. R.: A polymorphism in the agouti signaling protein gene is associated with human pigmentation. *Am. J. Hum. Genet.* 70: 770–775, 2002.

[28938] 2490. Klebig, M. L.; Wilkinson, J. E.; Geisler, J. G.; Woychik, R. P.: Ectopic expression of the agouti gene in transgenic mice causes obesity, features of type II diabetes, and yel-

low fur. Proc. Nat.Acad. Sci. 92: 4728–4732, 1995.

- [28939] 2491.Kwon, H. Y.; Bultman, S. J.; Loffler, C.; Chen, W.–J.; Furdon,P. J.; Powell, J. G.; Usala, A.–L.; Wilkison, W.; Hansmann, I.; Woychik,R. P.: Molecular structure and chromosomal mapping of the human homolog of the agouti gene. Proc. Nat. Acad. Sci. 91: 9760–9764, 1994.
- [28940] 2492.Manne, J.; Argeson, A. C.; Siracusa, L. D.: Mechanisms for the pleiotropic effects of the agouti gene. Proc. Nat. Acad. Sci. 92:4721–4724, 1995.
- [28941] 2493.Miltenberger, R. J.; Mynatt, R. L.; Bruce, B. D.; Wilkison, W.O.; Woychik, R. P.; Michaud, E. J.: An agouti mutation lacking the basic domain induces yellow pigmentation but not obesity in transgenic mice. Proc. Nat. Acad. Sci. 96: 8579–8584, 1999.
- [28942] 2494.Wilson, B. D.; Ollmann, M. M.; Kang, L.; Stoffel, M.; Bell, G.I.; Barsh, G. S.: Structure and function of ASP, the human homolog of the mouse agouti gene. Hum. Molec. Genet. 4: 223–230, 1995.
- [28943] 2495.Zemel, M. B.; Kim, J. H.; Woychik, R. P.; Michaud, E. J.; Kadwell,S. H.; Patel, I. R.; Wilkison, W. O.: Agouti regulation of intracellular calcium: role in the insulin resistance of viable yellow mice. Proc.Nat. Acad. Sci. 92: 4733–4737, 1995.

- [28944] 2496. Cardon, L. R.; Smith, S. D.; Fulker, D. W.; Kimberling, W. J.; Pennington, B. F.; DeFries, J. C.: Quantitative trait locus for reading disability on chromosome 6. *Science* 266: 276–279, 1994. Note: Erratum: *Science* 268: 1553 only, 1995.
- [28945] 2497. Field, L. L.; Kaplan, B. J.: Absence of linkage of phonological coding dyslexia to chromosome 6p23–p21.3 in a large family data set. *Am. J. Hum. Genet.* 63: 1448–1456, 1998.
- [28946] 2498. Fisher, S. E.; Francks, C.; Marlow, A. J.; MacPhie, I. L.; Newbury, D. F.; Cardon, L. R.; Ishikawa-Brush, Y.; Richardson, A. J.; Talcott, J. B.; Gayán, J.; Olson, R. K.; Pennington, B. F.; Smith, S. D.; DeFries, J. C.; Stein, J. F.; Monaco, A. P.: Independent genome-wide scans identify a chromosome 18 quantitative-trait locus influencing dyslexia. *Nature-Genet.* 30: 86–91, 2002.
- [28947] 2499. Fisher, S. E.; Marlow, A. J.; Lamb, J.; Maestrini, E.; Williams, D. F.; Richardson, A. J.; Weeks, D. E.; Stein, J. F.; Monaco, A. P.: A quantitative-trait locus on chromosome 6p influences different aspects of developmental dyslexia. *Am. J. Hum. Genet.* 64: 146–156, 1999.
- [28948] 2500. Gayán, J.; Smith, S. D.; Cherny, S. S.; Cardon, L. R.; Fulker, D. W.; Brower, A. M.; Olson, R. K.; Pennington, B. F.;

DeFries, J.C.: Quantitative-trait locus for specific language and reading deficit on chromosome 6p. *Am. J. Hum. Genet.* 64: 157–164, 1999.

[28949] 2501. Kaplan, D. E.; Gayan, J.; Ahn, J.; Won, T.-W.; Pauls, D.; Olson, R. K.; DeFries, J. C.; Wood, F.; Pennington, B. F.; Page, G. P.; Smith, S. D.; Gruen, J. R.: Evidence for linkage and association with reading disability, on 6p21.3–22. *Am. J. Hum. Genet.* 70: 1287–1298, 2002.

[28950] 2502. Smith, S. D.; Kimberling, W. J.; Pennington, B. F.: Screening for multiple genes influencing dyslexia. *Reading Writing* 3: 285–298, 1991.

[28951] 2503. Fazioli, F.; Minichiello, L.; Matoska, V.; Castagnino, P.; Miki, T.; Wong, W. T.; Di Fiore, P. P.: Eps8, a substrate for the epidermal growth factor receptor kinase, enhances EGF-dependent mitogenic signals. *EMBO J.* 12: 3799–3808, 1993.

[28952] 2504. Corvi, R.; Berger, N.; Balczon, R.; Romeo, G.: RET/PCM-1: a novel fusion gene in papillary thyroid carcinoma. *Oncogene* 19: 4236–4242, 2000.

[28953] 2505. Ohata, H.; Fujiwara, Y.; Koyama, K.; Nakamura, Y.: Mapping of the human autoantigen pericentriolar material 1 (PCM1) gene to chromosome 8p21.3–p22. *Genomics* 24: 404–406, 1994.

- [28954] 2506. Andresen, B. S.; Christensen, E.; Corydon, T. J.; Bross, P.; Pilgaard, B.; Wanders, R. J. A.; Ruiter, J. P. N.; Simonsen, H.; Winter, V.; Knudsen, I.; Schroeder, L. D.; Gregersen, N.; Skovby, F.: Iso-lated 2-methylbutyrylglycinuria caused by short/branched-chain acyl-CoA dehydrogenase deficiency: identification of a new enzyme defect, resolution of its molecular basis, and evidence for distinct acyl-CoA dehydrogenases in isoleucine and valine metabolism. *Am. J. Hum. Genet.* 67: 1095–1103, 2000.
- [28955] 2507. Takai, S.; Tanaka, M.; Sugimura, H.; Yamada, K.; Naito, Y.; Kino, I.; Matsuda, M.: Mapping of the human C3G gene coding a guanine nucleotide-releasing protein for Ras family to 9q34.3 by fluorescence in situ hybridization. *Hum. Genet.* 94: 549–550, 1994.
- [28956] 2508. Tanaka, S.; Morishita, T.; Hashimoto, Y.; Hattori, S.; Nakamura, S.; Shibuya, M.; Matsuoka, K.; Takenawa, T.; Kurata, T.; Nagashima, K.; Matsuda, M.: C3G, a guanine nucleotide-releasing protein expressed ubiquitously, binds to the Src homology 3 domains of CRK and GRB2/ASH proteins. *Proc. Nat. Acad. Sci.* 91: 3443–3447, 1994.
- [28957] 2509. Belich, M. P.; Glynn, R. J.; Senger, G.; Sheer, D.;

Trowsdale, J.: Proteasome components with reciprocal expression to that of the MHC-encoded LMP proteins. *Curr. Biol.* 4: 769–776, 1994.

[28958] 2510. McCusker, D.; Jones, T.; Sheer, D.; Trowsdale, J.: Genetic relationships of the genes encoding the human proteasome beta subunits and the proteasome PA28 complex. *Genomics* 45: 362–367, 1997.

[28959] 2511. Kumar, S.; Tomooka, Y.; Noda, M.: Identification of a set of genes with developmentally down-regulated expression in the mouse brain. *Biochem. Biophys. Res. Commun.* 185: 1155–1161, 1992.

[28960] 2512. Braybrooke, J. P.; Spink, K. G.; Thacker, J.; Hickson, I. D.: The RAD51 family member, RAD51L3, is a DNA-stimulated ATPase that forms a complex with XRCC2. *J. Biol. Chem.* 275: 29100–29106, 2000.

[28961] 2513. Johnson, R. D.; Liu, N.; Jasin, M.: Mammalian XRCC2 promotes the repair of DNA double-strand breaks by homologous recombination. *Nature* 401: 397–399, 1999.

[28962] 2514. Jones, N. J.; Cox, R.; Thacker, J.: Isolation and cross-sensitivity of x-ray-sensitive mutants of V79–4 hamster cells. *Mutat. Res.* 183: 279–286, 1987.

[28963] 2515. Jones, N. J.; Zhao, Y.; Siciliano, M. J.; Thompson, L. H.: Assignment of the XRCC2 human DNA repair gene to

chromosome 7q36 by complementation analysis. *Genomics* 26: 619–622, 1995.

- [28964] 2516. Kurumizaka, H.; Ikawa, S.; Nakada, M.; Enomoto, R.; Kagawa, W.; Kinebuchi, T.; Yamazoe, M.; Yokoyama, S.; Shibata, T.: Homologous pairing and ring and filament structure formation activities of the human Xrcc2–Rad51D complex. *J. Biol. Chem.* 277: 14315–14320, 2002.
- [28965] 2517. Liu, N.; Schild, D.; Thelen, M. P.; Thompson, L. H.: Involvement of Rad51C in two distinct protein complexes of Rad51 paralogs in human cells. *Nucleic Acids Res.* 30: 1009–1015, 2002.
- [28966] 2518. Masson, J.-Y.; Tarsounas, M. C.; Stasiak, A. Z.; Stasiak, A.; Shah, R.; McIlwraith, M. J.; Benson, F. E.; West, S. C.: Identification and purification of two distinct complexes containing the five RAD51 paralogs. *Genes Dev.* 15: 3296–3307, 2001.
- [28967] 2519. Tambini, C. E.; George, A. M.; Rommens, J. M.; Tsui, L.-C.; Scherer, S. W.; Thacker, J.: The XRCC2 DNA repair gene: identification of a positional candidate. *Genomics* 41: 84–92, 1997.
- [28968] 2520. Thacker, J.; Tambini, C. E.; Simpson, P. J.; Tsui, L.-C.; Scherer, S. W.: Localization to chromosome 7q36.1 of the human XRCC2 gene, determining sensitivity to

DNA-damaging agents. *Hum. Molec. Genet.* 4:113–120, 1995.

[28969] 2521.Kjeldsen, A. D.; Brusgaard, K.; Poulsen, L.; Kruse, T.; Rasmussen, K.; Green, A.: Mutations in the ALK-1 gene and the phenotype of hereditary hemorrhagic telangiectasia in two large Danish families. *Am. J. Med. Genet.* 98: 298–302, 2001.

[28970] 2522.Nakagawara, A.; Liu, X.-G.; Ikegaki, N.; White, P. S.; Yamashiro, D. J.; Nycum, L. M.; Biegel, J. A.; Brodeur, G. M.: Cloning and chromosomal localization of the human TRK-B tyrosine kinase receptor gene (NTRK2). *Genomics* 25:538–546, 1995.

[28971] 2523.Rico, B.; Xu, B.; Reichardt, L. F.: TrkB receptor signaling is required for establishment of GABAergic synapses in the cerebellum. *Nature Neurosci.* 5: 225–233, 2002.

[28972] 2524.Slaugenhaupt, S. A.; Blumenfeld, A.; Liebert, C. B.; Mull, J.; Lucente, D. E.; Monahan, M.; Breakefield, X. O.; Maayan, C.; Parada, L.; Axelrod, F. B.; Gusella, J. F.: The human gene for neurotrophic tyrosine kinase receptor type 2 (NTRK2) is located on chromosome 9 but is not the familial dysautonomia gene. *Genomics* 25: 730–732, 1995.

[28973] 2525.Soppet, D.; Escandon, E.; Maragos, J.; Middlemas, D. S.; Reid, S. W.; Blair, J.; Burton, L. E.; Stanton, B. R.; Kaplan,

D. R.; Hunter, T.; Nikolics, K.; Parada, L. F.: The neurotrophic factors brain-derived neurotrophic factor and neurotrophin-3 are ligands for the trkB tyrosine kinase receptor. *Cell* 65: 895–903, 1991.

[28974] 2526. Squinto, S. P.; Stitt, S. N.; Aldrich, T. H.; Davis, S.; Bianco, S. M.; Radziejewski, C.; Glass, D. J.; Masiakowski, P.; Furth, M. E.; Valenzuela, D. M.; DiStefano, P. S.; Yancopoulos, G. D.: trkB encodes a functional receptor for brain-derived neurotrophic factor and neurotrophin-3 but not nerve growth factor. *Cell* 65: 885–893, 1991.

[28975] 2527. Grun, F.; Hirose, Y.; Kawauchi, S.; Ogura, T.; Umesono, K.: Aldehyde dehydrogenase 6, a cytosolic retinaldehyde dehydrogenase prominently expressed in sensory neuroepithelia during development. *J. Biol. Chem.* 275: 41210–41218, 2000.

[28976] 2528. Hsu, L. C.; Chang, W.-C.; Hiraoka, L.; Hsieh, C.-L.: Molecular cloning, genomic organization, and chromosomal localization of an additional human aldehyde dehydrogenase gene, ALDH6. *Genomics* 24:333–341, 1994.

[28977] 2529. Kapfhamer, D.; Miller, D. E.; Lambert, S.; Bennett, V.; Glover, T. W.; Burmeister, M.: Chromosomal localization of the ankyrin-G gene (ANK3/Ank3) to human 10q21 and mouse 10. *Genomics* 27: 189–191, 1995.

- [28978] 2530.Bennett, M.; Reed, R.: Correspondence between a mammalian spliceosomecomponent and an essential yeast splicing factor. *Science* 262: 105–108,1993.
- [28979] 2531.Dresser, D. W.; Hacker, A.; Lovell–Badge, R.; Guerrier, D.: Thegenes for a spliceosome protein (SAP62) and the anti–Mullerian hormone(AMH) are contiguous. *Hum. Molec. Genet.* 4: 1613–1618, 1995.
- [28980] 2532.Burks, D. J.; Font de Mora, J.; Schubert, M.; Withers, D. J.; Myers,M. G.; Towery, H. H.; Altamuro, S. L.; Flint, C. L.; White, M. F.: IRS–2 pathways integrate female reproduction and energy homeostasis. *Nature* 407:377–382, 2000.
- [28981] 2533.Fritsche, A.; Madaus, A.; Renn, W.; Tschritter, O.; Teigeler, A.;Weisser, M.; Maerker, E.; Machicao, F.; Haring, H.; Stumvoll, M.:The prevalent Gly1057Asp polymorphism in the insulin receptor substrate–2gene is not associated with impaired insulin secretion. *J. Clin.Endocr. Metab.* 86: 4822–4825, 2001.
- [28982] 2534.Kubota, N.; Tobe, K.; Terauchi, Y.; Eto, K.; Yamauchi, T.; Suzuki,R.; Tsubamoto, Y.; Komeda, K.; Nakano, R.; Miki, H.; Satoh, S.; Sekihara,H.; Sciacchitano, S.; Lesniak, M.; Aizawa, S.; Nagai, R.; Kimura,S.; Akanuma, Y.; Taylor, S. I.; Kadowaki, T.: Disruption of insulinreceptor substrate

2 causes type 2 diabetes because of liver insulin resistance and lack of compensatory beta-cell hyperplasia. *Diabetes* 49:1880–1889, 2000.

[28983] 2535. Sun, X. J.; Wang, L.-M.; Zhang, Y.; Yenush, L.; Myers, M. G., Jr.; Glasheen, E.; Lane, W. S.; Pierce, J. H.; White, M. F.: Role of IRS-2 in insulin and cytokine signalling. *Nature* 377: 173–177, 1995.

[28984] 2536. Withers, D. J.; Burks, D. J.; Towery, H. H.; Altamuro, S. L.; Flint, C. L.; White, M. F.: Irs-2 coordinates Igf-1 receptor-mediated beta-cell development and peripheral insulin signalling. *Nature Genet.* 23:32–40, 1999.

[28985] 2537. Withers, D. J.; Gutierrez, J. S.; Towery, H.; Burks, D. J.; Ren, J.-M.; Previs, S.; Zhang, Y.; Bernal, D.; Pons, S.; Shulman, G. I.; Bonner-Weir, S.; White, M. F.: Disruption of IRS-2 causes type 2 diabetes in mice. *Nature* 391: 900–902, 1998.

[28986] 2538. Eberle, F.; Dubreuil, P.; Mattei, M.-G.; Devilard, E.; Lopez, M.: The human PRR2 gene, related to the human poliovirus receptor gene (PVR), is the true homolog of the murine Mph gene. *Gene* 159: 267–272, 1995.

[28987] 2539. Morrison, M. E.; Racaniello, V. R.: Molecular cloning and expression of a murine homolog of the human poliovirus receptor gene. *J. Virol.* 66:2807–2813, 1992.

- [28988] 2540.Wong, W. T.; Carlomagno, F.; Druck, T.; Barletta, C.; Croce, C.M.; Huebner, K.; Kraus, M. H.; Di Fiore, P. P.: Evolutionary conservation of the EPS8 gene and its mapping to human chromosome 12q23–q24. *Oncogene* 9:3057–3061, 1994.
- [28989] 2541.Avraham, K. B.; Levanon, D.; Negreanu, V.; Bernstein, Y.; Groner, Y.; Copeland, N. G.; Jenkins, N. A.: Mapping of the mouse homolog of the human runt domain gene, AML2, to the distal region of mouse chromosome 4. *Genomics* 25: 603–605, 1995.
- [28990] 2542.Bae, S.–C.; Takahashi, E.; Zhang, Y. W.; Ogawa, E.; Shigesada, K.; Namba, Y.; Satake, M.; Ito, Y.: Cloning, mapping and expression of PEBP2–alpha–C, a third gene encoding the mammalian Runt domain. *Gene* 159:245–248, 1995.
- [28991] 2543.Inoue, K.; Ozaki, S.; Shiga, T.; Ito, K.; Masuda, T.; Okado, N.; Iseda, T.; Kawaguchi, S.; Ogawa, M.; Bae, S.–C.; Yamashita, N.; Itohara, S.; Kudo, N.; Ito, Y.: Runx3 controls the axonal projection of proprioceptive dorsal root ganglion neurons. *Nature Neurosci.* 23Sept, 2002.
Note: Advance Electronic Publication.
- [28992] 2544.Li, Q.–L.; Ito, K.; Sakakura, C.; Fukamachi, H.; Inoue, K.; Chi, X.–Z.; Lee, K.–Y.; Nomura, S.; Lee, C.–W.; Han,

S.-B.; Kim, H.-M.; Kim, W.-J.; and 15 others: Causal relationship between the loss of RUNX3 expression and gastric cancer. *Cell* 109: 113–124, 2002.

[28993] 2545. Wijmenga, C.; Speck, N. A.; Dracopoli, N. C.; Hofker, M. H.; Liu, P.; Collins, F. S.: Identification of a new murine runt domain-containing gene, Cbfa3, and localization of the human homolog, CBFA3, to chromosome 1p35-pter. *Genomics* 26: 611–614, 1995.

[28994] 2546. Cody, J. D.; Hale, D. E.; Brkanac, Z.; Kaye, C. I.; Leach, R. J.: Growth hormone insufficiency associated with haploinsufficiency at 18q23. *Am. J. Med. Genet.* 71: 420–425, 1997.

[28995] 2547. Habert-Ortoli, E.; Amiranoff, B.; Loquet, I.; Laburthe, M.; Mayaux, J.-F.: Molecular cloning of a functional human galanin receptor. *Proc. Nat. Acad. Sci.* 91: 9780–9783, 1994.

[28996] 2548. Hecht, G.; Marrero, J. A.; Danilkovich, A.; Matkowskyj, K. A.; Savkovic, S. D.; Koutsouris, A.; Benya, R. V.: Pathogenic *Escherichia coli* increase Cl⁻ secretion from intestinal epithelia by upregulating galanin-1 receptor expression. *J. Clin. Invest.* 104: 253–262, 1999.

[28997] 2549. Beckman, L.; Beckman, G.; Mi, M. P.; De Simone, J.: The human placental amino acid naphthylamidases: their

molecular interrelations and correlations with perinatal factors. Hum. Hered. 19: 249–257, 1969.

[28998] 2550. Beckman, L.; Bjorling, G.; Christodoulou, C.: Pregnancy enzymes and placental polymorphism. II. Leucine aminopeptidase. Acta Genet. Statist. Med. 16: 122–131, 1966.

[28999] 2551. Horio, J.; Nomura, S.; Okada, M.; Katsumata, Y.; Nakanishi, Y.; Kumano, Y.; Takami, S.; Kinoshita, M.; Tsujimoto, M.; Nakazato, H.; Mizutani, S.: Structural organization of the 5-prime-end and chromosomal assignment of human placental leucine aminopeptidase/insulin-regulated membrane aminopeptidase gene. Biochem. Biophys. Res. Commun. 262:269–274, 1999.

[29000] 2552. Nagasaka, T.; Nomura, S.; Okamura, M.; Tsujimoto, M.; Nakazato, H.; Oiso, Y.; Nakashima, N.; Mizutani, S.: Immunohistochemical localization of placental leucine aminopeptidase/oxytocinase in normal human placental, fetal and adult tissues. Reprod. Fertil. Dev. 9: 747–753, 1997.

[29001] 2553. Røgi, T.; Tsujimoto, M.; Nakazato, H.; Mizutani, S.; Tomoda, Y.: Human placental leucine aminopeptidase/oxytocinase: a new member of type II membrane-spanning zinc metallopeptidase family. J. Biol. Chem. 271: 56–61,

1996.

- [29002] 2554.Scandalios, J. G.: Human serum leucine aminopeptidase: variation in pregnancy and in disease states. *J. Hered.* 58: 153–156, 1967.
- [29003] 2555.Levanon, D.; Negreanu, V.; Bernstein, Y.; Bar–Am, I.; Avivi, L.; Groner, Y.: AML1, AML2, and AML3, the human members of the runt domain gene–family: cDNA structure, expression, and chromosomal localization. *Genomics* 23:425–432, 1994.
- [29004] 2556.Bielinska, B.; Blaydes, S. M.; Buiting, K.; Yang, T.; Krajewska–Walasek, M.; Horsthemke, B.; Brannan, C. I.: De novo deletions of SNRPN exon1 in early human and mouse embryos result in a paternal to maternal imprint switch. *Nature Genet.* 25: 74–78, 2000. Note: Erratum: *Nature–Genet.* 25: 241 only, 2000.
- [29005] 2557.Raeymaekers, P.; Van Broeckhoven, C.; Backhovens, H.; Wehnert, A.; Muylle, L.; De Jonghe, P.; Gheuens, J.; Vandenberghe, A.: The Duffy blood group is linked to the alpha–spectrin locus in a large pedigree with autosomal dominant inheritance of Charcot–Marie–Tooth disease type 1. *Hum. Genet.* 78: 76–78, 1988.
- [29006] 2558.Eagle, L. R.; Yin, X.; Brothman, A. R.; Williams, B. J.; Atkin, N. B.; Prochownik, E. V.: Mutation of the MXI1 gene

in prostate cancer. *Nature Genet.* 9: 249–255, 1995.

- [29007] 2559. Edwards, A.; Hammond, H. A.; Jin, L.; Caskey, C. T.; Chakraborty, R.: Genetic variation at five trimeric and tetrameric tandem repeat loci in four human population groups. *Genomics* 12: 241–253, 1992.
- [29008] 2560. Ichikawa, T.; Ichikawa, Y.; Dong, J.; Hawkins, A. L.; Griffin, C. A.; Isaacs, W. B.; Oshimura, M.; Barrett, J. C.; Isaacs, J. T.: Localization of metastasis suppressor gene(s) for prostatic cancer to the short arm of human chromosome 11. *Cancer Res.* 52: 3486–3490, 1992.
- [29009] 2561. Ichikawa, T.; Ichikawa, Y.; Isaacs, J. T.: Genetic factors and suppression of metastatic ability of prostatic cancer. *Cancer Res.* 51: 3788–3792, 1991.
- [29010] 2562. Irvine, R. A.; Yu, M. C.; Ross, R. K.; Coetzee, G. A.: The CAG and GGC microsatellites of the androgen receptor gene are in linkage disequilibrium in men with prostate cancer. *Cancer Res.* 55: 1937–1940, 1995.
- [29011] 2563. Peters, M. A.; Ostrander, E. A.: Prostate cancer: more than two to tango. *Nature Genet.* 27: 134–135, 2001.
- [29012] 2564. Bansal, A.; Murray, D. K.; Wu, J. T.; Stephenson, R. A.; Middleton, R. G.; Meikle, A. W.: Heritability of prostate-specific antigen and relationship with zonal prostate volumes in aging twins. *J. Clin. Endocr. Metab.* 85:

1272–1276, 2000.

- [29013] 2565. Cleutjens, K. B. J. M.; van der Korput, H. A. G. M.; van Eekelen, C. C. E. M.; van Rooij, H. C. J.; Faber, P. W.; Trapman, J.: An androgen response element in a far upstream enhancer region is essential for high, androgen-regulated activity of the prostate-specific antigen promoter. *Molec. Endocr.* 11: 148–161, 1997.
- [29014] 2566. Nelson, R. J.; Demas, G. E.; Huang, P. L.; Fishman, M. C.; Dawson, V. L.; Dawson, T. M.; Snyder, S. H.: Behavioural abnormalities in male mice lacking neuronal nitric oxide synthase. *Nature* 378: 383–386, 1995.
- [29015] 2567. Henry, J. G.; Mitnick, M.; Dann, P. R.; Stewart, A. F.: Parathyroid hormone-related protein-(1–36) is biologically active when administered subcutaneously to humans. *J. Clin. Endocr. Metab.* 82: 900–906, 1997.
- [29016] 2568. Magnaghi, P.; Agazzi, A.; Semino, O.; Ferrari, M.; Barbui, T.; D'Angelo, A.; Taramelli, R.: A recombination event in the closely linked plasminogen and apolipoprotein(a) gene loci. *Clin. Genet.* 47: 285–289, 1995.
- [29017] 2569. Holick, M. F.; Ray, S.; Chen, T. C.; Tian, X.; Persons, K. S.: A parathyroid hormone antagonist stimulates epidermal proliferation and hair growth in mice. *Proc. Nat. Acad. Sci.* 91: 8014–8016, 1994.

- [29018] 2570.Lanske, B.; Amling, M.; Neff, L.; Guiducci, J.; Baron, R.; Kronenberg, H. M.: Ablation of the PTHrP gene or the PTH/PTHrP receptor gene leads to distinct abnormalities in bone development. *J. Clin. Invest.* 104:399–407, 1999.
- [29019] 2571.Mangin, M.; Ikeda, K.; Dreyer, B. E.; Broadus, A. E.: Isolation and characterization of the human parathyroid hormone–like peptide gene. *Proc. Nat. Acad. Sci.* 86: 2408–2412, 1989.
- [29020] 2572.Mangin, M.; Webb, A. C.; Dreyer, B. E.; Posillico, J. T.; Ikeda, K.; Weir, E. C.; Stewart, A. F.; Bander, N. H.; Milstone, L.; Barton, D. E.; Francke, U.; Broadus, A. E.: Identification of a cDNA encoding a parathyroid hormone–like peptide from a human tumor associated with humoral hypercalcemia of malignancy. *Proc. Nat. Acad. Sci.* 85: 597–601, 1988.
- [29021] 2573.Moseley, J. M.; Kubota, M.; Diefenbach–Jagger, H.; Wettenhall, R. E. H.; Kemp, B. E.; Suva, L. J.; Rodda, C. P.; Ebeling, P. R.; Hudson, P. J.; Zajac, J. D.; Martin, T. J.: Parathyroid hormone–related protein purified from a human lung cancer cell line. *Proc. Nat. Acad. Sci.* 84: 5048–5052, 1987.
- [29022] 2574.Philbrick, W. M.; Dreyer, B. E.; Nakchbandi, I. A.; Karaplis, A. C.: Parathyroid hormone–related protein is re–

quired for tooth eruption. Proc. Nat. Acad. Sci. 95: 11846–11851, 1998.

- [29023] 2575. Strewler, G. J.: The physiology of parathyroid hormone-related protein. New Eng. J. Med. 342: 177–185, 2000.
- [29024] 2576. Suva, L. J.; Winslow, G. A.; Wettenhall, R. E. H.; Hammonds, R. G.; Moseley, J. M.; Diefenbach-Jagger, H.; Rodda, C. P.; Kemp, B. E.; Rodriguez, H.; Chen, E. Y.; Hudson, P. J.; Martin, T. J.; Wood, W. I.: A parathyroid hormone-related protein implicated in malignant hypercalcemia: cloning and expression. Science 237: 893–896, 1987.
- [29025] 2577. Vortkamp, A.; Lee, K.; Lanske, B.; Segre, G. V.; Kronenberg, H. M.; Tabin, C. J.: Regulation of rate of cartilage differentiation by Indian hedgehog and PTH-related protein. Science 273: 613–622, 1996.
- [29026] 2578. Wysolmerski, J. J.; Cormier, S.; Philbrick, W. M.; Dann, P.; Zhang, J.-P.; Roume, J.; Delezoide, A.-L.; Silve, C.: Absence of functional type 1 parathyroid hormone (PTH)/PTH-related protein receptors in humans is associated with abnormal breast development and tooth impaction. J. Clin. Endocr. Metab. 86: 1788–1794, 2001.
- [29027] 2579. Wysolmerski, J. J.; Philbrick, W. M.; Dunbar, M. E.;

Lanske, B.; Kronenberg, H.; Broadus, A. E.: Rescue of the parathyroid hormone-related protein knockout mouse demonstrates that parathyroid hormone-related protein is essential for mammary gland development. *Development* 125:1285–1294, 1998.

[29028] 2580. Yasuda, T.; Banville, D.; Hendy, G. N.; Goltzman, D.: Characterization of the human parathyroid hormone-like peptide gene: functional and evolutionary aspects. *J. Biol. Chem.* 264: 7720–7725, 1989.

[29029] 2581. Mavrogiannis, L. A.; Antonopoulou, I.; Baxova, A.; Kutilek, S.; Kim, C. A.; Sugayama, S. M.; Salamanca, A.; Wall, S. A.; Morriss-Kay, G. M.; Wilkie, A. O. M.: Haploinsufficiency of the human homeobox gene ALX4 causes skull ossification defects. *Nature Genet.* 27: 17–18, 2001.

[29030] 2582. Qu, S.; Niswender, K. D.; Ji, Q.; van der Meer, R.; Keeney, D.; Magnuson, M. A.; Wisdom, R.: Polydactyly and ectopic ZPA formation in Alx-4 mutant mice. *Development* 124: 3999–4008, 1997.

[29031] 2583. Wu, Y.-Q.; Badano, J. L.; McCaskill, C.; Vogel, H.; Potocki, L.; Shaffer, L. G.: Haploinsufficiency of ALX4 as a potential cause of parietal foramina in the 11p11.2 contiguous gene-deletion syndrome. *Am. J. Hum. Genet.* 67: 1327–1332, 2000.

- [29032] 2584.Wuyts, W.; Cleiren, E.; Homfray, T.; Rasore-Quartino, A.; Vanhoenacker, F.; Van Hul, W.: The ALX4 homeobox gene is mutated in patients with ossification defects of the skull (foramina parietalia permagna, OMIM168500). J. Med. Genet. 37: 916-920, 2000.
- [29033] 2585.Deybach, J.-C.; de Verneuil, H.; Boulechfar, S.; Grandchamp, B.; Nordmann, Y.: Point mutations in the uroporphyrinogen III synthase gene in congenital erythropoietic porphyria (Gunther's disease). Blood 75:1763-1765, 1990.
- [29034] 2586.Lemyre, E.; Russo, P.; Melancon, S. B.; Gagne, R.; Potier, M.; Lambert, M.: Clinical spectrum of infantile free sialic acid storage disease. Am. J. Med. Genet. 82: 385-391, 1999.
- [29035] 2587.Mancini, G. M. S.; Beerens, C. E. M. T.; Aula, P. P.; Verheijen, F. W.: Sialic acid storage diseases: a multiple lysosomal transport defect for acidic monosaccharides. J. Clin. Invest. 87: 1329-1335, 1991.
- [29036] 2588.Schleutker, J.; Laine, A.-P.; Haataja, L.; Renlund, M.; Weissenbach, J.; Aula, P.; Peltonen, L.: Linkage disequilibrium utilized to establish a refined genetic position of the Salla disease locus on 6q14-q15. Genomics 27:286-292, 1995.

- [29037] 2589.Schleutker, J.; Leppanen, P.; Mansson, J.-E.; Erikson, A.; Weissenbach, J.; Peltonen, L.; Aula, P.: Lysosomal free sialic acid storage disorders with different phenotypic presentations--infantile-form sialic acid storage disease and Salla disease--represent allelic disorders on 6q14-15. *Am. J. Hum. Genet.* 57: 893-901, 1995.
- [29038] 2590.Tondeur, M.; Libert, J.; Vamos, E.; Van Hoof, F.; Thomas, G. H.; Strecker, G.: Infantile form of sialic acid storage disorder: clinical, ultrastructural, and biochemical studies in two siblings. *Europ. J. Pediatr.* 139: 142-147, 1982.
- [29039] 2591.Verheijen, F. W.; Verbeek, E.; Aula, N.; Beerens, C. E. M. T.; Havelaar, A. C.; Joosse, M.; Peltonen, L.; Aula, P.; Galjaard, H.; van der Spek, P. J.; Mancini, G. M. S.: A new gene, encoding an anion transporter, is mutated in sialic acid storage diseases. *Nature Genet.* 23:462-465, 1999.
- [29040] 2592.Krasnewich, D. M.; Tietze, F.; Krause, W.; Pretzlaff, R.; Wenger, D. A.; Diwadkar, V.; Gahl, W. A.: Clinical and biochemical studies in an American child with sialuria. *Biochem. Med. Metab. Biol.* 49:90-96, 1993.
- [29041] 2593.Leroy, J. G.; Seppala, R.; Huizing, M.; Dacremont, G.; De Simpel, H.; Van Coster, R. N.; Orvisky, E.; Krasnewich, D. M.; Gahl, W. A.: Dominant inheritance of sialuria, an in-

born error of feedback inhibition. *Am.J. Hum. Genet.* 68: 1419–1427, 2001.

[29042] 2594. Seppala, R.; Lehto, V.-P.; Gahl, W. A.: Mutations in the human UDP-N-acetylglucosamine 2-epimerase gene define the disease sialuria and the allosteric site of the enzyme. *Am. J. Hum. Genet.* 64: 1563–1569, 1999.

[29043] 2595. Murrell, J. R.; Spillantini, M. G.; Zolo, P.; Guazzelli, M.; Smith, M. J.; Hasegawa, M.; Redi, F.; Crowther, R. A.; Pietrini, P.; Ghetti, B.; Goedert, M.: Tau gene mutation G389R causes a tauopathy with abundant Pick body-like inclusions and axonal deposits. *J. Neuropath. Exp. Neurol.* 58: 1207–1226, 1999.

[29044] 2596. Nagase, T.; Ishikawa, K.; Kikuno, R.; Hirose, M.; Nomura, N.; Ohara, O.: Prediction of the coding sequences of unidentified human genes. XV. The complete sequences of 100 new cDNA clones from brain which code for large proteins in vitro. *DNA Res.* 6: 337–345, 1999.

[29045] 2597. Neumann, M.; Schulz-Schaeffer, W.; Crowther, R. A.; Smith, M. J.; Spillantini, M. G.; Goedert, M.; Kretzschmar, H. A.: Pick's disease associated with the novel tau gene mutation K369I. *Ann. Neurol.* 50: 503–513, 2001.

[29046] 2598. Neve, R. L.; Harris, P.; Kosik, K. S.; Kurnit, D. M.; Donlon, T. A.: Identification of cDNA clones for the human

microtubule-associated protein tau and chromosomal localization of the genes for tau and microtubule-associated protein 2. *Molec. Brain Res.* 1: 271–280, 1986.

- [29047] 2599. Pickering-Brown, S.; Baker, M.; Yen, S.-H.; Liu, W.-K.; Hasegawa, M.; Cairns, N.; Lantos, P. L.; Rossor, M.; Iwatsubo, T.; Davies, Y.; Allsop, D.; Furlong, R.; Owen, F.; Hardy, J.; Mann, D.; Hutton, M.: Pick's disease is associated with mutations in the tau gene. *Ann. Neurol.* 48: 859–867, 2000.
- [29048] 2600. Poorkaj, P.: Personal Communication. Seattle, Wash. 11/10/1998.
- [29049] 2601. Poorkaj, P.; Bird, T. D.; Wijsman, E.; Nemens, E.; Garruto, R.M.; Anderson, L.; Andreadis, A.; Wiederholt, W. C.; Raskind, M.; Schellenberg, G. D.: Tau is a candidate gene for chromosome 17 frontotemporal dementia. *Ann. Neurol.* 43: 815–825, 1998.
- [29050] 2602. Poorkaj, P.; Kas, A.; D'Souza, I.; Zhou, Y.; Pham, Q.; Stone, M.; Olson, M. V.; Schellenberg, G. D.: A genomic sequence analysis of the mouse and human microtubule-associated protein tau. *Mammalian Genome* 12: 700–712, 2001.
- [29051] 2603. Rapoport, M.; Dawson, H. N.; Binder, L. I.; Vitek, M. P.; Ferreira, A.: Tau is essential to beta-amyloid-induced

neurotoxicity. Proc.Nat. Acad. Sci. 99: 6364–6369, 2002.

[29052] 2604.Rizzu, P.; Van Swieten, J. C.; Joosse, M.; Hasegawa, M.; Stevens, M.; Tibben, A.; Niermeijer, M. F.; Hillebrand, M.; Ravid, R.; Oostra, B. A.; Goedert, M.; van Duijn, C. M.; Heutink, P.: High prevalence of mutations in the microtubule-associated protein tau in a population study of frontotemporal dementia in the Netherlands. Am. J. Hum. Genet. 64: 414–421, 1999.

[29053] 2605.Spillantini, M. G.; Murrell, J. R.; Goedert, M.; Farlow, M. R.; Klug, A.; Ghetti, B.: Mutation in the tau gene in familial multiple system tauopathy with presenile dementia. Proc. Nat. Acad. Sci. 95:7737–7741, 1998.

[29054] 2606.Spillantini, M. G.; Yoshida, H.; Rizzini, C.; Lantos, P. L.; Khan, N.; Rossor, M. N.; Goedert, M.; Brown, J.: A novel tau mutation (N296N) in familial dementia with swollen achromatic neurons and corticobasal inclusion bodies. Ann. Neurol. 48: 939–943, 2000.

[29055] 2607.Stambolic, V.; Ruel, L.; Woodgett, J. R.: Lithium inhibits glycogen synthase kinase-3 activity and mimics Wingless signalling in intact cells. Curr. Biol. 6: 1664–1668, 1996. Note: Erratum: Curr. Biol. 7: 196 only, 1997.

[29056] 2608.Stamer, K.; Vogel, R.; Thies, E.; Mandelkow, E.; Man-

delkow, E.-M.: Tau blocks traffic of organelles, neurofilaments, and APP vesicles in neurons and enhances oxidative stress. *J. Cell Biol.* 156: 1051–1063, 2002.

[29057] 2609. Varani, L.; Hasegawa, M.; Spillantini, M. G.; Smith, M. J.; Murrell, J. R.; Ghetti, B.; Klug, A.; Goedert, M.; Varani, G.: Structure of tau exon 10 splicing regulatory element RNA and destabilization by mutations of frontotemporal dementia and parkinsonism linked to chromosome 17. *Proc. Nat. Acad. Sci.* 96: 8229–8234, 1999.

[29058] 2610. Verpillat, P.; Camuzat, A.; Hannequin, D.; Thomas-Anterion, C.; Puel, M.; Belliard, S.; Dubois, B.; Didic, M.; Michel, B.-F.; Lacomblez, L.; Moreaud, O.; Sellal, F.; Golfier, V.; Campion, D.; Clerget-Darpoux, F.; Brice, A.: Association between the extended tau haplotype and frontotemporal dementia. *Arch. Neurol.* 59: 935–939, 2002.

[29059] 2611. Wilhelmsen, K. C.; Lynch, T.; Pavlou, E.; Higgins, M.; Hygaard, T. G.: Localization of disinhibition-dementia-parkinsonism-amyotrophy complex to 17q21–22. *Am. J. Hum. Genet.* 55: 1159–1165, 1994.

[29060] 2612. Yasuda, M.; Yokoyama, K.; Nakayasu, T.; Nishimura, Y.; Matsui, M.; Yokoyama, T.; Miyoshi, K.; Tanaka, C.: A Japanese patient with frontotemporal dementia and parkinsonism by a tau P301S mutation. *Neurology*

55:1224–1227, 2000.

- [29061] 2613. Zhukareva, V.; Mann, D.; Pickering–Brown, S.; Uryu, K.; Shuck, T.; Shah, K.; Grossman, M.; Miller, B. L.; Hulette, C. M.; Feinstein, S. C.; Trojanowski, J. Q.; Lee, V. M.–Y.: Sporadic Pick's disease: a tauopathy characterized by a spectrum of pathological tau isoforms in gray and white matter. *Ann. Neurol.* 51: 730–739, 2002.
- [29062] 2614. Weintraub, H.; Davis, R.; Tapscott, S.; Thayer, M.; Krause, M.; Benezra, R.; Blackwell, T. K.; Turner, D.; Rupp, R.; Hollenberg, S.; Zhuang, Y.; Lassar, A.: The myoD gene family: nodal point during specification of the muscle cell lineage. *Science* 251: 761–766, 1991.
- [29063] 2615. Amack, J. D.; Mahadevan, M. S.: The myotonic dystrophy expanded CUG repeat tract is necessary but not sufficient to disrupt C2C12 myoblast differentiation. *Hum. Molec. Genet.* 10: 1879–1887, 2001.
- [29064] 2616. Chen, S. L.; Dowhan, D. H.; Hosking, B. M.; Muscat, G. E. O.: The steroid receptor coactivator, GRIP–1, is necessary for MEF–2C–dependent gene expression and skeletal muscle differentiation. *Genes Dev.* 14: 1209–1228, 2000.
- [29065] 2617. Hasty, P.; Bradley, A.; Morris, J. H.; Edmondson, D. G.; Venuti, J. M.; Olson, E. N.; Klein, W. H.: Muscle defi–

ciency and neonatal death in mice with a targeted mutation in the myogenin gene. *Nature* 364:501–506, 1993.

[29066] 2618. Nabeshima, Y.; Hanaoka, K.; Hayasaka, M.; Esumi, E.; Li, S.; Nonaka, I.; Nabeshima, Y.-I.: Myogenin gene disruption results in perinatal lethality because of severe muscle defect. *Nature* 364: 532–535, 1993.

[29067] 2619. Braun, T.; Bober, E.; Winter, B.; Rosenthal, N.; Arnold, H. H.: Myf-6, a new member of the human gene family of myogenic determination factors: evidence for a gene cluster on chromosome 12. *EMBO J.* 9:821–831, 1990.

[29068] 2620. Braun, T.; Grzeschik, K.-H.; Bober, E.; Arnold, H.-H.: The MYF genes, a group of human muscle determining factors, are localized on different human chromosomes. (Abstract) *Cytogenet. Cell Genet.* 51:969, 1989.

[29069] 2621. Olson, E.; Edmondson, D.; Wright, W. E.; Lin, V. K.; Guenet, J.-L.; Simon-Chazottes, D.; Thompson, L. H.; Stallings, R. L.; Schroeder, W. T.; Duvic, M.; Brock, D.; Heilin, D.; Siciliano, M. J.: Myogenin is in an evolutionarily conserved linkage group on human chromosome 1q31–q41 and unlinked to other mapped muscle regulatory factor genes. *Genomics* 8:427–434, 1990.

[29070] 2622. Cupelli, L.; Renault, B.; Leblanc-Straceski, J.; Banks,

A.; Ward,D.; Kucherlapati, R. S.; Krauter, K.: Assignment of the human myogenicfactors 5 and 6 (MYF5, MYF6) gene cluster to 12q21 by in situ hybridizationand physical mapping of the locus between D12S350 and D12S106. Cytogenet.Cell Genet. 72: 250–251, 1996.

[29071] 2623.Ott, M.–O.; Bober, E.; Lyons, G.; Arnold, H.–H.; Buckingham, M.: Early expression of the myogenic regulatory gene, Myf–5, in precursorcells of skeletal muscle in the mouse embryo. Development 111:1097–1107, 1991.

[29072] 2624.Rudnicki, M. A.; Schnegelsberg, P. N. J.; Stead, R. H.; Braun,T.; Arnold, H.–H.; Jaenisch, R.: MyoD or Myf–5 is required for theformation of skeletal muscle. Cell 75: 1351–1359, 1993.

[29073] 2625.Akaboshi, E.: Cloning of the human myoglobin gene. Gene 33:241–249, 1985.

[29074] 2626.Boulton, F. E.; Huntsman, R. G.; Lehmann, H.; Lorkin, P. A.; Romero–Herrera,A. E.: Myoglobin variants. (Abstract) Biochem. J. 118: 39P only,1970.

[29075] 2627.Boulton, F. E.; Huntsman, R. G.; Lorkin, P. A.; Lehmann, H.: Abnormalhuman myoglobin: 53(D4) glutamic acid lysine. Nature 223: 832–833,1969.

[29076] 2628.Boulton, F. E.; Huntsman, R. G.; Yawson, G. I.; Romero–Herrera,A. E.; Lorkin, P. A.: The second variant of

human myoglobin: 138(H16)arginine to glutamine. Brit. J. Haemat. 20A: 69–74, 1971.

[29077] 2629. Boyer, S. H.: Similar incidence and non-randomness among human myoglobin and hemoglobin mutants in general populations: implications for the study of myoglobin in muscle disease. In: Pathogenesis of Human Muscular Dystrophies. Proc. Vth Int. Cong. of Muscular Dystrophy Assoc., Durango, Colo., June 21–25, 1976. Amsterdam: Excerpta Medica (pub.) 1977.

[29078] 2630. Boyer, S. H.; Fainer, D. C.; Naughton, M. A.: Myoglobin inherited structural variation in man. Science 140: 1228–1231, 1963.

[29079] 2631. Dozier, C.; Walbaum, S.; Leprince, D.; Stehelin, D.: EcoRI RFLP linked to the human myb gene. Nucleic Acids Res. 14: 1928 only, 1986.

[29080] 2632. Garry, D. J.; Ordway, G. A.; Lorenz, J. N.; Radford, N. B.; Chin, E. R.; Grange, R. W.; Bassel-Duby, R.; Williams, R. S.: Mice without myoglobin. Nature 395: 905–908, 1998.

[29081] 2633. Godecke, A.; Flogel, U.; Zanger, K.; Ding, Z.; Hirchenhain, J.; Decking, U. K. M.; Schrader, J.: Disruption of myoglobin in mice induces multiple compensatory mechanisms. Proc. Nat. Acad. Sci. 96: 10495–10500, 1999.

- [29082] 2634. Jeffreys, A.; Wilson, V.; Thein, S.: Hypervariable 'minisatellite' regions in human DNA. *Science* 314: 67–73, 1985.
- [29083] 2635. Jeffreys, A. J.; Wilson, V.; Blanchetot, A.; Weller, P.; Geurtsvan Kessel, A.; Spurr, N.; Solomon, E.; Goodfellow, P.: The human myoglobin gene: a third dispersed globin locus in the human genome. *Nucleic Acids Res.* 12: 3235–3243, 1984.
- [29084] 2636. McKeithan, T. W.; Rowley, J. D.; Shows, T. B.; Diaz, M. O.: Cloning of the chromosome translocation breakpoint junction of the t(14;19) in chronic lymphocytic leukemia. *Proc. Nat. Acad. Sci.* 84: 9257–9260, 1987.
- [29085] 2637. de Groot, N. G.; Otting, N.; Doxiadis, G. G. M.; Balla-Jhagjhoorsingh, S. S.; Heeney, J. L.; van Rood, J. J.; Gagneux, P.; Bontrop, R. E.: Evidence for an ancient selective sweep in the MHC class I gene repertoire of chimpanzees. *Proc. Nat. Acad. Sci.* 99: 11748–11753, 2002.
- [29086] 2638. Del Pozzo, G.; Guardiola, J.: Mummy DNA fragment identified. (Letter) *Nature* 339: 431–432, 1989.
- [29087] 2639. Dorf, M. E.; Balner, H.; DeGroot, M. L.; Benacerraf, B.: Histocompatibility-linked immune-response genes in the Rhesus monkey. *Transplant. Proc.* 6: 119–124, 1974.
- [29088] 2640. Edwards, J. H.; Allen, F. H.; Glenn, K. P.; Lamm, L. U.;

Robson, E. B.: The linkage relationships of HL-A. In: Histo-compatibility Testing. Baltimore: Williams and Wilkins (pub.) 1973.

[29089] 2641. Engelfriet, C. P.; Britten, A.: The cytotoxic test for leucocyte antibodies. A simple and reliable technique. Vox Sang. 10: 660–674, 1965.

[29090] 2642. Erlich, H.; Lee, J. S.; Petersen, J. W.; Bugawan, T.; De-Mars, R.: Molecular analysis of HLA class I and class II antigen loss mutants reveals a homozygous deletion of the DR, DQ, and part of the DP region: implications for class II gene order. Hum. Immun. 16: 205–219, 1986.

[29091] 2643. Erlich, H. A.; Stetler, D.; Sheng-Dong, R.; Ness, D.; Grumet, C.: Segregation and mapping analysis of polymorphic HLA class I restriction fragments: detection of a novel fragment. Science 222: 72–74, 1983.

[29092] 2644. Fellous, M.; Dausset, J.: Probable haploid expression of HL-A antigens on human spermatozoan. Nature 225: 191–193, 1970.

[29093] 2645. Figueroa, F.; Gunther, E.; Klein, J.: MHC polymorphism pre-dates speciation. (Letter) Nature 335: 265–267, 1988.

[29094] 2646. Francke, U.; Pellegrino, M. A.: Assignment of the major histocompatibility complex to a region of the short

arm of human chromosome 6. Proc.Nat. Acad. Sci. 74: 5776 only, 1977.

[29095] 2647.Francke, U.; Pellegrino, M. A.: Assignment of the major histocompatibilitycomplex to a region of the short arm of human chromosome 6. Proc.Nat. Acad. Sci. 74: 1147–1151, 1977.

[29096] 2648.Gill, T. J., III; Cramer, D. V.; Kunz, H. W.: The major histocompatibilitycomplex--comparison in the mouse, man, and the rat: a review. Am.J. Path. 90: 735–777, 1978.

[29097] 2649.Gladstone, P.; Furesz, L.; Pious, D.: Gene dosage and gene expression in the HLA region: evidence from deletion variants. Proc. Nat. Acad.Sci. 79: 1235–1239, 1982.

[29098] 2650.Gluecksohn-Waelsch, S.; Erickson, R. P.: The T-locus of the mouse:implications for mechanisms of development. Curr. Top. Dev. Biol. 5:281–316, 1970.

[29099] 2651.Goodfellow, P. N.; Jones, E.; Van Heyningen, V.; Solomon, E.;Kennett, R.; Bobrow, M.; Bodmer, W. F.: Linkage relationships ofthe HL-A system and beta-2-microglobulin. Birth Defects Orig. Art.Ser. 11(3): 162–167, 1975. Note: Alternate: Cytogenet. Cell Genet.14: 332–337, 1975.

- [29100] 2652.Gruen, J. R.; Nalabolu, S. R.; Chu, T. W.; Bowlus, C.; Fan, W.F.; Goei, V. L.; Wei, H.; Sivakamasundari, R.; Liu, Y.-C.; Xu, H.X.; Parimoo, S.; Nallur, G.; Ajioka, R.; Shukla, H.; Bray-Ward, P.;Pan, J.; Weissman, S. M.: A transcription map of the major histocompatibilitycomplex (MHC) class I region. *Genomics* 36: 70–85, 1996.
- [29101] 2653.Hedrick, S. M.: Dawn of the hunt for nonclassical MHC function. *Cell* 70:177–180, 1992.
- [29102] 2654.Helenius, A.; Morein, B.; Fries, E.; Simons, K.; Robinson, P.;Schirmacher, V.; Terhorst, C.; Strominger, J. L.: Human (HLA-A andHLA-B) and murine (H-2K and H-2D) histocompatibility antigens arecell surface receptors for Semliki Forest virus. *Proc. Nat. Acad.Sci.* 75: 3846–3850, 1978.
- [29103] 2655.Hirai, M.; Takahashi, E.; Ishida, T.; Hori, T.: Chromosomal localizationof the major histocompatibility complex (MHC) in the rhesus monkeyand chimpanzee by fluorecence in situ hybridization. *Cytogenet.Cell Genet.* 57: 204–205, 1991.
- [29104] 2656.Hood, L.; Steinmetz, M.; Goodenow, R.: Genes of the major histocompatibilitycomplex. *Cell* 28: 685–687, 1982.
- [29105] 2657.Jordan, B. R.; Bregegere, F.; Kourilsky, P.: Human HLA gene segmentisolated by hybridization with mouse

H-2 cDNA probes. *Nature* 290:521–523, 1981.

- [29106] 2658. Kissmeyer-Nielsen, F.; Jorgensen, F.; Lamm, L. U.: The HL-A system in clinical medicine. *Johns Hopkins Med. J.* 131: 385–400, 1972.
- [29107] 2659. Kissmeyer-Nielsen, F.; Svejgaard, A.; Ahrens, S.; Nielsen, L.S.: Crossing-over within the HL-A system. *Nature* 224: 75–76, 1969.
- [29108] 2660. Kissmeyer-Nielsen, F.; Svejgaard, A.; Hauge, M.: Genetics of the human HL-A transplantation system. *Nature* 219: 1116–1119, 1968.
- [29109] 2661. Kissmeyer-Nielsen, F.; Thorsby, E.: Human transplantation antigens. *Transplant. Rev.* 4: 1–176, 1970.
- [29110] 2662. Klein, J.: The major histocompatibility complex of the mouse. *Science* 203:516–521, 1979.
- [29111] 2663. Klein, J.; Figueroa, F.: Evolution of the major histocompatibility complex. *CRC Crit. Rev. Immun.* 6: 295–386, 1986.
- [29112] 2664. Kompf, J.; Bissbort, S.; Gohler, F.; Schunter, F.; Wernet, P.: Mapping of the linkage group GLO-Bf-HLA-B, C, A-PGM. I. Recombination frequencies. *Hum. Genet.* 44: 313–319, 1978.
- [29113] 2665. Lamm, L. U.; Friedrich, U.; Petersen, G. B.; Jorgensen, J.; Nielsen, J.; Therkelsen, A. J.; Kissmeyer-Nielsen,

F.: Assignment of the major histocompatibility complex to chromosome no. 6 in a family with a pericentric inversion. *Hum. Hered.* 24: 273–284, 1974.

- [29114] 2666. Lamm, L. U.; Kissmeyer-Nielsen, F.; Svejgaard, A.; Petersen, G. B.; Thorsby, E.; Mayr, W.; Hogman, C.: On the orientation of the HL-A region and the PGM(3) locus in the chromosome. *Tissue Antigens* 2:205–214, 1972.
- [29115] 2667. Lamm, L. U.; Svejgaard, A.; Kissmeyer-Nielsen, F.: Further evidence for PGM(3): HL-A is another linkage in man. *Nature N.B.* 231: 109–110, 1971.
- [29116] 2668. Lamm, L. U.; Thorsen, I.-L.; Petersen, G. B.; Jorgensen, J.; Henningsen, K.; Bech, B.; Kissmeyer-Nielsen, F.: Data on the HL-A linkage group. *Ann. Hum. Genet.* 38: 383–390, 1975.
- [29117] 2669. Lawlor, D. A.; Dickel, C. D.; Hauswirth, W. W.; Parham, P.: Ancient HLA genes from 7,500-year-old archaeological remains. *Nature* 349:785–788, 1991.
- [29118] 2670. Lawlor, D. A.; Ward, F. E.; Ennis, P. D.; Jackson, A. P.; Parham, P.: HLA-A and B polymorphisms predate the divergence of humans and chimpanzees. (Letter) *Nature* 335: 268–271, 1988.
- [29119] 2671. Lawrance, S. K.; Smith, C. L.; Srivastava, R.; Cantor, C. R.; Weissman, S. M.: Megabase-scale mapping of the

HLA gene complex by pulsed field gel electrophoresis. *Science* 235: 1387–1390, 1987.

[29120] 2672. Levine, F.; Erlich, H.; Mach, B.; Leach, R.; White, R.; Pious, D.: Deletion mapping of HLA and chromosome 6p genes. *Proc. Nat. Acad. Sci.* 82: 3741–3745, 1985.

[29121] 2673. Slate, D. L.; Ruddle, F. H.: Genetics of the interferon system. *Pharm. Therap.* 4: 221–230, 1979.

[29122] 2674. Stock, A. D.; Hsu, T. C.: Evolutionary conservatism in arrangement of genetic material. A comparative analysis of chromosome banding between the Rhesus macaque ($2n=42$, 84 arms) and the African green monkey ($2n=60$, 120 arms). *Chromosoma* 43: 211–224, 1973.

[29123] 2675. Streuli, M.; Nagata, S.; Weissmann, C.: At least three human type alpha interferons: structure of alpha-2. *Science* 209: 1343–1347, 1980.

[29124] 2676. Szabo, S. J.; Sullivan, B. M.; Stemmann, C.; Satoskar, A. R.; Sleckman, B. P.; Glimcher, L. H.: Distinct effects of T-bet in T(H)1 lineage commitment and IFN-gamma production in CD4 and CD8 T cells. *Science* 295: 338–342, 2002.

[29125] 2677. Takayanagi, H.; Ogasawara, K.; Hida, S.; Chiba, T.; Murata, S.; Sato, K.; Takaoka, A.; Yokochi, T.; Oda, H.; Tanaka, K.; Nakamura, K.; Taniguchi, T.: T-cell-mediated regulation of osteoclastogenesis by signalling cross-talk

between RANKL and IFN- γ . *Nature* 408:600–605, 2000.

[29126] 2678. Tan, Y. H.; Creagan, R. P.; Ruddle, F. H.: Assignment of the genes of the human interferon system to chromosomes 2 and 5. *Cytogenet. Cell Genet.* 13: 155–157, 1974.

[29127] 2679. Tan, Y. H.; Ke, Y. H.; Armstrong, J. A.; Ho, M.: The regulation of cellular interferon production: enhancement by antimetabolites. *Proc. Nat. Acad. Sci.* 67: 464–471, 1970.

[29128] 2680. Trent, J. M.; Olson, S.; Lawn, R. M.: Chromosomal localization of human leukocyte, fibroblast, and immune interferon genes by means of in situ hybridization. *Proc. Nat. Acad. Sci.* 79: 7809–7813, 1982.

[29129] 2681. Tzoneva, M.; Ganev, V.; Galabov, A.; Georgieva, K.: Selective immunodeficiency with defect in interferon- γ induction in two sibs with recurrent infections. *Clin. Genet.* 33: 454–456, 1988.

[29130] 2682. White, A. C., Jr.; Robinson, P.; Okhuysen, P. C.; Lewis, D. E.; Shahab, I.; Lahoti, S.; DuPont, H. L.; Chappell, C. L.: Interferon- γ expression in jejunal biopsies in experimental human cryptosporidiosis correlates with prior sensitization and control of oocyst excretion. *J. Infect. Dis.* 181: 701–709, 2000.

- [29131] 2683.Yip, Y. K.; Barrowclough, B. S.; Urban, C.; Vilcek, J.: Purification of two subspecies of human gamma (immune) interferon. *Proc. Nat.Acad. Sci.* 79: 1820–1824, 1982.
- [29132] 2684.Zimonjic, D. B.; Rezanka, L. J.; Evans, C. H.; Polymeropoulos, M. H.; Trent, J. M.; Popescu, N. C.: Mapping of the immune interferon gamma gene (IFNG) to chromosome band 12q14 by fluorescence in situ hybridization. *Cytogenet. Cell Genet.* 71: 247–248, 1995.
- [29133] 2685.Zohlhofer, D.; Richter, T.; Neumann, F.-J.; Nuhrenberg, T.; Wessely, R.; Brandl, R.; Murr, A.; Klein, C. A.; Baeuerle, P. A.: Transcriptome analysis reveals a role of interferon-gamma in human neointima formation. *Molec.Cell* 7: 1059–1069, 2001.
- [29134] 2686.Zoon, K. C.; Smith, M. E.; Bridgen, P. J.; Anfinsen, C. B.; Hunkapiller, M. W.; Hood, L. E.: Amino terminal sequence of the major component of human lymphoblastoid interferon. *Science* 207: 527–528, 1980.
- [29135] 2687.Clauss, I. M.; Wathelet, M. G.; Szpirer, J.; Content, J.; Islam, M. Q.; Levan, G.; Szpirer, C.; Huez, G. A.: Chromosomal localization of two human genes inducible by interferons, double-stranded RNA, and viruses. *Cytogenet. Cell Genet.* 53: 166–168, 1990.
- [29136] 2688.Itzhaki, J. E.; Barnett, M. A.; MacCarthy, A. B.; Buckle,

V. J.;Brown, W. R. A.; Porter, A. C. G.: Targeted breakage of a human chromosomemediated by cloned human telomeric DNA. *Nature Genet.* 2: 283–287,1992.

[29137] 2689.Itzhaki, J. E.; Porter, A. C. G.: Targeted disruption of a humaninterferon–inducible gene detected by secretion of human growth hormone. *NucleicAcids Res.* 19: 3835–3842, 1991.

[29138] 2690.Kelly, J. M.; Porter, A. C. G.; Chernajovsky, Y.; Gilbert, C. S.;Stark, G. R.; Kerr, I. M.: Characterization of a human gene inducibleby alpha– and beta–interferons and its expression in mouse cells. *EMBOJ.* 5: 1601–1606, 1986.

[29139] 2691.Porter, A. C. G.; Chernajovsky, Y.; Dale, T. C.; Gilbert, C. S.;Stark, G. R.; Kerr, I. M.: Interferon response element of the humangene 6–16. *EMBO J.* 7: 85–92, 1988.

[29140] 2692.Fu, X.–Y.; Schindler, C.; Improt, T.; Aebersold, R.; Darnell,J. E., Jr.: The proteins of ISGF–3, the interferon alpha–inducedtranscriptional activator, define a gene family involved in signaltransduction. *Proc. Nat. Acad. Sci.* 89: 7840–7843, 1992.

[29141] 2693.Levy, D. E.; Kessler, D. S.; Pine, R.; Darnell, J. E., Jr.: Cytoplasmicactivation of ISGF3, the positive regulator of interferon–alpha–stimulatedtranscription, reconstituted in

vitro. *Genes Dev.* 3: 1362–1371, 1989.

[29142] 2694. McCusker, D.; Wilson, M.; Trowsdale, J.: Organization of the genes encoding the human proteasome activators PA28- α and β . *Immunogenetics* 49:438–445, 1999.

[29143] 2695. Schindler, C.; Fu, X.-Y.; Imbrota, T.; Aebersold, R.; Darnell, J. E., Jr.: Proteins of transcription factor ISGF-3: one gene encodes the 91- and 84-kDa ISGF-3 proteins that are activated by interferon α . *Proc. Nat. Acad. Sci.* 89: 7836–7839, 1992.

[29144] 2696. Suhara, W.; Yoneyama, M.; Yonekawa, H.; Fujita, T.: Structure of mouse interferon stimulated gene factor 3 γ (ISGF3 γ /p48) cDNA and chromosomal localization of the gene. *J. Biochem.* 119:231–234, 1996.

[29145] 2697. Veals, S. A.; Schindler, C.; Leonard, D.; Fu, X.-Y.; Aebersold, R.; Darnell, J. E., Jr.; Levy, D. E.: Subunit of an α -interferon-responsive transcription factor is related to interferon regulatory factor and myb families of DNA-binding proteins. *Molec. Cell. Biol.* 12: 3315–3324, 1992.

[29146] 2698. Eason, D. D.; Shepherd, A. T.; Blanck, G.: Interferon regulatory factor 1 tryptophan 11 to arginine point mutation abolishes DNA binding. *Biochim. Biophys. Acta* 1446: 140–144, 1999.

- [29147] 2699.Savtchenko, E. S.; Tomic, M.; Ivker, R.; Blumenberg, M.: Threeparallel linkage groups of human acidic keratin genes. *Genomics* 7:394–407, 1990.
- [29148] 2700.Bowden, P. E.; Haley, J. L.; Kansky, A.; Rothnagel, J. A.; Jones,D. O.; Turner, R. J.: Mutation of a type II keratin gene (K6a) inpachyonychia congenita. *Nature Genet.* 10: 363–365, 1995.
- [29149] 2701.Rosenberg, M.; Fuchs, E.; Le Beau, M. M.; Eddy, R. L.; Shows, T.B.: Three epidermal and one simple epithelial type II keratin genesmap to human chromosome 12. *Cy-togenet. Cell Genet.* 57: 33–38, 1991.
- [29150] 2702.Takahashi, K.; Paladini, R. D.; Coulombe, P. A.: Cloning and characterizationof multiple human genes and cDNAs encoding highly related type IIkeratin 6 isoforms. *J. Biol. Chem.* 270: 18581–18592, 1995.
- [29151] 2703.Tyner, A.; Eichman, M.; Fuchs, E.: The sequence of a type II keratingene expressed in human skin: conservation of structure among allintermediate filament genes. *Proc. Nat. Acad. Sci.* 82: 4683–4687,1985.
- [29152] 2704.Wang, N.; Perkins, K. L.: Involvement of band 3p14 in t(3;8)hereditary renal carcinoma. *Cancer Genet. Cyto-genet.* 11: 479–481,1984.
- [29153] 2705.Schoenmakers, E. F. P. M.; Huysmans, C.; Van de

Ven, W. J. M.: Allelic knockout of novel splice variants of human recombination repair gene RAD51B in t(12;14) uterine leiomyomas. *Cancer Res.* 59: 19–23, 1999.

[29154] 2706. Funk, C. D.; Funk, L. B.; FitzGerald, G. A.; Samuelsson, B.: Characterization of human 12-lipoxygenase genes. *Proc. Nat. Acad. Sci.* 89: 3962–3966, 1992.

[29155] 2707. Yoshida, M. C.; Sasaki, M.; Mise, K.; Semba, K.; Nishizawa, M.; Yamamoto, T.; Toyoshima, K.: Regional mapping of the human proto-oncogene c-yes-1 to chromosome 18 at band q21.3. *Jpn. J. Cancer Res.* 76: 559–562, 1985.

[29156] 2708. Goossens, M.; Brauner, R.; Czernichow, P.; Duquesnoy, P.; Rappaport, R.: Isolated growth hormone (GH) deficiency type 1A associated with a double deletion in the human GH gene cluster. *J. Clin. Endocr. Metab.* 62: 712–716, 1986.

[29157] 2709. Harper, M. E.; Barrera-Saldana, H. A.; Saunders, G. F.: Chromosomal localization of the human placental lactogen-growth hormone gene cluster to 17q22–24. *Am. J. Hum. Genet.* 34: 227–234, 1982.

[29158] 2710. Harmer, D.; Evans, D. A. P.; Eze, L. C.; Jolly, M.; Whibley, E. J.: The relationship between the acetylator and the sparteine hydroxylation polymorphisms. *J. Med. Genet.*

23: 155–156, 1986.

- [29159] 2711.Hendershot, L. M.; Valentine, V. A.; Lee, A. S.; Morris, S. W.;Shapiro, D. N.: Localization of the gene encoding human BiP/GRP78,the endoplasmic reticulum cognate of the HSP70 family, to chromosome9q34. *Genomics* 20: 281–284, 1994.
- [29160] 2712.Law, M. L.; Seeliger, M. B.; Lee, A. S.; Kao, F. T.: Genetic mappingof the structural gene coding for a glucose–regulated protein (GRP78)of 78k–dalton to the long arm of human chromosome 9. (Abstract) *Cytogenet.Cell Genet.* 37: 518–519, 1984.
- [29161] 2713.Lee, A. S.; Delegeane, A.; Scharff, D.: Highly conserved glucose–regulatedprotein in hamster and chicken cells: preliminary characterizationof its cDNA clone. *Proc. Nat. Acad. Sci.* 78: 4922–4925, 1981.
- [29162] 2714.Lee, A. S.; Delegeane, A. M.; Baker, V.; Chow, P. C.: Transcriptionalregulation of two genes specifically induced by glucose starvationin hamster mutant fibroblast cell line. *J. Biol. Chem.* 258: 597–603,1983.
- [29163] 2715.Muresan, Z.; Arvan, P.: Enhanced binding to the molecular chaperoneBiP slows thyroglobulin export from the endoplasmic reticulum. *Molec.Endocr.* 12: 458–467, 1998.

- [29164] 2716. McCombie, R. R.; Dolphin, C. T.; Povey, S.; Phillips, I. R.; Shephard, E. A.: Localization of human flavin-containing monooxygenase genes FMO2 and FMO5 to chromosome 1q. *Genomics* 34: 426–429, 1996.
- [29165] 2717. Bevilacqua, M. P.; Nelson, R. M.: Selectins. *J. Clin. Invest.* 91:379–387, 1993.
- [29166] 2718. Florian, V.; Schluter, T.; Bohnensack, R.: A new member of the sorting nexin family interacts with the C-terminus of P-selectin. *Biochem. Biophys. Res. Commun.* 281: 1045–1050, 2001.
- [29167] 2719. Herrmann, S.-M.; Ricard, S.; Nicaud, V.; Mallet, C.; Evans, A.; Ruidavets, J.-B.; Arveiler, D.; Luc, G.; Cambien, F.: The P-selectin gene is highly polymorphic: reduced frequency of the pro715 allele carriers in patients with myocardial infarction. *Hum. Molec. Genet.* 7:1277–1284, 1998.
- [29168] 2720. Johnston, G. I.; Bliss, G. A.; Newman, P. J.; McEver, R. P.: Structure of the human gene encoding granule membrane protein-140, a member of the selectin family of adhesion receptors for leukocytes. *J. Biol. Chem.* 265: 21381–21385, 1990.
- [29169] 2721. Johnston, G. I.; Cook, R. G.; McEver, R. P.: Cloning of GMP-140, a granule membrane protein of platelets and

endothelium: sequence similarity to proteins involved in cell adhesion and inflammation. *Cell* 56:1033–1044, 1989.

[29170] 2722. Johnston, G. I.; Le Beau, M. M.; Lemons, R. S.; McEver, R. P.: Cloning of GMP-140: chromosomal localization, molecular heterogeneity and identification of cDNAs predicting both membrane bound and soluble proteins. (Abstract) *Blood* 72 (suppl.): 327a only, 1988.

[29171] 2723. Lages, B.; Shattil, S. J.; Bainton, D. F.; Weiss, H. J.: Decreased content and surface expression of alpha-granule membrane protein GMP-140 in one of two types of platelet alpha-delta storage pool deficiency. *J. Clin. Invest.* 87: 919–929, 1991.

[29172] 2724. Mayadas, T. N.; Johnson, R. C.; Rayburn, H.; Hynes, R. O.; Wagner, D. D.: Leukocyte rolling and extravasation are severely compromised in P selectin-deficient mice. *Cell* 74: 541–554, 1993.

[29173] 2725. Mazurov, A. V.; Vinogradov, D. V.; Khaspekova, S. G.; Krushinsky, A. V.; Gerdeva, L. V.; Vasiliev, S. A.: Deficiency of P-selectin in a patient with grey platelet syndrome. *Europ. J. Haemat.* 57:38–41, 1996.

[29174] 2726. Souttou, B.; Juhl, H.; Hackenbruck, J.; Rockseisen, M.; Klomp, H. J.; Raulais, D.; Vigny, M.; Wellstein, A.: Rela-

relationship between serum concentrations of the growth factor pleiotrophin and pleiotrophin-positive tumors. J. Nat. Cancer Inst. 90: 1468–1473, 1998.

[29175] 2727. Weber, D.; Klomp, H.-J.; Czubayko, F.; Wellstein, A.; Juhl, H.: Pleiotrophin can be rate-limiting for pancreatic cancer cell growth. Cancer Res. 60: 5284–5288, 2000.

[29176] 2728. Zhang, N.; Yeh, H.-J.; Zhong, R.; Li, Y.-S.; Deuel, T. F.: A dominant-negative pleiotrophin mutant introduced by homologous recombination leads to germ-cell apoptosis in male mice. Proc. Nat. Acad. Sci. 96: 6734–6738, 1999.

[29177] 2729. Eddy, R. L.; Kretschmer, P. J.; Fairhurst, J. L.; Shows, T. B.; Bohlen, P.; O'Hara, B.; Kovesdi, I.: A human gene family of neurite outgrowth-promoting proteins: heparin-binding neurite outgrowth promoting factor maps to 11p11–11p13. (Abstract) Cytogenet. Cell Genet. 58:1958 only, 1991.

[29178] 2730. Kaname, T.; Kuwano, A.; Murano, I.; Uehara, K.; Muramatsu, T.; Kajii, T.: Midkine gene (MDK), a gene for pre-natal differentiation and neuroregulation, maps to band 11p11.2 by fluorescence in situ hybridization. Genomics 17: 514–515, 1993.

[29179] 2731. Simon-Chazottes, D.; Matsubara, S.; Miyauchi, T.;

Muramatsu, T.;Guenet, J.-L.: Chromosomal localization of two cell surface-associated molecules of potential importance in development: midkine (Mdk) and basigin (Bsg). *Mammalian Genome* 2: 269–271, 1992.

- [29180] 2732.Cool, D. R.; Normant, E.; Shen, F.; Chen, H.-C.; Pannell, L.; Zhang, Y.; Loh, Y. P.: Carboxypeptidase E is a regulated secretory pathway sorting receptor: genetic obliteration leads to endocrine disorders in Cpe(fat) mice. *Cell* 88: 73–83, 1997.
- [29181] 2733.Beg, A. A.; Sha, W. C.; Bronson, R. T.; Ghosh, S.; Baltimore, D.: Embryonic lethality and liver degeneration in mice lacking the RelA component of NF- κ B. *Nature* 376: 167–170, 1995.
- [29182] 2734.Chen, L.; Fischle, W.; Verdin, E.; Greene, W. C.: Duration of nuclear NF- κ B action regulated by reversible acetylation. *Science* 293:1653–1657, 2001.
- [29183] 2735.Deloukas, P.; Dauwerse, J. G.; van Ommen, G. J. B.; van Loon, A.P. G. M.: The human NFKB3 gene encoding the p65 subunit of transcription factor NF- κ B is located on chromosome 11q12. *Genomics* 19: 592–594, 1994.
- [29184] 2736.Deloukas, P.; van Loon, A. P. G. M.: Genomic organization of the gene encoding the p65 subunit of NF-

kappa-B: multiple variants of the p65 protein may be generated by alternative splicing. Hum. Molec. Genet. 2: 1895–1900, 1993.

- [29185] 2737. Neurath, M. F.; Pettersson, S.; Myer zum Buschenfelde, K.-H.; Strober, W.: Local administration of antisense phosphorothioate oligonucleotides to the p65 subunit of NF-kappa-B abrogates established experimental colitis in mice. Nature Med. 2: 998–1004, 1996.
- [29186] 2738. Zhong, H.; Voll, R. E.; Ghosh, S.: Phosphorylation of NF-kappaB by PKA stimulates transcriptional activity by promoting a novel bivalent interaction with the coactivator CBP/p300. Molec. Cell 1: 661–671, 1998.
- [29187] 2739. Biamonti, G.; Buvoli, M.; Bassi, M. T.; Morandi, C.; Cobianchi, F.; Riva, S.: Isolation of an active gene encoding human hnRNP protein A1: evidence for alternative splicing. J. Molec. Biol. 207: 491–503, 1989.
- [29188] 2740. Buvoli, M.; Biamonti, G.; Tsoulfas, P.; Bassi, M. T.; Ghetti, A.; Riva, S.; Morandi, C.: cDNA cloning of human hnRNP protein A1 reveals the existence of multiple mRNA isoforms. Nucleic Acids Res. 16: 3751–3770, 1988.
- [29189] 2741. Michael, W. M.; Choi, M.; Dreyfuss, G.: A nuclear export signal in hnRNP A1: a signal-mediated, temperature-dependent nuclear protein export pathway. Cell 83:

415–422, 1995.

- [29190] 2742. Saccone, S.; Biamonti, G.; Maugeri, S.; Bassi, M. T.; Bunone, G.; Riva, S.; Della Valle, G.: Assignment of the human heterogeneous nuclear ribonucleoprotein A1 gene (HNRPA1) to chromosome 12q13.1 by cDNA competitive in situ hybridization. *Genomics* 12: 171–174, 1992.
- [29191] 2743. Pollard, A. J.; Sparey, C.; Robson, S. C.; Krainer, A. R.; Europe-Finner, G. N.: Spatio-temporal expression of the trans-acting splicing factors SF2/ASF and heterogeneous ribonuclear proteins A1/A1B in the myometrium of the pregnant human uterus: a molecular mechanism for regulating regional protein isoform expression in vivo. *J. Clin. Endocr. Metab.* 85:1928–1936, 2000.
- [29192] 2744. Nicoloso, M.; Caizergues-Ferrer, M.; Michot, B.; Azum, M.-C.; Bachellerie, J.-P.: U20, a novel small nuclear RNA, is encoded in an intron of the nucleolin gene in mammals. *Molec. Cell. Biol.* 14: 5766–5776, 1994.
- [29193] 2745. Srivastava, M.; Fleming, P. J.; Pollard, H. B.; Burns, A. L.: Cloning and sequencing of the human nucleolin cDNA. *FEBS Lett.* 250:99–105, 1989.
- [29194] 2746. Srivastava, M.; McBride, O. W.; Fleming, P. J.; Pollard, H. B.; Burns, A. L.: Genomic organization and chromosomal localization of the human nucleolin gene. *J. Biol. Chem.*

265: 14922–14931, 1990.

- [29195] 2747.Turc–Carel, C.; Pietrzak, E.; Kakati, S.; Kinniburgh, A. J.; Sandberg,A. A.: INT1 maps to 12q12–12q13. (Abstract) Cytogenet. Cell Genet. 46:706 only, 1987.
- [29196] 2748.van't Veer, L. J.; Geurts van Kessel, A. H. M.; van Heerikhuizen,H.; van Ooyen, A.; Nusse, R.: Molecular cloning and chromosomal assignmentof the human homolog of int–1, a mouse gene implicated in mammarytumorigenesis. Molec. Cell. Biol. 4: 2532–2534, 1984.
- [29197] 2749.van Ooyen, A.; Kwee, V.; Nusse, R.: The nucleotide sequence ofthe human int–1 mammary oncogene; evolutionary conservation of codingand non–coding sequences. EMBO J. 4: 2905–2909, 1985.
- [29198] 2750.Wolda, S. L.; Moon, R. T.: Cloning and developmental expressionin *Xenopus laevis* of seven additional members of the Wnt family. Oncogene 7:1941–1947, 1992.
- [29199] 2751.Satijn, D. P. E.; Gunster, M. J.; van der Vlag, J.; Hamer, K. M.;Schul, W.; Alkema, M. J.; Saurin, A. J.; Freemont, P. S.; van Driel,R.; Otte, A. P.: RING1 is associated with the polycomb group proteincomplex and acts as a transcriptional repressor. Molec. Cell. Biol. 17:4105–4113, 1997.
- [29200] 2752.Armstrong, B. C.; Krystal, G. W.: Isolation and char–

acterization of complementary DNA for N-cym, a gene encoded by the DNA strand opposite to N-myc. *Cell Growth Differ.* 3: 385–390, 1992.

[29201] 2753. Brodeur, G. M.; Seeger, R. C.: Gene amplification in human neuroblastomas: basic mechanisms and clinical implications. *Cancer Genet. Cytogenet.* 19:101–111, 1986.

[29202] 2754. Brodeur, G. M.; Seeger, R. C.; Schwab, M.; Varmus, H. E.; Bishop, J. M.: Amplification of N-myc in untreated human neuroblastomas correlates with advanced disease stage. *Science* 224: 1121–1124, 1984.

[29203] 2755. Campbell, G. R.; Zimmerman, K.; Blank, R. D.; Alt, F. W.; D'Eustachio, P.: Chromosomal location of N-myc and L-myc genes in the mouse. *Oncogene Res.* 4: 47–54, 1989.

[29204] 2756. Corvi, R.; Amler, L. C.; Savelyeva, L.; Gehring, M.; Schwab, M.: MYCN is retained in single copy at chromosome 2 band p23–24 during amplification in human neuroblastoma cells. *Proc. Nat. Acad. Sci.* 91:5523–5527, 1994.

[29205] 2757. Emanuel, B. S.; Balaban, G.; Boyd, J. P.; Grossman, A.; Negishi, M.; Parmiter, A.; Glick, M. C.: N-myc amplification in multiple homogeneously staining regions in two human neuroblastomas. *Proc. Nat. Acad. Sci.*

82:3736–3740, 1985.

- [29206] 2758. Garson, J. A.; van den Berghe, J. A.; Kemshead, J. T.: Novel non-isotopic in situ hybridization technique detects small (1 kb) unique sequences in routinely G-banded human chromosomes: fine mapping of N-myc and beta-NGF genes. *Nucleic Acids Res.* 15: 4761–4770, 1987.
- [29207] 2759. Guo, C.; White, P. S.; Weiss, M. J.; Hogarty, M. D.; Thompson, P. M.; Stram, D. O.; Gerbing, R.; Matthay, K. K.; Seeger, R. C.; Brodeur, G. M.; Maris, J. M.: Allelic deletion at 11q23 is common in MYCN single copy neuroblastomas. *Oncogene* 18: 4948–4957, 1999.
- [29208] 2760. Kanda, N.; Schreck, R.; Alt, F.; Bruns, G.; Baltimore, D.; Latt, S.: Isolation of amplified DNA sequences from IMR-32 human neuroblastoma cells: facilitation by fluorescence-activated flow sorting of metaphase chromosomes. *Proc. Nat. Acad. Sci.* 80: 4069–4073, 1983.
- [29209] 2761. Kohl, N. E.; Gee, C. E.; Alt, F. W.: Activated expression of the N-myc gene in human neuroblastomas and related tumors. *Science* 226: 1335–1337, 1984.
- [29210] 2762. Kohl, N. E.; Kanda, N.; Schreck, R. R.; Bruns, G.; Latt, S. A.; Gilbert, F.; Alt, F. W.: Transposition and amplification of oncogene-related sequences in human neuroblastomas. *Cell* 35: 359–367, 1983.

- [29211] 2763.Kohl, N. E.; Legouy, E.; DePinho, R. A.; Nisen, P. D.; Smith,R. K.; Gee, C. E.; Alt, F. W.: Human N-myc is closely related in organization and nucleotide sequence to c-myc. *Nature* 319: 73–77,1986.
- [29212] 2764.Lee, W.-H.; Murphree, A. L.; Benedict, W. F.: Expression and amplification of the N-myc gene in primary retinoblastoma. *Nature* 309:458–460, 1984.
- [29213] 2765.Michitsch, R. W.; Melera, P. W.: Nucleotide sequence of the 3-prime exon of the human N-myc gene. *Nucleic Acids Res.* 13: 2545–2558,1985.
- [29214] 2766.Ramsay, G.; Stanton, L.; Schwab, M.; Bishop, J. M.: Human proto-oncogene N-myc encodes nuclear proteins that bind DNA. *Molec. Cell. Biol.* 6:4450–4457, 1986.
- [29215] 2767.Reiter, J. L.; Brodeur, G. M.: High-resolution mapping of a 130-kb core region of the MYCN amplicon in neuroblastomas. *Genomics* 32:97–103, 1996.
- [29216] 2768.Schwab, M.: Amplification of N-myc in human neuroblastomas. *Trends Genet.* 1: 271–275, 1985.
- [29217] 2769.Schwab, M.; Alitalo, K.; Klempnauer, K.-H.; Varmus, H. E.; Bishop, J. M.; Gilbert, F.; Brodeur, G.; Goldstein, M.; Trent, J.: Amplified DNA with limited homology to myc cellular oncogene is shared by human neuroblastoma cell lines and a neuroblastoma tumour. *Nature* 305:245–248,

1983.

- [29218] 2770. Schwab, M.; Ellison, J.; Busch, M.; Rosenau, W.; Varmus, H. E.; Bishop, J. M.: Enhanced expression of the human gene N-myc consequent to amplification of DNA may contribute to malignant progression of neuroblastoma. Proc. Nat. Acad. Sci. 81: 4940–4944, 1984.
- [29219] 2771. Schwab, M.; Varmus, H. E.; Bishop, J. M.; Grzeschik, K.-H.; Naylor, S. L.; Sakaguchi, A. Y.; Brodeur, G.; Trent, J.: Chromosome localization in normal human cells and neuroblastomas of a gene related to c-myc. Nature 308:288–291, 1984.
- [29220] 2772. Seeger, R. C.; Brodeur, G. M.; Sather, H.; Dalton, A.; Siegel, S. E.; Wong, K. Y.; Hammond, D.: Association of multiple copies of the N-myc oncogene with rapid progression of neuroblastomas. New Eng. J. Med. 313: 1111–1116, 1985.
- [29221] 2773. Shiloh, Y.; Shipley, J.; Brodeur, G. M.; Bruns, G.; Korf, B.; Donlon, T.; Schreck, R. R.; Seeger, R.; Sakai, K.; Latt, S. A.: Differential amplification, assembly, and relocation of multiple DNA sequences in human neuroblastomas and neuroblastoma cell lines. Proc. Nat. Acad. Sci. 82: 3761–3765, 1985.
- [29222] 2774. Bahou, W. F.; Nierman, W. C.; Durkin, A. S.; Potter, C.

L.; Demetrick,D. J.: Chromosomal assignment of the human thrombin receptor gene:localization to region q13 of chromosome 5. Blood 82: 1532–1537,1993.

[29223] 2775.Coughlin, S. R.; Vu, T.–K. H.; Hung, D. T.; Wheaton, V. I.: Characterizationof a functional thrombin receptor: issues and opportunities. J. Clin.Invest. 89: 351–355, 1992.

[29224] 2776.Griffin, C. T.; Srinivasan, Y.; Zheng, Y.–W.; Huang, W.; Coughlin,S. R.: A role for thrombin receptor signaling in endothelial cellsduring embryonic development. Science 293: 1666–1670, 2001.

[29225] 2777.Poirier, C.; O'Brien, E. P.; Bueno Brunialti, A. L.; Chambard,J.–C.; Swank, R. T.; Guenet, J.–L.: The gene encoding the thrombinreceptor (Cf2r) maps to mouse chromosome 13. Mammalian Genome 7:322, 1996.

[29226] 2778.Schmidt, V. A.; Nierman, W. C.; Feldblyum, T. V.; Maglott, D. R.;Bahou, W. F.: The human thrombin receptor and proteinase activatedreceptor–2 genes are tightly linked on chromosome 5q13. Brit. J.Haemat. 97: 523–529, 1997.

[29227] 2779.Schmidt, V. A.; Vitale, E.; Bahou, W. F.: Genomic cloning andcharacterization of the human thrombin receptor gene: structural similarityto the proteinase acti–

vated receptor-2 gene. *J. Biol. Chem.* 271:9307–9312, 1996.

[29228] 2780. Vu, T.-K. H.; Hung, D. T.; Wheaton, V. I.; Coughlin, S. R.: Molecular cloning of a functional thrombin receptor reveals a novel proteolytic mechanism of receptor activation. *Cell* 64: 1057–1068, 1991.

[29229] 2781. Dean, G.: *The Porphyrrias. A Story of Inheritance and Environment.* Philadelphia: J. B. Lippincott (pub.) (2nd ed.): 1972.

[29230] 2782. Bennett, M. K.; Calakos, N.; Scheller, R. H.: Syntaxin: a synaptic protein implicated in docking of synaptic vesicles at presynaptic active zones. *Science* 257: 255–259, 1992.

[29231] 2783. Bennett, M. K.; Garcia-Arreaga, J. E.; Elferink, L. A.; Peterson, K.; Fleming, A. M.; Hazuka, C. D.; Scheller, R. H.: The syntaxin family of vesicular transport receptors. *Cell* 74: 863–873, 1993.

[29232] 2784. Fernandez, I.; Ubach, J.; Dulubova, I.; Zhang, X.; Sudof, T. C.; Rizo, J.: Three-dimensional structure of an evolutionarily conserved N-terminal domain of syntaxin 1A. *Cell* 94: 841–849, 1998.

[29233] 2785. Fisher, R. J.; Pevsner, J.; Burgoyne, R. D.: Control of fusion pore dynamics during exocytosis by Munc18. *Sci-*

ence 291: 875–878,2001.

- [29234] 2786.Martin–Martin, B.; Nabokina, S. M.; Lazo, P. A.; Mollinedo, F.: Co–expression of several human syntaxin genes in neutrophils and differentiating HL–60 cells: various isoforms and detection of syntaxin1. J. Leuko. Biol. 65: 397–406, 1999.
- [29235] 2787.Nakayama, T.; Fujiwara, T.; Miyazawa, A.; Asakawa, S.; Shimizu,N.; Shimizu, Y.; Mikoshiba, K.; Akagawa, K.: Mapping of the humanHPC–1/syntaxin 1A gene (STX1A) to chromosome 7 band q11.2. Genomics 42:173–176, 1997.
- [29236] 2788.Naren, A. P.; Di, A.; Cormet–Boyaka, E.; Boyaka, P. N.; McGhee,J. R.; Zhou, W.; Akagawa, K.; Fujiwara, T.; Thome, U.; Engelhardt,J. F.; Nelson, D. J.; Kirk, K. L.: Syntaxin 1A is expressed in airwayepithelial cells, where it modulates CFTR C1– currents. J. Clin.Invest. 105: 377–386, 2000.
- [29237] 2789.Richmond, J. E.; Weimer, R. M.; Jorgensen, E. M.: An open formof syntaxin bypasses the requirement for UNC–13 in vesicle priming. Nature 412:338–341, 2001.
- [29238] 2790.Zhang, R.; Maksymowych, A. B.; Simpson, L. L.: Cloning and sequenceanalysis of a cDNA encoding human syntaxin 1A, a polypeptide essentialfor exocytosis. Gene

159: 293–294, 1995.

- [29239] 2791. Shepley, M. P.; Sherry, B.; Weiner, H. L.: Monoclonal antibody identification of a 100-kDa membrane protein in HeLa cells and human spinal cord involved in poliovirus attachment. *Proc. Nat. Acad. Sci.* 85:7743–7747, 1988.
- [29240] 2792. Siddique, T.; Bartlett, R. J.; McKinney, R.; Hung, W.-Y.; Bruns, G.; Wilfert, C.; Roses, A. D.: The poliovirus sensitivity (PVS) is on chromosome 19q13-qter. (Abstract) *Cytogenet. Cell Genet.* 40:745 only, 1985.
- [29241] 2793. Siddique, T.; McKinney, R.; Hung, W.-Y.; Bartlett, R. J.; Bruns, G.; Mohandas, T. K.; Ropers, H.-H.; Wilfert, C.; Roses, A. D.: The poliovirus sensitivity (PVS) gene is on chromosome 19q12-q13.2. *Genomics* 3:156–160, 1988.
- [29242] 2794. Solecki, D. J.; Gromeier, M.; Mueller, S.; Bernhardt, G.; Wimmer, E.: Expression of the human poliovirus receptor/CD155 gene is activated by Sonic hedgehog. *J. Biol. Chem.* 277: 25697–25702, 2002.
- [29243] 2795. Morris, C. M.; Bodger, M. P.: Localization of the human poly(A)-binding protein gene (PAB1) to chromosomal regions 3q22-q25, 12q13-q14, and 13q12-q13 by in situ hybridization. *Genomics* 15: 209–211, 1993.
- [29244] 2796. Alkhatib, H. M.; Chen, D.; Cherney, B.; Bhatia, K.; Notario, V.; Giri, C.; Stein, G.; Slattery, E.; Roeder, R. G.;

Smulson, M. E.:Cloning and expression of cDNA for human poly(ADP-ribose) polymerase. Proc.Nat. Acad. Sci. 84: 1224-1228, 1987.

[29245] 2797.Auer, B.; Nagl, U.; Herzog, H.; Schneider, R.; Schweiger, M.:Human nuclear NAD⁺ ADP-riboseyltransferase(polymerizing): organizationof the gene. DNA 8: 575-580, 1989.

[29246] 2798.Baumgartner, M.; Schneider, R.; Auer, B.; Herzog, H.; Schweiger,M.; Hirsch-Kauffmann, M.: Fluorescence in situ mapping of the humannuclear NAD⁺ ADP-riboseyltransferase gene (ADPRT) and two secondarysites to human chromosomal bands 1q42, 13q34, and 14q24. Cytogenet.Cell Genet. 61: 172-174, 1992.

[29247] 2799.Bhatia, K. G.; Cherney, B. W.; Huppi, K.; Magrath, I. T.; Cossman,J.; Sausville, E.; Barriga, F.; Johnson, B.; Gause, B.; Bonney, G.;Neequayi, J.; DeBernardi, M.; Smulson, M.: A deletion linked to apoly(ADP-ribose) polymerase gene on chromosome 3q33-qter occurs frequentlyin the normal black population as well as in multiple tumor DNA. CancerRes. 50: 5406-5413, 1990.

[29248] 2800.Cherney, B. W.; McBride, O. W.; Chen, D.; Alkhatib, H.; Bhatia,K.; Hensley, P.; Smulson, M. E.: cDNA sequence, protein structure,and chromosomal location of the human

gene for poly(ADP-ribose) polymerase. Proc.Nat. Acad. Sci. 84: 8370-8374, 1987.

- [29249] 2801.d'Adda di Fagagna, F.; Hande, M. P.; Tong, W.-M.; Lansdorp, P.M.; Wang, Z.-Q.; Jackson, S. P.: Functions of poly(ADP-ribose) polymerase in controlling telomere length and chromosomal stability. Nature Genet. 23: 76-80, 1999.
- [29250] 2802.Doll, J. A.; Suarez, B. K.; Donis-Keller, H.: Association between prostate cancer in black Americans and an allele of the PADPRP pseudogene locus on chromosome 13. (Letter) Am. J. Hum. Genet. 58: 425-428, 1996.
- [29251] 2803.Flick, K.; Schneider, R.; Auer, B.; Hirsch-Kauffmann, M.; Schweiger, M.: No abnormalities in the NAD(+) ADP-ribosyltransferase (polymerizing) gene of transformed cells from a Fanconi's anemia patient. (Letter) Hum. Genet. 89: 690-691, 1992.
- [29252] 2804.Grube, K.; Burkle, A.: Poly(ADP-ribose) polymerase activity in mononuclear leukocytes of 13 mammalian species correlates with species-specific life span. Proc. Nat. Acad. Sci. 89: 11759-11763, 1992.
- [29253] 2805.Herzog, H.; Zabel, B.; Schneider, R.; Auer, B.; Hirsch-Kauffmann, M.; Schweiger, M.: Human NAD(+):protein ADP ribosyltransferase (ADPRT): synthesis of active enzyme in

E. coli, sequence of its cDNA from HeLa cells and chromosomal localization. *Biol. Chem. Hoppe-Seyler* 369:836–837, 1988.

[29254] 2806. Herzog, H.; Zabel, B. U.; Schneider, R.; Auer, B.; Hirsch-Kauffmann, M.; Schweiger, M.: Human nuclear NAD(+) ADP-ribosyltransferase: localization of the gene on chromosome 1q41–q42 and expression of an active human enzyme in *Escherichia coli*. *Proc. Nat. Acad. Sci.* 86: 3514–3518, 1989.

[29255] 2807. Keijzer, W.; Stefanini, M.; Bootsma, D.; Verkerk, A.; Geurts van Kessel, A. H. M.; Jongkind, J. F.; Westerveld, A.: Localization of a gene involved in complementation of the defect in xeroderma pigmentosum group A cells on human chromosome 1. *Exp. Cell Res.* 169: 490–501, 1987.

[29256] 2808. Kurosaki, T.; Ushiro, H.; Mitsuuchi, Y.; Suzuki, S.; Matsuda, M.; Matsuda, Y.; Katunuma, N.; Kangawa, K.; Matsuo, H.; Hirose, T.; Inayama, S.; Shizuta, Y.: Primary structure of human poly(ADP-ribose) synthetase as deduced from cDNA sequence. *J. Biol. Chem.* 262: 15990–15997, 1987.

[29257] 2809. Loetscher, P.; Alvarez-Gonzalez, R.; Althaus, F. R.: Poly(ADP-ribose) may signal changing metabolic conditions to the chromatin of mammalian cells. *Proc. Nat.*

Acad. Sci. 84: 1286–1289, 1987.

[29258] 2810.Lyn, D.; Cherney, B. W.; Lalande, M.; Berenson, J. R.; Lichtenstein,A.; Lupold, S.; Bhatia, K. G.; Smulson, M.: A duplicated region is responsible for the poly(ADP–ribose) polymerase polymorphism, on chromosome 13, associated with a predisposition to cancer. Am. J. Hum. Genet.

52:124–134, 1993.

[29259] 2811.McBride, O. W.; Cherney, B.; Takourdin, C.; Smulson, M. E.: Human poly(ADP–ribose) polymerase sequences are located on chromosomes 1,13, and 14. (Abstract) Cytogenet. Cell Genet. 46: 659–660, 1987.

[29260] 2812.Pieper, A. A.; Brat, D. J.; Krug, D. K.; Watkins, C. C.; Gupta,A.; Blackshaw, S.; Verma, A.; Wang, Z.–Q.; Snyder, S. H.: Poly(ADP–ribose)polymerase–deficient mice are protected from streptozotocin–induced diabetes. Proc. Nat. Acad. Sci. 96: 3059–3064, 1999.

[29261] 2813.Schweiger, M.; Auer, B.; Burtscher, H. J.; Hirsch–Kauffmann, M.;Klocker, H.; Schneider, R.: DNA repair in human cells: biochemistry of the hereditary diseases Fanconi's anaemia and Cockayne syndrome. Europ.J. Biochem. 165: 235–242, 1987.

[29262] 2814.Simbulan–Rosenthal, C. M.; Haddad, B. R.; Rosenthal, D. S.; Weaver,Z.; Coleman, A.; Luo, R.; Young, H. M.;

Wang, Z.-Q.; Ried, T.; Smulson, M. E.: Chromosomal aberrations in PARP $-/-$ mice: genome stabilization in immortalized cells by reintroduction of poly(ADP-ribose) polymerase cDNA. *Proc. Nat. Acad. Sci.* 96: 13191–13196, 1999.

[29263] 2815. Smithies, O.; Gregg, R. G.; Boggs, S. S.; Koralewski, M. A.; Kucherlapati, R. S.: *Nature (London)* 317: 230–234, 1985.

[29264] 2816. Bravo, R.: Synthesis of the nuclear protein cyclin (PCNA) and its relationship with DNA replication. *Exp. Cell Res.* 163: 287–293, 1986.

[29265] 2817. Hasan, S.; Hassa, P. O.; Imhof, R.; Hottiger, M. O.: Transcription coactivator p300 binds PCNA and may have a role in DNA repair synthesis. *Nature* 410: 387–391, 2001.

[29266] 2818. Hoege, C.; Pfander, B.; Moldovan, G.-L.; Pyrowolakis, G.; Jentsch, S.: RAD6-dependent DNA repair is linked to modification of PCNA by ubiquitin and SUMO. *Nature* 419: 135–141, 2002.

[29267] 2819. Ku, D.-H.; Travali, S.; Calabretta, B.; Huebner, K.; Baserga, R.: Human gene for proliferating cell nuclear antigen has pseudogenes and localizes to chromosome 20. *Somat. Cell Molec. Genet.* 15: 297–307, 1989.

[29268] 2820. Mann, M. J.; Gibbons, G. H.; Kernoff, R. S.; Diet, F. P.;

Tsao, P. S.; Cooke, J. P.; Kaneda, Y.; Dzaou, V. J.: Genetic engineering of vein grafts resistant to atherosclerosis. Proc. Nat. Acad. Sci. 92:4502–4506, 1995.

- [29269] 2821. Rao, V. V. N. G.; Schnittger, S.; Hansmann, I.: Chromosomal localization of the human proliferating cell nuclear antigen (PCNA) gene to or close to 20p12 by in situ hybridization. Cytogenet. Cell Genet. 56:169–170, 1991.
- [29270] 2822. Suzuka, I.; Daidoji, H.; Matsuoka, M.; Kadowaki, K.; Takasaki, Y.; Nakane, P. K.; Moriuchi, T.: Gene for proliferating-cell nuclear antigen (DNA polymerase delta auxiliary protein) is present in both mammalian and higher plant genomes. Proc. Nat. Acad. Sci. 86: 3189–3193, 1989.
- [29271] 2823. Taniguchi, Y.; Katsumata, Y.; Koido, S.; Suemizu, H.; Yoshimura, S.; Moriuchi, T.; Okumura, K.; Kagotani, K.; Taguchi, H.; Imanishi, T.; Gojobori, T.; Inoko, H.: Cloning, sequencing, and chromosomal localization of two tandemly arranged human pseudogenes for the proliferating cell nuclear antigen (PCNA). Mammalian Genome 7: 906–908, 1996.
- [29272] 2824. Travali, S.; Ku, D.-H.; Rizzo, M. G.; Ottavio, L.; Baserga, R.; Calabretta, B.: Structure of the human gene for the proliferating cell nuclear antigen. J. Biol. Chem. 264: 7466–7472, 1989.

- [29273] 2825. Webb, G.; Parsons, P.; Chenevix-Trench, G.: Localization of the gene for human proliferating nuclear antigen/cyclin by in situ hybridization. Hum.Genet. 86: 84–86, 1990.
- [29274] 2826. Fonatsch, C.; Duchrow, M.; Rieder, H.; Schluter, C.; Gerdes, J.: Assignment of the human Ki-67 gene (MKI67) to 10q25-qter. Genomics 11:476–477, 1991.
- [29275] 2827. Schluter, C.; Duchrow, M.; Wohlenberg, C.; Becker, M. H. G.; Key, G.; Flad, H.-D.; Gerdes, J.: The cell proliferation-associated antigen of antibody Ki-67: a very large, ubiquitous nuclear protein with numerous repeated elements, representing a new kind of cell cycle-maintaining proteins. J. Cell. Biol. 123: 513–522, 1993.
- [29276] 2828. Schonk, D. M.; Kuijpers, H. J. H.; van Drunen, E.; van Dalen, C. H.; Geurts van Kessel, A. H. M.; Verheijen, R.; Ramaekers, F. C. S.: Assignment of the gene(s) involved in the expression of the proliferation-related Ki-67 antigen to human chromosome 10. Hum. Genet. 83: 297–299, 1989.
- [29277] 2829. Traut, W.; Scholzen, T.; Winking, H.; Kubbutat, M. H. G.; Gerdes, J.: Assignment of the murine Ki-67 gene (Mki67) to chromosome band 7F3–F5 by in situ hybridization. Cytogenet. Cell Genet. 83: 12–13, 1998.

- [29278] 2830.Cooke, N. E.; Baxter, J. D.: Structural analysis of the prolactin gene suggests a separate origin for its 5-prime end. *Nature* 297:603–606, 1982.
- [29279] 2831.Cooke, N. E.; Coit, D.; Shine, J.; Baxter, J. D.; Martial, J. A.: Human prolactin: cDNA structural analysis and evolutionary comparisons. *J.Biol. Chem.* 256: 4007–4016, 1981.
- [29280] 2832.D'Alessandro, E.; Santiemma, V.; Lo Re, M. L.; Ligas, C.; Del Porto, G.: 6p23 deletion mosaicism in a woman with recurrent abortions and idiopathic hypoprolactinemia. *Am. J. Med. Genet.* 44: 220–222, 1992.
- [29281] 2833.DiMattia, G. E.: Personal Communication. London, Ontario, Canada 3/23/1998.
- [29282] 2834.DiMattia, G. E.; Gellersen, B.; Duckworth, M. L.; Friesen, H. G.: Human prolactin gene expression: the use of an alternative noncoding exon in decidua and the IM-9-P3 lymphoblast cell line. *J. Biol. Chem.* 265:16412–16421, 1990.
- [29283] 2835.Evans, A. M.; Petersen, J. W.; Sekhon, G. S.; DeMars, R. I.: Use of human lymphoblastoid deletion mutants to map the prolactin gene on human chromosome 6p. (Abstract) *Am. J. Hum. Genet.* 43: A143 only, 1988.
- [29284] 2836.Farid, N. R.; Noel, E. P.; Sampson, L.; Russell, N. A.:

Prolactin-secreting adenomata are possibly associated with HLA-B8. *Tissue Antigens* 15:333–335, 1980.

[29285] 2837. Gellersen, B.; Kempf, R.; Telgmann, R.; DiMattia, G. E.: Nonpituitary human prolactin gene transcription is independent of Pit-1 and differentially controlled in lymphocytes and in endometrial stroma. *Molec. Endocr.*

8:356–373, 1994.

[29286] 2838. Erdile, L. F.; Heyer, W.-D.; Kolodner, R.; Kelly, T. J.: Characterization of a cDNA encoding the 70-kDa single-stranded DNA-binding subunit of human replication protein A and the role of the protein in DNA replication. *J. Biol. Chem.* 266: 12090–12098, 1991.

[29287] 2839. Gomes, X. V.; Wold, M. S.: Functional domains of the 70-kilodalton subunit of human replication protein A. *Biochemistry* 35: 10558–10568, 1996.

[29288] 2840. Shen, L. X.; Basilion, J. P.; Stanton, V. P., Jr.: Single-nucleotide polymorphisms can cause different structural folds of mRNA. *Proc. Nat. Acad. Sci.* 96: 7871–7876, 1999.

[29289] 2841. Umbricht, C. B.; Erdile, L. F.; Jabs, E. W.; Kelly, T. J.: Cloning, overexpression, and genomic mapping of the 14-kDa subunit of human replication protein A. *J. Biol. Chem.* 268: 6131–6138, 1993.

[29290] 2842. Umbricht, C. B.; Griffin, C. A.; Hawkins, A. L.;

Grzeschik, K.H.; O'Connell, P.; Leach, R.; Green, E. D.; Kelly, T. J.: High-resolution genomic mapping of the three human replication protein A genes (RPA1, RPA2, and RPA3). *Genomics* 20: 249–257, 1994.

[29291] 2843. Bilbe, G.; Delabie, J.; Bruggen, J.; Richener, H.; As-selbergs, F. A. M.; Cerletti, N.; Sorg, C.; Odink, K.; Tarcsay, L.; Wiesendanger, W.; DeWolf-Peeters, C.; Shipman, R.: Restin: a novel intermediate filament-associated protein highly expressed in the Reed-Sternberg cells of Hodgkin's disease. *EMBO J.* 11: 2103–2113, 1992.

[29292] 2844. Delabie, J.; Shipman, R.; Bruggen, J.; De Strooper, B.; van Leuven, F.; Tarcsay, L.; Cerletti, N.; Odink, K.; Diehl, V.; Bilbe, G.; DeWolf-Peeters, C.: Expression of the novel intermediate filament-associated protein restin in Hodgkin's disease and anaplastic large-cell lymphoma. *Blood* 80:2891–2896, 1992.

[29293] 2845. Fukata, M.; Watanabe, T.; Noritake, J.; Nakagawa, M.; Yamaga, M.; Kuroda, S.; Matsuura, Y.; Iwamatsu, A.; Perez, F.; Kaibuchi, K.: Rac1 and Cdc42 capture microtubules through IQGAP1 and CLIP-170. *Cell* 109:873–885, 2002.

[29294] 2846. Griparic, L.; Keller, T. C. S., III: Identification and expression of two novel CLIP-170/restin isoforms expressed predominantly in muscle. *Biochim. Biophys. Acta* 1405:

35–46, 1998.

- [29295] 2847. Hilliker, C.; Delabie, J.; Speleman, F.; Bilbe, G.; Bruggen, J.; Van Leuven, F.; Van Den Berghe, H.: Localization of the gene (RSN) coding for restin, a marker for Reed–Sternberg cells in Hodgkin's disease, to human chromosome band 12q24.3 and YAC cloning of the locus. *Cytogenet. Cell Genet.* 65: 172–176, 1994.
- [29296] 2848. Durbin, H.; Rodrigues, N.; Bodmer, W. F.: Further characterization, isolation and identification of the epithelial cell–surface antigen defined by monoclonal antibody AUA1. *Int. J. Cancer* 45: 562–565, 1990.
- [29297] 2849. Meyaard, L.; van der Vuurst de Vries, A.–R.; de Ruiter, T.; Lanier, L. L.; Phillips, J. H.; Clevers, H.: The epithelial cellular adhesion molecule (Ep–CAM) is a ligand for the leukocyte–associated immunoglobulin–like receptor (LAIR). *J. Exp. Med.* 194: 107–112, 2001.
- [29298] 2850. Perez, M. S.; Walker, L. E.: Isolation and characterization of a cDNA encoding the KS1/4 epithelial carcinoma marker. *J. Immun.* 142: 3662–3667, 1989.
- [29299] 2851. Spurr, N. K.; Durbin, H.; Sheer, D.; Parkar, M.; Bobrow, L.; Bodmer, W. F.: Characterization and chromosomal assignment of a human cell surface antigen defined by the monoclonal antibody AUA1. *Int. J. Cancer* 38: 631–636,

1986.

- [29300] 2852.Strnad, J.; Hamilton, A. E.; Beavers, L. S.; Gamboa, G. C.; Apelgren, L. D.; Taber, L. D.; Sportsman, J. R.; Bumol, T. F.; Sharp, J. D.; Gadski, R. A.: Molecular cloning and characterization of a human adenocarcinoma/epithelial cell surface antigen complementary DNA. *Cancer Res.* 49: 314–317, 1989.
- [29301] 2853.Szala, S.; Froehlich, M.; Scollon, M.; Kasai, Y.; Steplewski, Z.; Koprowski, H.; Linnenbach, A. J.: Molecular cloning of cDNA for the carcinoma-associated antigen GA733–2. *Proc. Nat. Acad. Sci.* 87:3542–3546, 1990.
- [29302] 2854.Hitchins, M. P.; Monk, D.; Bell, G. M.; Ali, Z.; Preece, M. A.; Stanier, P.; Moore, G. E.: Maternal repression of the human GRB10 gene in the developing central nervous system; evaluation of the role for GRB10 in Silver–Russell syndrome. *Europ. J. Hum. Genet.* 9: 82–90, 2001.
- [29303] 2855.McCann, J. A.; Zheng, H.; Islam, A.; Goodyer, C. G.; Polychronakos, C.: Evidence against GRB10 as the gene responsible for Silver–Russell syndrome. *Biochem. Biophys. Res. Commun.* 286: 943–948, 2001.
- [29304] 2856.Miyoshi, N.; Kuroiwa, Y.; Kohda, T.; Shitara, H.; Yonekawa, H.; Kawabe, T.; Hasegawa, H.; Barton, S. C.; Surani, M. A.; Kaneko–Ishino, T.; Ishino, F.: Identification of

the Meg1/Grb10 imprinted gene on mouse proximal chromosome 11, a candidate for the Silver-Russell syndrome gene. Proc. Nat. Acad. Sci. 95: 1102–1107, 1998.

- [29305] 2857. Yoshihashi, H.; Maeyama, K.; Kosaki, R.; Ogata, T.; Tsukahara, M.; Goto, Y.; Hata, J.; Matsuo, N.; Smith, R. J.; Kosaki, K.: Imprinting of human GRB10 and its mutations in two patients with Russell-Silver syndrome. Am. J. Hum. Genet. 67: 476–482, 2000.
- [29306] 2858. Grob, P. M.; Ross, A. H.; Koprowski, H.; Bothwell, M.: Characterization of the human melanoma nerve growth factor receptor. J. Biol. Chem. 260: 8044–8049, 1985.
- [29307] 2859. Stanton, L. W.; Schwab, M.; Bishop, J. M.: Nucleotide sequence of the human N-myc gene. Proc. Nat. Acad. Sci. 83: 1772–1776, 1986.
- [29308] 2860. Brown, M. A.; Nicolai, H.; Xu, C.-F.; Griffiths, B. L.; Jones, K. A.; Solomon, E.; Hosking, L.; Trowsdale, J.; Black, D. M.; McFarlane, R.: Regulation of BRCA1. (Letter) Nature 372: 733 only, 1994.
- [29309] 2861. Brown, M. A.; Xu, C.-F.; Nicolai, H.; Griffiths, B.; Chambers, J. A.; Black, D.; Solomon, E.: The 5-prime end of the BRCA1 gene lies within a duplicated region of human chromosome 17q21. Oncogene 12: 2507–2513, 1996.

- [29310] 2862.Campbell, I. G.; Nicolai, H. M.; Foulkes, W. D.; Senger, G.; Stamp, G. W.; Allan, G.; Boyer, C.; Jones, K.; Bast, R. C., Jr.; Solomon, E.; Trowsdale, J.; Black, D. M.: A novel gene encoding a B-box protein within the BRCA1 region at 17q21.1. *Hum. Molec. Genet.* 3: 589–594, 1994.
- [29311] 2863.Kawashima, K.; Shikama, H.; Imoto, K.; Izawa, M.; Naruke, T.; Okabayashi, K.; Nishimura, S.: Close correlation between restriction fragment length polymorphism of the L-MYC gene and metastasis of human lung cancer to the lymph nodes and other organs. *Proc. Nat. Acad. Sci.* 85:2353–2356, 1988.
- [29312] 2864.Kaye, F.; Battey, J.; Nau, M.; Brooks, B.; Seifter, E.; De Greve, J.; Birrer, M.; Sausville, E.; Minna, J.: Structure and expression of the human L-myc gene reveal a complex pattern of alternative mRNA processing. *Molec. Cell. Biol.* 8: 186–195, 1988.
- [29313] 2865.McBride, O. W.; Kirsch, I.; Hollis, G.; Nau, M.; Battey, J.; Minna, J.: Human L-myc (MYCL) proto-oncogene is on chromosome 1p32.(Abstract) *Cytogenet. Cell Genet.* 40: 694 only, 1985.
- [29314] 2866.Nau, M. M.; Brooks, B. J.; Battey, J.; Sausville, E.; Gazdar, A.F.; Kirsch, I. R.; McBride, O. W.; Bertness, V.; Hollis, G. F.; Minna, J. D.: L-myc, a new myc-related gene

amplified and expressed in human small cell lung cancer.

Nature 318: 69–73, 1985.

[29315] 2867. Rouleau, G. A.; Bazanowski, A.; Gusella, J. F.; Haines, J. L.: A genetic map of chromosome 1: comparison of different data sets and linkage programs. Genomics 7: 313–318, 1990.

[29316] 2868. Speleman, F.; Van Camp, G.; Van Roy, N.: Reassignment of MYCL1 to human chromosome 1p34.3 by fluorescence in situ hybridization. Cytogenet. Cell Genet. 72: 189–190, 1996.

[29317] 2869. Van Roy, N.; Cheng, N. C.; Laureys, G.; Opdenakker, G.; Versteeg, R.; Speleman, F.: Molecular cytogenetic analysis of 1;17 translocations in neuroblastoma. Europ. J. Cancer 31A: 530–535, 1995.

[29318] 2870. Dean, M.; Park, M.; Le Beau, M. M.; Robins, T. S.; Diaz, M. O.; Rowley, J. D.; Blair, D. G.; Vande Woude, G. F.: The human met oncogene is related to the tyrosine kinase oncogenes. Nature 318: 385–388, 1985.

[29319] 2871. Akiyama, T.; Sudo, C.; Ogawara, H.; Toyoshima, K.; Yamamoto, T.: The product of the human c-erbB-2 gene: a 185-kilodalton glycoprotein with tyrosine kinase activity. Science 232: 1644–1646, 1986.

[29320] 2872. Ameyaw, M.-M.; Tayeb, M.; Thornton, N.; Folayan,

G.; Tariq, M.; Mobarek, A.; Evans, D. A. P.; Ofori-Adjei, D.; McLeod, H. L.: Ethnic variation in the HER-2 codon 655 genetic polymorphism previously associated with breast cancer. *J. Hum. Genet.* 47: 172–175, 2002.

[29321] 2873. Chan, J. Y. C.; Lerman, M. I.; Prabhakar, B. S.; Isozaki, O.; Santisteban, P.; Koppers, R. C.; Oates, E. L.; Notkins, A. L.; Kohn, L. D.: Cloning and characterization of a cDNA that encodes a 70-kDa novel human thyroid autoantigen. *J. Biol. Chem.* 264: 3651–3654, 1989.

[29322] 2874. Goedecke, W.; Eijpe, M.; Offenberg, H. H.; van Aalderen, M.; Heyting, C.: Mre11 and Ku70 interact in somatic cells, but are differentially expressed in early meiosis. *Nature Genet.* 23: 194–198, 1999.

[29323] 2875. Hartley, K. O.; Gell, D.; Smith, G. C. M.; Zhang, H.; Divecha, N.; Connelly, M. A.; Admon, A.; Lees-Miller, S. P.; Anderson, C. W.; Jackson, S. P.: DNA-dependent protein kinase catalytic subunit: a relative of phosphatidylinositol 3-kinase and the ataxia telangiectasia gene product. *Cell* 82: 849–856, 1995.

[29324] 2876. Koike, M.; Matsuda, Y.; Mimori, T.; Harada, Y.-N.; Shiomi, N.; Shiomi, T.: Chromosomal localization of the mouse and rat DNA double-strand break repair genes Ku p70 and Ku p80/XRCC5 and their mRNA expression in var-

ious mouse tissues. *Genomics* 38: 38–44, 1996.

- [29325] 2877. Kusano, K.; Johnson–Schlitz, D. M.; Engels, W. R.: Sterility of *Drosophila* with mutations in the Bloom syndrome gene—complementation by Ku70. *Science* 291: 2600–2602, 2001.
- [29326] 2878. Li, G. C.; Ouyang, H.; Li, X.; Nagasawa, H.; Little, J. B.; Chen, D. J.; Ling, C. C.; Fuks, Z.; Cordon–Cardo, C.: Ku70: a candidate tumor suppressor gene for murine T cell lymphoma. *Molec. Cell* 2:1–8, 1998.
- [29327] 2879. McBride, O. W.; Chan, J. Y. C.; Notkins, A. L.; Kohn, L. D.; Lerman, M.: The TSH receptor gene is located on human chromosome 22 and homologous sequences are present on chromosomes 1q, 8, 10, and Xq. (Abstract) *Am. J. Hum. Genet.* 41: A177, 1987.
- [29328] 2880. Mitchell, A. L.; Bale, A. E.; Chan, J.; Kohn, L.; Gonzalez, F.; McBride, O. W.: Localization of TSHR gene and cytochrome p450 IID subfamily on chromosome 22 by linkage analysis. (Abstract) *Cytogenet. Cell Genet.* 51: 1045, 1989.
- [29329] 2881. Reeves, W. H.; Sthoeger, Z. M.: Molecular cloning of cDNA encoding the p70 (Ku) lupus autoantigen. *J. Biol. Chem.* 264: 5047–5052, 1989.
- [29330] 2882. Takata, M.; Sasaki, M. S.; Sonoda, E.; Morrison, C.;

Hashimoto,M.; Utsumi, H.; Yamaguchi-Iwai, Y.; Shinohara, A.; Takeda, S.: Homologous recombination and non-homologous end-joining pathways of DNA double-strandbreak repair have overlapping roles in the maintenance of chromosomal integrity in vertebrate cells. *EMBO J.* 17: 5497–5508, 1998.

[29331] 2883.Takiguchi, Y.; Kurimasa, A.; Chen, F.; Pardington, P. E.; Kuriyama,T.; Okinaka, R. T.; Moyzis, R.; Chen, D. J.: Genomic structure and chromosomal assignment of the mouse Ku70 gene. *Genomics* 35: 129–135,1996.

[29332] 2884.Tuteja, N.; Tuteja, R.; Ochem, A.; Taneja, P.; Huang, N. W.; Simoncsits,A.; Susic, S.; Rahman, K.; Marusic, L.; Chen, J.; Zhang, J.; Wang,S.; Pongor, S.; Falaschi, A.: Human DNA helicase II: a novel DNAunwinding enzyme identified as the Ku autoantigen. *EMBO J.* 13: 4991–5001,1994.

[29333] 2885.Walker, J. R.; Corpina, R. A.; Goldberg, J.: Structure of theKu heterodimer bound to DNA and its implications for double-strandbreak repair. *Nature* 412: 607–614, 2001.

[29334] 2886.Baer, A. N.; Woosley, R. L.; Pincus, T.: Further evidence forthe lack of association between acetylator phenotype and systemiclupus erythematosus. *Arthritis*

Rheum. 29: 508–514, 1986.

- [29335] 2887. Esteller, M.; Garcia-Foncillas, J.; Andion, E.; Goodman, S. N.; Hidalgo, O. F.; Vanaclocha, V.; Baylin, S. B.; Herman, J. G.: Inactivation of the DNA-repair gene MGMT and the clinical response of gliomas to alkylating agents. *New Eng. J. Med.* 343: 1350–1354, 2000.
- [29336] 2888. Gardner, E.; Rydberg, B.; Karran, P.; Ponder, B. A. J.: Localization of the human O(6)-methylguanine-DNA methyltransferase gene to chromosome 10q24.33-qter. *Genomics* 11: 475–476, 1991.
- [29337] 2889. Natarajan, A. T.; Vermeulen, S.; Darroudi, F.; Valentine, M. B.; Brent, T. P.; Mitra, S.; Tano, K.: Chromosomal localization of human O(6)-methylguanine-DNA methyltransferase (MGMT) gene by in situ hybridization. *Mutagenesis* 7:83–85, 1992.
- [29338] 2890. Rydberg, B.; Spurr, N.; Karran, P.: cDNA cloning and chromosomal assignment of the human O(6)-methylguanine-DNA methyltransferase: cDNA expression in *Escherichia coli* and gene expression in human cells. *J. Biol. Chem.* 265: 9563–9569, 1990.
- [29339] 2891. Tano, K.; Shiota, S.; Collier, J.; Foote, R. S.; Mitra, S.: Isolation and structural characterization of a cDNA clone encoding the human DNA repair protein for

O(6)-alkylguanine. Proc. Nat. Acad. Sci. 87:686-690, 1990.

[29340] 2892. Zunino, A.; Arena, G.; Rossi, O.; Archidiacono, N.; Rocchi, M.; Romeo, G.; Abbondandolo, A.: Chromosomal assignment of human O(6)-methylguanine-DNA-methyltransferase gene by hamster-human somatic cell hybrids. Mutagenesis 6: 395-397, 1991.

[29341] 2893. Van Someren, H.; Westerveld, A.; Hagemeijer, A.; Mees, J. R.; Meera Khan, P.; Zaalberg, O. B.: Human antigen and enzyme markers in man-Chinese hamster somatic cell hybrids: evidence for synteny between the HL-A, PGM-3, ME-1, and IPO-B loci. Proc. Nat. Acad. Sci. 71:962-965, 1974.

[29342] 2894. Yoshida, H.; Mitarai, T.; Kawamura, T.; Kitajima, T.; Miyazaki, Y.; Nagasawa, R.; Kawaguchi, Y.; Kubo, H.; Ichikawa, I.; Sakai, O.: Role of the deletion polymorphism of the angiotensin converting enzyme gene in the progression and therapeutic responsiveness of IgA nephropathy. J. Clin. Invest. 96: 2162-2169, 1995.

[29343] 2895. Zheng, F.; Kundu, G. C.; Zhang, Z.; Ward, J.; DeMayo, F.; Mukherjee, A. B.: Uteroglobin is essential in preventing immunoglobulin A nephropathy in mice. Nature Med. 5:

1018–1025, 1999.

[29344] 2896. Anisowicz, A.; Zajchowski, D.; Stenman, G.; Sager, R.: Functional diversity of GRO gene expression in human fibroblasts and mammary epithelial cells. *Proc. Nat. Acad. Sci.* 85: 9645–9649, 1988.

[29345] 2897. Horuk, R.; Yansura, D. G.; Reilly, D.; Spencer, S.; Bourell, J.; Henzel, W.; Rice, G.; Unemori, E.: Purification, receptor binding analysis, and biological characterization of human melanoma growth stimulating activity (MGSA): evidence for a novel MGSA receptor. *J. Biol. Chem.* 268: 541–546, 1993.

[29346] 2898. Richmond, A.; Balentien, E.; Thomas, H. G.; Flaggs, G.; Barton, D. E.; Spiess, J.; Bordoni, R.; Francke, U.; Derynck, R.: Molecular characterization and chromosomal mapping of melanoma growth stimulatory activity, a growth factor structurally related to beta-thromboglobulin. *EMBO J.* 7: 2025–2033, 1988.

[29347] 2899. Sakaguchi, A. Y.; Lalley, P. A.; Ghosh Choudhury, G.; Martinez, L.; Han, E. S.; Killary, A. M.; Naylor, S. L.; Wang, L.-M.: Mouse melanoma growth stimulatory activity gene (Mgsa) is polymorphic and syntenic with the W, Patch, Rumpwhite, and recessive spotting loci on chromosome 5. *Genomics* 5: 629–632, 1989.

- [29348] 2900.Chen, S.-H.; Anderson, J. E.; Giblett, E. R.: Human red cell 2,3-diphosphoglyceratemutase and monophosphoglycerate mutase: genetic evidence for two separate-loci. *Am. J. Hum. Genet.* 29: 405-407, 1977.
- [29349] 2901.Daikh, D. I.; Douglass, J. O.; Adelman, J. P.: Structure and expressionof the human motilin gene. *DNA* 8: 615-621, 1989.
- [29350] 2902.Fan, Y.-S.; Eddy, R. L.; Byers, M. G.; Haley, L. L.; Henry, W.M.; Yano, H.; Seino, Y.; Fujita, J.; Yamada, Y.; Inagaki, N.; Takeda,J.; Imura, H.; Bell, G. I.; Shows, T. B.: Localization of the humanmotilin gene (MLN) to chromosome 6p21.2. (Abstract) *Cytogenet. CellGenet.* 51: 977 only, 1989.
- [29351] 2903.Feighner, S. D.; Tan, C. P.; McKee, K. K.; Palyha, O. C.; Hreniuk,D. L.; Pong, S.-S.; Austin, C. P.; Figueroa, D.; MacNeil, D.; Cascieri,M. A.; Nargund, R.; Bakshi, R.; Abramovitz, M.; Stocco, R.; Kargman,S.; O'Neill, G.; Van Der Ploeg, L. H. T.; Evans, J.; Patchett, A.A.; Smith, R. G.; Howard, A. D.: Receptor for motilin identifiedin the human gastrointestinal system. *Science* 284: 2184-2188, 1999.
- [29352] 2904.Gasparini, P.; Grifa, A.; Savasta, S.; Merlo, I.; Bisceglia, L.;Totaro, A.; Zelante, L.: The motilin gene: subregional localisation,tissue expression, DNA polymorphisms

and exclusion as a candidate gene for the HLA-associated
immotile cilia syndrome. *Hum. Genet.* 94:671–674, 1994.

[29353] 2905. Strissel, P. L.; Strick, R.; Tomek, R. J.; Roe, B. A.;
Rowley, J. D.; Zeleznik-Le, N. J.: DNA structural properties
of AF9 are similar to MLL and could act as recombination
hot spots resulting in MLL/AF9 translocations and leuke-
mogenesis. *Hum. Molec. Genet.* 9: 1671–1679, 2000.

[29354] 2906. Isnard, P.; Depetris, D.; Mattei, M.-G.; Ferrier, P.;
Djabali, M.: cDNA cloning, expression and chromosomal
localization of the murine AF-4 gene involved in human
leukemia. *Mammalian Genome* 9:1065–1068, 1998.

[29355] 2907. Lovett, B. D.; Lo Nigro, L.; Rappaport, E. F.; Blair, I.
A.; Osheroff, N.; Zheng, N.; Megonigal, M. D.; Williams, W.
R.; Nowell, P. C.; Felix, C. A.: Near-precise interchromoso-
mal recombination and functional DNA topoisomerase II
cleavage sites at MLL and AF-4 genomic breakpoints in
treatment-related acute lymphoblastic leukemia with
t(4;11) translocation. *Proc. Nat. Acad. Sci.* 98: 9802–9807,
2001.

[29356] 2908. Uckun, F. M.; Herman-Hatten, K.; Crotty, M.-L.;
Sensel, M. G.; Sather, H. N.; Tuel-Ahlgren, L.; Sarquis, M.
B.; Bostrom, B.; Nachman, J. B.; Steinherz, P. G.; Gaynon, P.
S.; Heerema, N.: Clinical significance of MLL-AF4 fusion

transcript expression in the absence of a cytogenetically-detectable t(4;11)(q21;q23) chromosomal translocation. Blood 92:810–821, 1998.

- [29357] 2909. Prasad, R.; Gu, Y.; Alder, H.; Nakamura, T.; Canaani, O.; Saito, H.; Huebner, K.; Gale, R. P.; Nowell, P. C.; Kuriyama, K.; Miyazaki, Y.; Croce, C. M.; Canaani, E.: Cloning of the ALL-1 fusion partner, the AF-6 gene, involved in acute myeloid leukemias with the t(6;11) chromosome translocation. Cancer Res. 53: 5624–5628, 1993.
- [29358] 2910. Saha, V.; Lillington, D. M.; Shelling, A. N.; Chaplin, T.; Yaspo, M.-L.; Ganesan, T. S.; Young, B. D.: AF6 gene on chromosome band 6q27 maps distal to the minimal region of deletion in epithelial ovarian cancer. Genes Chromosomes Cancer 14: 220–222, 1995.
- [29359] 2911. Gerling, I. C.; Karlsen, A. E.; Chapman, H. D.; Andersen, H. U.; Boel, E.; Cunningham, J. M.; Nerup, J.; Leiter, E. H.: The inducible nitric oxide synthase gene, Nos2, maps to mouse chromosome 11. Mammalian Genome 5: 318–320, 1994.
- [29360] 2912. Van Cong, N.; Moullec, J.: Linkage probable entre les groupes de phosphatase acide des globules rouges et le système Lewis. Ann. Genet. 14: 121–125, 1971.

- [29361] 2913.Wakita, Y.; Narahara, K.; Takahashi, Y.; Kikkawa, K.; Kimura,S.; Oda, M.; Kimoto, H.: Duplication of 2p25: confirmation of theassignment of soluble acid phosphatase (ACP1) locus to 2p25. Hum.Genet. 71: 259–260, 1985.
- [29362] 2914.Weitkamp, L. R.; Janzen, M. K.; Guttormsen, S. A.; Gershowitz,H.: Inherited pericentric inversion of chromosome number two: a linkagestudy. Ann. Hum. Genet. 33: 53–59, 1969.
- [29363] 2915.Weitkamp, L. R.; Lovrien, E. W.; Olaisen, B.; Fenger, K.; Gedde–Dahl,T., Jr.; Sorensen, S. A.; Conneally, P. M.; Bias, W. B.; Ott, J.:Linkage relations of the loci for the MN blood group and red cellphosphate. Birth Defects Orig. Art. Ser. 11(3): 276–280, 1975. Note:Alternate: Cytogenet. Cell Genet. 14: 446–450, 1975...
- [29364] 2916.Wo, Y.–Y. P.; McCormack, A. L.; Shabanowitz, J.; Hunt, D. F.;Davis, J. P.; Mitchell, G. L.; Van Etten, R. L.: Sequencing, cloning,and expression of human red cell–type acid phosphatase, a cytoplasmicphosphotyrosyl protein phosphatase. J. Biol. Chem. 267: 10856–10865,1992.
- [29365] 2917.Yoshihara, C. M.; Mohrenweiser, H. W.: Characterization of ACP1(TIC–1),an electrophoretic variant of erythrocyte acid phosphatase restrictedto the Ticuna Indians of Central Amazonas. Am. J. Hum. Genet. 32:898–907,

1980.

- [29366] 2918.Griffiths, L. R.: Authors response to 'Chromosome 17 and the inducible nitric oxide synthase gene in human essential hypertension' by Rutherford et al., Human Genetics, published on-line September 2001. (Letter) Hum.Genet. 110: 100–103, 2002.
- [29367] 2919.Jenkins, N. A.; Rothe, H.; Gilbert, D. J.; Copeland, N. G.; Kolb, H.: Mapping of the gene for inducible nitric oxide (NO) synthase of mouse macrophages to chromosome 11, close to Evi-2, nu, and Idd-4. Genomics 19:402–404, 1994.
- [29368] 2920.Johannesen, J.; Pie, A.; Pociot, F.; Kristiansen, O. P.; Karlsen, A. E.; Nerup, J. The Danish Study Group of Diabetes in Childhood; The Danish Insulin-Dependent Diabetes Mellitus Epidemiology and Genetics Group: Linkage of the human inducible nitric oxide synthase gene to type 1 diabetes. J. Clin. Endocr. Metab. 86: 2792–2796, 2001.
- [29369] 2921.Lowenstein, C. J.; Glatt, C. S.; Bredt, D. S.; Snyder, S. H.: Cloned and expressed macrophage nitric oxide synthase contrasts with the brain enzyme. Proc. Nat. Acad. Sci. 89: 6711–6715, 1992.
- [29370] 2922.Marsden, P. A.; Heng, H. H. Q.; Duff, C. L.; Shi, X.-M.; Tsui, L.-C.; Hall, A. V.: Localization of the human

gene for inducible nitric oxide synthase (NOS2) to chromosome 17q11.2–q12. *Genomics* 19:183–185, 1994.

[29371] 2923. Mehrabian, M.; Xia, Y.-R.; Wen, P.-Z.; Warden, C. H.; Herschman, H. R.; Luscis, A. J.: Localization of murine macrophage inducible nitric oxide synthase to mouse chromosome 11. *Genomics* 22: 646–647, 1994.

[29372] 2924. Morris, B. J.: Critique of 'Chromosome 17 and the inducible nitric oxide synthase gene in human essential hypertension' by Rutherford et al., *Human Genetics*, published online September 2001. (Letter) *Hum. Genet.* 110: 98–99, 2002.

[29373] 2925. Nathan, C.; Shiloh, M. U.: Reactive oxygen and nitrogen intermediates in the relationship between mammalian hosts and microbial pathogens. *Proc. Nat. Acad. Sci.* 97: 8841–8848, 2000.

[29374] 2926. Nicholson, S.; Bonecini-Almeida Mda, G; Lapa e Silva, J. R.; Nathan, C.; Xie, Q. W.; Mumford, R.; Weidner, J. R.; Calaycay, J.; Geng, J.; Boechat, N.; et al.; et al.: Inducible nitric oxide synthase in pulmonary alveolar macrophages from patients with tuberculosis. *J. Exp. Med.* 183:2293–2302, 1996.

[29375] 2927. Nozaki, Y.; Hasegawa, Y.; Ichiyama, S.; Nakashima, I.; Shimokata, K.: Mechanism of nitric oxide-dependent

killing of *Mycobacterium bovis* BCG in human alveolar macrophages. *Infect. Immun.* 65: 3644–3647, 1997.

- [29376] 2928. Vouldoukis, I.; Riveros–Moreno, V.; Dugas, B.; Ouaz, F.; Becherel, P.; Debre, P.; Moncada, S.; Mossalayi, M. D.: The killing of *Leishmania major* by human macrophages is mediated by nitric oxide induced after ligation of the Fc-epsilon-RII/CD23 surface antigen. *Proc. Nat. Acad. Sci.* 92: 7804–7808, 1995.
- [29377] 2929. Xu, W.; Charles, I. G.; Liu, L.; Moncada, S.; Emson, P.: Molecular cloning and structural organization of the human inducible nitric oxide synthase gene (NOS2). *Biochem. Biophys. Res. Commun.* 219: 784–788, 1996.
- [29378] 2930. Xu, W.; Charles, I. G.; Moncada, S.; Gorman, P.; Sheer, D.; Liu, L.; Emson, P.: Mapping of the genes encoding human inducible and endothelial nitric oxide synthase (NOS2 and NOS3) to the pericentric region of chromosome 17 and to chromosome 7, respectively. *Genomics* 21: 419–422, 1994.
- [29379] 2931. Bredt, D. S.; Hwang, P. M.; Glatt, C. E.; Lowenstein, C.; Reed, R. R.; Snyder, S. H.: Cloned and expressed nitric oxide synthase structurally resembles cytochrome P-450 reductase. *Nature* 351: 714–718, 1991.
- [29380] 2932. Brenman, J. E.; Chao, D. S.; Xia, H.; Aldape, K.; Bredt,

D. S.: Nitric oxide synthase complexed with dystrophin and absent from skeletal muscle sarcolemma in Duchenne muscular dystrophy. *Cell* 82:743–752, 1995.

[29381] 2933. Burnett, A. L.; Lowenstein, C. J.; Bredt, D. S.; Chang, T. S. K.; Snyder, S. H.: Nitric oxide: a physiologic mediator of penile erection. *Science* 257:401–403, 1992.

[29382] 2934. Day, B. J.; Patel, M.; Calavetta, L.; Chang, L.-Y.; Stamler, J. S.: A mechanism of paraquat toxicity involving nitric oxide synthase. *Proc. Nat. Acad. Sci.* 96: 12760–12765, 1999.

[29383] 2935. Deans, Z.; Dawson, S. J.; Xie, J.; Young, A. P.; Wallace, D.; Latchman, D. S.: Differential regulation of the two neuronal nitric-oxide synthase gene promoters by the Oct-2 transcription factor. *J. Biol. Chem.* 271:32153–32158, 1996.

[29384] 2936. Gu, Z.; Kaul, M.; Yan, B.; Kridel, S. J.; Cui, J.; Strongin, A.; Smith, J. W.; Liddington, R. C.; Lipton, S. A.: S-nitrosylation of matrix metalloproteinases: signaling pathway to neuronal cell death. *Science* 297: 1186–1190, 2002.

[29385] 2937. Kharazia, V. N.; Schmidt, H. H. H. W.; Weinberg, R. J.: Type I nitric oxide synthase fully accounts for NADPH-diaphorase in rat striatum, but not cortex. *Neuroscience*

62: 983–987, 1994.

- [29386] 2938.Kishimoto, J.; Spurr, N.; Liao, M.; Lizhi, L.; Emson, P.; Xu, W.: Localization of brain nitric oxide synthase (NOS) to human chromosome12. *Genomics* 14: 802–804, 1992.
- [29387] 2939.Kuo, R. C.; Baxter, G. T.; Thompson, S. H.; Stricker, S. A.; Patton,C.; Bonaventura, J.; Epel, D.: NO is necessary and sufficient foregg activation at fertilization. *Nature* 406: 633–636, 2000.
- [29388] 2940.Lee, C. G. L.; Gregg, A. R.; O'Brien, W. E.: Localiza-tion ofthe neuronal form of nitric oxide synthase to mouse chromosome 5. *MammalianGenome* 6: 56–57, 1995.
- [29389] 2941.Magee, T.; Fuentes, A. M.; Garban, H.; Rajavashisth, T.; Marquez,D.; Rodriguez, J. A.; Rajfer, J.; Gonzalez–Ca-david, N. F.: Cloningof a novel neuronal nitric oxide syn-thase expressed in penis and lowerurinary tract. *Biochem. Biophys. Res. Commun.* 226: 145–151, 1996.
- [29390] 2942.Devlin, R.; Henderson, H.; Monsalve, V.; Brunzell, J.; Deeb, S.;Hayden, M. R.: The molecular biology of hyper-triglyceridemia: characterizationof mutations in patients with lipoprotein lipase deficiency.(Abstract) *Am.J. Hum. Genet.* 45 (suppl.): A4, 1989.
- [29391] 2943.Craigen, W. J.: Leigh disease with deficiency of

lipoamide dehydrogenase:treatment failure with dichloroacetate. *Pediat. Neurol.* 14: 69–71,1996.

- [29392] 2944.Chynn, E. W.; Walton, D. S.; Hahn, L. B.; Dryja, T. P.: Norriedisease: diagnosis of a simplex case by DNA analysis. *Arch. Ophthal.* 114:1136–1138, 1996.
- [29393] 2945.Thomas, S. A.; Palmiter, R. D.: Impaired maternal behavior in mice lacking norepinephrine and epinephrine. *Cell* 91: 583–592, 1997.
- [29394] 2946.Thomas, S. A.; Palmiter, R. D.: Thermoregulatory and metabolic phenotypes of mice lacking noradrenaline and adrenaline. *Nature* 387:94–97, 1997.
- [29395] 2947.Weinshilboum, R. M.: Catecholamine biochemical genetics in human populations. In: Breakefield, X. O.: *Neurogenetics: Genetic Approaches to the Nervous System*. New York: Elsevier/North Holland (pub.) 1979. Pp. 257–282.
- [29396] 2948.Weinshilboum, R. M.; Schrott, H. G.; Raymond, F. A.; Weidman, W. H.; Elveback, L. R.: Inheritance of very low serum dopamine-beta-hydroxylase activity. *Am. J. Hum. Genet.* 27: 573–585, 1975.
- [29397] 2949.Wilson, A. F.; Elston, R. C.; Siervogel, R. M.; Tran, L. D.: Linkage of a gene regulating dopamine-beta-hydroxylase activity and the ABO blood group locus.

Am. J. Hum. Genet. 42: 160–166, 1988.

- [29398] 2950. Wilson, A. F.; Elston, R. C.; Siervogel, R. M.; Tran, L. D.: Linkage of a gene regulating dopamine-beta-hydroxylase activity and the ABO blood group locus. (Abstract) Am. J. Hum. Genet. 41: A191 only, 1987.
- [29399] 2951. Zabetian, C. P.; Anderson, G. M.; Buxbaum, S. G.; Elston, R. C.; Ichinose, H.; Nagatsu, T.; Kim, K.-S.; Kim, C.-H.; Malison, R. T.; Gelernter, J.; Cubells, J. F.: A quantitative-trait analysis of human plasma-dopamine beta-hydroxylase activity: evidence for a major functional polymorphism at the DBH locus. Am. J. Hum. Genet. 68: 515–522, 2001.
- [29400] 2952. Monros, E.; Molto, M. D.; Martinez, F.; Canizares, J.; Blanca, J.; Vilchez, J. J.; Prieto, F.; de Frutos, R.; Palau, F.: Phenotype correlation and intergenerational dynamics of the Friedreich ataxia GAA trinucleotide repeat. Am. J. Hum. Genet. 61: 101–110, 1997.
- [29401] 2953. Montermini, L.; Andermann, E.; Labuda, M.; Richter, A.; Pandolfo, M.; Cavalcanti, F.; Pianese, L.; Iodice, L.; Farina, G.; Monticelli, A.; Turano, M.; Filla, A.; De Michele, G.; Coccozza, S.: The Friedreich ataxia GAA triplet repeat: pre-mutation and normal alleles. Hum. Molec. Genet. 6: 1261–1266, 1997.

- [29402] 2954.Ohshima, K.; Montermini, L.; Wells, R. D.; Pandolfo, M.: Inhibitory effects of expanded GAA–TTC triplet repeats from intron I of the Friedreich ataxia gene on transcription and replication in vivo. *J. Biol. Chem.* 273:14588–14595, 1998.
- [29403] 2955.Pianese, L.; Cavalcanti, F.; De Michele, G.; Filla, A.; Campanella, G.; Calabrese, O.; Castaldo, I.; Monticelli, A.; Cocozza, S.: The effect of parental gender on the GAA dynamic mutation in the FRDA gene. (Letter) *Am. J. Hum. Genet.* 60: 460–463, 1997.
- [29404] 2956.Puccio, H.; Koenig, M.: Recent advances in the molecular pathogenesis of Friedreich ataxia. *Hum. Molec. Genet.* 9: 887–892, 2000.
- [29405] 2957.Robinson, B. H.; Sherwood, W. G.; Kahler, S.; O'Flynn, M. E.; Nadler, H.: Lipoamide dehydrogenase deficiency. (Letter) *New Eng. J. Med.* 304: 53–54, 1981.
- [29406] 2958.Rodius, F.; Duclos, F.; Wrogemann, K.; Le Paslier, D.; Ougen, P.; Billault, A.; Belal, S.; Musenger, C.; Brice, A.; Durr, A.; Mignard, C.; Sirugo, G.; Weissenbach, J.; Cohen, D.; Hentati, F.; Ben Hamida, M.; Mandel, J.–L.; Koenig, M.: Recombinations in individuals homozygous by descent localize the Friedreich ataxia locus in a cloned 450–kb interval. *Am. J. Hum. Genet.* 54: 1050–1059, 1994.

- [29407] 2959. Macdonald, D.; Aguiar, R. C.; Mason, P. J.; Goldman, J. M.; Cross, N. C.: A new myeloproliferative disorder associated with chromosomal translocations involving 8p11: a review. *Leukemia* 9: 1628–1630, 1995.
- [29408] 2960. Popovici, C.; Adelaide, J.; Ollendorff, V.; Chaffanet, M.; Guasch, G.; Jacrot, M.; Leroux, D.; Birnbaum, D.; Pebusque, M.-J.: Fibroblast growth factor receptor 1 is fused to FIM in stem-cell myeloproliferative disorder with t(8;13)(p12;q12). *Proc. Nat. Acad. Sci.* 95: 5712–5717, 1998.
- [29409] 2961. Popovici, C.; Zhang, B.; Gregoire, M.-J.; Jonveaux, P.; Lafage-Pochitaloff, M.; Birnbaum, D.; Pebusque, M.-J.: The t(6;8)(q27;p11) translocation in a stem cell myeloproliferative disorder fuses a novel gene, FOP, to fibroblast growth factor receptor 1. *Blood* 93: 1381–1389, 1999.
- [29410] 2962. Thomas, W.; Rubenstein, M.; Goto, M.; Drayna, D.: A genetic analysis of the Werner syndrome region on human chromosome 8p. *Genomics* 16: 685–690, 1993.
- [29411] 2963. Wood, S.; Schertzer, M.; Yaremko, M. L.: Sequence identity locates CEBPD and FGFR1 to mapped human loci within proximal 8p. *Cytogenet. Cell Genet.* 70: 188–191, 1995.
- [29412] 2964. Xiao, S.; Nalabolu, S. R.; Aster, J. C.; Ma, J.; Abruzzo,

L.;Jaffe, E. S.; Stone, R.; Weissman, S. M.; Hudson, T. J.; Fletcher,J. A.: FGFR1 is fused with a novel zinc-finger gene, ZNF198, in thet(8;13) leukaemia/lymphoma syndrome. *Nature Genet.* 18: 84–87, 1998.

[29413] 2965.Andersson, S. M.; Pispá, J. P.: Purification and properties of human liver tyrosine aminotransferase. *Clin. Chim. Acta* 125: 117–123,1982.

[29414] 2966.Barton, D. E.; Yang–Feng, T. L.; Francke, U.: The human tyrosineaminotransferase gene mapped to the long arm of chromosome 16 (region16q22–q24) by somatic cell hybrid analysis and in situ hybridization.*Hum. Genet.* 72: 221–224, 1986.

[29415] 2967.Chen, S.–H.; Giblett, E. R.: Genetic variation of soluble glutamic–oxaloacetictransaminase in man. *Am. J. Hum. Genet.* 23: 419–424, 1971.

[29416] 2968.Craig, I. W.; Tolley, E.; Bobrow, M.; van Heyningen, V.: Assignmentof a gene necessary for the expression of mitochondrial glutamic–oxaloacetictransaminase in human–mouse hybrid cells. *Cytogenet. Cell Genet.* 22:190–194, 1978.

[29417] 2969.Davidson, R. G.; Cortner, J. A.; Rattazzi, M. C.; Ruddle, F. H.;Lubs, H. A.: Genetic polymorphisms of human mitochondrial glutamicoxaloacetic transaminase. *Science*

169: 391–392, 1970.

- [29418] 1970. DeLorenzo, R. J.; Ruddle, F. H.: Glutamate transaminase (GOT) genetics in *mus musculus*: linkage, polymorphism, and phenotypes of the GOT-2 and GOT-1 loci. *Biochem. Genet.* 4: 259–273, 1970.
- [29419] 1971. Francke, U.; Weitkamp, L. R.: Report of the committee on the genetic constitution of chromosome 6. *Cytogenet. Cell Genet.* 25: 32–38, 1979.
- [29420] 1972. Jeremiah, S. J.; Povey, S.; Burley, M. W.; Kielty, C.; Lee, M.; Spowart, G.; Corney, G.; Cook, P. J. L.: Mapping studies on human mitochondrial glutamate oxaloacetate transaminase. *Ann. Hum. Genet.* 46: 145–152, 1982.
- [29421] 1973. Pol, S.; Bousquet-Lemerrier, B.; Pave-Preux, M.; Bulle, F.; Passage, E.; Hanoune, J.; Mattei, M. G.; Barouki, R.: Chromosomal localization of human aspartate aminotransferase genes by *in situ* hybridization. *Hum. Genet.* 83: 159–164, 1989.
- [29422] 1974. Pol, S.; Bousquet-Lemerrier, B.; Pave-Preux, M.; Pawlak, A.; Nalpas, B.; Berthelot, P.; Hanoune, J.; Barouki, R.: Nucleotide sequence and tissue distribution of the human mitochondrial aspartate aminotransferase mRNA. *Biochem. Biophys. Res. Commun.* 157: 1309–1315, 1988.
- [29423] 1975. Tolley, E.; van Heyningen, V.; Brown, R.; Bobrow, M.;

Craig, I.W.: Assignment to chromosome 16 of a gene necessary for the expression of human mitochondrial glutamate oxaloacetate transaminase (aspartate aminotransferase) (E.C. 2.6.1.1). *Biochem. Genet.* 18: 947–954, 1980.

[29424] 1976. Toyomasu, T.; Sakakibara, S.; Kagamiyama, H.; Matsumoto, H.: Genetic polymorphism of mitochondrial glutamate–oxaloacetate transaminase in Japanese. *Hum. Genet.* 66: 90–91, 1984.

[29425] 1977. Tsuji, S.; Kobayashi, H.; Uchida, Y.; Ihara, Y.; Miyatake, T.: Molecular cloning of human growth inhibitory factor cDNA and its down-regulation in Alzheimer's disease. *EMBO J.* 11: 4843–4850, 1992.

[29426] 1978. Uchida, Y.; Takio, K.; Titani, K.; Ihara, Y.; Tomonaga, M.: The growth inhibitory factor that is deficient in the Alzheimer's disease brain is a 68 amino acid metallothionein-like protein. *Neuron* 7:337–347, 1991.

[29427] 1979. Hoshino, S.; Miyazawa, H.; Enomoto, T.; Hanaoka, F.; Kikuchi, Y.; Kikuchi, A.; Ui, M.: A human homologue of the yeast GST1 gene codes for a GTP-binding protein and is expressed in a proliferation-dependent manner in mammalian cells. *EMBO J.* 8: 3807–3814, 1989.

[29428] 1980. Kikuchi, Y.; Shimatake, H.; Kikuchi, A.: A yeast gene

required for the G1-to-S transition encodes a protein containing an A-kinase target site and GTPase domain. *EMBO J.* 7: 1175–1182, 1988.

- [29429] 2981. Ozawa, K.; Murakami, Y.; Eki, T.; Yokoyama, K.; Soeda, E.; Hoshino, S.; Ui, M.; Hanaoka, F.: Mapping of the human GSPT1 gene, a human homolog of the yeast GST1 gene, to chromosomal band 16p13.1. *Somat. Cell Molec. Genet.* 18: 189–194, 1992.
- [29430] 2982. Harris, H.; Hopkinson, D. A.; Robson, E. B.: The incidence of rare alleles determining electrophoretic variants: data on 43 enzyme loci in man. *Ann. Hum. Genet.* 37: 237–253, 1974.
- [29431] 2983. Beutler, E.; Gelbart, T.; Kuhl, W.: Human red cell glucose-6-phosphate dehydrogenase: all active enzyme has sequence predicted by the X chromosome-encoded cDNA. *Cell* 62: 7–9, 1990.
- [29432] 2984. Henikoff, S.; Smith, J. M.: The human mRNA that provides the N-terminus of chimeric G6PD encodes GMP reductase. *Cell* 58: 1021–1022, 1989.
- [29433] 2985. Kanno, H.; Huang, I.-Y.; Kan, Y. W.; Yoshida, A.: Two structural genes on different chromosomes are required for encoding the major subunit of human red cell glucose-6-phosphate dehydrogenase. *Cell* 58: 595–606, 1989.

- [29434] 1986.Kondoh, T.; Kanno, H.; Chang, L.; Yoshida, A.: Genomic structure and expression of human guanosine monophosphate reductase. Hum.Genet. 88: 219–224, 1991.
- [29435] 1987.Kondoh, T.; Kanno, H.; Chang, L.; Yoshida, A.: Identification of common variant alleles of the human guanosine monophosphate reductase gene. Hum. Genet. 88: 225–227, 1991.
- [29436] 1988.Mason, P. J.; Bautista, J. M.; Vulliamy, T. J.; Turner, N.; Luzzatto, L.: Human red cell glucose–6–phosphate dehydrogenase is encoded only on the X chromosome. Cell 62: 9–10, 1990.
- [29437] 1989.Murano, I.; Tsukahara, M.; Kajii, T.; Yoshida, A.: Mapping of the human guanosine monophosphate reductase gene (GMPR) to chromosome 6p23 by fluorescence in situ hybridization. Genomics 19: 179–180, 1994.
- [29438] 1990.Yoshida, A.; Kan, Y. W.: Origin of 'fused' glucose–6–phosphate dehydrogenase. Cell 62: 11–12, 1990.
- [29439] 1991.Brady, W. A.; Kokoris, M. S.; Fitzgibbon, M.; Black, M. E.: Cloning, characterization, and modeling of mouse and human guanylate kinases. J.Biol. Chem. 271: 16734–16740, 1996.
- [29440] 1992.Dallapiccola, B.; Lungarotti, M. S.; Falorni, A.; Mag-

nani, M.;Dacha, M.: Evidence for the assignment of GUK1 gene locus to 1q32–q43segment from gene dosage effect. Ann. Genet. 23: 83–85, 1980.

[29441] 1993.Kajiwara, K.; Berson, E. L.; Dryja, T. P.: Digenic retinitispigmentosa due to mutations at the unlinked peripherin/RDS and ROM1loci. Science 264: 1604–1608, 1994.

[29442] 1994.Kajiwara, K.; Hahn, L. B.; Mukai, S.; Travis, G. H.; Berson, E.L.; Dryja, T. P.: Mutations in the human retinal degeneration slowgene in autosomal dominant retinitis pigmentosa. Nature 354: 480–483,1991.

[29443] 1995.Kajiwara, K.; Sandberg, M. A.; Berson, E. L.; Dryja, T. P.: Anull mutation in the human RDS/peripherin gene in a family with autosomaldominant retinitis punctata albescens. (Abstract) Am. J. Hum. Genet. 51(suppl.): A6 only, 1992.

[29444] 1996.Kajiwara, K.; Sandberg, M. A.; Berson, E. L.; Dryja, T. P.: Anull mutation in the human peripherin/RDS gene in a family with autosomaldominant retinitis punctata albescens. Nature Genet. 3: 208–212,1993.

[29445] 1997.Kedzierski, W.; Nusinowitz, S.; Birch, D.; Clarke, G.; McInnes,R. R.; Bok, D.; Travis, G. H.: Deficiency of rds/peripherin causesphotoreceptor death in mouse models of

digenic and dominant retinitis pigmentosa. Proc. Nat. Acad. Sci. 98: 7718–7723, 2001.

[29446] 1998. Keen, T. J.; Inglehearn, C. F.: Mutations and polymorphisms in the human peripherin-RDS gene and their involvement in inherited retinal degeneration. Hum. Mutat. 8: 297–303, 1996.

[29447] 1999. Kikawa, E.; Nakazawa, M.; Chida, Y.; Shiono, T.; Tamai, M.: A novel mutation (asn244-to-lys) in the peripherin/RDS gene causing autosomal dominant retinitis pigmentosa associated with bull's-eye maculopathy detected by nonradioisotopic SSCP. Genomics 20: 137–139, 1994.

[29448] 2000. Kim, R. Y.; Dollfus, H.; Keen, T. J.; Fitzke, F. W.; Arden, G. B.; Bhattacharya, S. S.; Bird, A. C.: Autosomal dominant pattern dystrophy of the retina associated with a 4-base pair insertion at codon 140 in the peripherin/RDS gene. Arch. Ophthalmol. 113: 451–455, 1995.

[29449] 2001. Kohl, S.; Christ-Adler, M.; Apfelstedt-Sylla, E.; Kellner, U.; Eckstein, A.; Zrenner, E.; Wissinger, B.: RDS/peripherin gene mutations are frequent causes of central retinal dystrophies. J. Med. Genet. 34: 620–626, 1997.

[29450] 2002. Ma, J.; Norton, J. C.; Allen, A. C.; Burns, J. B.; Hasel, K. W.; Burns, J. L.; Sutcliffe, J. G.; Travis, G. H.: Retinal degeneration slow (rds) in mouse results from simple inser-

tion of a t haplotype–specific element into protein–coding exon II. *Genomics* 28: 212–219, 1995.

- [29451] 3003. Nichols, B. E.; Drack, A. V.; Vandenburg, K.; Kimura, A. E.; Sheffield, V. C.; Stone, E. M.: A 2 base pair deletion in the RDS gene associated with butterfly–shaped pigment dystrophy of the fovea. *Hum. Molec. Genet.* 2: 601–603, 1993.
- [29452] 3004. Nichols, B. E.; Sheffield, V. C.; Vandenburg, K.; Drack, A. V.; Kimura, A. E.; Stone, E. M.: Butterfly–shaped pigment dystrophy of the fovea caused by a point mutation in codon 167 of the RDS gene. *Nature Genet.* 3: 202–207, 1993.
- [29453] 3005. Payne, A. M.; Downes, S. M.; Bessant, D. A. R.; Bird, A. C.; Bhattacharya, S. S.: Founder effect, seen in the British population, of the 172 peripherin/RDS mutation– and further refinement of genetic positioning of the peripherin/RDS gene. (Letter) *Am. J. Hum. Genet.* 192–195, 1998.
- [29454] 3006. Pendleton, J. W.; Violette, S. M.; Hunihan, L. W.; Greene, L. A.; Ruddle, F. H.: The peripherin gene maps to mouse chromosome 15. *Genomics* 9: 369–372, 1991.
- [29455] 3007. Reig, C.; Serra, A.; Gean, E.; Vidal, M.; Arumi, J.; De la Calzada, M. D.; Antich, J.; Carballo, M.: A point mutation

in the RDS-peripheringene in a Spanish family with central areolar choroidal dystrophy. *Ophthalmol.Genet.* 16: 35–44, 1995.

- [29456] 3008.Sarra, G.–M.; Stephens, C.; de Alwis, M.; Bainbridge, J. W. B.;Smith, A. J.; Thrasher, A. J.; Ali, R. R.: Gene replacement therapyin the retinal degeneration slow (rds) mouse: the effect on retinaldegeneration following partial transduction of the retina. *Hum. Molec.Genet.* 10: 2353–2361, 2001.
- [29457] 3009.Travis, G. H.; Brennan, M. B.; Danielson, P. E.; Kozak, C. A.;Sutcliffe, J. G.: Identification of a photoreceptor–specific mRNAencoded by the gene responsible for retinal degeneration slow (rds). *Nature* 338:70–73, 1989.
- [29458] 3010.Travis, G. H.; Christerson, L.; Danielson, P. E.; Klisak, I.;Sparkes, R. S.; Hahn, L. B.; Dryja, T. P.; Sutcliffe, J. G.: Thehuman retinal degeneration slow (RDS) gene: chromosome assignmentand structure of the mRNA. *Genomics* 10: 733–739, 1991.
- [29459] 3011.Travis, G. H.; Hepler, J. E.: A medley of retinal dystrophies. *NatureGenet.* 3: 191–192, 1993.
- [29460] 3012.Cramer, P.; Bushnell, D. A.; Fu, J.; Gnatt, A. L.; Maier–Davis,B.; Thompson, N. E.; Burgess, R. R.; Edwards, A. M.; David, P. R.;Kornberg, R. D.: Architecture of RNA

polymerase II and implications for the transcription mechanism. *Science* 288: 640–648, 2000.

[29461] 3013. Yudkovsky, N.; Ranish, J. A.; Hahn, S.: A transcription reinitiation intermediate that is stabilized by activator. *Nature* 408: 225–229, 2000.

[29462] 3014. Dammann, R.; Pfeifer, G. P.: Cloning and characterization of the human RNA polymerase I subunit hRPA40. *Biochim. Biophys. Acta* 1396: 153–157, 1998.

[29463] 3015. Fanciulli, M.; Bruno, T.; Di Padova, M.; De Angelis, R.; Lovari, S.; Floridi, A.; Passananti, C.: The interacting RNA polymerase II subunits, hRPB11 and hRPB3, are coordinately expressed in adult human tissues and down-regulated by doxorubicin. *FEBS Lett.* 427: 236–240, 1998.

[29464] 3016. Pati, U. K.; Weissman, S. M.: The amino acid sequence of the human RNA polymerase II 33-kDa subunit hRPB 33 is highly conserved among eukaryotes. *J. Biol. Chem.* 265: 8400–8403, 1990.

[29465] 3017. Becker, D. M.; Fikes, J. D.; Guarente, L.: A cDNA encoding a human CCAAT-binding protein cloned by functional complementation in yeast. *Proc. Nat. Acad. Sci.* 88: 1968–1972, 1991.

[29466] 3018. Henrion, A. A.; Martinez, A.; Mattei, M.-G.; Kahn, A.; Raymondjean, M.: Structure, sequence, and chromosomal

location of the gene for USF2 transcription factors in mouse. *Genomics* 25: 36–43, 1995.

- [29467] 3019. Gharani, N.; Waterworth, D. M.; Batty, S.; White, D.; Gilling-Smith, C.; Conway, G. S.; McCarthy, M.; Franks, S.; Williamson, R.: Association of the steroid synthesis gene CYP11a with polycystic ovary syndrome and hyperandrogenism. *Hum. Molec. Genet.* 6: 397–402, 1997.
- [29468] 3020. Jakimiuk, A. J.; Weitsman, S. R.; Magoffin, D. A.: 5- α -reductase activity in women with polycystic ovary syndrome. *J. Clin. Endocr. Metab.* 84: 2414–2418, 1999.
- [29469] 3021. Kuttann, F.; Couillin, P.; Girard, F.; Billaud, L.; Vincens, M.; Boucekkine, C.; Thalabard, J.-C.; Maudelonde, T.; Spritzer, P.; Mowszowicz, I.; Boue, A.; Mauvais-Jarvis, P.: Late-onset adrenal hyperplasia in hirsutism. *New Eng. J. Med.* 313: 224–231, 1985.
- [29470] 3022. Kuttann, F.; Mowszowicz, I.; Schaison, G.; Mauvais-Jarvis, P.: Androgen production and skin metabolism in hirsutism. *J. Endocr.* 75: 83–91, 1977.
- [29471] 3023. Kurtz, T. W.: The ACE of hearts. *Nature* 359: 588–589, 1992.
- [29472] 3024. Lindpaintner, K.; Lee, M.; Larson, M. G.; Rao, V. S.; Pfeiffer, M. A.; Ordovas, J. M.; Schaefer, E. J.; Wilson, A. F.; Wilson, P. W. F.; Vasan, R. S.; Myers, R. H.; Levy, D.: Ab-

sence of association or genetic linkage between the angiotensin-converting-enzyme gene and left ventricular mass. *New Eng. J. Med.* 334: 1023–1028, 1996.

[29473] 3025. Lindpaintner, K.; Pfeffer, M. A.; Kreutz, R.; Stampfer, M. J.; Grodstein, F.; LaMotte, F.; Buring, J.; Hennekens, C. H.: A prospective evaluation of an angiotensin-converting-enzyme gene polymorphism and the risk of ischemic heart disease. *New Eng. J. Med.* 332: 706–711, 1995.

[29474] 3026. Marre, M.; Bernadet, P.; Gallois, Y.; Savagner, F.; Guyene, T.–T.; Hallab, M.; Cambien, F.; Passa, P.; Alhenc-Gelas, F.: Relationships between angiotensin I converting enzyme gene polymorphism, plasma levels, and diabetic retinal and renal complications. *Diabetes* 43:384–388, 1994.

[29475] 3027. Marre, M.; Jeunemaitre, X.; Gallois, Y.; Rodier, M.; Chatellier, G.; Sert, C.; Dusselier, L.; Kahal, Z.; Chaillous, L.; Halimi, S.; Muller, A.; Sackmann, H.; Bauduceau, B.; Bled, F.; Passa, P.; Alhenc-Gelas, F.: Contribution of genetic polymorphism in the renin-angiotensin system to the development of renal complications in insulin-dependent diabetes. *J. Clin. Invest.* 99: 1585–1595, 1997.

[29476] 3028. Mattei, M.–G.; Hubert, C.; Alhenc-Gelas, F.; Roeckel, N.; Corvol, P.; Soubrier, F.: Angiotensin-I converting en-

zyme gene is on chromosome 17. (Abstract) Cytogenet. Cell Genet. 51: 1041, 1989.

- [29477] 3029. Montgomery, H. E.; Marshall, R.; Hemingway, H.; Myerson, S.; Clarkson, P.; Dollery, C.; Hayward, M.; Holliman, D. E.; Jubb, M.; World, M.; Thomas, E. L.; Brynes, A. E.; Saeed, N.; Barnard, M.; Bell, J. D.; Prasad, K.; Rayson, M.; Talmud, P. J.; Humphries, S. E.: Human gene for physical performance. (Letter) Nature 393: 221–222, 1998.
- [29478] 3030. Ohishi, M.; Fujii, K.; Minamino, T.; Hagaki, J.; Kamitani, A.; Rakugi, H.; Zhao, Y.; Mikami, H.; Miki, T.; Ogihara, T.: A potent genetic risk factor for restenosis. (Letter) Nature Genet. 5: 324–325, 1993.
- [29479] 3031. Oike, Y.; Hata, A.; Ogata, Y.; Numata, Y.; Shido, K.; Kondo, K.: Angiotensin converting enzyme as a genetic risk factor for coronary artery spasm: implication in the pathogenesis of myocardial infarction. J. Clin. Invest. 96: 2975–2979, 1995.
- [29480] 3032. Okabe, T.; Fushisawa, M.; Yotsumoto, M.; Takaru, F.; Lanzillo, J. J.; Fanburg, B. L.: Familial elevation of serum angiotensin-converting enzyme. Quart. J. Med. 216: 55–61, 1985.
- [29481] 3033. Montgomery, H. E.; Clarkson, P.; Dollery, C. M.; Prasad, K.; Losi, M.-A.; Hemingway, H.; Statters, D.; Jubb,

M.; Girvain, M.; Varnava,A.; World, M.; Deanfield, J.; Talmud, P.; McEwan, J. R.; McKenna,W. J.; Humphries, S.: Association of angiotensin–converting enzyme gene I/D polymorphism with change in left ventricular mass in response to physical training. *Circulation* 96: 741–747, 1997.

[29482] 3034. Pfeffer, M. A.; Braunwald, E.; Moye, L. A.; Basta, L.; Brown, E. J., Jr.; Cuddy, T. E.; Davis, B. R.; Geltman, E. M.; Goldman, S.; Flaker, G. C.; Klein, M.; Lamas, G. A.; Packer, M.; Rouleau, J.; Rouleau, J. L.; Rutherford, J.; Wertheimer, J. H.; Hawkins, C. M.: Effect of captopril on mortality and morbidity in patients with left ventricular dysfunction after myocardial infarction: results of the survival and ventricular enlargement trial. *New Eng. J. Med.* 327: 669–677, 1992.

[29483] 3035. Ramaraj, P.; Kessler, S. P.; Colmenares, C.; Sen, G. C.: Selective restoration of male fertility in mice lacking angiotensin–converting enzymes by sperm–specific expression of the testicular isozyme. *J. Clin. Invest.* 102: 371–378, 1998.

[29484] 3036. Rigat, B.; Hubert, C.; Alhenc–Gelas, F.; Cambien, F.; Corvol, P.; Soubrier, F.: An insertion/deletion polymorphism in the angiotensin–converting enzyme gene ac–

counting for half the variance of serum enzyme levels. *J. Clin. Invest.* 86: 1343–1346, 1990.

- [29485] 3037. Rigat, B.; Hubert, C.; Corvol, P.; Soubrier, F.: PCR detection of the insertion/deletion polymorphism of the human angiotensin converting enzyme gene (DCP1) (dipeptidylcarboxypeptidase 1). *Nucleic Acids Res.* 20: 1433, 1992.
- [29486] 3038. Ruiz, J.; Blanche, H.; Cohen, N.; Velho, G.; Cambien, F.; Cohen, D.; Passa, P.; Froguel, P.: Insertion/deletion polymorphism of the angiotensin-converting enzyme gene is strongly associated with coronary heart disease in non-insulin-dependent diabetes mellitus. *Proc. Nat. Acad. Sci.* 91: 3662–3665, 1994.
- [29487] 3039. Schachter, F.; Faure-Delanef, L.; Guenot, F.; Rouger, H.; Froguel, P.; Lesueur-Ginot, L.; Cohen, D.: Genetic associations with human longevity at the APOE and ACE loci. *Nature Genet.* 6: 29–32, 1994.
- [29488] 3040. Schmidt, S.; Schone, N.; Ritz, E.; Giesel, R.; Bergis, K.; Strojek, K.; Greszczak, W.; Schroter, W.; Willms, B. H. L.; Petzold, R.; Henrichs, H. R.; Rambauser, M.; Schwarzbeck, A.; Kohr, B.; Schneider, P.; Bosch, A.: Association of ACE gene polymorphism and diabetic nephropathy? *Kidney Int.* 47: 1176–1181, 1995.

- [29489] 3041.Schunkert, H.; Hense, H.-W.; Holmer, S. R.; Stender, M.; Perz,S.; Keil, U.; Lorell, B. H.; Riegger, G. A. J.: Association between a deletion polymorphism of the angiotensin-converting-enzyme gene and left ventricular hypertrophy. *New Eng. J. Med.* 330: 1634-1638,1994.
- [29490] 3042.Singer, D. R. J.; Missouris, C. G.; Jeffery, S.: Angiotensin-converting enzyme gene polymorphism: what to do about all the confusion? (Editorial) *Circulation* 94:236-239, 1996.
- [29491] 3043.Soubrier, F.; Alhenc-Gelas, F.; Hubert, C.; Allegrini, J.; John,M.; Tregear, G.; Corvol, P.: Two putative active centers in human angiotensin I-converting enzyme revealed by molecular cloning. *Proc.Nat. Acad. Sci.* 85: 9386-9390, 1988.
- [29492] 3044.Tarnow, L.; Cambien, F.; Rossing, P.; Nielsen, F. S.; Hansen,B. V.; Lecerf, L.; Poirier, O.; Danilov, S.; Parving, H.-H.: Lack of relationship between an insertion/deletion polymorphism in the angiotensin I-converting enzyme gene and diabetic nephropathy and proliferative retinopathy in IDDM patients. *Diabetes* 44: 489-494,1995.
- [29493] 3045.Tiret, L.; Rigat, B.; Visvikis, S.; Breda, C.; Corvol, P.; Cambien,F.; Soubrier, F.: Evidence, from combined segregation and linkage analysis, that a variant of the an-

giotensin I-converting enzyme (ACE)gene controls plasma ACE levels. Am. J. Hum. Genet. 51: 197–205,1992.

[29494] 3046.Vleming, L. J.; van der Pijl, J. W.; Lemkes, H. H. P. J.; Westendorp,R. G. J.; Maassen, J. A.; Daha, M. R.; van Es, L. A.; van Kooten,C.: The DD genotype of the ACE gene polymorphism is associated withprogression of diabetic nephropathy to end stage renal failure inIDDM. Clin. Nephrol. 51: 133–140, 1999.

[29495] 3047.Williams, A. G.; Rayson, M. P.; Jubb, M.; World, M.; Woods, D.R.; Hayward, M.; Martin, J.; Humphries, S. E.; Montgomery, H. E.:The ACE gene and muscle performance. Nature 403: 614 only, 2000.

[29496] 3048.Reddi, A. H.: BMP–1: resurrection as procollagen C–proteinase. Science 271:5–6, 1996.

[29497] 3049.Takahara, K.; Lee, S.; Wood, S.; Greenspan, D. S.: Structuralorganization and genetic localization of the human bone morphogeneticprotein 1/mammalian tolloid gene. Genomics 29: 9–15, 1995.

[29498] 3050.Takahara, K.; Lyons, G. E.; Greenspan, D. S.: Bone morphogeneticprotein–1 and a mammalian tolloid homologue (mTld) are encoded byalternatively spliced transcripts which are differentially expressedin some tissues. J. Biol. Chem. 269: 32572–32578, 1994.

- [29499] 3051.Scott, I. C.; Blitz, I. L.; Pappano, W. N.; Imamura, Y.; Clark,T. G.; Steiglitiz, B. M.; Thomas, C. L.; Maas, S. A.; Takahara, K.;Cho, K. W. Y.; Greenspan, D. S.: Mammalian BMP-1/Tolloid-related metalloproteinases, including novel family member mammalian Tolloid-like2, have differential enzymatic activities and distributions of expression relevant to patterning and skeletogenesis. *Dev. Biol.* 213: 283-300,1999.
- [29500] 3052.Yoshiura, K.; Tamura, T.; Hong, H.-S.; Ohta, T.; Soejima, H.;Kishino, T.; Jinno, Y.; Niikawa, N.: Mapping of the bone morphogeneticprotein 1 gene (BMP1) to 8p21: removal of BMP1 from candidacy forthe bone disorder in Langer-Giedion syndrome. *Cytogenet. Cell Genet.* 64:208-209, 1993.
- [29501] 3053.Hahn, G. V.; Cohen, R. B.; Wozney, J. M.; Levitz, C. L.; Shore,E. M.; Zasloff, M. A.; Kaplan, F. S.: A bone morphogenetic proteinsubfamily: chromosomal localization of human genes for BMP5, BMP6,and BMP7. *Genomics* 14: 759-762, 1992.
- [29502] 3054.Lyons, K.; Graycar, J. L.; Lee, A.; Hashmi, S.; Lindquist, P. B.;Chen, E. Y.; Hogan, B. L. M.; Derynck, R.: Vgr-1, a mammalian generelated to *Xenopus* Vg-1, is a member of the transforming growth factorbeta gene su-

perfamily. Proc. Nat. Acad. Sci. 86: 4554–4558, 1989.

[29503] 3055. Rickard, D. J.; Hofbauer, L. C.; Bonde, S. K.; Gori, F.; Spelsberg, T. C.; Riggs, B. L.: Bone morphogenetic protein-6 production in human osteoblastic cell lines: selective regulation by estrogen. J. Clin. Invest. 101: 413–422, 1998.

[29504] 3056. Marker, P. C.; King, J. A.; Copeland, N. G.; Jenkins, N. A.; Kingsley, D. M.: Chromosomal localization, embryonic expression, and imprinting tests for Bmp7 on distal mouse chromosome 2. Genomics 28: 576–580, 1995.

[29505] 3057. Ozkaynak, E.; Rueger, D. C.; Drier, E. A.; Corbett, C.; Ridge, R. J.; Sampath, T. K.; Oppermann, H.: OP-1 cDNA encodes an osteogenic protein in the TGF- β family. EMBO J. 9: 2085–2093, 1990.

[29506] 3058. Solursh, M.; Langille, R. M.; Wood, J.; Sampath, T. K.: Osteogenic protein-1 is required for mammalian eye development. Biochem. Biophys. Res. Commun. 218: 438–443, 1996.

[29507] 3059. Modi, W. S.; Dean, M.; Pollock, D. D.; Seuanez, H. N.; Christakos, S.: Chromosomal localization of the calbindin gene. (Abstract) Cytogenet. Cell Genet. 58: 1930 only, 1991.

[29508] 3060. Parmentier, M.; De Vijlder, J. J. M.; Muir, E.; Szpirer,

C.; Islam, M. Q.; Geurts van Kessel, A.; Lawson, D. E. M.; Vassart, G.: The human calbindin 27 kDa gene: structural organization of the 5-prime and 3-prime regions, chromosomal assignment and restriction fragment length polymorphism. *Genomics* 4: 309–319, 1989.

[29509] 3061. Parmentier, M.; Lawson, D. E. M.; Vassart, G.: Human 27-kDa calbindin complementary DNA sequence: evolutionary and functional implications. *Europ. J. Biochem.* 170: 207–215, 1987.

[29510] 3062. Parmentier, M.; Passage, E.; Vassart, G.; Mattei, M.-G.: The human calbindin D28k (CALB1) and calretinin (CALB2) genes are located at 8q21.3–q22.1 and 16q22–q23, respectively, suggesting a common duplication with the carbonic anhydrase isozyme loci. *Cytogenet. Cell Genet.* 57:41–43, 1991.

[29511] 3063. Parmentier, M.; Vassart, G.: HindIII RFLP on chromosome 8 detected with a calbindin 27 kDa cDNA probe, HBSC21. *Nucleic Acids Res.* 16:9373 only, 1988.

[29512] 3064. Seto-Ohshima, A.; Emson, P. C.; Lawson, E.; Mountjoy, C. Q.; Carrasco, L. H.: Loss of matrix calcium-binding protein-containing neurons in Huntington's disease. *Lancet* i: 1252–1254, 1988.

[29513] 3065. Parmentier, M.; Szpirer, J.; Levan, G.; Vassart, G.:

The humangenes for calbindin 27 and 29 kDa proteins are located on chromosomes 8 and 16, respectively. Cytogenet. Cell Genet. 52: 85–87, 1989.

- [29514] 3066. Schiffmann, S. N.; Cheron, G.; Lohof, A.; d'Alcantara, P.; Meyer, M.; Parmentier, M.; Schurmans, S.: Impaired motor coordination and Purkinje cell excitability in mice lacking calretinin. Proc. Nat. Acad. Sci. 96: 5257–5262, 1999.
- [29515] 3067. Davies, A. A.; Moss, S. E.; Crompton, M. R.; Jones, T. A.; Spurr, N. K.; Sheer, D.; Kozak, C.; Crompton, M. J.: The gene coding for the p68 calcium-binding protein is localized to bands q32–q34 of human chromosome 5, and to mouse chromosome 11. Hum. Genet. 82: 234–238, 1989.
- [29516] 3068. Smith, P. D.; Davies, A.; Crompton, M. J.; Moss, S. E.: Structure of the human annexin VI gene. Proc. Nat. Acad. Sci. 91: 2713–2717, 1994.
- [29517] 3069. Sudhof, T. C.; Slaughter, C. A.; Leznicki, I.; Barjon, P.; Reynolds, G. A.: Human 67-kDa calyculin contains a duplication of four repeats found in 35-kDa lipocortins. Proc. Nat. Acad. Sci. 85: 664–668, 1988.
- [29518] 3070. Blow, J. J.; Laskey, R. A.: A role for the nuclear envelope in controlling DNA replication within the cell cycle. Nature 332: 546–548, 1988.
- [29519] 3071. Burkhardt, R.; Schulte, D.; Hu, D.; Musahl, C.;

Gohring, F.; Knippers, R.: Interactions of human nuclear proteins P1Mcm3 and P1Cdc46. *Europ.J. Biochem.* 228: 431–438, 1995.

[29520] 3072. Chong, J. P.; Mahbubani, H. M.; Khoo, C. Y.; Blow, J. J.: Purification of an MCM-containing complex as a component of the DNA replication licensing system. *Nature* 375: 418–421, 1995.

[29521] 3073. Hu, B.; Burkhardt, R.; Schulte, D.; Musahl, C.; Knippers, R.: The P1 family: a new class of nuclear mammalian proteins related to the yeast Mcm replication proteins. *Nucleic Acids Res.* 21: 5289–5293, 1993.

[29522] 3074. Kubota, Y.; Mimura, S.; Nishimoto, S.; Takisawa, H.; Nojima, H.: Identification of the yeast MCM3-related protein as a component of *Xenopus* DNA replication licensing factor. *Cell* 81: 601–609, 1995.

[29523] 3075. Labib, K.; Tercero, J. A.; Diffley, J. F. X.: DNA replication fork progression requires uninterrupted MCM2–7 function. *Science* 288: 1643–1647, 2000.

[29524] 3076. Madine, M. A.; Khoo, C. Y.; Mills, A. D.; Laskey, R. A.: MCM3 complex required for cell cycle regulation of DNA replication in vertebrate cells. *Nature* 375: 421–424, 1995.

[29525] 3077. Mincheva, A.; Todorov, I.; Werner, D.; Fink, T. M.; Lichter, P.: The human gene for nuclear protein BM28

(CDCL1), a new member of the early S-phase family of proteins, maps to chromosome band 3q21. *Cytogenet. Cell Genet.* 65: 276–277, 1994.

- [29526] 3078. Tsuruga, H.; Yabuta, N.; Hashizume, K.; Ikeda, M.; Endo, Y.; Nojima, H.: Expression, nuclear localization and interactions of human MCM/P1 proteins. *Biochem. Biophys. Res. Commun.* 236: 118–125, 1997.
- [29527] 3079. Adams, M. D.; Dubnick, M.; Kerlavage, A. R.; Moreno, R.; Kelley, J. M.; Utterback, T. R.; Nagle, J. W.; Fields, C.; Venter, J. C.: Sequence identification of 2,375 human brain genes. *Nature* 355: 632–634, 1992.
- [29528] 3080. Tugendreich, S.; Boguski, M. S.; Seldin, M. S.; Hieter, P.: Linking yeast genetics to mammalian genomes: identification and mapping of the human homolog of CDC27 via the expressed sequence tag (EST) database. *Proc. Nat. Acad. Sci.* 90: 10031–10035, 1993.
- [29529] 3081. Demetrick, D. J.; Beach, D. H.: Chromosome mapping of human CDC25A and CDC25B phosphatases. *Genomics* 18: 144–147, 1993.
- [29530] 3082. Fauman, E. B.; Cogswell, J. P.; Lovejoy, B.; Rocque, W. J.; Holmes, W.; Montana, V. G.; Piwnicka-Worms, H.; Rink, M. J.; Saper, M. A.: Crystal structure of the catalytic domain of the human cell cycle control phosphatase, Cdc25A. *Cell*

93: 617–625, 1998.

- [29531] 3083.Galaktionov, K.; Lee, A. K.; Eckstein, J.; Draetta, G.; Meckler, J.; Loda, M.; Beach, D.: CDC25 phosphatases as potential human oncogenes. *Science* 269:1575–1577, 1995.
- [29532] 3084.Mailand, N.; Falck, J.; Lukas, C.; Syljuasen, R. G.; Welcker, M.; Bartek, J.; Lukas, J.: Rapid destruction of human Cdc25A in response to DNA damage. *Science* 288: 1425–1429, 2000.
- [29533] 3085.Lane, S. A.; Baker, E.; Sutherland, G. R.; Tonks, I.; Hayward, N.; Ellem, K.: The human cell cycle gene CDC25B is located at 20p13. *Genomics* 15:693–694, 1993.
- [29534] 3086.Lincoln, A. J.; Wickramasinghe, D.; Stein, P.; Schultz, R. M.; Palko, M. E.; De Miguel, M. P.; Tessarollo, L.; Donovan, P. J.: Cdc25b phosphatase is required for resumption of meiosis during oocyte maturation. *Nature Genet.* 30: 446–449, 2002.
- [29535] 3087.Dullaart, R. P.; Hoogenberg, K.; Riemens, S. C.; Groener, J. E.; van Tol, A. Sluiter, W. J.; Stulp, B. K.: Cholesteryl ester transfer protein gene polymorphism is a determinant of HDL cholesterol and of the lipoprotein response to a lipid-lowering diet in type 1 diabetes. *Diabetes* 46:2082–2087, 1997.

- [29536] 3088.Durlach, A.; Clavel, C.; Girard-Globa, A.; Durlach, V.: Sex-dependent association of a genetic polymorphism of cholesteryl ester transfer protein with high-density lipoprotein cholesterol and macrovascular pathology in type II diabetic patients. *J. Clin. Endocr. Metab.* 84:3656–3659, 1999.
- [29537] 3089.Freeman, D. J.; Griffin, B. A.; Holmes, A. P.; Lindsay, G. M.; Gaffney, D.; Packard, C. J.; Shepherd, J.: Regulation of plasma HDL cholesterol and subfraction distribution by genetic and environmental factors: associations between the TaqI B RFLP in the CETP gene and smoking and obesity. *Arterioscler. Thromb.* 14: 336–344, 1994.
- [29538] 3090.Hannuksela, M. L.; Liinamaa, M. J.; Kesaniemi, Y. A.; Savolainen, M. J.: Relation of polymorphisms in the cholesteryl ester transfer protein gene to transfer protein activity and plasma lipoprotein levels in alcohol drinkers. *Atherosclerosis* 110: 35–44, 1994.
- [29539] 3091.Herrera, V. L. M.; Makrides, S. C.; Xie, H. X.; Adari, H.; Krauss, R. M.; Ryan, U. S.; Ruiz-Opazo, N.: Spontaneous combined hyperlipidemia, coronary heart disease and decreased survival in Dahl salt-sensitive hypertensive rats transgenic for human cholesteryl ester transfer protein. *Nature Med.* 5: 1383–1389, 1999.

- [29540] 3092.Fumeron, F.; Betoulle, D.; Luc, G.; Behague, I.; Ricard, S.; Poirier, O.; Jemaa, R.; Evans, A.; Arveiler, D.; Marques-Vidal, P.; Bard, J.-M.; Fruchart, J.-C.; Ducimetiere, P.; Apfelbaum, M.; Cambien, F.: Alcohol intake modulates the effect of a polymorphism of the cholesteryl ester transfer protein gene on plasma high density lipoprotein and the risk of myocardial infarction. *J. Clin. Invest.* 96: 1664-1671, 1995.
- [29541] 3093.Inazu, A.; Jiang, X.-C.; Haraki, T.; Yagi, K.; Kamon, N.; Koizumi, J.; Mabuchi, H.; Takeda, R.; Takata, K.; Moriyama, Y.; Doi, M.; Tall, A.: Genetic cholesteryl ester transfer protein deficiency caused by two prevalent mutations as a major determinant of increased levels of high density lipoprotein cholesterol. *J. Clin. Invest.* 94: 1872-1882, 1994.
- [29542] 3094.Kondo, I.; Berg, K.; Drayna, D.; Lawn, R.: DNA polymorphism at the locus for human cholesteryl ester transfer protein (CETP) is associated with high density lipoprotein cholesterol and apolipoprotein levels. *Clin. Genet.* 35: 49-56, 1989.
- [29543] 3095.Kuivenhoven, J. A.; de Knijff, P.; Boer, J. M. A.; Smalheer, H. A.; Botma, G. J.; Seidell, J. C.; Kastelein, J. J.; Pritchard, P. H.: Heterogeneity at the CETP gene locus: in-

fluence on plasma CETP concentrations and HDL cholesterol levels. *Arterioscler. Thromb. Vasc. Biol.* 17: 560–568, 1997.

[29544] 3096. Kuivenhoven, J. A.; Jukema, J. W.; Zwinderman, A. H.; de Knijff, P.; McPherson, R.; Bruschke, A. V. G.; Lie, K. I.; Kastelein, J. J. P.: The role of a common variant of the cholesteryl ester transfer protein gene in the progression of coronary atherosclerosis. *New Eng. J. Med.* 338: 86–93, 1998.

[29545] 3097. Lusis, A. J.; Zollman, S.; Sparkes, R. S.; Klisak, I.; Mohandas, T.; Drayna, D.; Lawn, R. M.: Assignment of the human gene for cholesteryl ester transfer protein to chromosome 16q12–16q21. *Genomics* 1: 232–242, 1987.

[29546] 3098. Bentley, K. L.; Ferguson-Smith, A. C.; Miki, T.; Kidd, K. K.; Ruddle, F. H.: Physical linkage of Hox 2.1 and nerve growth factor receptor. (Abstract) *Cytogenet. Cell Genet.* 51: 961 only, 1989.

[29547] 3099. Bibel, M.; Barde, Y.-A.: Neurotrophins: key regulators of cell fate and cell shape in the vertebrate nervous system. *Genes Dev.* 14: 2919–2937, 2000.

[29548] 3100. Bothwell, M.: p75(NTR): a receptor after all. *Science* 272: 506–507, 1996.

[29549] 3101. Carter, B. D.; Kaltschmidt, C.; Kaltschmidt, B.; Of-

fenhauser, N.;Bohm–Matthaei, R.; Baeuerle, P. A.; Barde, Y.–A.: Selective activationof NF–kappa–B by nerve growth factor through the neurotrophin receptorp75. Science 272: 542–545, 1996.

[29550] 3102.Carter, B. D.; Lewin, G. R.: Neurotrophins live or let die: doesp75(NTR) decide? Neuron 18: 187–190, 1997.

[29551] 3103.Chao, M. V.; Bothwell, M. A.; Ross, A. H.; Koprowski, H.; Lanahan,A. A.; Buck, C. R.; Sehgal, A.: Gene transfer and molecular cloningof the human NGF receptor. Science 232: 518–521, 1986.

[29552] 3104.Dobrowsky, R. T.; Werner, M. H.; Castellino, A. M.; Chao, M. V.;Hannun, Y. A.: Activation of the sphingomyelin cycle through the low–affinity neurotrophin receptor. Science 265: 1596–1599, 1994.

[29553] 3105.Colonna, M.; Bresnahan, M.; Bahram, S.; Strominger, J. L.; Spies,T.: Allelic variants of the human putative peptide transporter involvedin antigen processing. Proc. Nat. Acad. Sci. 89: 3932–3936, 1992.

[29554] 3106.Huh, G. S.; Boulanger, L. M.; Du, H.; Riquelme, P. A.; Brotz, T.M.; Shatz, C. J.: Functional requirement for class I MHC in CNS developmentand plasticity. Science 290: 2155–2159, 2000.

[29555] 3107.Karttunen, J. T.; Lehner, P. J.; Gupta, S. S.; Hewitt, E.

W.; Cresswell, P.: Distinct functions and cooperative interaction of the subunits of the transporter associated with antigen processing (TAP). *Proc. Nat. Acad. Sci.* 98: 7431–7436, 2001.

[29556] 3108. Cullen, M.; Erlich, H.; Klitz, W.; Carrington, M.: Molecular mapping of a recombination hotspot located in the second intron of the human TAP2 locus. *Am. J. Hum. Genet.* 56: 1350–1358, 1995.

[29557] 3109. de la Salle, H.; Donato, L.; Zimmer, J.; Plebani, A.; Hanau, D.; Bonneville, M.; Tongio, M.–M.: HLA class I deficiencies. In: Ochs, H. D.; Smith, C. I. E.; Puck, J. M. (eds.): *Primary Immunodeficiency Diseases: A Molecular and Genetic Approach*. New York: Oxford University Press 1999. Pp. 181–188.

[29558] 3110. de la Salle, H.; Hanau, D.; Fricker, D.; Urlacher, A.; Kelly, A.; Salamero, J.; Powis, S. H.; Donato, L.; Bausinger, H.; Laforet, M.; Jeras, M.; Spehner, D.; Bieber, T.; Falkenrodt, A.; Cazenave, J.–P.; Trowsdale, J.; Tongio, M.–M.: Homozygous human TAP peptide transporter mutation in HLA class I deficiency. *Science* 265: 237–241, 1994.

[29559] 3111. Jeffreys, A. J.; Ritchie, A.; Neumann, R.: High resolution analysis of haplotype diversity and meiotic crossover in the human TAP2 recombination hotspot. *Hum. Molec.*

Genet. 9: 725–733, 2000.

- [29560] 3112.Powis, S. H.; Mockridge, I.; Kelly, A.; Kerr, L.-A.; Glynn, R.; Gileadi, U.; Beck, S.; Trowsdale, J.: Polymorphism in a second ABC transporter gene located within the class II region of the human major histocompatibility complex. *Proc. Nat. Acad. Sci.* 89: 1463–1467, 1992.
- [29561] 3113.Glauder, J.; Ragg, H.; Rauch, J.; Engels, J. W.: Human peptidylglycine α -amidating monooxygenase: cDNA, cloning and functional expression of a truncated form in COS cells. *Biochem. Biophys. Res. Commun.* 169:551–558, 1990.
- [29562] 3114.Lossie, A. C.; Eipper, B. A.; Hand, T. A.; Camper, S. A.: Localization of the peptidylglycine α -amidating monooxygenase gene (Pam) introduces a region of homology between human chromosome 5q and mouse chromosome 1. *Mammalian Genome* 5: 738–739, 1994.
- [29563] 3115.Ouafik, L'H.; Mattei, M. G.; Giraud, P.; Oliver, C.; Eipper, B.A.; Mains, R. E.: Localization of the gene encoding peptidylglycine α -amidating monooxygenase (PAM) to human chromosome 5q14–5q21. *Genomics* 18: 319–321, 1993.
- [29564] 3116.Jackson, R. S.; Creemers, J. W. M.; Ohagi, S.; Raffin-Sanson, M.-L.; Sanders, L.; Montague, C. T.; Hutton, J. C.;

O'Rahilly, S.: Obesity and impaired prohormone processing associated with mutations in the human prohormone convertase 1 gene. *Nature Genet.* 16: 303–306, 1997.

[29565] 3117. Naggert, J. K.; Fricker, L. D.; Varlamov, O.; Nishina, P. M.; Rouille, Y.; Steiner, D. F.; Carroll, R. J.; Paigen, B. J.; Leiter, E. H.: Hyperproinsulinaemia in obese fat/fat mice associated with a carboxypeptidase E mutation which reduces enzyme activity. *Nature Genet.* 10: 135–142, 1995.

[29566] 3118. O'Rahilly, S.; Gray, H.; Humphreys, P. J.; Krook, A.; Polonsky, K. S.; White, A.; Gibson, S.; Taylor, K.; Carr, C.: Brief report: impaired processing of prohormones associated with abnormalities of glucose homeostasis and adrenal function. *New Eng. J. Med.* 333: 1386–1390, 1995.

[29567] 3119. Ohagi, S.; Sakaguchi, H.; Sanke, T.; Tatsuta, H.; Hanabusa, T.; Nanjo, K.: Human prohormone convertase 3 gene: exon–intron organization and molecular scanning for mutations in Japanese subjects with NIDDM. *Diabetes* 45: 897–901, 1996.

[29568] 3120. Furuta, M.; Carroll, R.; Martin, S.; Swift, H.; Ravazzola, M.; Orci, L.; Steiner, D.: Incomplete processing of proinsulin to insulin accompanied by elevation of Des-31,32 proinsulin intermediates in islets of mice lacking active PC2. *J. Biol. Chem.* 273: 3431–3437, 1998.

- [29569] 3121. Furuta, M.; Yano, H.; Zhou, A.; Rouille, Y.; Holst, J.; Carroll, R.; Ravazzola, M.; Orci, L.; Furuta, H.; Steiner, D.: Defective prohormone processing and altered pancreatic islet morphology in mice lacking active SPC2. *Proc. Nat. Acad. Sci.* 94: 6646–6651, 1997.
- [29570] 3122. Furuta, M.; Zhou, A.; Webb, G.; Carroll, R.; Ravazzola, M.; Orci, L.; Steiner, D. F.: Severe defect in proglucagon processing in islet A-cells of prohormone convertase 2 null mice. *J. Biol. Chem.* 276:27197–27202, 2001.
- [29571] 3123. Gabreels, B. A. T. F.; Swaab, D. F.; de Kleijn, D. P. V.; Seidah, N. G.; Van de Loo, J.-W.; Van de Ven, W. J. M.; Martens, G. J. M. and van Leeuwen, F. W.: Attenuation of the polypeptide 7B2, prohormone convertase PC2, and vasopressin in the hypothalamus of some Prader-Willi patients: indications for a processing defect. *J. Clin. Endocr. Metab.* 83:591–599, 1998.
- [29572] 3124. Maglott, D. R.; Feldblyum, T. V.; Durkin, A. S.; Nierman, W. C.: Radiation hybrid mapping of SNAP, PCSK2, and THBD (human chromosome 20p). *Mammalian Genome* 7: 400–401, 1996.
- [29573] 3125. Ohagi, S.; LaMendola, J.; LeBeau, M. M.; Espinosa, R., III; Takeda, J.; Smeekens, S. P.; Chan, S. J.; Steiner, D. F.:

Identification and analysis of the gene encoding human PC2, a prohormone convertase expressed in neuroendocrine tissues. *Proc. Nat. Acad. Sci.* 89: 4977–4981, 1992.

[29574] 3126. Taylor, N. A.; Shennan, K. I. J.; Cutler, D. F.; Docherty, K.: Mutations within the propeptide, the primary cleavage site or the catalytic site, or deletion of C-terminal sequences, prevents secretion of proPC2 from transfected COS-7 cells. *Biochem. J.* 321: 367–373, 1997.

[29575] 3127. Chan, W.-Y.; Liu, Q.-R.; Borjigin, J.; Busch, H.; Rennert, O. M.; Tease, L. A.; Chan, P.-K.: Characterization of the cDNA encoding human nucleophosmin and studies of its role in normal and abnormal growth. *Biochemistry* 28: 1033–1039, 1989.

[29576] 3128. Cheng, G.-X.; Zhu, X.-H.; Men, X.-Q.; Wang, L.; Huang, Q.-H.; Jin, X. L.; Xiong, S.-M.; Zhu, J.; Guo, W.-M.; Chen, J.-Q.; Xu, S.-F.; So, E.; Chan, L.-C.; Waxman, S.; Zelen, A.; Chen, G.-Q.; Dong, S.; Liu, J.-X.; Chen, S.-J.: Distinct leukemia phenotypes in transgenic mice and different corepressor interactions generated by promyelocytic leukemia variant fusion genes PLZF-RAR- α and NPM-RAR- α . *Proc. Nat. Acad. Sci.* 96: 6318–6323, 1999.

[29577] 3129. Dutta, S.; Akey, I. V.; Dingwall, C.; Hartman, K. L.;

Laue, T.; Nolte, R. T.; Head, J. F.; Akey, C. W.: The crystal structure of nucleoplasmin-core: implications for histone binding and nucleosome assembly. *Molec. Cell* 8: 841–853, 2001.

- [29578] 3130.Okuda, M.; Horn, H. F.; Tarapore, P.; Tokuyama, Y.; Smulian, A.G.; Chan, P.-K.; Knudsen, E. S.; Hofmann, I. A.; Snyder, J. D.; Bove,K. E.; Fukasawa, K.: Nucleophosmin/B23 is a target of CDK2/cyclinE in centrosome duplication. Cell 103: 127–140, 2000.
- [29579] 3131.Redner, R. L.; Rush, E. A.; Faas, S.; Rudert, W. A.; Corey, S.J.: The t(5;17) variant of acute promyelocytic leukemia expressesa nucleophosmin–retinoic acid receptor fusion. Blood 87: 882–886,1996.
- [29580] 3132.Moore, M. D.; Cooper, N. R.; Tack, B. F.; Nemerow, G. R.: Molecularcloning of the cDNA encoding the Epstein–Barr virus/C3d receptor (complementreceptor type 2) of human B lymphocytes. Proc. Nat. Acad. Sci. 84:9194–9198, 1987.
- [29581] 3133.Prota, A. E.; Sage, D. R.; Stehle, T.; Fingerroth, J. D.: The crystalstructure of human CD21: implications for Epstein–Barr virus and C3dbinding. Proc. Nat. Acad. Sci. 99: 10641–10646, 2002.
- [29582] 3134.Rodriguez de Cordoba, S.; Rubinstein, P.: Quantitative variationsof the C3b/C4b receptor (CR1) in human erythrocyte are controlledby genes within the regulator of complement activation (RCA) genecluster. J. Exp. Med. 164: 1274–1283, 1986.

- [29583] 3135.Szakonyi, G.; Guthridge, J. M.; Li, D.; Young, K.; Holers, V. M.;Chen, X. S.: Structure of complement receptor 2 in complex with itsC3d ligand. *Science* 292: 1725–1728, 2001.
- [29584] 3136.Weis, J. J.; Tedder, T. F.; Fearon, D. T.: Identification of a145,000 M(r) membrane protein as the C3d receptor (CR2) of human Blymphocytes. *Proc. Nat. Acad. Sci.* 81: 881–885, 1984.
- [29585] 3137.Yefenof, E.; Klein, G.; Jondal, M.; Oldstone, M. B. A.: Surfacemarkers on human B– and T–lymphocytes. IX. Two color immunofluorescencestudies on the association between EBV receptors and complement receptoron the surface of lymphoid cell lines. *Int. J. Cancer* 17: 693–700,1976.
- [29586] 3138.Potter, E.; Behan, D. P.; Fischer, W. H.; Linton, E. A.; Lowry,P. J.; Vale, W. W.: Cloning and characterization of the cDNAs forhuman and rat corticotropin releasing factor–binding proteins. *Nature* 349:423–426, 1991.
- [29587] 3139.Vamvakopoulos, N. C.; Sioutopoulou, T. O.; Durkin, S. A.; Nierman,W. C.; Wasmuth, J. J.; McPherson, J. D.: Mapping the human corticotropinreleasing hormone binding protein gene (CRHBP) to the long arm ofchromosome 5 (5q11.2–q13.3). *Genomics* 25: 325–327, 1995.

- [29588] 3140. Arbiser, J. L.; Morton, C. C.; Bruns, G. A. P.; Majzoub, J. A.: Human corticotropin releasing hormone gene is located on the long arm of chromosome 8. *Cytogenet. Cell Genet.* 47: 113–116, 1988.
- [29589] 3141. Behan, D. P.; Heinrichs, S. C.; Troncoso, J. C.; Liu, X.-J.; Kawas, C. H.; Ling, N.; De Souza, E. B.: Displacement of corticotropin releasing factor from its binding protein as a possible treatment for Alzheimer's disease. *Nature* 378: 284–287, 1995.
- [29590] 3142. Cheng, Y.-H.; Nicholson, R. C.; King, B.; Chan, E.-C.; Fitter, J. T.; Smith, R.: Corticotropin-releasing hormone gene expression in primary placental cells is modulated by cyclic adenosine 3'-prime, 5'-prime-monophosphate. *J. Clin. Endocr. Metab.* 85: 1239–1244, 2000.
- [29591] 3143. Habib, K. E.; Weld, K. P.; Rice, K. C.; Pushkas, J.; Champoux, M.; Listwak, S.; Webster, E. L.; Atkinson, A. J.; Schulkin, J.; Contoreggi, C.; Chrousos, G. P.; McCann, S. M.; Suomi, S. J.; Higley, J. D.; Gold, P. W.: Oral administration of a corticotropin-releasing hormone receptor antagonist significantly attenuates behavioral, neuroendocrine, and autonomic responses to stress in primates. *Proc. Nat. Acad. Sci.* 97: 6079–6084, 2000.
- [29592] 3144. Inder, W. J.; Prickett, T. C. R.; Ellis, M. J.; Hull, L.;

Reid,R.; Benny, P. S.; Livesey, J. H.; Donald, R. A.: The utility of plasmaCRH as a predictor of preterm delivery. J. Clin. Endocr. Metab. 86:5706–5710, 2001.

[29593] 3145.Kellogg, J.; Luty, J. A.; Thompson, R.; Luo, X. Y.; Magenis, R.E.; Litt, M.: Corticotropin releasing hormone (CRH) maps to human chromosome 8 and identifies a TaqI RFLP. Cytogenet. Cell Genet. 51:1022, 1989.

[29594] 3146.Knapp, L. T.; Keegan, C. E.; Seasholtz, A. F.; Camper, S. A.:Corticotropin–releasing hormone (Crh) maps to mouse chromosome 3. MammalianGenome 4: 615–617, 1993.

[29595] 3147.Kyllo, J. H.; Collins, M. M.; Vetter, K. L.; Cuttler, L.; Rosenfield,R. L.; Donohoue, P. A.: Linkage of congenital isolated adrenocorticotrophic hormone deficiency to the corticotropin releasing hormone locus using simple sequence repeat polymorphisms. Am. J. Med. Genet. 62: 262–267,1996.

[29596] 3148.Majzoub, J. A.: Personal Communication. Boston, Mass. 3/3/1995.

[29597] 3149.Mandel, H.; Berant, M.; Gotfried, E.; Hochberg, Z.: Autosomalrecessive hypothalamic corticotropin deficiency: a new entity andits metabolic consequences. (Abstract) Am. J. Hum. Genet. 47 (suppl.):A66, 1990.

- [29598] 3150. McLean, M.; Bisits, A.; Davies, J.; Woods, R.; Lowry, P.; Smith, R.: A placental clock controlling the length of human pregnancy. *Nature Med.* 1: 460–463, 1995.
- [29599] 3151. Muglia, L.; Jacobson, L.; Dikkes, P.; Majzoub, J. A.: Corticotropin–releasing hormone deficiency reveals major fetal but not adult glucocorticoid need. *Nature* 373: 427–432, 1995.
- [29600] 3152. Robinson, B. G.; Emanuel, R. L.; Frim, D. M.; Majzoub, J. A.: Glucocorticoid stimulates expression of corticotropin–releasing hormone gene in human placenta. *Proc. Nat. Acad. Sci.* 85: 5244–5248, 1988.
- [29601] 3153. Scatena, C. D.; Adler, S.: Trans–acting factors dictate the species–specific placental expression of corticotropin–releasing factor genes in choriocarcinoma cell lines. *Endocrinology* 137: 3000–3008, 1996.
- [29602] 3154. Scatena, C. D.; Adler, S.: Characterization of a human–specific regulator of placental corticotropin–releasing hormone. *Molec. Endocr.* 12: 1228–1240, 1998.
- [29603] 3155. Shibahara, S.; Morimoto, Y.; Furutani, Y.; Notake, M.; Takahashi, H.; Shimizu, S.; Horikawa, S.; Numa, S.: Isolation and sequence analysis of the human corticotropin–releasing factor precursor gene. *EMBO J.* 2: 775–779, 1983.
- [29604] 3156. Stratakis, C. A.; Chrousos, G. P.: Neuroendocrinol–

ogy and pathophysiology of the stress system. Ann. N.Y. Acad. Sci. 771: 1–18, 1995.

[29605] 3157. Stratakis, C. A.; Sarlis, N. J.; Berrettini, W. H.; Badner, J. A.; Chrousos, G. P.; Gershon, E. S.; Detera-Wadleigh, S. D.: Lack of linkage between the corticotropin-releasing hormone (CRH) gene and bipolar affective disorder. Molec. Psychiat. 2: 483–485, 1997.

[29606] 3158. Xu, B.; Sano, T.; Yamada, S.; Li, C. C.; Hirokawa, M.: Expression of corticotropin-releasing hormone messenger ribonucleic acid in human pituitary corticotroph adenomas associated with proliferative potential. J. Clin. Endocr. Metab. 85: 1220–1225, 2000.

[29607] 3159. Zouboulis, C. C.; Seltmann, H.; Hiroi, N.; Chen, W.; Young, M.; Oeff, M.; Scherbaum, W. A.; Orfanos, C. E.; McCann, S. M.; Bornstein, S. R.: Corticotropin-releasing hormone: an autocrine hormone that promotes lipogenesis in human sebocytes. Proc. Nat. Acad. Sci. 99: 7148–7153, 2002.

[29608] 3160. Chen, R.; Lewis, K. A.; Perrin, M. H.; Vale, W. W.: Expression cloning of a human corticotropin-releasing-factor receptor. Proc. Nat. Acad. Sci. 90: 8967–8971, 1993.

[29609] 3161. Dieterich, K. D.; Gundelfinger, E. D.; Ludecke, D. K.; Lehnert, H.: Mutation and expression analysis of corti-

corticotropin-releasing factor₁ receptor in adrenocorticotropin-secreting pituitary adenomas. *J. Clin. Endocr. Metab.* 83: 3327–3331, 1998.

- [29610] 3162. Grammatopoulos, D.; Dai, Y.; Chen, J.; Karteris, E.; Papadopoulou, N.; Easton, A. J.; Hillhouse, E. W.: Human corticotropin-releasing hormone receptor: differences in subtype expression between pregnant and nonpregnant myometria. *J. Clin. Endocr. Metab.* 83: 2539–2544, 1998.
- [29611] 3163. Leproult, R.; Colecchia, E. F.; L'Hermite-Baleriaux, M.; Van Cauter, E.: Transition from dim to bright light in the morning induces an immediate elevation of cortisol levels. *J. Clin. Endocr. Metab.* 86: 151–157, 2001.
- [29612] 3164. Liaw, C. W.; Grigoriadis, D. E.; Lovenberg, T. W.; De Souza, E. B.; Maki, R. A.: Localization of ligand-binding domains of human corticotropin-releasing factor receptor: a chimeric receptor approach. *Molec. Endocr.* 11: 980–985, 1997.
- [29613] 3165. Lublin, D. M.; Mallinson, G.; Poole, J.; Reid, M. E.; Thompson, E. S.; Ferdman, B. R.; Telen, M. J.; Anstee, D. J.; Tanner, M. J. A.: Molecular basis of reduced or absent expression of decay-accelerating factor in Crohn's blood group phenotypes. *Blood* 84: 1276–1282, 1994.
- [29614] 3166. Medof, M. E.; Lublin, D. M.; Holers, V. M.; Ayers, D.

J.; Getty, R. R.; Leykam, J. F.; Atkinson, J. P.; Tykocinski, M. L.: Cloning and characterization of cDNAs encoding the complete sequence of decay-accelerating factor of human complement. *Proc. Nat. Acad. Sci.* 84: 2007–2011, 1987.

[29615] 3167. Reid, M. E.; Chandrasekaran, V.; Sausais, L.; Pierre, J.; Bullock, R.: Disappearance of antibodies to Cromer blood group system antigens during mid pregnancy. *Vox Sang.* 71: 48–50, 1996.

[29616] 3168. Wang, L.; Uchikawa, M.; Tsuneyama, H.; Tokunaga, K.; Tadokoro, K.; Juji, T.: Molecular cloning and characterization of decay-accelerating factor deficiency in Cromer blood group Inab phenotype. *Blood* 91: 680–684, 1998.

[29617] 3169. Danielson, K. G.; Fazzio, A.; Cohen, I.; Cannizzaro, L. A.; Eichstetter, I.; Iozzo, R. V.: The human decorin gene: intron-exon organization, discovery of two alternatively spliced exons in the 5-prime untranslated region, and mapping of the gene to chromosome 12q23. *Genomics* 15: 146–160, 1993.

[29618] 3170. Dyne, K. M.; Valli, M.; Forlino, A.; Mottes, M.; Kresse, H.; Cetta, G.: Deficient expression of the small proteoglycan decorin in a case of severe/lethal osteogenesis imperfecta. *Am. J. Med. Genet.* 63: 161–166, 1996.

[29619] 3171. Ion, A.; Crosby, A. H.; Kremer, H.; Kenmochi, N.; Van

Reen, M.;Fenske, C.; Van Der Burgt, I.; Brunner, H. G.; Montgomery, K.: Detailedmapping, mutation analysis, and intragenic polymorphism identificationin candidate Noo–nan syndrome genes MYL2, DCN, EPS8, and RPL6. J.Med. Genet. 37: 884–886, 2000.

[29620] 3172.McBride, O. W.; Fisher, L. W.; Young, M. F.: Localiza–tion of PGI(biglycan, BGN) and PGII (decorin, DCN, PG–40) genes on human chromosomesXq13–qter and 12q, re–spectively. Genomics 6: 219–255, 1990.

[29621] 3173.Moscatello, D. K.; Santra, M.; Mann, D. M.; McQuil–lan, D. J.; Wong,A. J.; Iozzo, R. V.: Decorin suppresses tu–mor cell growth by activatingthe epidermal growth factor receptor. J. Clin. Invest. 101: 406–412,1998.

[29622] 3174.Pulkkinen, L.; Alitalo, T.; Krusius, T.; Peltonen, L.: Expressionof decorin in human tissues and cell lines and defined chromosomalassignment of the gene locus (DCN). Cytogenet. Cell Genet. 60: 107–111,1992.

[29623] 3175.Pulkkinen, L.; Kainulainen, K.; Krusius, T.; Makinen, P.; Schollin,J.; Gustavsson, K.–H.; Peltonen, L.: Deficient expression of thegene coding for decorin in a lethal form of Marfan syndrome. J. Biol.Chem. 265: 17780–17785, 1990.

[29624] 3176.Schollin, J.; Bjarke, B.; Gustavson, K.–H.: Probable

homozygotic form of the Marfan syndrome in a newborn child. *Acta Paediat. Scand.* 77:452–456, 1988.

- [29625] 3177. Scholzen, T.; Solursh, M.; Suzuki, S.; Reiter, R.; Morgan, J. L.; Buchberg, A. M.; Siracusa, L. D.; Iozzo, R. V.: The murine decorin: complete cDNA cloning, genomic organization, chromosomal assignment, and expression during organogenesis and tissue differentiation. *J. Biol. Chem.* 269: 28270–28281, 1994.
- [29626] 3178. Vogel, K. G.; Clark, P. E.: Small proteoglycan synthesis by skin fibroblasts cultured from elderly donors and patients with defined defects in types I and III collagen metabolism. *Europ. J. Cell Biol.* 49:236–243, 1989.
- [29627] 3179. Comer, K. A.; Falany, J. L.; Falany, C. N.: Cloning and expression of human liver dehydroepiandrosterone sulphotransferase. *Biochem. J.* 289: 233–240, 1993.
- [29628] 3180. Durocher, F.; Morissette, J.; Dufort, I.; Simard, J.; Luu-The, V.: Genetic linkage mapping of the dehydroepiandrosterone sulfotransferase (STD) gene on the chromosome 19q13.3 region. *Genomics* 29: 781–783, 1995.
- [29629] 3181. Kong, A.-N. T.; Yang, L.; Ma, M.; Tao, D.; Bjornsson, T. D.: Molecular cloning of the alcohol/hydroxysteroid form (hSTa) of sulfotransferase from human liver. *Biochem.*

Biophys. Res. Commun. 187: 448–454, 1992.

- [29630] 3182.Otterness, D.; Mohrenweiser, H. W.; Brandriff, B. F.; Weinshilboum, R. M.: Dehydroepiandrosterone sulfotransferase gene (STD): localization to human chromosome 19q13.3. Cytogenet. Cell Genet. 70: 45–47, 1995.
- [29631] 3183.Otterness, D. M.; Her, C.; Aksoy, S.; Kimura, S.; Wieben, E. D.; Weinshilboum, R. M.: Human dehydroepiandrosterone sulfotransferase gene: molecular cloning and structural characterization. DNA Cell Biol. 14: 331–341, 1995.
- [29632] 3184.Otterness, D. M.; Wieben, E. D.; Wood, T. C.; Watson, W. G.; Madden, B. J.; McCormick, D. J.; Weinshilboum, R. M.: Human liver dehydroepiandrosterone sulfotransferase: molecular cloning and expression of cDNA. Molec. Pharm. 41: 865–872, 1992.
- [29633] 3185.von Lindern, M.; Fornerod, M.; van Baal, S.; Jaegle, M.; de Wit, T.; Buijs, A.; Grosveld, G.: The translocation (6;9), associated with a specific subtype of acute myeloid leukemia, results in the fusion of two genes, dek and can, and the expression of a chimeric, leukemia-specific dek-can mRNA. Molec. Cell. Biol. 12: 1687–1697, 1992.
- [29634] 3186.Schulte-Körne, G.; Grimm, T.; Nothen, M. M.; Müller-Myhsok, B.; Cichon, S.; Vogt, I. R.; Propping, P.;

Remschmidt, H.: Evidence for linkage of spelling disability to chromosome 15. (Letter) *Am. J. Hum. Genet.* 63: 279–282, 1998.

[29635] 3187. Yoon, J.-W.; Yoon, C.-S.; Lim, H.-W.; Huang, Q. Q.; Kang, Y.; Pyun, K. H.; Hirasawa, K.; Sherwin, R. S.; Jun, H.-S.: Control of autoimmune diabetes in NOD mice by GAD expression or suppression in beta cells. *Science* 284: 1183–1187, 1999.

[29636] 3188. Nagase, T.; Ishikawa, K.; Suyama, M.; Kikuno, R.; Hirose, M.; Miyajima, N.; Tanaka, A.; Kotani, H.; Nomura, N.; Ohara, O.: Prediction of the coding sequences of unidentified human genes. XII. The complete sequences of 100 new cDNA clones from brain which code for large proteins in vitro. *DNA Res.* 5: 355–364, 1998.

[29637] 3189. Clancy, K. P.; Berger, R.; Cox, M.; Bleskan, J.; Walton, K. A.; Hart, I.; Patterson, D.: Localization of the L-glutamine synthetase gene to chromosome 1q23. *Genomics* 38: 418–420, 1996.

[29638] 3190. Gibbs, C. S.; Campbell, K. E.; Wilson, R. H.: Sequence of a human glutamine synthetase cDNA. *Nucleic Acids Res.* 15: 6293 only, 1987.

[29639] 3191. Gunnersen, D.; Haley, B.: Detection of glutamine synthetase in the cerebrospinal fluid of Alzheimer diseased

patients: a potential diagnostic biochemical marker. Proc. Nat. Acad. Sci. 89: 11949–11953, 1992.

- [29640] 3192. Helou, K.; Das, A. T.; Lamers, W. H.; Hoovers, J. M. N.; Szpirer, C.; Szpirer, J.; Klinga-Levan, K.; Levan, G.: FISH mapping of three ammonia metabolism genes (Glul, Cps1, Glud1) in rat, and the chromosomal localization of GLUL in human and Cps1 in mouse. Mammalian Genome 8:362–364, 1997.
- [29641] 3193. Pesole, G.; Bozzetti, M. P.; Lanave, C.; Preparata, G.; Saccone, C.: Glutamine synthetase gene evolution: a good molecular clock. Proc. Nat. Acad. Sci. 88: 522–526, 1991.
- [29642] 3194. Wang, Y.; Kudoh, J.; Kubota, R.; Asakawa, S.; Minoshima, S.; Shimizu, N.: Chromosomal mapping of a family of human glutamine synthetase genes: functional gene (GLUL) on 1q25, pseudogene (GLULP) on 9p13, and three related genes (GLULL1, GLULL2, GLULL3) on 5q33, 11p15, and 11q24. Genomics 37: 195–199, 1996.
- [29643] 3195. Smith, M. D.; Dawson, S. J.; Latchman, D. S.: Inhibition of neuronal process outgrowth and neuronal specific gene activation by the Brn-3b transcription factor. J. Biol. Chem. 272: 1382–1388, 1997.
- [29644] 3196. Horisberger, M. A.; Wathélet, M.; Szpirer, J.; Szpirer, C.; Islam, Q.; Levan, G.; Huez, G.; Content, J.: cDNA cloning

and assignment to chromosome 21 of IFI-78K gene, the human equivalent of murine Mx gene. *Somat. Cell Molec. Genet.* 14: 123–131, 1988.

[29645] 3197. Huber, P.; Aebi, M.; Grob, R.; Pravtcheva, D.; Ruddle, F.; Haller, O.: Chromosomal localization of two human Mx genes. (Abstract) *Experientia* 44:A84 only, 1988.

[29646] 3198. Li, Y.; Youssoufian, H.: MxA overexpression reveals a common genetic link in four Fanconi anemia complementation groups. *J. Clin. Invest.* 100:2873–2880, 1997.

[29647] 3199. Winqvist, R.; Lundstrom, K.; Salminen, M.; Laatikainen, M.; Ulmanen, I.: Mapping of human catechol-O-methyltransferase gene to 22q11.2 and detection of a frequent RFLP with BglI. (Abstract) *Cytogenet. Cell Genet.* 58: 2051 only, 1991.

[29648] 3200. Bu, L.; Jin, Y.; Shi, Y.; Chu, R.; Ban, A.; Eiberg, H.; Andres, L.; Jiang, H.; Zheng, G.; Qian, M.; Cui, B.; Xia, Y.; Liu, J.; Hu, L.; Zhao, G.; Hayden, M. R.; Kong, X.: Mutant DNA-binding domain of HSF4 is associated with autosomal dominant lamellar and Marner cataract. *Nature Genet.* 31: 276–278, 2002.

[29649] 3201. Eiberg, H.; Marner, E.; Rosenberg, T.; Mohr, J.: Marner's cataract (CAM) assigned to chromosome 16: linkage to haptoglobin. *Clin. Genet.* 34:272–275, 1988.

- [29650] 3202.Marner, E.: A family with eight generations of hereditary cataract. *ActaOphthal.* 27: 537–551, 1949.
- [29651] 3203.Pokutta, S.; Weis, W. I.: Structure of the dimerization and beta-catenin-bindingregion of alpha-catenin. *Molec. Cell* 5: 533–543, 2000.
- [29652] 3204.Bailey, A.; Norris, A. L.; Leek, J. P.; Clissold, P. M.; Carr,I. M.; Ogilvie, D. J.; Morrison, J. F. J.; Meredith, D. M.; Markham,A. F.: Yeast artificial chromosome cloning of the beta-catenin locus on human chromosome 3p21–22. *Chromosome Res.* 3: 201–203, 1995.
- [29653] 3205.Blaker, H.; Hofmann, W. J.; Rieker, R. J.; Penzel, R.; Graf, M.;Otto, H. F.: Beta-catenin accumulation and mutation of the CTNNB1gene in hepatoblastoma. *Genes Chromosomes Cancer* 25: 399–402, 1999.
- [29654] 3206.Chan, T. A.; Wang, Z.; Dang, L. H.; Vogelstein, B.; Kinzler, K.W.: Targeted inactivation of CTNNB1 reveals unexpected effects of beta-catenin mutation. *Proc. Nat. Acad. Sci.* 99: 8265–8270, 2002.
- [29655] 3207.Chenn, A.; Walsh, C. A.: Regulation of cerebral cortical size by control of cell cycle exit in neural precursors. *Science* 297:365–369, 2002.
- [29656] 3208.Eastman, Q.; Grosschedl, R.: Regulation of LEF-1/TCF transcription factors by Wnt and other signals. *Curr.*

Opin. Cell Biol. 11: 233–240,1999.

- [29657] 3209.Gat, U.; DasGupta, R.; Degenstein, L.; Fuchs, E.: De novo hairfollicle morphogenesis and hair tumors in mice expressing a truncatedbeta–catenin in skin. Cell 95: 605–614, 1998.
- [29658] 3210.Asakura, H.; Zwain, I. H.; Yen, S. S. C.: Expression of genesencoding corticotropin–releasing factor (CRF), type 1 CRF receptor,and CRF–binding protein and localization of the gene products in thehuman ovary. J. Clin. Endocr. Metab. 82: 2720–2725, 1997.
- [29659] 3211.Lynch, E. D.; Lee, M. K.; Morrow, J. E.; Welcsh, P. L.; Leon, P.E.; King, M.–C.: Nonsyndromic deafness DFNA1 associated with mutationof the human homolog of the Drosophila gene diaphanous. Science 278:1315–1318, 1997.
- [29660] 3212.Hamann, J.; Vogel, B.; van Schijndel, G. M.; van Lier, R. A.:The seven–span transmembrane receptor CD97 has a cellular ligand (CD55,DAF). J. Exp. Med. 184: 1185–1189, 1996.
- [29661] 3213.Hourcade, D.; Garcia, A. D.; Post, T. W.; Taillon–Miller, P.; Holers,V. M.; Wagner, L. M.; Bora, N. S.; Atkinson, J. P.: Analysis of thehuman regulators of complement activation (RCA) gene cluster withyeast artificial chromo–

somes (YACs). *Genomics* 12: 289–300, 1992.

- [29662] 3214. Lemons, R. S.; Le Beau, M. M.; Lublin, D. M.; Holers, V. M.; Tykocinski, M. L.; Medof, M. E.; Atkinson, J. P.: The gene encoding decay-accelerating factor (DAF) is located in the complement regulatory locus on the long arm of chromosome 1. (Abstract) *Cytogenet. Cell Genet.* 46:646–647, 1987.
- [29663] 3215. Lublin, D. M.; Lemons, R. S.; Le Beau, M. M.; Holers, V. M.; Tykocinski, M. L.; Medof, M. E.; Atkinson, J. P.: The gene encoding decay-accelerating factor (DAF) is located in the complement regulatory locus on the long arm of chromosome 1. (Abstract) *Clin. Res.* 35: 460A only, 1987.
- [29664] 3216. Swanson, J. M.; Flodman, P.; Kennedy, J.; Spence, M. A.; Moyzis, R.; Schuck, S.; Murias, M.; Moriarty, J.; Barr, C.; Smith, M.; Posner, M.: Dopamine genes and ADHD. *Neurosci. Behav. Rev.* 24: 21–25, 2000.
- [29665] 3217. Swanson, J. M.; Sunohara, G. A.; Kennedy, J. L.; Regino, R.; Fineberg, E.; Wigal, T.; Lerner, M.; Williams, L.; LaHoste, G. J.; Wigal, S.: Association of the dopamine receptor D4 (DRD4) gene with a refined phenotype of attention deficit hyperactivity disorder (ADHD): a family-based approach. *Molec. Psychiat.* 3: 38–41, 1998.
- [29666] 3218. Tomitaka, M.; Tomitaka, S.; Otuka, Y.; Kim, K.;

Matuki, H.; Sakamoto, K.; Tanaka, A.: Association between novelty seeking and dopamine receptor D4 (DRD4) exon III polymorphism in Japanese subjects. *Am. J. Med. Genet. (Neuropsychiat. Genet.)* 88: 469–471, 1999.

[29667] 3219. Van Tol, H. H. M.; Bunzow, J. R.; Guan, H. C.; Sunahara, R. K.; Seeman, P.; Niznik, H. B.; Civelli, O.: Cloning of the gene for a human dopamine D4 receptor with high affinity for the antipsychotic clozapine. *Nature* 350: 610–614, 1991.

[29668] 3220. Van Tol, H. H. M.; Wu, C. M.; Guan, H.-C.; Ohara, K.; Bunzow, J. R.; Civelli, O.; Kennedy, J.; Seeman, P.; Niznik, H. B.; Jovanovic, V.: Multiple dopamine D4 receptor variants in the human population. *Nature* 358: 149–152, 1992.

[29669] 3221. Wiesbeck, G. A.; Mauerer, C.; Thome, J.; Jakob, F.; Boening, J.: Neuroendocrine support for a relationship between 'novelty seeking' and dopaminergic function in alcohol-dependent men. *Psychoneuroendocrinology* 20: 755–761, 1995.

[29670] 3222. Beischlag, T. V.; Marchese, A.; Meador-Woodruff, J. H.; Damask, S. P.; O'Dowd, B. F.; Tyndale, R. F.; Van Tol, H. H. M.; Seeman, P.; Niznik, H. B.: The human dopamine D5 receptor gene: cloning and characterization of the 5-prime

flanking and promoter region. *Biochemistry* 34:
5960–5970, 1995.

- [29671] 3223. Eubanks, J. H.; Altherr, M.; Wagner-McPherson, C.; McPherson, J. D.; Wasmuth, J. J.; Evans, G. A.: Localization of the D5 dopaminereceptor gene to human chromosome 4p15.1–p15.3, centromeric to the Huntington's disease locus. *Genomics* 12: 510–516, 1992.
- [29672] 3224. Grandy, D. K.; Allen, L. J.; Zhang, Y.; Magenis, R. E.; Civelli, O.: Chromosomal localization of three human D5 dopamine receptorgenes. *Genomics* 13: 968–973, 1992.
- [29673] 3225. Grandy, D. K.; Zhang, Y.; Bouvier, C.; Zhou, Q.–Y.; Johnson, R. A.; Allen, L.; Buck, K.; Bunzow, J. R.; Salon, J.; Civelli, O.: Multiple human D5 dopamine receptor genes: a functional receptor and two pseudogenes. *Proc. Nat. Acad. Sci.* 88: 9175–9179, 1991.
- [29674] 3226. Grosson, C. L. S.; MacDonald, M. E.; Duyao, M. P.; Ambrose, C. M.; Roffler-Tarlov, S.; Gusella, J. F.: Syntenic conservation of the Huntington's disease gene and surrounding loci on mouse chromosome 5. *Mammalian Genome* 5: 424–428, 1994.
- [29675] 3227. Liu, F.; Wan, Q.; Pristupa, Z. B.; Yu, X.–M.; Wang, Y. T.; Niznik, H. B.: Direct protein–protein coupling enables cross–talk between dopamine D5 and gamma–

aminobutyric acid A receptors. *Nature* 403:274–280, 2000.

- [29676] 3228. Misbahuddin, A.; Placzek, M. R.; Chaudhuri, K. R.; Wood, N. W.; Bhatia, K. P.; Warner, T. T.: A polymorphism in the dopamine receptor DRD5 is associated with blepharospasm. *Neurology* 58: 124–126, 2002.
- [29677] 3229. Placzek, M. R.; Misbahuddin, A.; Chaudhuri, K. R.; Wood, N. W.; Bhatia, K. P.; Warner, T. T.: Cervical dystonia is associated with a polymorphism in the dopamine (D5) receptor gene. *J. Neurol. Neurosurg. Psychiat.* 71: 262–264, 2001.
- [29678] 3230. Polymeropoulos, M. H.; Xiao, H.; Merrill, C. R.: The human D5 dopamine receptor (DRD5) maps on chromosome 4. *Genomics* 11: 777–778, 1991.
- [29679] 3231. Sherrington, R.; Mankoo, B.; Attwood, J.; Kalsi, G.; Curtis, D.; Buetow, K.; Povey, S.; Gurling, H.: Cloning of the human dopamine D5 receptor gene and identification of a highly polymorphic microsatellite for the DRD5 locus that shows tight linkage to the chromosome 4p reference marker RAF1P1. *Genomics* 18: 423–425, 1993.
- [29680] 3232. Tiberi, M.; Jarvie, K. R.; Silvia, C.; Falardeau, P.; Gingrich, J. A.; Godinot, N.; Bertrand, L.; Yang-Feng, T. L.; Freneau, R. T., Jr.; Caron, M. G.: Cloning, molecular char-

acterization, and chromosomal assignment of a gene encoding a second D-1 dopamine receptor subtype: differential expression pattern in rat brain compared with the D-1A receptor. *Proc. Nat. Acad. Sci.* 88: 7491-7495, 1991.

- [29681] 3233. Weinshank, R. L.; Adham, N.; Macchi, M.; Olsen, M. A.; Branchek, T. A.; Hartig, P. R.: Molecular cloning and characterization of a high affinity dopamine receptor (D-1-beta) and its pseudogene. *J. Biol. Chem.* 266: 22427-22435, 1991.
- [29682] 3234. Byerley, W.; Hoff, M.; Holik, J.; Caron, M. G.; Giros, B.: VNTR polymorphism for the human dopamine transporter gene (DAT1). *Hum. Molec. Genet.* 2: 335, 1993.
- [29683] 3235. Cook, E. H., Jr.; Stein, M. A.; Krasowski, M. D.; Cox, N. J.; Olkon, D. M.; Kieffer, J. E.; Leventhal, B. L.: Association of attention-deficit disorder and the dopamine transporter gene. *Am. J. Hum. Genet.* 56:993-998, 1995.
- [29684] 3236. Doucette-Stamm, L.; Blakely, D. J.; Tian, J.; Mockus, S.; Mao, J.: Population genetic study of the human dopamine transporter gene (DAT1). *Genet. Epidemiol.* 12: 303-308, 1995.
- [29685] 3237. Gainetdinov, R. R.; Wetsel, W. C.; Jones, S. R.; Levin, E. D.; Jaber, M.; Caron, M. G.: Role of serotonin in the

paradoxical calming effect of psychostimulants on hyperactivity. *Science* 283: 397–401, 1999.

- [29686] 3238. Gelernter, J.; Vandenbergh, D.; Kruger, S. D.; Pauls, D. L.; Kurlan, R.; Pakstis, A. J.; Kidd, K. K.; Uhl, G.: The dopamine transporter protein gene (SLC6A3): primary linkage mapping and linkage studies in Tourette syndrome. *Genomics* 30: 459–463, 1995.
- [29687] 3239. Gill, M.; Daly, G.; Heron, S.; Hawi, Z.; Fitzgerald, M.: Confirmation of association between attention deficit hyperactivity disorder and a dopamine transporter polymorphism. *Molec. Psychiat.* 2: 311–313, 1997.
- [29688] 3240. Giros, B.; El Mestikawy, S.; Godinot, N.; Zheng, K.; Han, H.; Yang-Feng, T.; Caron, M. G.: Cloning, pharmacological characterization, and chromosome assignment of the human dopamine transporter. *Molec. Pharm.* 42: 383–390, 1992.
- [29689] 3241. Giros, B.; Jaber, M.; Jones, S. R.; Wightman, R. M.; Caron, M. G.: Hyperlocomotion and indifference to cocaine and amphetamine in mice lacking the dopamine receptor. *Nature* 370: 606–612, 1996.
- [29690] 3242. Goldman, D.: Dopamine transporter, alcoholism and other diseases. *Nature Med.* 1: 624–625, 1995.
- [29691] 3243. Korn, W. T.; Schatzki, S. C.; DiSciullo, A. J.; Scully, R.

E.: Papillary cystadenoma of the broad ligament in von Hippel–Lindaudisease. *Am. J. Obstet. Gynec.* 163: 596–598, 1990.

[29692] 3244.Isnard, R.; Kalotka, H.; Durr, A.; Cossee, M.; Schmitt, M.; Pousset,F.; Thomas, D.; Brice, A.; Koenig, M.; Komajda, M.: Correlation betweenleft ventricular hypertrophy and GAA trinucleotide repeat length inFriedreich's ataxia. *Circulation* 95: 2247–2249, 1997.

[29693] 3245.Koutnikova, H.; Campuzano, V.; Foury, F.; Dolle, P.; Cazzalini,O.; Koenig, M.: Studies of human, mouse and yeast homologues indicatea mitochondrial function for frataxin. *Nature Genet.* 16: 345–351,1997.

[29694] 3246.Kromberg, J. G. R.; Jenkins, T.: Albinism in the South AfricanNegro. III. Genetic counselling issues. *J. Biosoc. Sci.* 16: 99–108,1984.

[29695] 3247.Kugelman, T. P.; Van Scott, E. J.: Tyrosinase activity in melanocytesof human albinos. *J. Invest. Derm.* 37: 73–76, 1961.

[29696] 3248.Lee, S.–T.; Nicholls, R. D.; Bunday, S.; Laxova, R.; Musarella,M.; Spritz, R. A.: Mutations of the P gene in oculocutaneous albinism,ocular albinism, and Prader–Willi syndrome plus albinism. *New Eng.J. Med.* 330: 529–534, 1994.

- [29697] 3249.Lee, S.-T.; Nicholls, R. D.; Jong, M. T. C.; Fukai, K.; Spritz, R. A.: Organization and sequence of the human P gene and identification of a new family of transport proteins. *Genomics* 26: 354–363, 1995.
- [29698] 3250.Lee, S.-T.; Nicholls, R. D.; Schnur, R. E.; Guida, L. C.; Lu-Kuo, J.; Spinner, N. B.; Zackai, E. H.; Spritz, R. A.: Diverse mutations of the P gene among African-Americans with type II (tyrosinase-positive) oculocutaneous albinism (OCA2). *Hum. Molec. Genet.* 3: 2047–2051, 1994.
- [29699] 3251.Ludowese, C. J.; Thompson, K. J.; Sekhon, G. S.; Pauli, R. M.: Absence of predictable phenotypic expression in proximal 15q duplications. *Clin. Genet.* 40: 194–201, 1991.
- [29700] 3252.Manga, P.: Identification and molecular characterization of the genes for brown and rufous oculocutaneous albinism in southern Africa. Ph.D. Thesis: University of the Witwatersrand, Johannesburg , 1997.
- [29701] 3253.Manga, P.; Kromberg, J. G. R.; Turner, A.; Jenkins, T.; Ramsay, M.: In southern Africa, brown oculocutaneous albinism (BOCA) maps to the OCA2 locus on chromosome 15q: P-gene mutations identified. *Am. J. Hum. Genet.* 68: 782–787, 2001.
- [29702] 3254.Baker, E.; Chen, L. Z.; Smith, C. A.; Callen, D. F.;

Goodwin, R.;Sutherland, G. R.: Chromosomal location of the human tumor necrosisfactor receptor genes. Cyto-genet. Cell Genet. 57: 117–118, 1991.

[29703] 3255.Chan, F. K.–M.; Chun, H. J.; Zheng, L.; Siegel, R. M.; Bui, K.L.; Lenardo, M. J.: A domain in TNF receptors that mediates ligand–independentreceptor assembly and sig-naling. Science 288: 2351–2354, 2000.

[29704] 3256.Atuk, N. O.; McDonald, T.; Wood, T.; Carpenter, J. T.; Walzak,M. P.; Donaldson, M.; Gillenwater, J. Y.; Turner, S. M.; Westfall,V.: Familial pheochromocytoma, hypercal-cemia, and von Hippel–Lindaudisease: a ten–year study of a large family. Medicine 58: 209–218,1979.

[29705] 3257.Bender, B. U.; Altehofer, C.; Januszewicz, A.; Gart-ner, R.; Schmidt,H.; Hoffmann, M. M.; Heidemann, P. H.; Neumann, H. P. H.: Functioningthoracic paraganglioma: association with Von Hippel–Lindau syndrome. J.Clin. En-docr. Metab. 82: 3356–3360, 1997.

[29706] 3258.Bender, B. U.; Eng, C.; Olschewski, M.; Berger, D. P.; Laubenberger,J.; Altehofer, C.; Kirste, G.; Orszagh, M.; van Velthoven, V.; Miosczka,H.; Schmidt, D.; Neumann, H. P. H.: VHL c.505 T–C mutation confers a high age related penetrance but no increased overall mortality. J.Med. Genet. 38: 508–514, 2001.

- [29707] 3259.Bender, B. U.; Gutsche, M.; Glasker, S.; Muller, B.; Kirste, G.;Eng, C.; Neumann, H. P. H.: Differential genetic alterations in vonHippel–Lindau syndrome–associated and sporadic pheochromocytomas. *J.Clin. Endocr. Metab.* 85: 4568–4574, 2000.
- [29708] 3260.Bonnet, P.; Dechaume, J.; Blanc, E.: L'anevrisme cirsoide de l'aretine l'anevrisme racemeux, ses relations avec l'anevrisme cirsoide de la face et l'anevrisme cirsoide du cerveau. *Bull. Soc. Ophtal.Franc.* 51: 521–524, 1938.
- [29709] 3261.Bradley, J. F.; Collins, D. L.; Schimke, R. N.; Parrott, H. N.;Rothberg, P. G.: Two distinct phenotypes caused by two different missense mutations in the same codon of the VHL gene. *Am. J. Med.Genet.* 87: 163–167, 1999.
- [29710] 3262.Brauch, H.; Kishida, T.; Glavac, D.; Chen, F.; Pausch, F.; Hofler, H.; Latif, F.; Lerman, M. I.; Zbar, B.; Neumann, H. P. H.: Von Hippel–Lindau(VHL) disease with pheochromocytoma in the Black Forest region of Germany: evidence for a founder effect. *Hum. Genet.* 95: 551–556, 1995.
- [29711] 3263.Brown, D. G.; Hilal, S. K.; Tenner, M. S.: Wyburn–Mason syndrome. *Arch.Neurol.* 28: 67–68, 1973.
- [29712] 3264.Cahill, G. F.; Melicow, M. M.; Guerry, D.: The renal lesions in von Hippel–Lindau's disease. *Trans. Am. Assoc. Genitourinary Surg.* 35:271–281, 1942.

- [29713] 3265.Cameron, S. J.; Doig, A.: Cerebellar tumors presented with clinical features of pheochromocytoma. *Lancet* I: 492–494, 1970.
- [29714] 3266.Chapman, R. C.; Diaz–Perez, R.: Pheochromocytoma associated with cerebellar hemangioblastoma. *J.A.M.A.* 182: 1014–1017, 1962.
- [29715] 3267.Chen, F.; Slife, L.; Kishida, T.; Mulvihill, J.; Tisherman, S.E.; Zbar, B.: Genotype–phenotype correlation in von Hippel–Lindau disease: identification of a mutation associated with VHL type 2A. *J.Med. Genet.* 33: 716–717, 1996.
- [29716] 3268.Christoferson, L. A.; Gustafson, M. B.; Petersen, A. G.: VonHippel–Lindau's disease. *J.A.M.A.* 178: 280–282, 1961.
- [29717] 3269.Chen, F.; Kishida, T.; Yao, M.; Hustad, T.; Glavac, D.; Dean, M.; Gnarr, J. R.; Orcutt, M. L.; Duh, F. M.; Glenn, G.; Green, J.; Hsia, Y. E.; Lamiell, J.; Li, H.; Wei, M. H.; Schmidt, L.; Tory, K.; Kuzman, I.; Stackhouse, T.; Latif, F.; Linehan, W. M.; Lerman, M.; Zbar, B.: Germline mutations in the von Hippel–Lindau disease tumor suppressor gene: correlations with phenotype. *Hum. Mutat.* 5: 66–75, 1995.
- [29718] 3270.Collins, E. T.: Intra–ocular growths (two cases, brother and sister, with peculiar vascular new growth,

probably primarily retinal,affecting both eyes). Trans. Ophthal. Soc. U.K. 14: 141–149, 1894.

- [29719] 3271.Crossey, P. A.; Foster, K.; Richards, F. M.; Phipps, M. E.; Latif,F.; Tory, K.; Jones, M. H.; Bentley, E.; Kumar, R.; Lerman, M. I.;Zbar, B.; Affara, N. A.; Ferguson–Smith, M. A.; Maher, E. R.: Moleculargenetic investigations of the mechanism of tumourigenesis in von Hippel–Lindaudisease: analysis of allele loss in VHL tumours. Hum. Genet. 93:53–58, 1994.
- [29720] 3272.Crossey, P. A.; Richards, F. M.; Foster, K.; Green, J. S.; Prowse,A.; Latif, F.; Lerman, M. I.; Zbar, B.; Affara, N. A.; Ferguson–Smith,M. A.; Maher, E. R.: Identification of intragenic mutations in thevon Hippel–Lindau disease tumour suppressor gene and correlation withdisease phenotype. Hum. Molec. Genet. 3: 1303–1308, 1994.
- [29721] 3273.Cushing, H.; Bailey, P.: Hemangiomas of cerebellum and retina(Lindau's disease), with the report of a case. Arch. Ophthal. 57:447–463, 1928.
- [29722] 3274.Davies, D. R.; Norman, A. M.; Whitehouse, R. W.; Evans, D. G.R.: Non–expression of von Hippel–Lindau phenotype in an obligategene carrier. Clin. Genet. 45: 104–106, 1994.
- [29723] 3275.Decker, H.–J. H.; Neumann, H. P. H.; Walter, T. A.;

Sandberg,A. A.: 3p involvement in a renal cell carcinoma in von Hippel–Lindausyndrome: region of tumor break–point clustering on 3p? Cancer Genet.Cytogenet. 33: 59–65, 1988.

- [29724] 3276.Duan, D. R.; Humphrey, J. S.; Chen, D. Y. T.; Weng, Y.; Sukegawa,J.; Lee, S.; Gnarra, J. R.; Linehan, W. M.; Klausner, R. D.: Characterizationof the VHL tumor sup–pressor gene product: localization, complex forma–tion,and the effect of natural inactivating mutations. Proc. Nat. Acad.Sci. 92: 6459–6463, 1995.
- [29725] 3277.Duan, D. R.; Pause, A.; Burgess, W. H.; Aso, T.; Chen, D. Y. T.;Garrett, K. P.; Conaway, R. C.; Conaway, J. W.; Linehan, W. M.; Klausner,R. D.: Inhibition of transcription elongation by the VHL tumor suppressorprotein. Science 269: 1402–1406, 1995.
- [29726] 3278.Schoof, E.; Girstl, M.; Frobenius, W.; Kirschbaum, M.; Dorr, H.G.; Rascher, W.; Dotsch, J.: Decreased gene ex–pression of 11–beta–hydroxysteroiddehydrogenase type 2 and 15–hydroxyprostaglandin dehydrogenase inhuman placenta of patients with preeclampsia. J. Clin. Endocr. Metab. 86:1313–1317, 2001.
- [29727] 3279.Hurvitz, J. R.; Suwairi, W. M.; Van Hul, W.; El–Shanti, H.; Superti–Furga,A.; Roudier, J.; Holderbaum, D.; Pauli, R.

M.; Herd, J. K.; Van Hul, E.; Rezai-Delui, H.; Legius, E.; Le Merrer, M.; Al-Alami, J.; Bahabri, S. A.; Warman, M. L.: Mutations in the CCN gene family member WISP3 cause progressive pseudorheumatoid dysplasia. *Nature Genet.* 23:94–98, 1999.

[29728] 3280. Spranger, J.; Albert, C.; Schilling, F.; Bartsocas, C.; Stoss, H.: Progressive pseudorheumatoid arthritis of childhood (PPAC): a hereditary disorder simulating rheumatoid arthritis. *Europ. J. Pediatr.* 140:34–40, 1983.

[29729] 3281. Bahabri, S. A.; Suwairi, W. M.; Laxer, R. M.; Polinkovsky, A.; Dalaan, A. A.; Warman, M. L.: The camptodactyly–arthropathy–coxavara–pericarditis syndrome: clinical features and genetic mapping to human chromosome 1. *Arthritis Rheum.* 41: 730–735, 1998.

[29730] 3282. Marcelino, J.; Carpten, J. D.; Suwairi, W. M.; Gutierrez, O. M.; Schwartz, S.; Robbins, C.; Sood, R.; Makalowska, I.; Baxevanis, A.; Johnstone, B.; Laxer, R. M.; Zemel, L.; and 13 others: CACP, encoding a secreted proteoglycan, is mutated in camptodactyly–arthropathy–coxavara–pericarditis syndrome. *Nature Genet.* 23: 319–322, 1999.

[29731] 3283. Tannin, G. M.; Agarwal, A. K.; Monder, C.; New, M. I.; White, P. C.: The human gene for 11–beta–hydroxysteroid dehydrogenase: structure, tissue distribution, and chro–

mosomal localization. J. Biol. Chem. 266:16653–16658, 1991.

- [29732] 3284.Andrechek, E. R.; Hardy, W. R.; Girgis–Gabardo, A. A.; Perry, R.L. S.; Butler, R.; Graham, F. L.; Kahn, R. C.; Rudnicki, M. A.; Muller,W. J.: ErbB2 is required for muscle spindle and myoblast cell survival. Molec.Cell. Biol. 22: 4714–4722, 2002.
- [29733] 3285.Bargmann, C. I.; Hung, M.–C.; Weinberg, R. A.: The NEU oncogeneencodes an epidermal growth factor receptor–related protein. Nature 319:226–230, 1986.
- [29734] 3286.Chan, R.; Hardy, W. R.; Laing, M. A.; Hardy, S. E.; Muller, W.J.: The catalytic activity of the ErbB–2 receptor tyrosine kinaseis essential for embryonic development. Molec. Cell. Biol. 22: 1073–1078,2002.
- [29735] 3287.Coussens, L.; Yang–Feng, T. L.; Liao, Y.–C.; Chen, E.; Gray, A.;McGrath, J.; Seeburg, P. H.; Libermann, T. A.; Schlessinger, J.; Francke,U.; Levinson, A.; Ullrich, A.: Tyrosine kinase receptor with extensivehomology to EGF receptor shares chromosomal location with NEU oncogene. Science 230:1132–1139, 1985.
- [29736] 3288.Crone, S. A.; Zhao, Y.–Y.; Fan, L.; Gu, Y.; Minami–sawa, S.; Liu,Y.; Peterson, K. L.; Chen, J.; Kahn, R.; Con–dorelli, G.; Ross, J.,Jr.; Chien, K. R.; Lee, K.–F.: ErbB2 is es–

sential in the prevention of dilated cardiomyopathy. *Nature Med.* 8: 459–465, 2002.

[29737] 3289. Dankort, D.; Maslikowski, B.; Warner, N.; Kanno, N.; Kim, H.; Wang, Z.; Moran, M. F.; Oshima, R. G.; Cardiff, R. D.; Muller, W. J.: Grb2 and Shc adapter proteins play distinct roles in Neu (ErbB-2) induced mammary tumorigenesis: implications for human breast cancer. *Molec. Cell. Biol.* 21: 1540–1551, 2001.

[29738] 3290. De Boer, J. G.: A new mutator phenotype in breast cancer? (Commentary) *Proc. Nat. Acad. Sci.* 99: 3368–3369, 2002.

[29739] 3291. De Placido, S.; Carlomagno, C.; De Laurentiis, M.; Bianco, A. R.: C-erbB2 expression predicts tamoxifen efficacy in breast cancer patients. *Breast Cancer Res. Treat.* 52: 55–64, 1998.

[29740] 3292. Di Fiore, P. P.; Pierce, J. H.; Kraus, M. H.; Segatto, O.; King, C. R.; Aaronson, S. A.: erbB-2 is a potent oncogene when overexpressed in NIH/3T3 cells. *Science* 237: 178–182, 1987.

[29741] 3293. Doherty, J. K.; Bond, C.; Jardim, A.; Adelman, J. P.; Clinton, G. M.: The HER-2/neu receptor tyrosine kinase gene encodes a secreted autoinhibitor. *Proc. Nat. Acad. Sci.* 96: 10869–10874, 1999.

- [29742] 3294.Ehsani, A.; Low, J.; Wallace, R. B.; Wu, A. M.: Characterization of a new allele of the human ERBB2 gene by allele-specific competition hybridization. *Genomics* 15: 426–429, 1993.
- [29743] 3295.Francke, U.: Personal Communication. New Haven, Connecticut 4/1988.
- [29744] 3296.Fukushige, S.-I.; Matsubara, K.-I.; Yoshida, M.; Sasaki, M.; Suzuki, T.; Semba, K.; Toyoshima, K.; Yamamoto, T.: Localization of a novel v-erbB-related gene, c-erbB-2, on human chromosome 17 and its amplification in a gastric cancer cell line. *Molec. Cell. Biol.* 6: 955–958, 1986.
- [29745] 3297.Kaneko, Y.; Homma, C.; Maseki, N.; Sakurai, M.; Toyoshima, K.; Yamamoto, T.: Human c-erbB-2 remains on chromosome 17 in band q21 in the 15;17 translocation associated with acute promyelocytic leukemia. *Jpn.J. Cancer Res.* 78: 16–19, 1987.
- [29746] 3298.Lin, W.; Sanchez, H. B.; Deerinck, T.; Morris, J. K.; Ellisman, M.; Lee, K. F.: Aberrant development of motor axons and neuromuscular synapses in erbB2-deficient mice. *Proc. Nat. Acad. Sci.* 97: 1299–1304, 2000.
- [29747] 3299.Liu, S.; Liu, W.; Jakubczak, J. L.; Erexson, G. L.; Tindall, K.R.; Chan, R.; Muller, W. J.; Adhya, S.; Garges, S.;

Merlino, G.:Genetic instability favoring transversions associated with ErbB2–inducedmammary tumorigenesis. Proc. Nat. Acad. Sci. 99: 3770–3775, 2002.

[29748] 3300.Mehta, R. R.; McDermott, J. H.; Hieken, T. J.; Marler, K. C.;Patel, M. K.; Wild, L. D.; Das Gupta, T. K.: Plasma c–erbB2 levelsin breast cancer patients: prognostic significance in predicting responseto chemotherapy. J. Clin. Oncol. 16: 2409–2416, 1998.

[29749] 3301.Morris, J. K.; Lin, W.; Hauser, C.; Marchuk, Y.; Getman, D.; Lee,K.–F.: Rescue of the cardiac defect in ErbB2 mutant mice revealsessential roles of ErbB2 in peripheral nervous system development. Neuron 23:273–283, 1999.

[29750] 3302.Muleris, M.; Almeida, A.; Malfoy, B.; Dutrillaux, B.: Assignmentof v–erb–b2 avian erythroblastic leukemia viral oncogene homolog 2(ERBB2) to human chromosome band 17q21.1 by in situ hybridization. Cytogenet.Cell Genet. 76: 34–35, 1997.

[29751] 3303.Ozcelik, C.; Erdmann, B.; Pilz, B.; Wettschureck, N.; Britsch,S.; Hubner, N.; Chien, K. R.; Birchmeier, C.; Garratt, A. N.: Conditionalmutation of the ErbB2 (HER2) receptor in cardiomyocytes leads to dilatedcardiomyopathy. Proc. Nat. Acad. Sci. 99: 8880–8885, 2002.

[29752] 3304.Papewalis, J.; Nikitin, A. Y.; Rajewsky, M. F.: G to A

polymorphism at amino acid codon 655 of the human erbB-2/HER2 gene. *Nucleic Acids Res.* 19: 5452 only, 1991.

[29753] 3305. Pegram, M. D.; Finn, R. S.; Arzoo, K.; Beryt, M.; Pietras, R.J.; Slamon, D. J.: The effect of HER-2/neu over-expression on chemotherapeutic drug sensitivity in human breast and ovarian cancer cells. *Oncogene* 15:537-547, 1997.

[29754] 3306. Pietras, R. J.; Pegram, M. D.; Finn, R. S.; Maneval, D. A.; Slamon, D. J.: Remission of human breast cancer xenografts on therapy with humanized monoclonal antibody to HER-2 receptor and DNA-reactive drugs. *Oncogene* 17:2235-2249, 1998.

[29755] 3307. Popescu, N. C.; King, C. R.; Kraus, M. H.: Localization of the human erbB-2 gene on normal and rearranged chromosomes 17 to bands q12-21.32. *Genomics* 4: 362-366, 1989.

[29756] 3308. Qiu, Y.; Ravi, L.; Kung, H.-J.: Requirement of ErbB2 for signalling by interleukin-6 in prostate carcinoma cells. *Nature* 393: 83-85, 1998.

[29757] 3309. Semba, K.; Kamata, N.; Toyoshima, K.; Yamamoto, T.: A v-erbB-related protooncogene, c-erbB-2, is distinct from the c-erbB-1/epidermal growth factor-receptor gene

and is amplified in a human salivary gland adenocarcinoma. Proc.Nat. Acad. Sci. 82: 6497–6501, 1985.

[29758] 3310.Slamon, D. J.; Godolphin, W.; Jones, L. A.; Holt, J. A.; Wong,S. G.; Keith, D. E.; Levin, W. J.; Stuart, S. G.; Udove, J.; Ullrich,A.; Press, M. F.: Studies of the HER-2/neu proto-oncogene in humanbreast and ovarian cancer. Science 244: 707–712, 1989.

[29759] 3311.Slamon, D. J.; Leyland-Jones, B.; Shak, S.; Fuchs, H.; Paton,V.; Bajamonde, A.; Fleming, T.; Eiermann, W.; Wolter, J.; Pegram,M.; Baselga, J.; Norton, L.: Use of chemotherapy plus a monoclonalantibody against HER2 for metastatic breast cancer that overexpressesHER2. New Eng. J. Med. 344: 783–792, 2001.

[29760] 3312.Beutler, E.; Kuhl, W.; Gelbart, T.: 6-Phosphogluconolactonasedeficiency, a hereditary erythrocyte enzyme deficiency: possible interactionwith glucose-6-phosphate dehydrogenase deficiency. Proc. Nat. Acad.Sci. 82: 3876–3878, 1985.

[29761] 3313.Benkmann, H.-G.; Paik, Y. K.; Chen, L. Z.; Goedde, H. W.: Polymorphismof 6-PGD in South Korea: a new genetic variant 6-PGD Korea. Hum.Genet. 74: 204–205, 1986.

[29762] 3314.Blake, N. M.; Kirk, R. L.: New genetic variant of

6-phosphogluconate dehydrogenase in Australian aborigines. *Nature* 221: 278 only, 1969.

- [29763] 3315. Bowman, J. E.; Carson, P. E.; Frischer, H.; De Garay, A. L.: Genetic of starch-gel electrophoretic variants of human 6-phosphogluconate dehydrogenase: population and family studies in the United States and in Mexico. *Nature* 210: 811-812, 1966.
- [29764] 3316. Brewer, G. J.; Dern, R. J.: A new inherited enzymatic deficiency of human erythrocytes: 6-phosphogluconate dehydrogenase deficiency. *Am. J. Hum. Genet.* 16: 472-476, 1964.
- [29765] 3317. Burgerhout, W.; Van Someren, H.; Bootsma, D.: Cytological mapping of the genes assigned to the human A1 chromosome by use of radiation-induced chromosome breakage in a human-Chinese hamster hybrid cell line. *Humangenetik* 20: 159-162, 1973.
- [29766] 3318. Davidson, R. G.: Electrophoretic variants of human 6-phosphogluconate dehydrogenase: population and family studies and description of a new variant. *Ann. Hum. Genet.* 30: 355-362, 1967.
- [29767] 3319. Dern, R. J.; Brewer, G. J.; Tashian, R. E.; Shows, T. B.: Hereditary variation of erythrocytic 6-phosphogluconate dehydrogenase. *J. Lab. Clin. Med.* 67: 255-264, 1966.

- [29768] 3320. Douglas, G. R.; McAlpine, P. J.; Hamerton, J. L.: Regional localization of loci for human PGM-1 and 6PGD on human chromosome 1 by use of hybrids of Chinese hamster-human somatic cells. *Proc. Nat. Acad. Sci.* 70:2737-2740, 1973.
- [29769] 3321. Fildes, R. A.; Parr, C. W.: Human red-cell phosphogluconate dehydrogenases. *Nature* 200:890-891, 1963.
- [29770] 3322. Nelson, M. S.: Biochemical and genetic characterization of the Lowell variant: a new phenotype of 6-phosphogluconate dehydrogenase. *Hum. Genet.* 62: 333-336, 1982.
- [29771] 3323. Nevo, S.: A new rare PGD variant, PGD Mediterranean. *Hum. Genet.* 81:199 only, 1989.
- [29772] 3324. Parr, C. W.: Erythrocyte phosphogluconate dehydrogenase polymorphism. *Nature* 210:487-489, 1966.
- [29773] 3325. Parr, C. W.; Fitch, L. I.: Inherited quantitative variations of human phosphogluconate dehydrogenase. *Ann. Hum. Genet.* 30: 339-353, 1967.
- [29774] 3326. Ritter, H.; Toriverdian, G.; Wendt, G. G.; Zilch, I.: Genetic and linkage analysis on 6-PGD. *Humangenetik* 14: 73-75, 1971.
- [29775] 3327. Tariverdian, G.; Ropers, H.-H.; Op't Hof, J.; Ritter, H.: Zur Genetik der 6-Phosphogluconatdehydrogenase (EC:

1.1.1.44): Eine neue Variante F (Freiburg). Humangenetik 10: 355–357, 1970.

[29776] 3328. Weitkamp, L. R.: Genetic linkage relationships of the ADA and 6-PGD loci in 'Humangenetik.' (Letter) Humangenetik 15: 359–360, 1972.

[29777] 3329. Weitkamp, L. R.; Guttormsen, S. A.; Greendyke, R. M.: Genetic linkage between a locus for 6-PGD and the Rh locus: evaluation of possible heterogeneity in the recombination fraction between sexes and among families. Am. J. Hum. Genet. 23: 462–470, 1971.

[29778] 3330. Weitkamp, L. R.; Guttormsen, S. A.; Shreffler, D. C.; Sing, C. F.; Napier, J. A.: Genetic linkage relations of the loci for 6-phosphogluconate dehydrogenase and adenosine deaminase in man. Am. J. Hum. Genet. 22: 216–220, 1970.

[29779] 3331. Westerveld, A.; Meera Khan, P.: Evidence for linkage between human loci for 6-phosphogluconate dehydrogenase and phosphoglucomutase(1) in man–Chinese hamster somatic cell hybrids. Nature 236: 30–32, 1972.

[29780] 3332. McEver, R. P.; Beckstead, J. H.; Moore, K. L.; Marshall-Carlson, L.; Bainton, D. F.: GMP-140, a platelet alpha-granule membrane protein, is also synthesized by vascular endothelial cells and is localized in Weibel–Palade

bodies. J. Clin. Invest. 84: 92–99, 1989.

[29781] 3333.Thomas, K. R.; Capecchi, M. R. :Cell 51: 503–512, 1987.

[29782] 3334.Scita, G.; Nordstrom, J.; Carbone, R.; Tenca, P.; Giardina, G.;Gutkind, S.; Bjarnegard, M.; Betsholtz, C.; Di Fiore, P. P.: EPS8and E3B1 transduce signals from Ras to Rac. Nature 401: 290–293,1999.

[29783] 3335.Remmers, E. F.; Goldmuntz, E. A.; Cash, J. M.; Croford, L. J.;Misiewicz–Poltorak, B.; Zha, H.; Wilder, R. L.: Genetic map of ninepolymorphic loci comprising a single linkage group on rat chromosome10: evidence for linkage conservation with human chromosome 17 andmouse chromosome 11. Genomics 14: 618–623, 1992.

[29784] 3336.Sarraf, P.; Mueller, E.; Smith, W. M.; Wright, H. M.; Kum, J.B.; Aaltonen, L. A.; de la Chapelle, A.; Spiegelman, B. M.; Eng, C.: Loss–of–function mutations in PPAR–gamma associated with human coloncancer. Molec. Cell 3: 799–804, 1999.

[29785] 3337.Scheidler, S.; Fredericks, W. J.; Rauscher, F. J., III; Barr,F. G.; Vogt, P. K.: The hybrid PAX3–FKHR fusion protein of alveolarrhabdomyosarcoma transforms fibroblasts in culture. Proc. Nat. Acad.Sci. 93: 9805–9809, 1996.

[29786] 3338.Sublett, J. E.; Jeon, I–S.; Shapiro, D. N.: The alveolar

rhabdomyosarcomaPAX3/FKHR fusion protein is a transcriptional activator. *Oncogene* 11:545–552, 1995.

[29787] 3339.Whang–Peng, J.; Knutsen, T.; Theil, K.; Horowitz, M. E.; Triche,T.: Cytogenetic studies in subgroups of rhabdomyosarcoma. *GenesChromosomes Cancer* 5: 299–310, 1992.

[29788] 3340.Richard, I.; Broux, O.; Chiannilkulchai, N.; Fougere–ousse, F.; Allamand,V.; Bourg, N.; Brenguier, L.; Devaud, C.; Pasturaud, P.; Roudaut,C.; Lorenzo, F.; Sebastiani–Katchis, C.; Schultz, R. A.; Polymeropoulos,M. H.; Gyapay, G.; Auffray, C.; Beckmann, J. S.: Regional localizationof human chromosome 15 loci. *Genomics* 23: 619–627, 1994.

[29789] 3341.Zuniga, A.; Haramis, A.–P. G.; McMahon, A. P.; Zeller, R.: Signalrelay by BMP antagonism controls the SHH/FGF4 feedback loop in vertebratelimb buds. *Nature* 401: 598–602, 1999.

[29790] 3342.Bao, L.; Gerard, N. P.; Eddy, R. L., Jr.; Shows, T. B.; Gerard,C.: Mapping of genes for the human C5a receptor (C5AR), human FMLPreceptor (FPR), and two FMLP receptor homologue orphan receptors (FPRH1,FPRH2) to chromosome 19. *Genomics* 13: 437–440, 1992.

[29791] 3343.Cohen–Haguenauer, O.; Van Cong, N.; Prud'homme,

J. F.; Jegou-Foubert, C.; Gross, M. S.; De Tand, M. F.; Milgrom, E.; Frezal, J.: A gene expressed in human breast cancer and regulated by estrogen in MCF-7 cells is located on chromosome 21. (Abstract) Cytogenet. Cell Genet. 40:606 only, 1985.

[29792] 3344. Jakowlew, S. B.; Breathnach, R.; Jeltsch, J.-M.; Masiakowski, P.; Chambon, P.: Sequence of the pS2 mRNA induced by estrogen in the human breast cancer cell line MCF-7. Nucleic Acids Res. 12: 2861-2878, 1984.

[29793] 3345. Lefebvre, O.; Chenard, M.-P.; Masson, R.; Linares, J.; Dierich, A.; LeMeur, M.; Wendling, C.; Tomasetto, C.; Chambon, P.; Rio, M.-C.: Gastric mucosa abnormalities and tumorigenesis in mice lacking the pS2 trefoil protein. Science 274: 259-262, 1996.

[29794] 3346. Moisan, J.-P.; Mattei, M.-G.; Mandel, J.-L.: Chromosome localization and polymorphism of an oestrogen-inducible gene specifically expressed in some breast cancers. Hum. Genet. 79: 168-171, 1988.

[29795] 3347. Moisan, J. P.; Mattei, M. G.; Baeteman-Volkel, M. A.; Mattei, J. F.; Brown, A. M. C.; Garnier, J. M.; Jeltsch, J. M.; Masiakowsky, P.; Roberts, M.; Mandel, J. L.: A gene expressed in human mammary tumor cells under estrogen control (BCEI) is located in 21q22.3 and defines an RFLP.

(Abstract) Cytogenet. Cell Genet. 40: 701–702,1985.

- [29796] 3348.Roberts, M.; Wallace, J.; Jeltsch, J.–M.; Berry, M.: The 5–primeflanking region of the human pS2 gene mediates its transcriptionalactivation by estrogen in MCF–7 cells. Biochem. Biophys. Res. Commun. 151:306–313, 1988.
- [29797] 3349.Seib, T.; Blin, N.; Hilgert, K.; Seifert, M.; Theisinger, B.;Engel, M.; Dooley, S.; Zang, K.–D.; Welter, C.: The three human trefoilgenes TFF1, TFF2, and TFF3 are located within a region of 55 kb onchromosome 21q22.3. Genomics 40: 200–202, 1997.
- [29798] 3350.Watkins, P. C.; Tanzi, R. E.; Roy, J.; Stuart, N.; Stanislovitis,P.; Gusella, J. F.: A cosmid genetic linkage map of chromosome 21and localization of the breast cancer estrogen–inducible (BCEI) gene.(Abstract) Am. J. Hum. Genet. 41: A189 only, 1987.
- [29799] 3351.Ponnambalam, S.; Jackson, A. P.; LeBeau, M. M.; Pravtcheva, D.;Ruddle, F. H.; Alibert, C.; Parham, P.: Chromosomal location andsome structural features of human clathrin light–chain genes (CLTAand CLTB). Genomics 24: 440–444, 1994.
- [29800] 3352.Andersen, T. I.; Wooster, R.; Laake, K.; Collins, N.; Warren, W.;Skrede, M.; Eeles, R.; Tveit, K. M.; Johnston, S. R. D.; Dowsett,M.; Olsen, A. O.; Moller, P.; Stratton, M. R.;

Borresen–Dale, A.–L.: Screening for ESR mutations in breast and ovarian cancer patients. *Hum.Mutat.* 9: 531–536, 1997.

[29801] 3353.Auboeuf, D.; Honig, A.; Berget, S. M.; O'Malley, B. W.: Coordinateregulation of transcription and splicing by steroid receptor coregulators. *Science* 298:416–419, 2002.

[29802] 3354.Balleine, R. L.; Hunt, S. M. N.; Clarke, C. L.: Coexpression ofalternatively spliced estrogen and progesterone receptor transcripts in human breast cancer. *J. Clin. Endocr. Metab.* 84: 1370–1377, 1999.

[29803] 3355.Becherini, L.; Gennari, L.; Masi, L.; Mansani, R.; Massart, F.;Morelli, A.; Falchetti, A.; Gonnelli, S.; Fiorelli, G.; Tanini, A.;Brandi, M. L.: Evidence of a linkage disequilibrium between polymorphisms in the human estrogen receptor–alpha gene and their relationship to bone mass variation in postmenopausal Italian women. *Hum. Molec.Genet.* 9: 2043–2050, 2000.

[29804] 3356.Bord, S.; Horner, A.; Beavan, S.; Compston, J.: Estrogen receptors alpha and beta are differentially expressed in developing human bone. *J.Clin. Endocr. Metab.* 86: 2309–2314, 2001.

[29805] 3357.Castagnoli, A.; Maestri, I.; Bernardi, F.; Del Senno, L.:

PvuII RFLP inside the human estrogen receptor gene. *Nucleic Acids Res.* 15:866 only, 1987.

- [29806] 3358. Chaidarun, S. S.; Alexander, J. M.: A tumor-specific truncated estrogen receptor splice variant enhances estrogen-stimulated gene expression. *Molec. Endocr.* 12: 1355–1366, 1998.
- [29807] 3359. Chiang, C.-H.; Cheng, K. W.; Igarashi, S.; Nathwani, P. S.; Leung, P. C. K.: Hormonal regulation of estrogen receptor alpha and beta gene expression in human granulosa-luteal cells in vitro. *J. Clin. Endocr. Metab.* 85: 3828–3839, 2000.
- [29808] 3360. Clark, G. M.; McGuire, W. L.: Steroid receptors and other prognostic factors in primary breast cancer. *Semin. Oncol.* 15 (suppl. 1): 20–25, 1988.
- [29809] 3361. Couse, J. F.; Hewitt, S. C.; Bunch, D. O.; Sar, M.; Walker, V. R.; Davis, B. J.; Korach, K. S.: Postnatal sex reversal of the ovaries in mice lacking estrogen receptors alpha and beta. *Science* 286:2328–2331, 1999.
- [29810] 3362. Davis, V. L.; Chan, C.-C.; Schoen, T. J.; Couse, J. F.; Chader, G. J.; Korach, K. S.: An estrogen receptor repressor induces cataract formation in transgenic mice. *Proc. Nat. Acad. Sci.* 99: 9427–9432, 2002.
- [29811] 3363. Esmaili, B.; Harvey, J. T.; Hewlett, B.: Immunohisto-

chemicalevidence for estrogen receptors in meibomian glands. *Ophthalmology* 107:180–184, 2000.

- [29812] 3364.Fan, S.; Wang, J.-A.; Yuan, R.; Ma, Y.; Meng, Q.; Er-dos, M. R.; Pestell, R. G.; Yuan, F.; Auborn, K. J.; Goldberg, I. D.; Rosen, E.M.: BRCA1 inhibition of estrogen receptor signaling in transfected cells. *Science* 284: 1354–1356, 1999.
- [29813] 3365.Fuqua, S. A. W.; Chamness, G. C.; McGuire, W. L.: Estrogen receptormutations in breast cancer. *J. Cell. Biochem.* 51: 135–139, 1993.
- [29814] 3366.Gosden, J. R.; Middleton, P. G.; Rout, D.: Localization of thehuman oestrogen receptor gene to chromosome 6q24–q27 by in situ hybridization. *Cytogenet.Cell Genet.* 43: 218–220, 1986.
- [29815] 3367.Green, S.; Walter, P.; Kumar, V.; Krust, A.; Bornert, J.-M.; Argos, P.; Chambon, P.: Human oestrogen receptor cDNA: sequence, expressionand homology to v-erb-A. *Nature* 320: 134–139, 1986.
- [29816] 3368.Greene, G. L.; Gilna, P.; Waterfield, M.; Baker, A.; Hort, Y.; Shine, J.: Sequence and expression of human es-trogen receptor complementaryDNA. *Science* 231: 1150–1154, 1986.
- [29817] 3369.Heine, P. A.; Taylor, J. A.; Iwamoto, G. A.; Lubahn, D.

B.; Cooke, P. S.: Increased adipose tissue in male and female estrogen receptor- α knockout mice. *Proc. Nat. Acad. Sci.* 97: 12729–12734, 2000.

- [29818] 3370. Herrington, D. M.; Howard, T. D.; Hawkins, G. A.; Reboussin, D. M.; Xu, J.; Zheng, S. L.; Brosnihan, K. B.; Meyers, D. A.; Bleecker, E. R.: Estrogen-receptor polymorphisms and effects of estrogen replacement on high-density lipoprotein cholesterol in women with coronary disease. *New Eng. J. Med.* 346: 967–974, 2002.
- [29819] 3371. Issa, J.-P. J.; Ottaviano, Y. L.; Celano, P.; Hamilton, S. R.; Davidson, N. E.; Baylin, S. B.: Methylation of the estrogen receptor CpG island links ageing and neoplasia in human colon. *Nature Genet.* 7: 536–540, 1994.
- [29820] 3372. Jeltsch, J. M.; Roberts, M.; Schatz, C.; Garnier, J. M.; Brown, A. M. C.; Chambon, P.: Structure of the human estrogen-responsive gene pS2. *Nucleic Acids Res.* 15: 1401–1414, 1987.
- [29821] 3373. Bing, D. H.; Almeda, S.; Isliker, H.; Lahav, J.; Hynes, R. O.: Fibronectin binds to the C1q component of complement. *Proc. Nat. Acad. Sci.* 79: 4198–4201, 1982.
- [29822] 3374. Bittner, M.; Meltzer, P.; Chen, Y.; Jiang, Y.; Seftor, E.; Hendrix, M.; Radmacher, M.; Simon, R.; Yakhini, Z.; Bendor, A.; Sampas, N.; Dougherty, E.; and 16 others: Molecu-

lar classification of cutaneous malignant melanoma by gene expression profiling. *Nature* 406: 536–540, 2000.

- [29823] 3375. Clark, E. A.; Golub, T. R.; Lander, E. S.; Hynes, R. O.: Genomic analysis of metastasis reveals an essential role for RhoC. *Nature* 406: 532–535, 2000.
- [29824] 3376. Clemmensen, I.: Fibronectin and its role in connective tissue diseases. (Editorial) *Europ. J. Clin. Invest.* 11: 145–146, 1981.
- [29825] 3377. Croce, C. M.: Personal Communication. Philadelphia, Pa. 1/12/1983.
- [29826] 3378. Eun, C. K.; Klinger, H. P.: Human chromosome 11 affects the expression of fibronectin fibers in human–mouse cell hybrids. *Cytogenet. Cell Genet.* 27: 57–65, 1980.
- [29827] 3379. Gutman, A.; Kornblihtt, A. R.: Identification of a third region of cell-specific alternative splicing in human fibronectin mRNA. *Proc. Nat. Acad. Sci.* 84: 7179–7182, 1987.
- [29828] 3380. Henry, I.; Jeanpierre, M.; Weil, D.; Grzeschik, K. H.; Ramirez, F.; Junien, C.: The structural gene for fibronectin (FN) maps to 2q323–qter. (Abstract) *Cytogenet. Cell Genet.* 40: 650 only, 1985.
- [29829] 3381. Hirano, H.; Yamada, Y.; Sullivan, M.; de Crombrug–

ghe, B.; Pastan, I.; Yamada, K. M.: Isolation of genomic DNA clones spanning the entire fibronectin gene. Proc. Nat. Acad. Sci. 80: 46–50, 1983.

[29830] 3382. Jhanwar, S. C.; Jensen, J. T.; Kaelbling, M.; Chaganti, R. S.K.; Klinger, H. P.: In situ localization of human fibronectin (FN) genes to chromosome regions 2p14–p16, 2q34–q36, and 11q12.1–q13.5 in germ line cells, but to chromosome 2 sites only in somatic cells. Cytogenet. Cell Genet. 41: 47–53, 1986.

[29831] 3383. Koch, G. A.; Schoen, R. C.; Klebe, R. J.; Shows, T. B.: Assignment of a fibronectin gene to human chromosome 2 using monoclonal antibodies. Exp. Cell Res. 141: 293–302, 1982.

[29832] 3384. Kornblihtt, A. R.; Umezawa, K.; Vibe-Pedersen, K.; Baralle, F.E.: Primary structure of human fibronectin: differential splicing may generate at least 10 polypeptides from a single gene. EMBO J. 4: 1755–1759, 1985.

[29833] 3385. Kornblihtt, A. R.; Vibe-Pedersen, K.; Baralle, F. E.: Isolation and characterization of cDNA clones for human and bovine fibronectins. Proc. Nat. Acad. Sci. 80: 3218–3222, 1983.

[29834] 3386. Kornblihtt, A. R.; Vibe-Pedersen, K.; Baralle, F. E.: Human fibronectin: molecular cloning evidence for two

mRNA species differing by an internal segment coding for a structural domain. EMBO J. 3: 221–226, 1984.

[29835] 3387. Kurkinen, M.; Vartio, T.; Vaheri, A.: Polypeptides of human plasma fibronectin are similar but not identical. Biochim. Biophys. Acta 624:490–498, 1980.

[29836] 3388. Matsuura, H.; Takio, K.; Titani, K.; Greene, T.; Levery, S. B.; Salyan, M. E. K.; Hakomori, S.: The oncofetal structure of human fibronectin defined by monoclonal antibody FDC-6. J. Biol. Chem. 263:3314–3322, 1988.

[29837] 3389. McDonagh, J.: Fibronectin: a molecular glue. Arch. Path. Lab. Med. 105: 393–396, 1981.

[29838] 3390. Mosesson, M. W.; Amrani, D. L.: The structure and biologic activities of plasma fibronectin. Blood 56: 145–158, 1980.

[29839] 3391. Odermatt, E.; Tamkun, J. W.; Hynes, R. O.: Repeating modular structure of the fibronectin gene: relationship to protein structure and subunit variation. Proc. Nat. Acad. Sci. 82: 6571–6575, 1985.

[29840] 3392. Owerbach, D.; Doyle, D.; Shows, T. B.: Genetics of the large, external, transformation-sensitive (LETS) protein: assignment of a gene coding for expression of LETS to human chromosome 8. Proc. Nat. Acad. Sci. 75: 5640–5644, 1978.

- [29841] 3393.Prowse, K.; Tricoli, J.; Klebe, R.; Shows, T.: Chromosome 2 assignment of the structural gene for fibronectin (FN) using a cloned probe.(Abstract) Cytogenet. Cell Genet. 40: 724 only, 1985.
- [29842] 3394.Prowse, K. R.; Tricoli, J. V.; Klebe, R. J.; Shows, T. B.: Assignment of the human fibronectin structural gene to chromosome 2. Cytogenet.Cell Genet. 41: 42–46, 1986.
- [29843] 3395.Rennard, S. I.; Church, R. L.; Rohrbach, D. H.; Shupp, D. E.; Abe, S.; Hewitt, A. T.; Murray, J. C.; Martin, G. R.: Localization of the human fibronectin (FN) gene on chromosome 8 by a specific enzyme immunoassay. Biochem. Genet. 19: 551–566, 1981.
- [29844] 3396.Ridley, A.: Molecular switches in metastasis. Nature 406: 466–467, 2000.
- [29845] 3397.Li, Q. J.; Ashraf, M. F.; Shen, D.; Green, W. R.; Stark, W. J.; Chan, C.-C; O'Brien, T. P.: The role of apoptosis in the pathogenesis of Fuchs endothelial dystrophy of the cornea. Arch. Ophthalmol. 119:1597–1604, 2001.
- [29846] 3398.McCurley, R. S.; Recinos, A., III; Olsen, A. S.; Gingrich, J. C.; Szczepaniak, D.; Cameron, H. S.; Krauss, R.; Weston, B. W.: Physical maps of human alpha(1,3)fucosyltransferase genes FUT3–FUT6 on chromosomes 19p13.3 and 11q21. Genomics 26: 142–146, 1995.

- [29847] 3399. Weston, B. W.; Nair, R. P.; Larsen, R. D.; Lowe, J. B.: Isolation of a novel human $\alpha(1,3)$ fucosyltransferase gene and molecular comparison to the human Lewis blood group $\alpha(1,3/1,4)$ fucosyltransferase gene: syntenic, homologous, nonallelic genes encoding enzymes with distinct acceptor substrate specificities. *J. Biol. Chem.* 267: 4152–4160, 1992.
- [29848] 3400. Weston, B. W.; Smith, P. L.; Kelly, R. J.; Lowe, J. B.: Molecular cloning of a fourth member of a human $\alpha(1,3)$ fucosyltransferase gene family: multiple homologous sequences that determine expression of the Lewis x, sialyl Lewis x, and difucosyl sialyl Lewis x epitopes. *J. Biol. Chem.* 267: 24575–24584, 1992.
- [29849] 3401. Brinkman–Van der Linden, E. C. M.; Mollicone, R.; Oriol, R.; Larson, G.; Van den Eijnden, D. H.; Van Dijk, W.: A missense mutation in the FUT6 gene results in total absence of α -3-fucosylation of human α -1-acid glycoprotein. *J. Biol. Chem.* 271: 14492–14495, 1996.
- [29850] 3402. Billardon, C.; Van Cong, N.; Picard, J. Y.; Dekaouel, C.; Rebourcet, R.; Weil, D.; Feingold, J.; Frezal, J.: Linkage studies of enzyme markers in man–mouse somatic cell hybrids. *Ann. Hum. Genet.* 36: 273–284, 1973.
- [29851] 3403. Cook, P. J. L.; Hamerton, J. L.: Report of the commit–

tee on the genetic constitution of chromosome 1. Cytogenet. Cell Genet. 25:9–20, 1979.

- [29852] 3404. Zhou, X.-P.; Smith, W. M.; Gimm, O.; Mueller, E.; Gao, X.; Sarraf, P.; Prior, T. W.; Plass, C.; van Deimling, A.; Black, P. M.; Yates, A. J.; Eng, C.: Over-representation of PPAR-gamma sequence variants in sporadic cases of glioblastoma multiforme: preliminary evidence for common low penetrance modifiers for brain tumour risk in the general population. J. Med. Genet. 37: 410–414, 2000.
- [29853] 3405. Bingham, C.; Bulman, M. P.; Ellard, S.; Allen, L. I. S.; Lipkin, G. W.; van't Hoff, W. G.; Woolf, A. S.; Rizzoni, G.; Novelli, G.; Nicholls, A. J.; Hattersley, A. T.: Mutations in the hepatocyte nuclear factor-1-beta gene are associated with familial hypoplastic glomerulocystic kidney disease. Am. J. Hum. Genet. 68: 219–224, 2001.
- [29854] 3406. Kaplan, B. S.; Gordon, I.; Pincott, J.; Barratt, T. M.: Familial hypoplastic glomerulocystic kidney disease: a definite entity with dominant inheritance. Am. J. Med. Genet. 34: 569–573, 1989.
- [29855] 3407. Rizzoni, G.; Loirat, C.; Levy, M.; Milanesi, C.; Zachello, G.; Mathieu, H.: Familial hypoplastic glomerulocystic kidney: a new entity? Clin. Nephrol. 18: 263–268, 1982.

- [29856] 3408.Karasawa, M.; Zwacka, R. M.; Reuter, A.; Fink, T.; Hsieh, C. L.;Lichter, P.; Francke, U.; Weiher, H.: The human homolog of the glomerulosclerosisgene Mpv17: structure and genomic organization. Hum. Molec. Genet. 2:1829–1834, 1993.
- [29857] 3409.Fong, H. K. W.; Yoshimoto, K. K.; Eversole–Cire, P.; Simon, M.I.: Identification of a GTP–binding protein alpha subunit that lacksan apparent ADP–ribosylation site for pertussis toxin. Proc. Nat.Acad. Sci. 85: 3066–3070, 1988.
- [29858] 3410.Matsuoka, M.; Itoh, H.; Kozasa, T.; Kaziro, Y.: Sequence analysisof cDNA and genomic DNA for a putative pertussis toxin–insensitiveguanine nucleotide–binding regulatory protein alpha subunit. Proc.Nat. Acad. Sci. 85: 5384–5388, 1988.
- [29859] 3411.Wilkie, T. M.; Gilbert, D. J.; Olsen, A. S.; Chen, X.–N.; Amatruda,T. T.; Korenberg, J. R.; Trask, B. J.; de Jong, P.; Reed, R. R.; Simon,M. I.; Jenkins, N. A.; Copeland, N. G.: Evolution of the mammalianG protein alpha subunit multi–gene family. Nature Genet. 1: 85–91,1992.
- [29860] 3412.Magovcevic, I.; Ang, S.–L.; Seidman, J. G.; Tolman, C. J.; Neer,E. J.; Morton, C. C.: Regional localization of the human G proteinalpha(i2) (GNAI2) gene: assignment to

3p21 and a related sequence(GNAI2L) to 12p12–p13. Genomics 12: 125–129, 1992.

- [29861] 3413.Neer, E. J.; Michel, T.; Eddy, R.; Shows, T.; Seidman, J. G.:Genes for two homologous G–protein alpha subunits map to differenthuman chromosomes. Hum. Genet. 77: 259–262, 1987.
- [29862] 3414.Colombo, M. P.; Martinotti, A.; Howard, T. A.; Schneider, C.; D'Eustachio,P.; Seldin, M. F.: Localization of growth arrest–specific genes onmouse chromosomes 1, 7, 8, 11, 13, and 16. Mammalian Genome 2: 130–134,1992.
- [29863] 3415.Del Sal, G.; Collavin, L.; Ruaro, M. E.; Edomi, P.; Saccone, S.;Della Valle, G.; Schneider, C.: Structure, function, and chromosomemapping of the growth–suppressing human homologue of the murine gas1gene. Proc. Nat. Acad. Sci. 91: 1848–1852, 1994.
- [29864] 3416.Del Sal, G.; Ruaro, M. E.; Philipson, L.; Schneider, C.: The growtharrest–specific gene, gas1, is involved in growth suppression. Cell 70:595–607, 1992.
- [29865] 3417.Evdokiou, A.; Webb, G. C.; Peters, G. B.; Dobrovic, A.; O'Keefe,D. S.; Forbes, I. J.; Cowled, P. A.: Localization of the human growtharrest–specific gene (GAS1) to chromosome bands 9q21.3–q22, a regionfrequently deleted in myeloid malignancies. Genomics 18: 731–733,1993.

- [29866] 3418.Schneider, C.; King, R. M.; Philipson, L.: Genes specifically expressed at growth arrest of mammalian cells. Cell 54: 787–793,1988.
- [29867] 3419.Webb, G. C.; Cowled, P. A.; Evdokiou, A.; Ford, J. H.; Forbes,I. J.: Assignment, by in situ hybridization, of the growth arrest–specific gene, Gas–1, to mouse chromosome 13, bands B3–C2. Genomics 14: 548–549,1992.
- [29868] 3420.Lebo, R. V.; Cheung, M.–C.; Bruce, B. D.: Rapid gene mapping by dual laser chromosome sorting and spot blot DNA analysis. (Abstract) Am.J. Hum. Genet. 36: 101S only, 1984.
- [29869] 3421.Kudo, J.; Chao, L.–Y.; Narni, F.; Saunders, G. F.: Structure of the human gene encoding the invariant gamma–chain of class II histocompatibility antigens. Nucleic Acids Res. 13: 8827–8841, 1985.
- [29870] 3422.O'Sullivan, D. M.; Larhammar, D.; Wilson, M. C.; Peterson, P. A.; Quaranta, V.: Structure of the human Ia–associated invariant (gamma)–chain gene: identification of 5–prime sequences shared with major histocompatibility–complex class II genes. Proc. Nat. Acad. Sci. 83: 4484–4488, 1986.
- [29871] 3423.Riese, R. J.; Shi, G.–P.; Villadangos, J.; Stetson, D.; Driessen,C.; Lennon–Dumenil, A.–M.; Chu, C.–L.; Naumov,

Y.; Behar, S. M.; Ploegh, H.; Locksley, R.; Chapman, H. A.: Regulation of CD1 function and NK1.1+ T cell selection and maturation by cathepsin S. *Immunity* 15:909–919, 2001.

[29872] 3424. Topilski, I.; Harmelin, A.; Flavell, R. A.; Levo, Y.; Shachar, I.: Preferential Th1 immune response in invariant chain-deficient mice. *J. Immun.* 168: 1610–1617, 2002.

[29873] 3425. Abderrahim, H.; Sambucy, J.-L.; Iris, F.; Ougen, P.; Billault, A.; Chumakov, I. M.; Dausset, J.; Cohen, D.; Le Paslier, D.: Cloning the human major histocompatibility complex in YACs. *Genomics* 23:520–527, 1994.

[29874] 3426. Adman, R.; Pious, D. A.: Isoantigenic variants: isolation from human diploid cells in culture. *Science* 168: 370–372, 1970.

[29875] 3427. Arnaiz-Villena, A.; Festenstein, H.: HLA genotyping by using spermatozoa: evidence for haploid gene expression. *Lancet* II: 707–709, 1976.

[29876] 3428. Auffray, C.; Strominger, J. L.: Molecular genetics of the human major histocompatibility complex. *Adv. Hum. Genet.* 15: 197–247, 1986.

[29877] 3429. Awdeh, Z. L.; Raum, D.; Yunis, E. J.; Alper, C. A.: Extended HLA/complement allele haplotypes: evidence for T/t-like complex in man. *Proc. Nat. Acad. Sci.* 80: 259–263,

1983.

- [29878] 3430. Bach, F. H.; Amos, D. B.: Hu-1 major histocompatibility locus in man. *Science* 156: 1506-1508, 1967.
- [29879] 3431. Bach, M. L.; Bach, F. H.: The genetics of histocompatibility. *Hosp. Practice* 5(8): 33-44, 1970.
- [29880] 3432. Balner, H.: The major histocompatibility complex of primates: evolutionary aspects and comparative histogenetics. *Phil. Trans. Roy. Soc. London B* 292: 109-119, 1981.
- [29881] 3433. Balner, H.; D'Amaro, J.; Visser, T. P.: Tissue typing of chimpanzees: I. Evidence for two allelic series of leukocyte antigens. *Transplant. Proc.* 6: 141-149, 1974.
- [29882] 3434. Berger, R.; Bernheim, A.; Sasportes, M.; Hauptmann, G.; Hors, J.; Legrand, L.; Fellous, M.: Regional mapping of the HLA on the short arm of chromosome 6. *Clin. Genet.* 15: 245-251, 1979.
- [29883] 3435. Bernard, J.: La decouverte du systeme principal d'histocompatibilite de l'homme. (Editorial) *Presse Med.* 75: 2369 only, 1967.
- [29884] 3436. Bodmer, W. F.: Personal Communication. London, England 9/26/1986.
- [29885] 3437. Bodmer, W. F.; Bodmer, J. G.; Adler, S.; Payne, R.; Bialek, J.: Genetics of '4' and 'LA' human leukocyte groups.

Ann. N.Y. Acad.Sci. 129: 473–489, 1966.

- [29886] 3438.Bodmer, W. F.; Bodmer, J. G.; Tripp, M.: Recombination between the LA and 4 loci of the HL-A system. In: Histocompatibility Testing 1970. Copenhagen: Munksgaard (pub.) 1970. Pp. 187–191.
- [29887] 3439.Bodmer, W. F. (ed.): Evolutionary significance of the HL-A system. Nature 237:139–145, 1972.
- [29888] 3440.Bodmer, W. F. (ed.): The HLA System (17 papers). Brit. Med.Bull. 34(3): 213–316, 1978.
- [29889] 3441.Borgaonkar, D. S.; Bias, W. B.: HL-A loci and chromosome 6. Birth Defects Orig. Art. Ser. X(3): 67–68, 1974.
- [29890] 3442.Borgaonkar, D. S.; Bias, W. B.; Chase, G. A.; Sadasivan, G.; Herr, H. M.; Golomb, H. M.; Bahr, G. F.; Kunkel, L. M.: Identification of C6–G21 translocation chromosome by the Q–M and Giemsa banding techniques in a patient with Down's syndrome, with possible assignment of Gm locus. Clin. Genet. 4: 53–57, 1973.
- [29891] 3443.Breuning, M. H.; van den Berg–Loonen, E. M.; Bernini, L. F.; Bijlsma, J. B.; van Loghem, E.; Meera Khan, P.; Nijenhuis, L. E.: Localization of HLA on the short arm of chromosome 6. Hum. Genet. 37: 131–139, 1977.
- [29892] 3444.Bull, R. W.; Benson, J. W.; Pearson, G.; Mann, J. D.: HLA substantiation of a trisomic human chromosome 6.

Transplant. Proc. 10: 747–748,1978.

- [29893] 3445.Campbell, R. D.; Trowsdale, J.: Map of the human MHC. Immun.Today 14: 349–352, 1993.
- [29894] 3446.Cardoso, C. S.; Alves, H.; Mascarenhas, M.; Goncalves, R.; Oliveira,P.; Rodrigues, P.; Cruz, E.; de Sousa, M.; Porto, G.: Co-selectionof the H63D mutation and the HLA-A29 allele: a new paradigm of linkage disequilibrium? Immunogenetics 53: 1002–1008, 2002.
- [29895] 3447.Carrington, M.; Nelson, G. W.; Martin, M. P.; Kissner, T.; Vlahov,D.; Goedert, J. J.; Kaslow, R.; Buchbinder, S.; Hoots, K.; O'Brien,S. J.: HLA and HIV-1: heterozygote advantage and B*35-Cw*04 disadvantage. Science 283:1748–1752, 1999.
- [29896] 3448.Ceppellini, R.; Van Rood, J. J.: The HL-A system. I. Geneticsand molecular biology. Seminars Hemat. 11: 233–252, 1974.
- [29897] 3449.Cohen, D.; Paul, P.; Font, M.-P.; Cohen, O.; Sayagh, B.; Marcadet,A.; Busson, M.; Mahouy, G.; Cann, H. M.; Dausset, J.: Analysis ofHLA class I genes with restriction endonuclease fragments: implicationsfor polymorphism of the human major histocompatibility complex. Proc.Nat. Acad. Sci. 80: 6289–6292, 1983.
- [29898] 3450.Colombani, J.: Changing the name of the major his-

to compatibility complex. Res. Immun. 143: 411–417, 1992.

[29899] 3451. Dausset, J.: The major histocompatibility complex in man: past, present, and future concepts. Science 213: 1469–1474, 1981.

[29900] 3452. Dausset, J.: Personal Communication. Paris, France 1/12/1983.

[29901] 3453. Dausset, J.: Similarities between the HL-A system and other immunogenetic systems. (Editorial) Vox Sang. 23: 153–164, 1972.

[29902] 3454. Ion, R.; Telvi, L.; Chaussain, J.-L.; Barbet, J. P.; Nunes, M.; Safar, A.; Rethore, M.-O.; Fellous, M.; McElreavey, K.: Failure of testicular development associated with a rearrangement of 9p24.1 proximal to the SNF2 gene. Hum. Genet. 102: 151–156, 1998.

[29903] 3455. Matsushime, H.; Jinno, A.; Takagi, N.; Shibuya, M.: A novel mammalian protein kinase gene (mak) is highly expressed in testicular germ cells at and after meiosis. Molec. Cell. Biol. 10: 2261–2268, 1990.

[29904] 3456. Taketo, M.; Jinno, A.; Yamaguchi, S.; Matsushime, H.; Shibuya, M.; Seldin, M. F.: Mouse Mak gene for male germ cell-associated kinase maps to chromosome 13. Genomics 19: 397–398, 1994.

- [29905] 3457.Cohen, P. T. W.; Omenn, G. S.: Genetic variation of the cytoplasmic and mitochondrial malic enzymes in the monkey: *Macaca nemestrina*. *Biochem.Genet.* 7: 289–301, 1972.
- [29906] 3458.Gonzalez–Manchon, C.; Butta, N.; Ferrer, M.; Ayuso, M. S.; Parrilla,R.: Molecular cloning and functional characterization of the human cytosolic malic enzyme promoter: thyroid hormone responsiveness. *DNACell Biol.* 16: 533–544, 1997.
- [29907] 3459.Loeber, G.; Dworkin, M. B.; Infante, A.; Ahorn, H.: Characterization of cytosolic malic enzyme in human tumor cells. *FEBS Lett.* 344:181–186, 1994.
- [29908] 3460.Meera Khan, P.; Hagemeijer, A.; Wijnen, L. M. M.; van der Goes,R. G. M.: PGM3 and ME1 are probably in the 6pter–q12 region. (Abstract) *Cytogenet.Cell Genet.* 37: 537 only, 1984.
- [29909] 3461.Nass, S. J.; Olowson, M.; Miyashita, N.; Moriwaki, K.; Balling,R.; Imai, K.: Mapping of the Mod–1 locus on mouse chromosome 9. *MammalianGenome* 4: 333–337, 1993.
- [29910] 3462.Povey, S.; Wilson, D. E., Jr.; Harris, H.; Gormley, I. P.; Perry,P.; Buckton, K. E.: Sub–unit structure of soluble and mitochondrial malic enzyme: demonstration of human mitochondrial enzyme in human–mouse hybrids. *Ann. Hum.*

Genet. 39: 203–212, 1975.

- [29911] 3463. Itoh, T.; Tanaka, T.; Nagai, R.; Kamiya, T.; Sawayama, T.; Nakayama, T.; Tomoike, H.; Sakurada, H.; Yazaki, Y.; Nakamura, Y.: Genomic organization and mutational analysis of HERG, a gene responsible for familial long QT syndrome. Hum. Genet. 102: 435–439, 1998.
- [29912] 3464. Jiang, C.; Atkinson, D.; Towbin, J. A.; Splawski, I.; Lehmann, M. H.; Li, H.; Timothy, K.; Taggart, R. T.; Schwartz, P. J.; Vincent, G. M.; Moss, A. J.; Keating, M. T.: Two long QT syndrome loci map to chromosomes 3 and 7 with evidence for further heterogeneity. Nature Genet. 8: 141–147, 1994.
- [29913] 3465. Jongbloed, R. J. E.; Wilde, A. A. M.; Geelen, J. L. M. C.; Doevendans, P.; Schaap, C.; Van Langen, I.; van Tintelen, J. P.; Cobben, J. M.; Beaufort-Krol, G. C. M.; Geraedts, J. P. M.; Smeets, H. J. M.: Novel KCNQ1 and HERG missense mutations in Dutch long-QT families. Hum. Mutat. 13: 301–310, 1999.
- [29914] 3466. Kagan, A.; Yu, Z.; Fishman, G. I.; McDonald, T. V.: The dominant negative LQT2 mutation A561V reduces wild-type HERG expression. J. Biol. Chem. 275: 11241–11248, 2000.
- [29915] 3467. Kuperschmidt, S.; Yang, T.; Chanthaphaychith, S.;

Wang, Z.; Towbin, J. A.; Roden, D. M.: Defective human ether- α -go-go-related gene trafficking linked to an endoplasmic reticulum retention signal in the C terminus. *J. Biol. Chem.* 277: 27442–27448, 2002.

[29916] 3468. Larsen, L. A.; Svendsen, I. H.; Jensen, A. M.; Kanter, J. K.; Andersen, P. S.; Møller, M.; Sørensen, S. A.; Sandoe, E.; Jacobsen, J. R.; Vuust, J.; Christiansen, M.: Long QT syndrome with a high mortality rate caused by a novel G572R missense mutation in KCNH2. *Clin. Genet.* 57: 125–130, 2000.

[29917] 3469. Li, X.; Xu, J.; Li, M.: The human δ -1261 mutation of the HERG potassium channel results in a truncated protein that contains a subunit interaction domain and decreases the channel expression. *J. Biol. Chem.* 272: 705–708, 1997.

[29918] 3470. Miller, C.: The inconstancy of the human heart. *Nature* 379: 767–768, 1996.

[29919] 3471. Moss, A. J.; Zareba, W.; Kaufman, E. S.; Gattman, E.; Peterson, D. R.; Benhorin, J.; Towbin, J. A.; Keating, M. T.; Priori, S. G.; Schwartz, P. J.; Vincent, G. M.; Robinson, J. L.; Andrews, M. L.; Feng, C.; Hall, W. J.; Medina, A.; Zhang, L.; Wang, Z.: Increased risk of arrhythmic events in long-QT syndrome with mutations in the pore region of the human

ether-a-go-go-related gene potassium channel. *Circulation* 105:794–799, 2002.

[29920] 3472. Nakajima, T.; Kurabayashi, M.; Ohyama, Y.; Kaneko, Y.; Furukawa, T.; Itoh, T.; Taniguchi, Y.; Tanaka, T.; Nakamura, Y.; Hiraoka, M.; Nagai, R.: Characterization of S818L mutation in HERG C-terminus in LQT2: modification of activation-deactivation gating properties. *FEBS Lett.* 481: 197–203, 2000.

[29921] 3473. Priori, S. G.; Barhanin, J.; Hauer, R. N. W.; Haverkamp, W.; Jongsma, H. J.; Kleber, A. G.; McKenna, W. J.; Roden, D. M.; Rudy, Y.; Schwartz, K.; Schwartz, P. J.; Towbin, J. A.; Wilde, A. M.: Genetic and molecular basis of cardiac arrhythmias: impact on clinical management. Parts I and II. *Circulation* 99: 518–528, 1999.

[29922] 3474. Priori, S. G.; Barhanin, J.; Hauer, R. N. W.; Haverkamp, W.; Jongsma, H. J.; Kleber, A. G.; McKenna, W. J.; Roden, D. M.; Rudy, Y.; Schwartz, K.; Schwartz, P. J.; Towbin, J. A.; Wilde, A. M.: Genetic and molecular basis of cardiac arrhythmias: impact on clinical management. Part III. *Circulation* 99: 674–681, 1999.

[29923] 3475. Priori, S. G.; Napolitano, C.; Schwartz, P. J.: Low penetrance in the long QT syndrome: clinical impact. *Circulation* 99: 529–533, 1999.

- [29924] 3476.Rajamani, S.; Anderson, C. L.; Anson, B. D.; January, C. T.:Pharmacological rescue of human K⁺ channel long-QT2 mutations. *Circulation* 105:2830–2835, 2002.
- [29925] 3477.Sanguinetti, M. C.; Jiang, C.; Curran, M. E.; Keating, M. T.:A mechanistic link between an inherited and an acquired cardiac arrhythmia:HERG encodes the I(Kr) potassium channel. *Cell* 81: 299–307, 1995.
- [29926] 3478.Satler, C. A.; Vesely, M. R.; Duggal, P.; Ginsburg, G. S.; Beggs,A. H.: Multiple different missense mutations in the pore region ofHERG in patients with long QT syndrome. *Hum. Genet.* 102: 265–272,1998.
- [29927] 3479.Satler, C. A.; Walsh, E. P.; Vesely, M. R.; Plummer, M. H.; Ginsburg,G. S.; Jacob, H. J.: Novel missense mutation in the cyclic nucleotide-bindingdomain of HERG causes long QT syndrome. *Am. J. Med. Genet.* 65: 27–35,1996.
- [29928] 3480.Smith, P. L.; Baukrowtiz, T.; Yellen, G.: The inward rectificationmechanism of the HERG cardiac potassium channel. *Nature* 379: 833–835,1996.
- [29929] 3481.Splawski, I.; Shen, J.; Timothy, K. W.; Vincent, G. M.; Lehmann,M. H.; Keating, M. T.: Genomic structure of three long QT syndromegenes: KVLQT1, HERG, and KCNE1. *Genomics* 51: 86–97, 1998.
- [29930] 3482.Tanaka, T.; Nagai, R.; Tomoike, H.; Takata, S.; Yano,

K.; Yabuta, K.; Haneda, N. Nakano, O.; Shibata, A.; Sawayama, T.; Kasai, H.; Yazaki, Y.; Nakamura, Y.: Four novel KVLQT1 and four novel HERG mutations in familial long-QT syndrome. *Circulation* 95: 565–567, 1997.

[29931] 3483. Thomas, P. J.; Qu, B.-H.; Pedersen, P. L.: Defective protein folding as a basis of human disease. *Trends Biochem. Sci.* 20: 456–459, 1995.

[29932] 3484. Trudeau, M. C.; Warmke, J. W.; Ganetzky, B.; Robertson, G. A.: HERG, a human inward rectifier in the voltage-gated potassium channel family. *Science* 269: 92–95, 1995.

[29933] 3485. Warmke, J. W.; Ganetzky, B.: A family of potassium channel genes related to eag in *Drosophila* and mammals. *Proc. Nat. Acad. Sci.* 91: 3438–3442, 1994.

[29934] 3486. Yoshida, H.; Horie, M.; Otani, H.; Kawashima, T.; Onishi, Y.; Sasayama, S.: Bradycardia-induced long QT syndrome caused by a de novo missense mutation in the S2–S3 inner loop of HERG. *Am. J. Med. Genet.* 98: 348–352, 2001.

[29935] 3487. Zareba, W.; Moss, A. J.; Schwartz, P. J.; Vincent, G. M.; Robinson, J. L.; Priori, S. G.; Benhorin, J.; Locati, E. H.; Towbin, J. A.; Keating, M. T.; Lehmann, M. H.; Hall, W. J.; International Long-QT Syndrome Registry Research Group:

Influence of the genotype on the clinical course of the long-QT syndrome. *New Eng. J. Med.* 339: 960–965, 1998.

[29936] 3488. Zhou, Z.; Gong, Q.; Epstein, M. L.; January, C. T.: HERG channel dysfunction in human long QT syndrome: intracellular transport and functional defects. *J. Biol. Chem.* 273: 21061–21066, 1998.

[29937] 3489. Rosengard, A. M.; Krutzsch, H. C.; Shearn, A.; Biggs, J. R.; Barker, E.; Margulies, I. M. K.; King, C. R.; Liotta, L. A.; Steeg, P. S.: Reduced nm23/awd protein in tumour metastasis and aberrant Drosophila development. *Nature* 342: 177–180, 1989.

[29938] 3490. Steeg, P. S.; Bevilacqua, G.; Kopper, L.; Thorgeirsson, U. P.; Talmadge, J. E.; Liotta, L. A.; Sobel, M. E.: Evidence for a novel gene associated with a low tumor metastatic potential. *J. Nat. Cancer Inst.* 80: 200–204, 1988.

[29939] 3491. Subramanian, C.; Cotter, M. A., II; Robertson, E. S.: Epstein–Barr virus nuclear protein EBNA–3C interacts with the human metastasis suppressor Nm23–H1: a molecular link to cancer metastasis. *Nature Med.* 7: 350–355, 2001.

[29940] 3492. Varesco, L.; Caligo, M. A.; Simi, P.; Black, D. M.; Nardini, V.; Casarino, L.; Rocchi, M.; Ferrara, G.; Solomon, E.; Bevilacqua, G.: The NM23 gene maps to human chromosome band 17q22 and shows a restriction fragment length

polymorphism with BglII. *Genes Chromosomes Cancer* 4:84–88, 1992.

[29941] 3493. Backer, J. M.; Mendola, C. E.; Kovesdi, I.; Fairhurst, J. L.; O'Hara, B.; Eddy, R. L., Jr.; Shows, T. B.; Mathew, S.; Murty, V. V. V. S.; Chaganti, R. S. K.: Chromosomal localization and nucleoside diphosphate kinase activity of human metastasis-suppressor genes NM23-1 and NM23-2. *Oncogene* 8:497–502, 1993.

[29942] 3494. Chandrasekharappa, S. C.; Gross, L. A.; King, S. E.; Collins, F. S.: The human NME2 gene lies within 18kb of NME1 in chromosome 17. *Genes Chromosomes Cancer* 6: 245–248, 1993.

[29943] 3495. Gilles, A. M.; Presecan, E.; Vonica, A.; Lascu, I.: Nucleosidediphosphate kinase from human erythrocytes: structural characterization of the two polypeptide chains responsible for heterogeneity of the hexameric enzyme. *J. Biol. Chem.* 266: 8784–8789, 1991.

[29944] 3496. Kelsell, D. P.; Black, D. M.; Solomon, E.; Spurr, N. K.: Localization of a second NM23 gene, NME2, to chromosome 17q21–q22. *Genomics* 17:522–524, 1993.

[29945] 3497. Stahl, J. A.; Leone, A.; Rosengard, A. M.; Porter, L.; King, C. R.; Steeg, P. S.: Identification of a second human nm23 gene, nm23-H2. *Cancer Res.* 51: 445–449, 1991.

- [29946] 3498. Boyes, J.; Bird, A.: DNA methylation inhibits transcription indirectly via a methyl-CpG binding protein. *Cell* 64: 1123–1134, 1991.
- [29947] 3499. Cross, S. H.; Meehan, R. R.; Nan, X.; Bird, A.: A component of the transcriptional repressor MeCP1 shares a motif with DNA methyltransferase and HRX proteins. *Nature Genet.* 16: 256–259, 1997.
- [29948] 3500. Hendrich, B.; Abbott, C.; McQueen, H.; Chambers, D.; Cross, S.; Bird, A.: Genomic structure and chromosomal mapping of the murine and human Mbd1, Mbd2, Mbd3, and Mbd4 genes. *Mammalian Genome* 10:906–912, 1999.
- [29949] 3501. Hempstead, B. L.; Martin-Zanca, D.; Kaplan, D. R.; Parada, L.F.; Chao, M. V.: High-affinity NGF binding requires coexpression of the trk proto-oncogene and the low-affinity NGF receptor. *Nature* 350:678–683, 1991.
- [29950] 3502. Huebner, K.; Isobe, M.; Chao, M.; Bothwell, M.; Ross, A. H.; Finan, J.; Hoxie, J. A.; Sehgal, A.; Buck, C. R.; Lahan, A.; Nowell, P.C.; Koprowski, H.; Croce, C. M.: The nerve growth factor receptor gene is at human chromosome region 17q12–17q22, distal to the chromosome 17 breakpoint in acute leukemias. *Proc. Nat. Acad. Sci.* 83: 1403–1407, 1986.
- [29951] 3503. Ip, N. Y.; Stitt, T. N.; Tapley, P.; Klein, R.; Glass, D.

J.; Fandl, J.; Greene, L. A.; Barbacid, M.; Yancopoulos, G. D.: Similarities and differences in the way neurotrophins interact with the Trk receptors in neuronal and nonneuronal cells. *Neuron* 10: 137–149, 1993.

[29952] 3504. Johnson, D.; Lanahan, A.; Buck, C. R.; Sehgal, A.; Morgan, C.; Mercer, E.; Bothwell, M.; Chao, M.: Expression and structure of the human NGF receptor. *Cell* 47: 545–554, 1986.

[29953] 3505. Lee, K. F.; Li, E.; Huber, J.; Landis, S. C.; Sharpe, A. H.; Chao, M. V.; Jaenisch, R.: Targeted mutation of the gene encoding the low affinity NGF receptor p75 leads to deficits in the peripheral sensory nervous system. *Cell* 69: 737–749, 1992.

[29954] 3506. Mischel, P. S.; Smith, S. G.; Vining, E. R.; Valletta, J. S.; Mobley, W. C.; Reichardt, L. F.: The extracellular domain of p75(NTR) is necessary to inhibit neurotrophin-3 signaling through TrkA. *J. Biol. Chem.* 276: 11294–11301, 2001.

[29955] 3507. Rettig, W. J.; Thomson, T. M.; Spengler, B. A.; Biedler, J. L.; Old, L. J.: Assignment of human nerve growth factor receptor gene to chromosome 17 and regulation of receptor expression in somatic cell hybrids. *Somat. Cell Molec. Genet.* 12: 441–447, 1986.

[29956] 3508. Tuffereau, C.; Benejean, J.; Blondel, D.; Kieffer, B.;

Flamand,A.: Low-affinity nerve-growth factor receptor (p75NTR) can serve as a receptor for rabies virus. EMBO J. 17: 7250–7259, 1998.

[29957] 3509.Welcher, A. A.; Bitler, C. M.; Radeke, M. J.; Shooter, E. M.:Nerve growth factor binding domain of the nerve growth factor receptor. Proc.Nat. Acad. Sci. 88: 159–163, 1991.

[29958] 3510.Breakefield, X. O.; Orloff, G.; Castiglione, C.; Coussens, L.;Axelrod, F. B.; Ullrich, A.: Structural gene for beta-nerve growthfactor not defective in familial dysautonomia. Proc. Nat. Acad. Sci. 81:4213–4216, 1984.

[29959] 3511.Carrier, A.; Rosier, M.–F.; Guillemot, F.; Goguel, A.–F.; Pulcini,F.; Bernheim, A.; Auffray, C.; Devignes, M.–D.: Integrated physical,genetic, and genic map covering 3 Mb around the human NGF gene (NGFB)at 1p13. Genomics 31: 80–89, 1996.

[29960] 3512.Chuang, H.; Prescott, E. D.; Kong, H.; Shields, S.; Jordt, S.–E.;Basbaum, A. I.; Chao, M. V.; Julius, D.: Bradykinin and nerve growthfactor release the capsaicin receptor from PtdIns(4,5)P₂–mediatedinhibition. Nature 411: 957–962, 2001.

[29961] 3513.Darby, J. K.; Feder, J.; Selby, M.; Riccardi, V.; Ferrell, R.;Siao, D.; Goslin, K.; Rutter, W.; Shooter, E. M.; Cavalli–

Sforza, L. L.: A discordant sibship analysis between beta-NGF and neurofibromatosis. *Am.J. Hum. Genet.* 37: 52–59, 1985.

[29962] 3514. Darby, J. K.; Kidd, J. R.; Pakstis, A. J.; Sparkes, R. S.; Cann, H. M.; Ferrell, R. E.; Gerhard, D. G.; Riccardi, V.; Ege-land, J. A.; Shooter, E. M.; Cavalli-Sforza, L. L.; Kidd, K. K.: Linkage relationships of the gene for the beta subunit of nerve growth factor (NGFB) with other chromosome 1 marker loci. *Cytogenet. Cell Genet.* 39: 158–160, 1985.

[29963] 3515. Dracopoli, N. C.: Personal Communication. Cambridge, Mass. 4/29/1988.

[29964] 3516. Dracopoli, N. C.; Rose, E.; Whitfield, G. K.; Guidon, P. T.; Bale, S. J.; Chance, P. A.; Kourides, I. A.; Housman, D. E.: Two thyroid hormone regulated genes, the beta-subunits of nerve growth factor (NGFB) and thyroid stimulating hormone (TSHB), are located less than 310 kb apart in both human and mouse genomes. *Genomics* 3: 161–167, 1988.

[29965] 3517. Francke, U.; de Martinville, B.; Coussens, L.; Ullrich, A.: The human gene for the beta subunit of nerve growth factor is located on the proximal short arm of chromosome 1. *Science* 222: 1248–1251, 1983.

[29966] 3518. Garson, J. A.; van den Berghe, J. A.; Kemshead, J. T.:

Novel non-isotopic in situ hybridization technique detects small (1 kb) unique sequences in routinely G-banded human chromosomes: fine mapping of N-myc and beta-NGF genes. *Nucleic Acids Res.* 15: 4761–4770, 1987.

[29967] 3519. Lee, R.; Kermani, P.; Teng, K. K.; Hempstead, B. L.: Regulation of cell survival by secreted proneurotrophins. *Science* 294: 1945–1948, 2001.

[29968] 3520. Levi-Montalcini, R.: The nerve growth factor thirty-five years later. *Science* 237: 1154–1162, 1987.

[29969] 3521. MacInnis, B. L.; Campenot, R. B.: Retrograde support of neuronal survival without retrograde transport of nerve growth factor. *Science* 295: 1536–1539, 2002.

[29970] 3522. Middleton-Price, H.; van den Berghe, J.; Harding, A.; Scott, J.; Malcolm, S.: Analysis of markers on chromosome 1. (Abstract) *Cytogenet. Cell Genet.* 46: 662, 1987.

[29971] 3523. Mitchell, E. L. D.; Jones, D.; White, G. R. M.; Varley, J. M.; Santibanez Koref, M. F.: Determination of the gene order of the three loci CD2, NGFB, and NRAS at human chromosome band 1p13 and refinement of their localisation at the subband level by fluorescence in situ hybridization. *Cytogenet. Cell Genet.* 70: 183–185, 1995.

[29972] 3524. Munke, M.; Lindgren, V.; de Martinville, B.; Francke, U.: Comparative analysis of mouse-human hybrids with re-

arranged chromosomes 1 by insitu hybridization and Southern blotting: high-resolution mapping of NRAS, NGFB, and AMY on human chromosome 1. *Somat. Cell Molec. Genet.* 10: 589–599, 1984.

[29973] 3525. Ramer, M. S.; Priestley, J. V.; McMahon, S. B.: Functional regeneration of sensory axons into the adult spinal cord. *Nature* 403: 312–316, 2000.

[29974] 3526. Sanico, A. M.; Stanis, A. M.; Gleeson, T. D.; Bora, S.; Proud, D.; Bienenstock, J.; Koliatsos, V. E.; Togias, A.: Nerve growth factor expression and release in allergic inflammatory disease of the upper airways. *Am. J. Resp. Crit. Care Med.* 161: 1631–1635, 2000.

[29975] 3527. Ullrich, A.; Gray, A.; Berman, C.; Dull, T. J.: Human beta-nerve growth factor gene sequence highly homologous to that of mouse. *Nature* 303: 821–825, 1983.

[29976] 3528. Beauchemin, N.; Draber, P.; Dveksler, G.; Gold, P.; Gray-Owen, S.; Grunert, F.; Hammarstrom, S.; Holmes, K. V.; Karlsson, A.; Kuroki, M.; Lin, S.-H.; Lucka, L.; and 13 others: Redefined nomenclature for members of the carcinoembryonic antigen family. *Exp. Cell Res.* 252: 243–249, 1999.

[29977] 3529. Inazawa, J.; Abe, T.; Inoue, K.; Misawa, S.; Oikawa, S.; Nakazato, H.; Yoshida, M. C.: Regional assignment of

nonspecific cross-reacting antigen (NCA) of the CEA gene family to chromosome 19 at band q13.2. Cytogenet. Cell Genet. 52: 28–31, 1989.

[29978] 3530. Willcocks, T. C.; Craig, S. P.; Craig, I. W.: Assignment of the coding sequence for carcinoembryonic antigen (CEA) and normal cross-reacting antigen (NCA) to human chromosome 19q13. Ann. Hum. Genet. 53: 141–148, 1989.

[29979] 3531. Qian, F.; Kruse, U.; Lichter, P.; Sippel, A. E.: Chromosomal localization of the four genes (NFIA, B, C, and X) for the human transcription factor nuclear factor I by FISH. Genomics 28: 66–73, 1995.

[29980] 3532. Santoro, C.; Mermod, N.; Andrews, P. C.; Tjian, R.: A family of human CCAAT-box-binding proteins active in transcription and DNA replication: cloning and expression of multiple cDNAs. Nature 334: 218–224, 1988.

[29981] 3533. Scherthan, H.; Seisenberger, C.; Greulich, K.; Winnacker, E.-L.: Mapping of the murine nuclear factor I/X gene (Nfix) to mouse chromosome 8 C1–2 by FISH. Genomics 22: 247–249, 1994.

[29982] 3534. Seisenberger, C.; Winnacker, E.-L.; Scherthan, H.: Localisation of the human nuclear factor I/X (NFI/X) gene to chromosome 19p13 and detection of five other related loci at 1p21–22, 1q42–43, 5q15, 11p13 and 20q13 by

FISH. Hum. Genet. 91: 535–537, 1993.

- [29983] 3535. Auphan, N.; DiDonato, J.A.; Rosette, C.; Helmberg, A.; Karin, M.: Immunosuppression by glucocorticoids: inhibition of NF- κ B activity through induction of I- κ B synthesis. Science 270: 286–290, 1995.
- [29984] 3536. Baeuerle, P. A.: I- κ B--NF- κ B structures: at the interface of inflammation control. Cell 95: 729–731, 1998.
- [29985] 3537. Haskill, S.; Beg, A. A.; Tompkins, S. M.; Morris, J. S.; Yurochko, A. D.; Sampson-Johannes, A.; Mondal, K.; Ralph, P.; Baldwin, A. S., Jr.: Characterization of an immediate-early gene induced in adherent monocytes that encodes I- κ B-like activity. Cell 65: 1281–1289, 1991.
- [29986] 3538. Huxford, T.; Huang, D.-B.; Malek, S.; Ghosh, G.: The crystal structure of the I- κ B- α /NF- κ B complex reveals mechanisms of NF- κ B activation. Cell 95: 759–770, 1998.
- [29987] 3539. Ito, C. Y.; Adey, N.; Bautch, V. L.; Baldwin, A. S., Jr.: Structure and evolution of the human IKBA gene. Genomics 29: 490–495, 1995.
- [29988] 3540. Jacobs, M. D.; Harrison, S. C.: Structure of an I- κ B- α /NF- κ B complex. Cell 95: 749–758, 1998.

- [29989] 3541.Jung, M.; Zhang, Y.; Lee, S.; Ditschilo, A.: Correction of radiationsensitivity in ataxia telangiectasia cells by a truncated I-kappa-B-alpha. *Science* 268:1619–1621, 1995.
- [29990] 3542.Le Beau, M. M.; Ito, C.; Cogswell, P.; Espinosa, R., III; Fernald,A. A.; Baldwin, A. S., Jr.: Chromosomal localization of the genesencoding the p50/p105 subunits of NF-kappa-B (NFKB2) and the I-kappa-B/MAD-3(NFKBI) inhibitor of NF-kappa-B to 4q24 and 14q13, respectively. *Genomics* 14:529–531, 1992.
- [29991] 3543.Neish, A. S.; Gewirtz, A. T.; Zeng, H.; Young, A. N.; Hobert,M. E.; Karmali, V.; Rao, A. S.; Madara, J. L.: Prokaryotic regulationof epithelial responses by inhibition of I-kappa-B-alpha ubiquitination. *Science* 289:1560–1563, 2000.
- [29992] 3544.Rupec, R. A.; Poujol, D.; Grosgeorge, J.; Carle, G. F.; Livolsi,A.; Peyron, J.-F.; Schmid, R. M.; Baeuerle, P. A.; Messer, G.: Structuralanalysis, expression, and chromosomal localization of the mouse ikbagene. *Immunogenetics* 49: 395–403, 1999.
- [29993] 3545.Scheinman, R. I.; Cogswell, P. C.; Lofquist, A. K.; Baldwin, A.S., Jr.: Role of transcriptional activation of I-kappa-B-alpha inmediation of immunosuppression by

glucocorticoids. *Science* 270:283–286, 1995.

[29994] 3546.Compton, D. A.; Szilak, I.; Cleveland, D. W.: Primary structure of NuMA, an intranuclear protein that defines a novel pathway for segregation of proteins at mitosis. *J. Cell Biol.* 116: 1395–1408, 1992.

[29995] 3547.Lydersen, B. K.; Pettijohn, D. E.: Human-specific nuclear protein that associates with the polar region of the mitotic apparatus: distribution in a human/hamster hybrid cell. *Cell* 22: 489–499, 1980.

[29996] 3548.Merdes, A.; Ramyar, K.; Vechio, J. D.; Cleveland, D. W.: A complex of NuMA and cytoplasmic dynein is essential for mitotic spindle assembly. *Cell* 87:447–458, 1996.

[29997] 3549.Sparks, C. A.; Bangs, P. L.; McNeil, G. P.; Lawrence, J. B.; Fey, E. G.: Assignment of the nuclear mitotic apparatus protein NuMA gene to human chromosome 11q13. *Genomics* 17: 222–224, 1993.

[29998] 3550.Wells, R. A.; Catzavelos, C.; Kamel-Reid, S.: Fusion of retinoic acid receptor alpha to NuMA, the nuclear mitotic apparatus protein, by a variant translocation in acute promyelocytic leukaemia. *Nature Genet.* 17: 109–113, 1997.

[29999] 3551.Wiese, C.; Wilde, A.; Moore, M. S.; Adam, S. A.; Merdes, A.; Zheng, Y.: Role of importin-beta in coupling

Ran to downstream targets in microtubule assembly. Science 291: 653–656, 2001.

[30000] 3552. Yang, C. H.; Lambie, E. J.; Snyder, M.: NuMA: an unusually long coiled-coil related protein in the mammalian nucleus. J. Cell Biol. 116:1303–1317, 1992.

[30001] 3553. Halila, R.; Apostolou, S.; Winqvist, R.; Callen, D.; Prockop, D. J.; Peltonen, L.: Isolation and genomic assignment of a candidate cDNA clone for type III procollagen N-proteinase. (Abstract) Am. J. Hum. Genet. 51 (suppl.): A128 only, 1992.

[30002] 3554. Halila, R.; Peltonen, L.; Prockop, D. J.: Isolation of a candidate cDNA clone for type III procollagen N-proteinase from human placental cDNA library. (Abstract) Am. J. Hum. Genet. 45 (suppl.): A192 only, 1989.

[30003] 3555. Nomura, N.; Nagase, T.; Miyajima, N.; Sazuka, T.; Tanaka, A.; Sato, S.; Seki, N.; Kawarabayashi, Y.; Ishikawa, K.; Tabata, S.: Prediction of the coding sequences of unidentified human genes. II. The coding sequences of 40 new genes (KIAA0041–KIAA0080) deduced by analysis of cDNA clones from human cell line KG-1. DNA Res. 1: 223–229, 1994.

[30004] 3556. Freedman, D. A.; Epstein, C. B.; Roth, J. C.; Levine, A. J.: A genetic approach to mapping the p53 binding site in

the MDM2 protein. *Molec.Med.* 3: 248–259, 1997.

- [30005] 3557.Fuchs, S. Y.; Adler, V.; Buschmann, T.; Yin, Z.; Wu, X.; Jones,S. N.; Ronai, Z.: JNK targets p53 ubiquitination and degradationin nonstressed cells. *Genes Dev.* 12: 2658–2663, 1998.
- [30006] 3558.Mayo, L. D.; Donner, D. B.: A phosphatidylinositol 3–kinase/Aktpathway promotes translocation of Mdm2 from the cytoplasm to the nucleus. *Proc.Nat. Acad. Sci.* 98: 11598–11603, 2001.
- [30007] 3559.Mitchell, E. L. D.; White, G. R. M.; Santibanez–Koref, M. F.;Varley, J. M.; Heighway, J.: Mapping of gene loci in the q13–q15region of chromosome 12. *Chromosome Res.* 3: 261–262, 1995.
- [30008] 3560.Momand, J.; Zambetti, G. P.; Olson, D. C.; George, D. L.; Levine,A. J.: The mdm–2 oncogene product forms a complex with the p53 proteinand inhibits p53–mediated transactivation. *Cell* 69: 1237–1245, 1992.
- [30009] 3561.Oliner, J. D.; Kinzler, K. W.; Meltzer, P. S.; George, D. L.;Vogelstein, B.: Amplification of a gene encoding a p53–associatedprotein in human sarcomas. *Nature* 358: 80–83, 1992.
- [30010] 3562.Ries, S.; Biederer, C.; Woods, D.; Shifman, O.; Shira–sawa, S.;Sasazuki, T.; McMahon, M.; Oren, M.; McCormick,

F.: Opposing effects of Ras on p53: transcriptional activation of mdm2 and induction of p19(ARF). *Cell* 103: 321–330, 2000.

- [30011] 3563. Shenoy, S. K.; McDonald, P. H.; Kohout, T. A.; Lefkowitz, R. J.: Regulation of receptor fate by ubiquitination of activated beta-2-adrenergic receptor and beta-arrestin. *Science* 294: 1307–1313, 2001.
- [30012] 3564. Shieh, S.-Y.; Ikeda, M.; Taya, Y.; Prives, C.: DNA damage-induced phosphorylation of p53 alleviates inhibition by MDM2. *Cell* 91: 325–334, 1997.
- [30013] 3565. Testa, J. R.; Bellacosa, A.: AKT plays a central role in tumorigenesis. *Proc. Nat. Acad. Sci.* 98: 10983–10985, 2001.
- [30014] 3566. Xiao, Z.-X.; Chen, J.; Levine, A. J.; Modjtahedi, N.; Xing, J.; Sellers, W. R.; Livingston, D. M.: Interaction between the retinoblastoma protein and the oncoprotein MDM2. *Nature* 375: 694–698, 1995.
- [30015] 3567. Zhang, Y.; Xiong, Y.: A p53 amino-terminal nuclear export signal inhibited by DNA damage-induced phosphorylation. *Science* 292: 1910–1915, 2001.
- [30016] 3568. Balazs, I.: Personal Communication. New York, N. Y. 8/23/1983.
- [30017] 3569. Bezieau, S.; Devilder, M.-C.; Avet-Loiseau, H.; Mel-

lerin, M.-P.; Puthier, D.; Pennarun, E.; Rapp, M.-J.; Harousseau, J.-L.; Moisan, J.-P.; Bataille, R.: High incidence of N and K-Ras activating mutations in multiple myeloma and primary plasma cell leukemia at diagnosis. Hum. Mutat. 18: 212-224, 2001.

- [30018] 3570. Bos, J. L.; Toksoz, D.; Marshall, C. J.; Verlaan-de Vries, M.; Veeneman, G. H.; van der Eb, A. J.; van Boom, J. H.; Janssen, J. W. G.; Steenvoorden, A. C. M.: Amino-acid substitutions at codon 13 of the N-ras oncogene in human acute myeloid leukaemia. Nature 315: 726-730, 1985.
- [30019] 3571. Davis, M.; Malcolm, S.; Hall, A.: The N-ras oncogene is located on the short arm of chromosome 1. (Abstract) Cytogenet. Cell Genet. 37: 448-449, 1984.
- [30020] 3572. Davis, M.; Malcolm, S.; Hall, A.; Marshall, C. J.: Localisation of the human N-ras oncogene to chromosome 1cen-p21 by in situ hybridisation. EMBO J. 2: 2281-2283, 1983.
- [30021] 3573. Deka, R.; Majumder, P. P.; Warren, A. C.; Surti, U.; Hoffner, L.; Hauselman, E.; Antonarakis, S. E.; Ferrell, R. E.; Chakravarti, A.: Gene-centromere mapping using ovarian teratomas: results from chromosomes 1p, 13q and 21q. (Abstract) Am. J. Hum. Genet. 45 (suppl.): A137 only, 1989.

- [30022] 3574.de Martinville, B.; Cunningham, J. M.; Murray, M. J.; Francke, U.: The N-ras oncogene assigned to chromosome 1 (p31-cen) by somatic cell hybrid analysis. (Abstract) Cytogenet. Cell Genet. 37: 531 only, 1984.
- [30023] 3575.Hall, A.; Brown, R.: Human N-ras: cDNA cloning and gene structure. Nucleic Acids Res. 13: 5255–5268, 1985.
- [30024] 3576.Hall, A.; Marshall, C. J.; Spurr, N. K.; Weiss, R. A.: Identification of transforming gene in two human sarcoma cell lines as a new member of the ras gene family located on chromosome 1. Nature 303: 396–400, 1983.
- [30025] 3577.Linder, D.; McCaw, B. F.; Hecht, F.: Parthenogenic origin of benign ovarian teratomas. New Eng. J. Med. 292: 63–66, 1975.
- [30026] 3578.Nitta, N.; Ochiai, M.; Nagao, M.; Sugimura, T.: Amino-acid substitution at codon 13 of the N-ras oncogene in rectal cancer in a Japanese patient. Jpn. J. Cancer Res. 78: 21–26, 1987.
- [30027] 3579.Nobori, T.; Hexdall, L. E.; Carson, D. A.: A polymorphic region defined by pCN2 (the 3-prime nontranslated region of N-ras) maps to chromosome 9cen-p12. Hum. Genet. 87: 433–437, 1991.
- [30028] 3580.Parrington, J. M.; West, L. F.; Povey, S.: The origin of ovarian teratomas. J. Med. Genet. 21: 4–12, 1984.

- [30029] 3581. Popescu, N. C.; Amsbaugh, S. C.; DiPaolo, J. A.; Tronick, S. R.; Aaronson, S. A.; Swan, D. C.: Chromosomal localization of three human ras genes by in situ molecular hybridization. *Somat. Cell Molec. Genet.* 11: 149–155, 1985.
- [30030] 3582. Povey, S.; Morton, N. E.; Sherman, S. L.: Report of the committee on the genetic constitution of chromosomes 1 and 2 (HGM8). *Cytogenet. Cell Genet.* 40: 67–106, 1985.
- [30031] 3583. Rabin, M.; Watson, M.; Barker, P.; Ryan, J.; Breg, W. R.; Ruddle, F. H.: Chromosomal assignment of human c-fos and N-ras oncogenes. (Abstract) *Am. J. Hum. Genet.* 35: 148A only, 1983.
- [30032] 3584. Rabin, M.; Watson, M.; Barker, P. E.; Ryan, J.; Breg, W. R.; Ruddle, F. H.: NRAS transforming gene maps to region p11–p13 on chromosome 1 by in situ hybridization. *Cytogenet. Cell Genet.* 38: 70–72, 1984.
- [30033] 3585. Ryan, J.; Barker, P. E.; Shimizu, K.; Wigler, M.; Ruddle, F. H.: Chromosomal assignment of a family of human oncogenes. *Proc. Nat. Acad. Sci.* 80: 4460–4463, 1983.
- [30034] 3586. Ashkar, S.; Weber, G. F.; Panoutsakopoulou, V.; Sanchirico, M. E.; Jansson, M.; Zawaideh, S.; Rittling, S. R.; Denhardt, D. T.; Glimcher, M. J.; Cantor, H.: Eta-1 (osteopontin): an early component of type–

1(cell-mediated) immunity. Science 287: 860–864, 2000.

- [30035] 3587.Baccarini-Contrì, M.; Taparelli, F.; Pasquali-Ronchetti, I.: Osteopontin is a constitutive component of normal elastic fibers in human skin and aorta. Matrix Biol. 14: 553–560, 1994.
- [30036] 3588.Beck, G. R., Jr.; Zerler, B.; Moran, E.: Phosphate is a specific signal for induction of osteopontin gene expression. Proc. Nat. Acad.Sci. 97: 8352–8357, 2000.
- [30037] 3589.Crosby, A. H.; Lyu, M. S.; Lin, K.; McBride, O. W.; Kerr, J. M.; Aplin, H. M.; Fisher, L. W.; Young, M. F.; Kozak, C. A.; Dixon, M.J.: Mapping of the human and mouse bone sialoprotein and osteopontin loci. Mammalian Genome 7: 149–151, 1996.
- [30038] 3590.Fisher, L. W.; McBride, O. W.; Termine, J. D.; Young, M. F.: Human bone sialoprotein: deduced protein sequence and chromosomal localization. J.Biol. Chem. 265: 2347–2351, 1990.
- [30039] 3591.Graf, K.; Do, Y. S.; Ashizawa, N.; Meehan, W. P.; Giachelli, C.M.; Marboe, C. C.; Fleck, E.; Hsueh, W. A.: Myocardial osteopontin expression is associated with left ventricular hypertrophy. Circulation 96:3063–3071, 1997.
- [30040] 3592.Kiefer, M. C.; Bauer, D. M.; Barr, P. J.: The cDNA and derived amino acid sequence for human osteopontin. Nu-

cleic Acids Res. 17:3306, 1989.

[30041] 3593.Kim, J.-H.; Skates, S. J.; Uede, T.; Wong, K.; Schorge, J. O.;Feltmate, C. M.; Berkowitz, R. S.; Cramer, D. W.; Mok, S. C.: Osteopontinas a potential diagnostic biomarker for ovarian cancer. J.A.M.A. 287:1671–1679, 2002.

[30042] 3594.Kohri, K.; Nomura, S.; Kitamura, Y.; Nagata, T.; Yoshioka, K.;Iguchi, M.; Yamate, T.; Umekawa, T.; Suzuki, Y.; Sinohara, H.; Kurita,T.: Structure and expression of the mRNA encoding urinary stone protein(osteopontin). J. Biol. Chem. 268: 15180–15184, 1993.

[30043] 3595.Kohri, K.; Suzuki, Y.; Yoshida, K.; Yamamoto, K.; Amasaki, N.;Yamate, T.; Umekawa, T.; Iguchi, M.; Sino-hara, H.; Kurita, T.: Molecularcloning and sequencing of cDNA encoding urinary stone protein, whichis identical to osteopontin. Biochem. Biophys. Res. Commun. 184:859–864, 1992.

[30044] 3596.Liaw, L.; Birk, D. E.; Ballas, C. B.; Whitsitt, J. S.; Davidson,J. M.; Hogan, B. L. M.: Altered wound healing in mice lacking a functionalosteopontin gene (spp1). J. Clin. Invest. 101: 1468–1478, 1998.

[30045] 3597.Morimoto, I.; Sasaki, Y.; Ishida, S.; Imai, K.; Tokino, T.: Identificationof the osteopontin gene as a direct target of TP53. Genes ChromosomesCancer 33: 270–278, 2002.

- [30046] 3598.Reinholt, F. P.; Hultenby, K.; Oldberg, A.; Heinegard, D.: Osteopontin--a possible anchor of osteoclasts to bone. Proc. Nat. Acad. Sci. 87:4473-4475, 1990.
- [30047] 3599.Singh, K.; Balligand, J.-L.; Fischer, T. A.; Smith, T. W.; Kelly, R. A.: Glucocorticoids increase osteopontin expression in cardiac myocytes and microvascular endothelial cells: role in regulation of inducible nitric oxide synthase. J. Biol. Chem. 270: 28471-28478, 1995.
- [30048] 3600.Weber, G. F.; Ashkar, S.; Glimcher, M. J.; Cantor, H.: Receptor-ligand interaction between CD44 and osteopontin (Eta-1). Science 271: 509-512, 1996.
- [30049] 3601.Staal, A.; van Wijnen, A. J.; Birkenhager, J. C.; Pols, H. A.P.; Pahl, J.; DeLuca, H.; Gaub, M.-P.; Lian, J. B.; Stein, G. S.; van Leeuwen, J. P. T. M.; Stein, J. L.: Distinct conformations of vitamin D receptor/retinoid X receptor-alpha heterodimers are specified by dinucleotide differences in the vitamin D-responsive elements of the osteocalcin and osteopontin genes. Molec. Endocr. 10: 1444-1456, 1996.
- [30050] 3602.Young, M. F.; Kerr, J. M.; Termine, J. D.; Wewer, U. M.; Wang, M. G.; McBride, O. W.; Fisher, L. W.: cDNA cloning, mRNA distribution and heterogeneity, chromosomal location and RFLP analysis of human osteopontin. Genomics 7: 491-502, 1990.

- [30051] 3603.Vogelstein, B.; Fearon, E. R.; Hamilton, S. R.; Kern, S. E.; Preisinger,A. C.; Leppert, M.; Nakamura, Y.; White, R.; Smits, A. M. M.; Bos,J. L.: Genetic alterations during colorectal-tumor development. *NewEng. J. Med.* 319: 525–532, 1988.
- [30052] 3604.Zhao, C.; Takita, J.; Tanaka, Y.; Setou, M.; Nakagawa, T.; Takeda,S.; Yang, H. W.; Terada, S.; Nakata, T.; Takei, Y.; Saito, M.; Tsuji,S.; Hayashi, Y.; Hirokawa, N.: Charcot-Marie-Tooth disease type 2Acaused by mutation in a microtubule motor KIF1B-beta. *Cell* 105:587–597, 2001.
- [30053] 3605.Maemura, K.; Kurihara, H.; Kurihara, Y.; Oda, H.; Ishikawa, T.;Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Yazaki, Y.: Sequenceanalysis, chromosomal location, and developmental expression of themouse preproendothelin-1 gene. *Genomics* 31: 177–184, 1996.
- [30054] 3606.Maggi, M.; Barni, T.; Fantoni, G.; Mancina, R.; Pupilli, C.; Luconi,M.; Crescioli, C.; Serio, M.; Vannelli, G. B.: Expression and biologicaleffects of endothelin-1 in human gonadotropin-releasing hormone-secretingneurons. *J. Clin. Endocr. Metab.* 85: 1658–1665, 2000.
- [30055] 3607.Napolitano, M.; Miceli, F.; Calce, A.; Vacca, A.; Gulino, A.;Apa, R.; Lanzone, A.: Expression and relationship between endothelin-1messenger ribonucleic acid

(mRNA) and inducible/endothelial nitricoxide synthase mRNA isoforms from normal and preeclamptic placentas. J.Clin. Endocr. Metab. 85: 2318–2323, 2000.

- [30056] 3608.Okafor, M. C.; Delamere, N. A.: The inhibitory influence of endothelinon active sodium–potassium transport in porcine lens. Invest. Ophthal.Vis. Sci. 42: 1018–1023, 2001.
- [30057] 3609.Pache, M.; Kaiser, H. J.; Haufschild, T.; Lubeck, P.; Flammer,J.: Increased endothelin–1 plasma levels in giant cell arteritis:a report on four patients. Am. J. Ophthal. 133: 160–162, 2002.
- [30058] 3610.Pages, J.–C.; Drieu, C.; Blanche, H.; Beckmann, J.; Cann, H. M.: A short tandem repeat polymorphism at the endothelin 1 (EDN1) locus. Hum.Molec. Genet. 2: 90, 1993.
- [30059] 3611.Pezzetti, F.; Scapoli, L.; Martinelli, M.; Carinci, F.; Brunelli,G.; Carls, F. P.; Palomba, F.; Gombos, F.; Carinci, P.; Tognon, M.: Linkage analysis of candidate endothelin pathway genes in nonsyndromicfamilial orofacial cleft. Ann. Hum. Genet. 64: 341–347, 2000.
- [30060] 3612.Yanagisawa, H.; Hammer, R. E.; Richardson, J. A.; Williams, S.C.; Clouthier, D. E.; Yanagisawa, M.: Role of endothelin–1/endothelin–Areceptor–mediated signaling

pathway in the aortic arch patterning in mice. *J. Clin. Invest.* 102: 22–33, 1998.

- [30061] 3613. Yanagisawa, H.; Yanagisawa, M.; Kapur, R. P.; Richardson, J. A.; Williams, S. C.; Clouthier, D. E.; de Wit, D.; Emoto, N.; Hammer, R. E.: Dual genetic pathways of endothelin-mediated intercellular signaling revealed by targeted disruption of endothelin converting enzyme-1 gene. *Development* 125: 825–836, 1998.
- [30062] 3614. Zeidel, M. L.; Brady, H. R.; Kone, B. C.; Gullans, S. R.; Brenner, B. M.: Endothelin, a peptide inhibitor of Na(+)-K(+)-ATPase in intact renal tubular epithelial cells. *Am. J. Physiol.* 257: C1101–C1107, 1989.
- [30063] 3615. Bloch, K. D.; Hong, C. C.; Eddy, R. L.; Shows, T. B.; Quertermous, T.: cDNA cloning and chromosomal assignment of the endothelin 2 gene: vasoactive intestinal contractor peptide is rat endothelin 2. *Genomics* 10: 236–242, 1991.
- [30064] 3616. Deng, A. Y.; Dene, H.; Pravenec, M.; Rapp, J. P.: Genetic mapping of two new blood pressure quantitative trait loci in the rat by genotyping endothelin system genes. *J. Clin. Invest.* 93: 2701–2709, 1994.
- [30065] 3617. Ohkubo, S.; Ogi, K.; Hosoya, M.; Matsumoto, H.; Suzuki, N.; Kimura, C.; Onda, H.; Fujino, M.: Specific ex-

pression of human endothelin-2(ET-2) gene in a renal adenocarcinoma cell line: molecular cloning of cDNA encoding the precursor of ET-2 and its characterization. FEBS Lett. 274: 136-140, 1990.

- [30066] 3618. Baynash, A. G.; Hosoda, K.; Giaid, A.; Richardson, J. A.; Emoto, N.; Hammer, R. E.; Yanagisawa, M.: Interaction of endothelin-3 with endothelin-B receptor is essential for development of epidermal melanocytes and enteric neurons. Cell 79: 1277-1285, 1994.
- [30067] 3619. Gros, P.; Croop, J.; Housman, D.: Mammalian multidrug resistance gene: complete cDNA sequence indicates strong homology to bacterial transport proteins. Cell 47: 371-380, 1986.
- [30068] 3620. Gros, P.; Neriah, Y. B.; Croop, J. M.; Housman, D. E.: Isolation and expression of a complementary DNA that confers multidrug resistance. Nature 323: 728-731, 1986.
- [30069] 3621. Lerman, C.; Caporaso, N. E.; Audrain, J.; Main, D.; Bowman, E. D.; Lockshin, B.; Boyd, N. R.; Shields, P. G.: Evidence suggesting the role of specific genetic factors in cigarette smoking. Health Psych. 18: 14-20, 1999.
- [30070] 3622. Lossie, A. C.; Vandenberg, D. J.; Uhl, G. R.; Camper, S. A.: Localization of the dopamine transporter gene, Dat1, on mouse chromosome

- [30071] 3623.Mammalian Genome 5: 117–118, 1994.13. Sabol, S. Z.; Nelson, M. L.; Fisher, C.; Gunzerath, L.; Brody,C. L.; Hu, S.; Sirota, L. A.; Marcus, S. E.; Greenberg, B. D.; Lucas,F. R., IV; Benjamin, J.; Murphy, D. L.; Hamer, D. H.: A genetic associationfor cigarette smoking behavior. Health Psych. 18: 7–13, 1999.
- [30072] 3624.Tiihonen, J.; Kuikka, J.; Bergstrom, K.; Hakola, P.; Karhu, J.;Ryynanen, O.–P.; Fohr, J.: Altered striatal dopamine re–uptake sitedensities in habitually violent and non–violent alcoholics. NatureMed. 1: 654–657, 1995.
- [30073] 3625.Vandenbergh, D. J.; Persico, A. M.; Hawkins, A. L.; Griffin, C.A.; Li, X.; Jabs, E. W.; Uhl, G. R.: Human dopamine transporter gene(DAT1) maps to chromosome 5p15.3 and displays a VNTR. Genomics 14:1104–1106, 1992.
- [30074] 3626.Vandenbergh, D. J.; Persico, A. M.; Uhl, G. R.: A human dopaminetransporter cDNA predicts reduced glyco–sylation, displays a novelrepetitive element and provides racially–dimorphic Taql RFLPs. Molec.Brain Res. 15: 161–166, 1992.
- [30075] 3627.Waldman, I. D.; Rowe, D. C.; Abramowitz, A.; Kozel, S. T.; Mohr,J. H.; Sherman, S. L.; Cleveland, H. H.; Sanders, M. L.; Gard, J.M. C.; Stever, C.: Association and linkage of

the dopamine transporter gene and attention-deficit hyperactivity disorder in children: heterogeneity owing to diagnostic subtype and severity. *Am. J. Hum. Genet.* 63:1767–1776, 1998.

- [30076] 3628. Huang, H.; Mahler-Araujo, B. M.; Sankila, A.; Chimelli, L.; Yonekawa, Y.; Kleihues, P.; Ohgaki, H.: APC mutations in sporadic medulloblastomas. *Am. J. Path.* 156: 433–437, 2000.
- [30077] 3629. Behan, D. P.; Potter, E.; Lewis, K. A.; Jenkins, N. A.; Copeland, N.; Lowry, P. J.; Vale, W. W.: Cloning and structure of the human corticotropin releasing factor-binding protein gene (CRHBP). *Genomics* 16:63–68, 1993.
- [30078] 3630. Lublin, D. M.; Lemons, R. S.; Le Beau, M. M.; Holers, V. M.; Tykocinski, M. L.; Medof, M. E.; Atkinson, J. P.: The gene encoding decay-accelerating factor (DAF) is located in the complement-regulatory locus on the long arm of chromosome 1. *J. Exp. Med.* 165: 1731–1736, 1987.
- [30079] 3631. von Boehmer, H.; Sarukhan, A.: GAD, a single autoantigen for diabetes. *Science* 284: 1135–1136, 1999.
- [30080] 3632. Williamson, E. A.; Ince, P. G.; Harrison, D.; Kendall-Taylor, P.; Harris, P. E.: G-protein mutations in human pituitary adrenocorticotrophic hormone-secreting adenomas. *Europ. J. Clin. Invest.* 25: 128–131, 1995.

[30081] 3633.Marsden, V. S.; O'Connor, L.; O'Reilly, L. A.; Silke, J.; Metcalf,D.; Ekert, P. G.; Huang, D. C. S.; Cecconi, F.; Kuida, K.; Tomaselli,K. J.; Roy, S.; Nicholson, D. W.; Vaux, D. L.; Bouillet, P.; Adams,J. M.; Strasser, A.: Apoptosis initiated by Bcl-2-regulated caspaseactivation independently of the cytochrome c/Apaf-1/caspase-9 apoptosome. *Nature* 419:634-637, 2002.

[30082] 3634.Huelsken, J.; Vogel, R.; Erdmann, B.; Cotsarelis, G.; Birchmeier,W.: Beta-catenin controls hair follicle morphogenesis and stem celldifferentiation in the skin. *Cell* 105: 533-545, 2001.

[30083] 3635.Ilyas, M.; Tomlinson, I. P. M.; Rowan, A.; Pignatelli, M.; Bodmer,W. F.: Beta-catenin mutations in cell lines established from humancolorectal cancers. *Proc. Nat. Acad. Sci.* 94: 10330-10334, 1997.

[30084] 3636.Iwao, K.; Nakamori, S.; Kameyama, M.; Imaoka, S.; Kinoshita, M.;Fukui, T.; Ishiguro, S.; Nakamura, Y.; Miyoshi, Y.: Activation ofthe beta-catenin gene by interstitial deletions involving exon 3 inprimary colorectal carcinomas without adenomatous polyposis coli mutations. *CancerRes.* 58: 1021-1026, 1998.

[30085] 3637.Kawasaki, Y.; Senda, T.; Ishidata, T.; Koyama, R.; Morishita,T.; Iwayama, Y.; Higuchi, O.; Akiyama, T.: Asef, a

link between the tumor suppressor APC and G-protein signaling. *Science* 289: 1194–1197, 2000.

[30086] 3638. Koch, A.; Denkhaus, D.; Albrecht, S.; Leuschner, I.; von Schweinitz, D.; Pietsch, T.: Childhood hepatoblastomas frequently carry a mutated degradation targeting box of the beta-catenin gene. *Cancer Res.* 59:269–273, 1999.

[30087] 3639. Korinek, V.; Barker, N.; Morin, P. J.; van Wichen, D.; de Weger, R.; Kinzler, K. W.; Vogelstein, B.; Clevers, H.: Constitutive transcriptional activation by a beta-catenin–Tcf complex in APC–/– colon carcinoma. *Science* 275:1784–1787, 1997.

[30088] 3640. Kraus, C.; Liehr, T.; Hulsken, J.; Behrens, J.; Birchmeier, W.; Grzeschik, K.–H.; Ballhausen, W. G.: Localization of the human beta-catenin gene (CTNNB1) to 3p21: a region implicated in tumor development. *Genomics* 23:272–274, 1994.

[30089] 3641. Legoix, P.; Bluteau, O.; Bayer, J.; Perret, C.; Balabaud, C.; Belghiti, J.; Franco, D.; Thomas, G.; Laurent–Puig, P.; Zucman–Rossi, J.: Beta-catenin mutations in hepatocellular carcinoma correlate with a low rate of loss of heterozygosity. *Oncogene* 18: 4044–4046, 1999.

[30090] 3642. Lin, S.–Y.; Xia, W.; Wang, J. C.; Kwong, K. Y.; Spohn, B.; Wen, Y.; Pestell, R. G.; Hung, M.–C.: Beta-catenin, a

novel prognostic marker for breast cancer: its roles in cyclin D1 expression and cancer progression. *Proc. Nat. Acad. Sci.* 97: 4262–4266, 2000.

- [30091] 3643. McCrea, P. D.; Turck, C. W.; Gumbiner, B.: A homolog of the armadillo protein in *Drosophila* (plakoglobin) associated with E-cadherin. *Science* 254:1359–1361, 1991.
- [30092] 3644. Morin, P. J.; Sparks, A. B.; Korinek, V.; Barker, N.; Clevers, H.; Vogelstein, B.; Kinzler, K. W.: Activation of beta-catenin–Tcf signaling in colon cancer by mutations in beta-catenin or APC. *Science* 275:1787–1790, 1997.
- [30093] 3645. Murase, S.; Mosser, E.; Schuman, E. M.: Depolarization drives beta-catenin into neuronal spines promoting changes in synaptic structure and function. *Neuron* 35: 91–105, 2002.
- [30094] 3646. Nollet, F.; Berx, G.; Molemans, F.; van Roy, F.: Genomic organization of the human beta-catenin gene (CTNNB1). *Genomics* 32: 413–424, 1996.
- [30095] 3647. Peifer, M.: Cancer, catenins, and cuticle pattern: a complex connection. *Science* 262: 1667–1668, 1993.
- [30096] 3648. Rodova, M.; Islam, M. R.; Maser, R. L.; Calvet, J. P.: The polycystic kidney disease–1 promoter is a target of the beta-catenin/T-cell factor pathway. *J. Biol. Chem.* 277:

29577–29583, 2002.

- [30097] 3649. Roose, J.; Huls, G.; van Beest, M.; Moerer, P.; van der Horn, K.; Goldschmeding, R.; Logtenberg, T.; Clevers, H.: Synergy between tumor suppressor APC and the beta-catenin–Tcf4 target Tcf1. *Science* 285:1923–1926, 1999.
- [30098] 3650. Rubinfeld, B.; Robbins, P.; El-Gamil, M.; Albert, I.; Porfiri, E.; Polakis, P.: Stabilization of beta-catenin by genetic defects in melanoma cell lines. *Science* 275: 1790–1792, 1997.
- [30099] 3651. Saadi-Kheddouchi, S.; Berrebi, D.; Romagnolo, B.; Cluzeaud, F.; Peuchmaur, M.; Kahn, A.; Vandewalle, A.; Perret, C.: Early development of polycystic kidney disease in transgenic mice expressing an activated mutant of the beta-catenin gene. *Oncogene* 20: 5972–5981, 2001.
- [30100] 3652. Sagae, S.; Kobayashi, K.; Nishioka, Y.; Sugimura, M.; Ishioka, S.; Nagata, M.; Terasawa, K.; Tokino, T.; Kudo, R.: Mutational analysis of beta-catenin gene in Japanese ovarian carcinomas: frequent mutations in endometrioid carcinomas. *Jpn. J. Cancer Res.* 90: 510–515, 1999.
- [30101] 3653. Trent, J. M.; Wiltshire, R.; Su, L.-K.; Nicolaides, N. C.; Vogelstein, B.; Kinzler, K. W.: The gene for the APC-binding protein beta-catenin (CTNNB1) maps to chromosome 3p22, a region frequently altered in human malignancies.

nancies. Cytogenet. Cell Genet. 71: 343–344, 1995.

[30102] 3654.van Hengel, J.; Nollet, F.; Berx, G.; van Roy, N.; Speleman, F.;van Roy, F.: Assignment of the human beta-catenin gene (CTNNB1) to3p22–p21.3 by fluorescence in situ hybridization. Cytogenet. CellGenet. 70: 68–70, 1995.

[30103] 3655.Chan, S. J.; San Segundo, B.; McCormick, M. B.; Steiner, D. F.: Nucleotide and predicted amino acid sequences of cloned human andmouse preprocathepsin B cDNAs. Proc. Nat. Acad. Sci. 83: 7721–7725,1986.

[30104] 3656.Deussing, J.; Roth, W.; Rommerskirch, W.; Wiederanders, B.; vonFigura, K.; Peters, C.: The genes of the lysosomal cysteine proteinasescathepsin B, H, L, and S map to different mouse chromosomes. MammalianGenome 8: 241–245, 1997.

[30105] 3657.Shigemitsu, K.; Sekido, Y.; Usami, N.; Mori, S.; Sato, M.; Horio,Y.; Hasegawa, Y.; Bader, S. A.; Gazdar, A. F.; Minna, J. D.; Hida,T.; Yoshioka, H.; Imaizumi, M.; Ueda, Y.; Takahashi, M.; Shimokata,K.: Genetic alteration of the beta-catenin gene (CTNNB1) in humanlung cancer and malignant mesothelioma and identification of a new3p21.3 homozygous deletion. Oncogene 20: 4249–4257, 2001.

[30106] 3658.Geck, P.; Sonnenschein, C.; Soto, A. M.: The

D13S171 marker, misannotated to BRCA2, links the AS3 gene to various cancers. (Letter) Am. J. Hum. Genet. 69: 461–463, 2001.

- [30107] 3659. Geck, P.; Szelei, J.; Jimenez, J.; Sonnenschein, C.; Soto, A. M.: Early gene expression during androgen-induced inhibition of proliferation of prostate cancer cells: a new suppressor candidate on chromosome 13, in the BRCA2–Rb1 locus. J. Steroid Biochem. Molec. Biol. 68:41–50, 1999.
- [30108] 3660. Kuske, M. D. A.; Johnson, J. P.: Assignment of the human PHLDA1 gene to chromosome 12q15 by radiation hybrid mapping. Cytogenet. Cell Genet. 89: 1 only, 2000.
- [30109] 3661. Reddy, P. H.; Stockburger, E.; Gillevet, P.; Tagle, D. A.: Mapping and characterization of novel (CAG)_n repeat cDNAs from adult human brain derived by the oligo capture method. Genomics 46: 174–182, 1997.
- [30110] 3662. Bontekoe, C. J. M.; McIlwain, K. L.; Nieuwenhuizen, I. M.; Yuva-Paylor, L. A.; Nellis, A.; Willemsen, R.; Fang, Z.; Kirkpatrick, L.; Bakker, C. E.; McAninch, R.; Cheng, N. C.; Merriweather, M.; Hoogeveen, A. T.; Nelson, D.; Paylor, R.; Oostra, B. A.: Knockout mouse model for Fxr2: a model for mental retardation. Hum. Molec. Genet. 11: 487–498, 2002.

- [30111] 3663.Tamanini, F.; Willemsen, R.; van Unen, L.; Bontekoe, C.; Galjaard,H.; Oostra, B. A.; Hoogeveen, A. T.: Differential expression of FMR1,FXR1 and FXR2 proteins in human brain and testis. *Hum. Molec. Genet.* 6:1315–1322, 1997.
- [30112] 3664.Zhang, Y.; O'Connor, J. P.; Siomi, M. C.; Srinivasan, S.; Dutra,A.; Nussbaum, R. L.; Dreyfuss, G.: The fragile X mental retardationsyndrome protein interacts with novel homologs FXR1 and FXR2. *EMBOJ.* 14: 5358–5366, 1995.
- [30113] 3665.Lee, J. K.; Bhakta, S.; Rosen, S. D.; Hemmerich, S.: Cloning andcharacterization of a mammalian N-acetylglucosamine–6–sulfotransferasethat is highly restricted to intestinal tissue. *Biochem. Biophys.Res. Commun.* 263: 543–549, 1999.
- [30114] 3666.Raymond, C. S.; Parker, E. D.; Kettlewell, J. R.; Brown, L. G.;Page, D. C.; Kusz, K.; Jaruzelska, J.; Reinberg, Y.; Fletjer, W. L.;Bardwell, V. J.; Hirsch, B.; Zarkower, D.: A region of human chromosome9p required for testis development contains two genes related to knownsexual regulators. *Hum. Molec. Genet.* 8: 989–996, 1999.
- [30115] 3667.Colonna, M.; Samaridis, J.: Cloning of immunoglobulin–superfamilymembers associated with HLA–C and HLA–B recognition by human naturalkiller cells. *Science* 268: 405–408, 1995.

- [30116] 3668.Lanier, L. L.: Turning on natural killer cells. J. Exp. Med. 191:1259–1262, 2000.
- [30117] 3669.Lanier, L. L.: Natural killer cells: from no receptors to toomany. Immunity 6: 371–378, 1997.
- [30118] 3670.Lanier, L. L.: Follow the leader: NK cell receptors for classicaland nonclassical MHC class I. Cell 92: 705–707, 1998.
- [30119] 3671.Ljunggren, H. G.; Karre, K.: In search of the 'missing self':MHC molecules and NK cell recognition. Immun. To-day 11: 237–244,1990.
- [30120] 3672.Rajalingam, R.; Krausa, P.; Shilling, H. G.; Stein, J. B.; Balamurugan,A.; McGinnis, M. D.; Cheng, N. W.; Mehra, N. K.; Parham, P.: DistinctiveKIR and HLA diversity in a panel of north Indian Hindus. Immunogenetics 53:1009–1019, 2002.
- [30121] 3673.Wagtmann, N.; Biassoni, R.; Cantoni, C.; Verdiani, S.; Malnati,M. S.; Vitale, M.; Bottino, C.; Moretta, L.; Moretta, A.; Long, E.O.: Molecular clones of the p58 NK cell receptor reveal immunoglobulin–relatedmolecules with diversity in both the extra– and intracellular domains. Immunity 2:439–449, 1995.
- [30122] 3674.Wagtmann, N.; Rajagopalan, S.; Winter, C. C.; Peruzzi, M.; Long,E. O.: Killer cell inhibitory receptors spe–

cific for HLA-C and HLA-B identified by direct binding and by functional transfer. *Immunity* 3:801–809, 1995.

- [30123] 3675. Wende, H.; Colonna, M.; Ziegler, A.; Volz, A.: Organization of the leukocyte receptor cluster (LRC) on human chromosome 19q13.4. *Mammalian Genome* 10: 154–160, 1999.
- [30124] 3676. Dohring, C.; Samaridis, J.; Colonna, M.: Alternatively spliced forms of human killer inhibitory receptors. *Immunogenetics* 44: 227–230, 1996.
- [30125] 3677. Matsumoto, N.; Laub, F.; Aldabe, R.; Zhang, W.; Ramirez, F.; Yoshida, T.; Terada, M.: Cloning the cDNA for a new human zinc finger protein defines a group of closely related Kruppel-like transcription factors. *J. Biol. Chem.* 273: 28229–28237, 1998.
- [30126] 3678. Okazaki, I.; Kinoshita, K.; Muramatsu, M.; Yoshikawa, K.; Honjo, T.: The AID enzyme induces class switch recombination in fibroblasts. *Nature* 416:340–345, 2002.
- [30127] 3679. Petersen-Mahrt, S. K.; Harris, R. S.; Neuberger, M. S.: AID mutates *E. coli* suggesting a DNA deamination mechanism for antibody diversification. *Nature* 418:99–104, 2002.
- [30128] 3680. Revy, P.; Muto, T.; Levy, Y.; Geissmann, F.; Plebani,

A.; Sanal, O.; Catalan, N.; Forveille, M.; Dufourcq-Lagelouse, R.; Gennery, A.; Tezcan, I.; Ersoy, F.; and 9 others: Activation-induced cytidine deaminase (AID) deficiency causes the autosomal recessive form of the hyper-IgM syndrome (HIGM2). *Cell* 102: 565–575, 2000.

[30129] 3681. Yoshikawa, K.; Okazaki, I.; Eto, T.; Kinoshita, K.; Muramatsu, M.; Nagaoka, H.; Honjo, T.: AID enzyme-induced hypermutation in an actively transcribed gene in fibroblasts. *Science* 296: 2033–2036, 2002.

[30130] 3682. Geiszt, M.; Kopp, J. B.; Varnai, P.; Leto, T. L.: Identification of Renox, an NAD(P)H oxidase in kidney. *Proc. Nat. Acad. Sci.* 97: 8010–8014, 2000.

[30131] 3683. Shiose, A.; Kuroda, J.; Tsuruya, K.; Hirai, M.; Hirakata, H.; Naito, S.; Hattori, M.; Sakaki, Y.; Sumimoto, H.: A novel superoxide-producing NAD(P)H oxidase in kidney. *J. Biol. Chem.* 276: 1417–1423, 2001.

[30132] 3684. Kokame, K.; Kato, H.; Miyata, T.: Homocysteine-responsive genes in vascular endothelial cells identified by differential display analysis: GRP78/BiP and novel genes. *J. Biol. Chem.* 271: 29659–29665, 1996.

[30133] 3685. Park, H.; Adams, M. A.; Lachat, P.; Bosman, F.; Pang, S. C.; Graham, C. H.: Hypoxia induces the expression of a 43-kDa protein (PROXY-1) in normal and malignant cells.

Biochem. Biophys. Res. Commun. 276:321–328, 2000.

- [30134] 3686. Shimono, A.; Okuda, T.; Kondoh, H.: N-myc-dependent repression of Ndr1, a gene identified by direct subtraction of whole mouse embryonic DNAs between wild type and N-myc mutant. *Mech. Dev.* 83: 39–52, 1999.
- [30135] 3687. van Belzen, N.; Dinjens, W. N. M.; Diesveld, M. P. G.; Groen, N. A.; van der Made, A. C. J.; Nozawa, Y.; Vliestra, R.; Trapman, J.; Bosman, F. T.: A novel gene which is up-regulated during colon epithelial cell differentiation and down-regulated in colorectal neoplasms. *Lab. Invest.* 77: 85–92, 1997.
- [30136] 3688. Zhou, D.; Salnikow, K.; Costa, M.: Cap43, a novel gene specifically induced by Ni²⁺ compounds. *Cancer Res.* 58: 2182–2189, 1998.
- [30137] 3689. Janz, R.; Sudhof, T. C.: Cellugyrin, a novel ubiquitous form of synaptogyrin that is phosphorylated by pp60(c-src). *J. Biol. Chem.* 273: 2851–2857, 1998.
- [30138] 3690. Ludwig, J.; Kersch, S.; Brandt, U.; Pfeiffer, K.; Getlawi, F.; Apps, D. K.; Schagger, H.: Identification and characterization of a novel 9.2-kDa membrane sector-associated protein of vacuolar proton-ATPase from chromaffin granules. *J. Biol. Chem.* 273: 10939–10947, 1998.
- [30139] 3691. Nanji, M.; Coronado, V. A.; Cox, D. W.: ATP6H, a

subunit of vacuolarATPase involved in metal transport: evaluation in canine copper toxicosis. *MammalianGenome* 12: 617–621, 2001.

- [30140] 3692.Arama, E.; Yanai, A.; Kilfin, G.; Bernstein, A.; Motro, B.: MurineNIMA-related kinases are expressed in patterns suggesting distinctfunctions in gametogenesis and a role in the nervous system. *Oncogene* 16:1813–1823, 1998.
- [30141] 3693.Fry, A. M.; Meraldi, P.; Nigg, E. A.: A centrosomal function forthe human Nek2 protein kinase, a member of the NIMA family of cellcycle regulators. *EMBO J.* 17: 470–481, 1998.
- [30142] 3694.Schultz, S. J.; Fry, A. M.; Sutterlin, C.; Ried, T.; Nigg, E. A.: Cell cycle–dependent expression of Nek2, a novel human protein kinaserelated to the NIMA mitotic regulator of *Aspergillus nidulans*. *CellGrowth Diff.* 5: 625–635, 1994.
- [30143] 3695.Schultz, S. J.; Nigg, E. A.: Identification of 21 novel humanprotein kinases, including 3 members of a family related to the cellcycle regulator nimA of *Aspergillus nidulans*. *Cell Growth Diff.* 4:821–830, 1993.
- [30144] 3696.Tamari, M.; Daigo, Y.; Nakamura, Y.: Isolation and characterizationof a novel serine threonine kinase gene on chromosome 3q22–21.3. *J.Hum. Genet.* 44: 116–120,

1999.

- [30145] 3697. Tamari, M.; Daigo, Y.; Ishikawa, S.; Nakamura, Y.: Genomic structure of a novel human gene (XYLB) on chromosome 3p22–p21.3 encoding a xylulokinase-like protein. *Cytogenet. Cell Genet.* 82: 101–104, 1998.
- [30146] 3698. Migone, T.–S.; Zhang, J.; Luo, X.; Zhuang, L.; Chen, C.; Hu, B.; Hong, J. S.; Perry, J. W.; Chen, S.–F.; Zhou, J. X. H.; Cho, Y. H.; Ullrich, S.; and 14 others: TL1A is a TNF-like ligand for DR3 and TR6/DcR3 and functions as a T cell costimulator. *Immunity* 16: 479–492, 2002.
- [30147] 3699. Zhai, Y.; Ni, J.; Jiang, G.–W.; Lu, J.; Xing, L.; Lincoln, C.; Carter, K. C.; Janat, F.; Kozak, D.; Xu, S.; Rojas, L.; Aggarwal, B. B.; Ruben, S.; Li, L.–Y.; Gentz, R.; Yu, G.–L.: VEGI, a novel cytokine of the tumor necrosis factor family, is an angiogenesis inhibitor that suppresses the growth of colon carcinomas in vivo. *FASEB J.* 13:181–189, 1999.
- [30148] 3700. Yue, T.–L.; Ni, J.; Romanic, A. M.; Gu, J.–L.; Keller, P.; Wang, C.; Kumar, S.; Yu, G.; Hart, T. K.; Wang, X.; Xia, Z.; DeWolf, W.E., Jr.; Feuerstein, G. Z.: TL1, a novel tumor necrosis factor-like cytokine, induces apoptosis in endothelial cells: involvement of activation of stress protein kinases (stress-activated protein kinase and p38 mitogen-activated protein kinase) and caspase-3-like

protease. J.Biol. Chem. 274: 1479–1486, 1999.

[30149] 3701.Hertzel, A. V.; Bernlohr, D. A.: Cloning and chromosomal location of the murine keratinocyte lipid-binding protein gene. Gene 221:235–243, 1998.

[30150] 3702.Madsen, P.; Rasmussen, H. H.; Leffers, H.; Honore, B.; Celis, J.E.: Molecular cloning and expression of a novel keratinocyte protein(psoriasis-associated fatty acid-binding protein [PA-FABP]) that is highly up-regulated in psoriatic skin and that shares similarity to fatty acid-binding proteins. J. Invest. Derm. 99: 299–305, 1992.

[30151] 3703.Siegenthaler, G.; Hotz, R.; Chatellard-Gruaz, D.; Didierjean, L.; Hellman, U.; Saurat, J. H.: Purification and characterization of the human epidermal fatty acid-binding protein: localization during epidermal cell differentiation in vivo and in vitro. Biochem J. 302:363–371, 1994.

[30152] 3704.Gu, Z.; Flemington, C.; Chittenden, T.; Zambetti, G. P.: El24, a p53 response gene involved in growth suppression and apoptosis. Molec.Cell. Biol. 20: 233–241, 2000.

[30153] 3705.Gu, Z.; Gilbert, D. J.; Valentine, V. A.; Jenkins, N. A.; Copeland, N. G.; Zambetti, G. P.: The p53-inducible gene El24/PIG8 localizes to human chromosome 11q23 and the proximal region of mouse chromosome 9. Cytogenet. Cell

Genet. 89: 230–233, 2000.

- [30154] 3706. Polyak, K.; Xia, Y.; Zweier, J. L.; Kinzler, K. W.; Vogelstein, B.: A model for p53-induced apoptosis. *Nature* 389: 300–305, 1997.
- [30155] 3707. Contente, A.; Dittmer, A.; Koch, M. C.; Roth, J.; Dobbelstein, M.: A polymorphic microsatellite that mediates induction of PIG3 by p53. *Nature Genet.* 30: 315–320, 2002.
- [30156] 3708. Hernandez, M.-C.; Andres-Barquin, P. J.; Holt, I.; Israel, M. A.: Cloning of human ENC-1 and evaluation of its expression and regulation in nervous system tumors. *Exp. Cell Res.* 242: 470–477, 1998.
- [30157] 3709. Hernandez, M.-C.; Andres-Barquin, P. J.; Israel, M. A.: Assignment of the ectodermal-neural cortex 1 gene (Enc1) to mouse chromosome band 13D1 by fluorescence in situ hybridization. *Cytogenet. Cell Genet.* 89: 158–159, 2000.
- [30158] 3710. Hernandez, M.-C.; Andres-Barquin, P. J.; Kuo, W. L.; Israel, M. A.: Assignment of the ectodermal-neural cortex 1 gene (ENC1) to human chromosome band 5q13 by in situ hybridization. *Cytogenet. Cell Genet.* 87: 89–90, 1999.
- [30159] 3711. Kim, T.-A.; Lim, J.; Ota, S.; Raja, S.; Rogers, R.; Rivenay, B.; Avraham, H.; Avraham, S.: NRP/B, a novel nuclear

matrix protein, associates with p110(RB) and is involved in neuronal differentiation. *J. Cell Biol.* 141: 553–566, 1998.

[30160] 3712. Rosenfeld, M. G.; Mermod, J.-J.; Amara, S. G.; Swanson, L. W.; Sawchenko, P. E.; Rivier, J.; Vale, W. W.; Evans, R. M.: Production of a novel neuropeptide encoded by the calcitonin gene via tissue-specific RNA processing. *Nature* 304: 129–135, 1983.

[30161] 3713. Saller, B.; Feldmann, G.; Haupt, K.; Broecker, M.; Janssen, O.E.; Roggendorf, M.; Mann, K.; Lu, M.: RT-PCR-based detection of circulating calcitonin-producing cells in patients with advanced medullary thyroid cancer. *J. Clin. Endocr. Metab.* 87: 292–296, 2002.

[30162] 3714. Simpson, N. E.; Goodfellow, P. J.; Riddell, D. C.; Hamerton, J.L.; Holden, J. J. A.; White, B. N.: Assignment of the calcitonin gene to chromosome 11 and probable exclusion of linkage between the gene and the locus for multiple endocrine neoplasia type 2. (Abstract) *Am. J. Hum. Genet.* 36: 153S, 1984.

[30163] 3715. Struthers, A. D.; Brown, M. J.; Macdonald, D. W. R.; Beacham, J. L.; Stevenson, J. C.; Morris, H. R.; MacIntyre, I.: Human calcitonin gene related peptide: a potent endogenous vasodilator in man. *Clin. Sci.* 70: 389–393, 1986.

[30164] 3716. Testa, J. R.: Personal Communication. Baltimore, Md.

3/1984.

- [30165] 3717.Tiller–Borcich, J. K.; Capili, H.; Gordan, G. S.: Human braincalcitonin gene–related peptide (CGRP) is concentrated in the locuscaeruleus. *Neuropeptides* 11: 55–61, 1988.
- [30166] 3718.Tippins, J. R.: CGRP: a novel neuropeptide from the calcitoningene is the most potent vasodilator known. *J. Hypertension* 4 (suppl.5): S102–S105, 1986.
- [30167] 3719.Tschopp, F. A.; Henke, H.; Petermann, J. B.; Tobler, P. H.; Janzer,R.; Hokfelt, T.; Lundberg, J. M.; Cuello, C.; Fischer, J. A.: Calcitoningene–related peptide and its binding sites in the human central nervoussystem and pituitary. *Proc. Nat. Acad. Sci.* 82: 248–252, 1985.
- [30168] 3720.Christopoulos, G.; Perry, K. J.; Morfis, M.; Tilakaratne, N.; Gao,Y.; Fraser, N. J.; Main, M. J.; Foord, S. M.; Sexton, P. M.: Multipleamylin receptors arise from receptor activity–modifying protein interactionwith the calcitonin receptor gene product. *Molec. Pharm.* 56: 235–242,1999.
- [30169] 3721.de Vernejoul, M.–C.: Personal Communication. Paris, France 1/19/1999.
- [30170] 3722.Gorn, A. H.; Lin, H. Y.; Yamin, M.; Auron, P. E.; Flannery, M.R.; Tapp, D. R.; Manning, C. A.; Lodish, H. F.;

Krane, S. M.; Goldring, S. R.: Cloning, characterization, and expression of a human calcitonin receptor from an ovarian carcinoma cell line. *J. Clin. Invest.* 90:1726–1735, 1992.

[30171] 3723. Chung, C. D.; Liao, J.; Liu, B.; Rao, X.; Jay, P.; Berta, P.; Shuai, K.: Specific inhibition of Stat3 signal transduction by PIAS3. *Science* 278:1803–1805, 1997.

[30172] 3724. Hagen, F. S.; Grant, F. J.; Kuijper, J. L.; Slaughter, C. A.; Moomaw, C. R.; Orth, K.; O'Hara, P. J.; Munford, R. S.: Expression and characterization of recombinant human acyloxyacyl hydrolase, a leukocyte enzyme that deacylates bacterial lipopolysaccharides. *Biochemistry* 30: 8415–8423, 1991.

[30173] 3725. Whitmore, T. E.; Mathewes, S. L.; O'Hara, P. J.; Durnam, D. M.: Chromosomal localization of the acyloxyacyl hydrolase (AOAH) gene to 7p14–p12 using fluorescence in situ hybridization. *Genomics* 21:457–458, 1994.

[30174] 3726. Greenberg, F.; Faucett, A.; Rose, E.; Bancalari, L.; Kardon, N. B.; Mizejewski, G.; Haddow, J. E.; Alpert, E.: Congenital deficiency of alpha-fetoprotein. *Am. J. Obstet. Gynec.* 167: 509–511, 1992.

[30175] 3727. Greenberg, F.; Rose, E.; Alpert, E.: Hereditary persistence of alpha-fetoprotein. *Gastroenterology* 98: 1083–1085, 1990.

- [30176] 3728.Hammer, R. E.; Krumlauf, R.; Camper, S. A.; Brinster, R. L.; Tilghman,S. M.: Diversity of alpha-fetoprotein gene expression in mice is generated by a combination of separate enhancer elements. *Science* 235:53–58, 1986.
- [30177] 3729.Ingram, R. S.; Scott, R. W.; Tilghman, S. M.: Alpha-fetoprotein and albumin genes are in tandem in the mouse genome. *Proc. Nat. Acad.Sci.* 78: 4694–4698, 1981.
- [30178] 3730.Jagodzinski, L. L.; Sargent, T. D.; Yang, M.; Glackin, C.; Bonner,J.: Sequence homology between RNAs encoding rat alpha-fetoprotein and rat serum albumin. *Proc. Nat. Acad. Sci.* 78: 3521–3525, 1981.
- [30179] 3731.Magenis, R. E.; Luo, X. Y.; Dugaiczyk, A.; Ryan, S. C.; Oosterhuis,J. E.: Chromosomal localization of the albumin and alpha-fetoprotein genes in the orangutan (*Pongo pygmaeus*) and gorilla (*Gorilla gorilla*). (Abstract) *Cytogenet. Cell Genet.* 51: 1037, 1989.
- [30180] 3732.McVey, J. H.; Michaelides, K.; Hansen, L. P.; Ferguson-Smith,M.; Tilghman, S.; Krumlauf, R.; Tuddenham, E. G. D.: A G-to-A substitution in an HNF I binding site in the human alpha-fetoprotein gene is associated with hereditary persistence of alpha-fetoprotein (HPAFP). *Hum. Molec.Genet.* 2: 379–384, 1993.
- [30181] 3733.Morinaga, T.; Sakai, M.; Wegmann, T. G.; Tamaoki,

T.: Primary structures of human alpha-fetoprotein and its mRNA. *Proc. Nat. Acad.Sci.* 80: 4604–4608, 1983.

[30182] 3734.Motulsky, A. G.: Personal Communication. Seattle, Wash. 1983.

[30183] 3735.Olsson, M.; Lindahl, G.; Ruoslahti, E.: Genetic control of alpha-fetoproteinsynthesis in the mouse. *J. Exp. Med.* 145: 819–827, 1977.

[30184] 3736.Rose, E.; Greenberg, F.; Alpert, E.: Hereditary persistence of alpha fetoprotein. (Abstract) *Am. J. Hum. Genet.* 45 (suppl.): A61, 1989.

[30185] 3737.Ruoslahti, E.; Terry, W. D.: Alpha fetoprotein and serum albumin show sequence homology. *Nature* 260: 804–805, 1976.

[30186] 3738.Sakai, M.; Morinaga, T.; Urano, Y.; Watanabe, K.; Wegmann, T.G.; Tamaoki, T.: The human alpha-fetoprotein gene: sequence organization and the 5-prime flanking region. *J. Biol. Chem.* 260: 5055–5060, 1985.

[30187] 3739.Staples, J.: Alpha fetoprotein, cancer, and benign conditions. (Letter) *Lancet* II: 1277, 1986.

[30188] 3740.Szpirer, J.; Levan, G.; Thorn, M.; Szpirer, C.: Gene mapping in the rat by mouse-rat somatic cell hybridization: synteny of the albumin and alpha-fetoprotein genes and assignment to chromosome 14. *Cytogenet. Cell Genet.*

38: 142–149, 1984.

[30189] 3741.Tilghman, S. M.: Personal Communication. Princeton, N. J. 8/12/1992.

[30190] 3742.Tilghman, S. M.; Belayew, A.: Transcriptional control of themurine albumin/alpha-fetoprotein locus during development. Proc.Nat. Acad. Sci. 79: 5254–5257, 1982.

[30191] 3743.Urano, Y.; Sakai, M.; Watanabe, K.; Tamaoki, T.: Tandem arrangementof the albumin and alpha-feto-protein genes in the human genome. Gene 32:255–261, 1984.

[30192] 3744.Vogt, T. F.; Solter, D.; Tilghman, S. M.: Raf, a trans-actinglocus, regulates the alpha-fetoprotein gene in a cell-autonomous manner. Science 236:301–303, 1987.

[30193] 3745.Voigtlander, T.; Vogel, F.: Low alpha-fetoprotein and serum albuminlevels in Morbus Down may point to a common regulatory mechanism. Hum.Genet. 71: 276–277, 1985.

[30194] 3746.Watanabe, K.; Saito, A.; Tamaoki, T.: Cell-specific enhanceractivity in a far upstream region of the human al-p-ha-fetoprotein gene. J.Biol. Chem. 262: 4812–4818, 1987.

[30195] 3747.Abbott, C. A.; Baker, E.; Sutherland, G. R.; Mc-Caughan, G. W.:Genomic organization, exact localization,

and tissue expression of the human CD26 (dipeptidyl peptidase IV) gene. *Immunogenetics* 40:331–338, 1994.

[30196] 3748. Callebaut, C.; Krust, B.; Jacotot, E.; Hovanessian, A. G.: T cell activation antigen, CD26, as a cofactor for entry of HIV in CD4⁺ cells. *Science* 262:2045–2050, 1993.

[30197] 3749. Darmoul, D.; Lacasa, M.; Chantret, I.; Swallow, D. M.; Trugnan, G.: Isolation of a cDNA probe for the human intestinal dipeptidylpeptidase IV and assignment of the gene locus DPP4 to chromosome 2. *Ann. Hum. Genet.* 54: 191–197, 1990.

[30198] 3750. Herbschleb-Voogt, E.; Grzeschik, K.-H.; Pearson, P. L.; Meera Khan, P.: Assignment of adenosine deaminase complexing protein (ADCP) gene(s) to human chromosome 2 in rodent–human somatic cell hybrids. *Hum. Genet.* 59: 317–323, 1981.

[30199] 3751. Kameoka, J.; Tanaka, T.; Nojima, Y.; Schlossman, S. F.; Morimoto, C.: Direct association of adenosine deaminase with a T cell activation antigen, CD26. *Science* 261: 466–469, 1993.

[30200] 3752. Koch, G. A.; Shows, T. B.: Genes on human chromosomes 2 and 6 are required for expression of the adenosine deaminase complexing protein (ADCP) in human–mouse somatic cell hybrids. (Abstract) *Cytogenet. Cell*

Genet. 25: 174, 1979.

- [30201] 3753. Marguet, D.; Baggio, L.; Kobayashi, T.; Bernard, A.-M.; Pierres, M.; Nielsen, P. F.; Ribel, U.; Watanabe, T.; Drucker, D. J.; Wagtmann, N.: Enhanced insulin secretion and improved glucose tolerance in mice lacking CD26. Proc. Nat. Acad. Sci. 97: 6874–6879, 2000.
- [30202] 3754. Mathew, S.; Morrison, M. E.; Murty, V. V. V. S.; Houghton, A. N.; Chaganti, R. S. K.: Assignment of the DPP4 gene encoding adenosine deaminase binding protein (CD26/dipeptidylpeptidase IV) to 2q23. Genomics 22:211–212, 1994.
- [30203] 3755. Misumi, Y.; Hayashi, Y.; Arakawa, F.; Ikehara, Y.: Molecular cloning and sequence analysis of human dipeptidyl peptidase IV, a serine proteinase on the cell surface. Biochim. Biophys. Acta 1131:333–336, 1992.
- [30204] 3756. Morrison, M. E.; Vijayasaradhi, S.; Engelstein, D.; Albino, A. P.; Houghton, A. N.: A marker for neoplastic progression of human melanocytes is a cell surface ectopeptidase. J. Exp. Med. 117: 1135–1143, 1993.
- [30205] 3757. Van Cong, N.; Weil, D.; Gross, M.-S.; Foubert, C.; Jami, J.; Frezal, J.: Contrôle génétique et épigénétique de l'expression de l'adenosine deaminase. Analyse des cellules humaines et hybrides homme-rongeur. Ann. Genet.

24: 141–147, 1981.

- [30206] 3758.Boison, D.; Scheurer, L.; Zumsteg, V.; Rulicke, T.; Litynski, P.;Fowler, B.; Brandner, S.; Mohler, H.: Neonatal hepatic steatosisby disruption of the adenosine kinase gene. *Proc. Nat. Acad. Sci.* 99:6985–6990, 2002.
- [30207] 3759.Chan, T.–S.; Cregan, R. P.; Reardon, M. P.: Adenosine kinase asa new selective marker in somatic cell genetics: isolation of adenosinekinase–deficient mouse cell lines and human–mouse hybrid cell linescontaining adenosine kinase. *Somat. Cell Genet.* 4: 1–12, 1978.
- [30208] 3760.Francke, U.; Thompson, L.: Regional mapping, by exclusion, ofadenosine kinase (ADK) on human chromosome 10 using the gene dosageapproach. (Abstract) *Cytogenet. Cell Genet.* 25: 156, 1979.
- [30209] 3761.Bundey, S.: Recent views on genetic factors in retinoblastoma.(Abstract) *J. Med. Genet.* 17: 386–387, 1980.
- [30210] 3762.Bundey, S.; Morten, J. E. N.: An unusual pedigree with retinoblastoma.Does it shed light on the delayed mutation and host resistance theories? *Hum.Genet.* 59: 434–436, 1981.
- [30211] 3763.Cance, W. G.; Brennan, M. F.; Dudas, M. E.; Huang, C.–M.; Cordon–Cardo,C.: Altered expression of the

retinoblastoma gene product in humansarcomas. New Eng. J. Med. 323: 1457–1462, 1990.

[30212] 3764.Canning, S.; Dryja, T. P.: Short, direct repeats at the breakpointsof deletions of the retinoblastoma gene. Proc. Nat. Acad. Sci. 86:5044–5048, 1989.

[30213] 3765.Carlson, E. A.; Desnick, R. J.: Mutational mosaicism and geneticcounseling in retinoblastoma. Am. J. Med. Genet. 4: 365–381, 1979.

[30214] 3766.Cavenee, W. K.: The genetic basis of neoplasia: the retinoblastomaparadigm. Trends Genet. 2: 299–300, 1986.

[30215] 3767.Cavenee, W. K.; Dryja, T. P.; Phillips, R. A.; Benedict, W. F.;Godbout, R.; Gallie, B. L.; Murphree, A. L.; Strong, L. C.; White,R. L.: Expression of recessive alleles by chromosomal mechanismsin retinoblastoma. Nature 305: 779–784, 1983.

[30216] 3768.Cavenee, W. K.; Hansen, M. F.; Nordenskjold, M.; Kock, E.; Maumenee,I.; Squire, J. A.; Phillips, R. A.; Gallie, B. L.: Genetic originof mutations predisposing to retinoblastoma. Science 228: 501–503,1985.

[30217] 3769.Cavenee, W. K.; Murphree, A. L.; Shull, M. M.; Benedict, W. F.;Sparkes, R. S.; Kock, E.; Nordenskjold, M.: Prediction of familialpredisposition to retinoblastoma. New

Eng. J. Med. 314: 1201–1207,1986.

[30218] 3770.Chano, T.; Ikegawa, S.; Kontani, K.; Okabe, H.; Baldini, N.; Saeki,Y.: Identification of RB1CC1, a novel human gene that can induceRB1 in various human cells. Oncogene 21: 1295–1298, 2002.

[30219] 3771.Chauveinc, L.; Mosseri, V.; Quintana, E.; Desjardins, L.; Schlienger,P.; Doz, F.; Dutrillaux, B.: Osteosarcoma following retinoblastoma:age at onset and latency period. Ophthalmic Genet. 22: 77–88, 2001.

[30220] 3772.Chen, P.–L.; Scully, P.; Shew, J.–Y.; Wang, J. Y. J.; Lee, W.–H.: Phosphorylation of the retinoblastoma gene product is modulatedduring the cell cycle and cellular differentiation. Cell 58: 1193–1198,1989.

[30221] 3773.Connolly, M. J.; Payne, R. H.; Johnson, G.; Gallie, B. L.; Alderdice,P. W.; Marshall, W. H.; Lawton, R. D.: Familial, EsD–linked, retinoblastomawith reduced penetrance and variable expressivity. Hum. Genet. 65:122–124, 1983.

[30222] 3774.Cowell, J. K.; Bia, B.: A novel missense mutation in patientsfrom a retinoblastoma pedigree showing only mild expression of thetumor phenotype. Oncogene 16: 3211–3213, 1998.

[30223] 3775.Cowell, J. K.; Rutland, P.; Hungerford, J.; Jay, M.: Deletionof chromosome region 13q14 is transmissible

and does not always predispose to retinoblastoma. Hum. Genet. 80: 43–45, 1988.

[30224] 3776. Cowell, J. K.; Rutland, P.; Jay, M.; Hungerford, J.: Deletion of the esterase D locus from a survey of 200 retinoblastoma patients. Hum. Genet. 72: 164–167, 1986.

[30225] 3777. Cowell, J. K.; Smith, T.; Bia, B.: Frequent constitutional C to T mutations in CGA–arginine codons in the RB1 gene produce premature stop codons in patients with bilateral (hereditary) retinoblastoma. Europ. J. Hum. Genet. 2: 281–290, 1994.

[30226] 3778. Dahiya, A.; Wong, S.; Gonzalo, S.; Gavin, M.; Dean, D. C.: Linking the Rb and Polycomb pathways. Molec. Cell 8: 557–568, 2001.

[30227] 3779. Davison, E. V.; Gibbons, B.; Aherne, G. E. S.; Roberts, D. F.: Chromosomes in retinoblastoma patients. Clin. Genet. 15: 505–508, 1979.

[30228] 3780. DeCaprio, J. A.; Ludlow, J. W.; Lynch, D.; Furukawa, Y.; Griffin, J.; Piwnicka-Worms, H.; Huang, C.-M.; Livingston, D. M.: The product of the retinoblastoma susceptibility gene has properties of a cell cycle regulatory element. Cell 58: 1085–1095, 1989.

[30229] 3781. de Grouchy, J.; Turleau, C.; Cabanis, M. O.; Richardet, J. M.: Retinoblastome et deletion intercalaire du

chromosome 13. Arch.Franc. Pediat. 37: 531–535, 1980.

- [30230] 3782.Dryja, T.; Cavenee, W.; Epstein, J.; Rapaport, J.; Goorin, A.;Koufos, A.: Chromosome 13 homozygosity in osteogenic sarcoma withoutretinoblastoma. (Abstract) Am. J. Hum. Genet. 36: 28S, 1984.
- [30231] 3783.Dryja, T. P.; Bruns, G. A. P.; Gallie, B.; Petersen, R.; Green,W.; Rapaport, J. M.; Albert, D. M.; Gerald, P. S.: Low incidenceof deletion of the esterase D locus in retinoblas–toma patients. Hum.Genet. 64: 151–155, 1983.
- [30232] 3784.Dryja, T. P.; Cavenee, W.; White, R.; Rapaport, J. M.; Petersen,R.; Albert, D. M.; Bruns, G. A. P.: Homozygosity of chromosome 13in retinoblastoma. New Eng. J. Med. 310: 550–553, 1984.
- [30233] 3785.Dryja, T. P.; Friend, S.; Weinberg, R. A.: Isolation of a cDNAfragment derived from human retina mRNA which detects a locus within13q14 often deleted in retinoblas–tomas. (Abstract) Am. J. Hum. Genet. 39:A29, 1986.
- [30234] 3786.Dryja, T. P.; Mukai, S.; Petersen, R.; Rapaport, J. M.; Walton,D.; Yandell, D. W.: Parental origin of mutations of the retinoblastomagene. Nature 339: 556–558, 1989.
- [30235] 3787.Dryja, T. P.; Mukai, S.; Rapaport, J. M.; Yandell, D. W.: Parentalorigin of mutations of the retinoblastoma gene. (Abstract) Am. J.Hum. Genet. 45 (suppl.): A19,

1989.

- [30236] 3788.Messer Peters, P. G.; Kamarck, M. E.; Hemler, M. E.; Strominger, J. L.; Ruddle, F. H.: Genetic and biochemical characterization of a human surface determinant on somatic cell hybrids: the 4F2 antigen. *Somat.Cell Genet.* 8: 825–834, 1982.
- [30237] 3789.Markert, C. L.; Silvers, W. K.: The effects of genotype and cell environment on melanoblast differentiation in the house mouse. *Genetics* 41:429–450, 1956.
- [30238] 3790.Van Cong, N.; Moreau–Gachelin, F.; Ray, D.; Gross, M. S.; de Tand, M. F.; Tavitian, A.; Frezal, J.: Assignment of SPI1 oncogene to chromosome 11 (somatic cell hybrid analysis), region p11.22 (in situ hybridization). (Abstract) *Cytogenet. Cell Genet.* 51: 1097 only, 1989.
- [30239] 3791.Bell, L. R.; Maine, E. M.; Schedl, P.; Cline, T. W.: Sex-lethal, a *Drosophila* sex determination switch gene, exhibits sex-specific RNA splicing and sequence similarity to RNA binding proteins. *Cell* 55:1037–1046, 1988.
- [30240] 3792.Fletcher, C. F.; Okano, H. J.; Gilbert, D. J.; Yang, Y.; Yang, C.; Copeland, N. G.; Jenkins, N. A.; Darnell, R. B.: Mouse chromosomal locations of nine genes encoding homologs of human paraneoplastic neurologic disorder antigens. *Genomics* 45: 313–319, 1997.

- [30241] 3793.King, P. H.; Levine, T. D.; Fremeau, R. T., Jr.; Keene, J. D.:Mammalian homologs of Drosophila elav localized to a neuronal subsetcan bind in vitro to the 3-prime UTR of mRNA encoding the Id transcriptionalrepressor. J. Neu-rosce. 14: 1943–1952, 1994.
- [30242] 3794.Levine, T. D.; Gao, F.; King, P. H.; Andrews, L. G.; Keene, J.D.: He1–N1: an autoimmune RNA-binding protein with specificity for3-prime uridylate-rich untranslated re-gions of growth factor mRNAs. Molec.Cell. Biol. 13: 3494–3504, 1993.
- [30243] 3795.Muresu, R.; Baldini, A.; Gress, T.; Posner, J. B.; Furneaux, H.M.; Siniscalco, M.: Mapping of the gene cod-ing for a paraneoplasticencephalomyelitis antigen (HuD) to human chromosome site 1p34. Cytogenet.Cell Genet. 65: 177–178, 1994.
- [30244] 3796.Robinow, S.; Campos, A. R.; Yao, K.–M.; White, K.: The elav geneproduct of Drosophila, required in neurons, has three RNP consensusmotifs. Science 242: 1570–1572, 1988.
- [30245] 3797.Szabo, A.; Dalmau, J.; Manley, G.; Rosenfeld, M.; Wong, E.; Henson,J.; Posner, J. B.; Furneaux, H. M.: HuD, a paraneoplastic encephalomyelitisantigen, contains RNA-binding domains and is homologous to elav andsex–

lethal. Cell 67: 325–333, 1991.

[30246] 3798. Clinton, M.; Frangou–Lazaridis, M.; Panneerselvam, C.; Horecker, B. L.: The sequence of human parathymosin deduced from a cloned humankidney cDNA. Biochem. Biophys. Res. Commun. 158: 855–862, 1989.

[30247] 3799. Szabo, P.; Clinton, M.; Macera, M.; Horecker, B. L.: Localization of the gene coding for parathymosin to chromosome 17 in humans. Cytogenet. Cell Genet. 50: 91–92, 1989.

[30248] 3800. Arnold, A.; Kim, H. G.; Gaz, R. D.; Eddy, R. L.; Fukushima, Y.; Byers, M. G.; Shows, T. B.; Kronenberg, H. M.: Molecular cloning and chromosomal mapping of DNA rearranged with the parathyroid hormone gene in a parathyroid adenoma. J. Clin. Invest. 83: 2034–2040, 1989.

[30249] 3801. Mehta, A. D.; Rock, R. S.; Rief, M.; Spudich, J. A.; Mooseker, M. S.; Cheney, R. E.: Myosin–V is a processive actin–based motor. Nature 400: 590–593, 1999.

[30250] 3802. Mercer, J. A.; Seperack, P. K.; Strobel, M. C.; Copeland, N. G.; Jenkins, N. A.: Novel myosin heavy chain encoded by murine dilute coat colour locus. Nature 349: 709–713, 1991.

[30251] 3803. Moore, K. J.; Testa, J. R.; Francke, U.; Milatovich, A.;

Copeland, N. G.; Jenkins, N. A.: Cloning and regional assignment of the human myosin heavy chain 12 (MYH12) gene to chromosome band 15q21. *Cytogenet. Cell Genet.* 69: 53–58, 1995.

[30252] 3804. Pastural, E.; Barrat, F. J.; Dufourcq-Lagelouse, R.; Certain, S.; Sanal, O.; Jabado, N.; Seger, R.; Griscelli, C.; Fischer, A.; de Saint Basile, G.: Griscelli disease maps to chromosome 15q21 and is associated with mutations in the myosin-Va gene. *Nature Genet.* 16:289–292, 1997.
Note: Erratum: *Nature Genet.* 23: 373 only, 1999.

[30253] 3805. Rief, M.; Rock, R. S.; Mehta, A. D.; Mooseker, M. S.; Cheney, R. E.; Spudich, J. A.: Myosin-V stepping kinetics: a molecular model for processivity. *Proc. Nat. Acad. Sci.* 97: 9482–9486, 2000.

[30254] 3806. Russell, E. S.: A quantitative histological study of the pigment found in the coat-color mutants of the house mouse. IV. The nature of the effects of genic substitution in five major allelic series. *Genetics* 34:146–166, 1949.

[30255] 3807. Musahl, C.; Schulte, D.; Burkhart, R.; Knippers, R.: A human homologue of the yeast replication protein Cdc21: interactions with other MCM proteins. *Europ. J. Biochem.* 230: 1096–1101, 1995.

[30256] 3808. Kim, N.-S.; Kato, T.; Abe, N.; Kato, S.: Nucleotide se-

quence of human cDNA encoding eukaryotic initiation factor 4A1. *Nucleic Acids Res.* 21: 2012 only, 1993.

[30257] 3809. Kukimoto, I.; Watanabe, S.; Taniguchi, K.; Ogata, T.; Yoshiike, K.; Kanda, T.: Characterization of the cloned promoter of the human initiation factor 4A1 gene. *Biochem. Biophys. Res. Commun.* 233:844–847, 1997.

[30258] 3810. Nielsen, P. J.; McMaster, G. K.; Trachsel, H.: Cloning of eukaryotic protein synthesis initiation factor genes: isolation and characterization of cDNA clones encoding factor eIF-4A. *Nucleic Acids Res.* 13: 6867–6880, 1985.

[30259] 3811. Anderson, D. M.; Maraskovsky, E.; Billingsley, W. L.; Dougall, W. C.; Tometsko, M. E.; Roux, E. R.; Teepe, M. C.; DuBose, R. F.; Cosman, D.; Galibert, L.: A homologue of the TNF receptor and its ligand enhance T-cell growth and dendritic-cell function. *Nature* 390:175–179, 1997.

[30260] 3812. Croucher, P. I.; Shipman, C. M.; Lippitt, J.; Perry, M.; Asosingh, K.; Hijzen, A.; Brabbs, A. C.; van Beek, E. J. R.; Holen, I.; Skerry, T. M.; Dunstan, C. R.; Russell, G. R.; Van Camp, B.; Vanderkerken, K.: Osteoprotegerin inhibits the development of osteolytic bone disease in multiple myeloma. *Blood* 98: 3534–3540, 2001.

[30261] 3813. Fata, J. E.; Kong, Y.-Y.; Li, J.; Sasaki, T.; Irie-Sasaki, J.; Moorehead, R. A.; Elliott, R.; Scully, S.; Voura, E. B.;

Lacey, D.L.; Boyle, W. J.; Khokha, R.; Penninger, J. M.: The osteoclast differentiation factor osteoprotegerin–ligand is essential for mammary gland development. *Cell* 103:41–50, 2000.

[30262] 3814. Kim, N.; Odgren, P. R.; Kim, D.–K.; Marks, S. C., Jr.; Choi, Y.: Diverse roles of the tumor necrosis factor family member TRANCE in skeletal physiology revealed by TRANCE deficiency and partial rescue by a lymphocyte–expressed TRANCE transgene. *Proc. Nat. Acad. Sci.* 97:10905–10910, 2000.

[30263] 3815. Kong, Y.–Y.; Feige, U.; Sarosi, I.; Bolon, B.; Tafuri, A.; Morony, S.; Capparelli, C.; Li, J.; Elliott, R.; McCabe, S.; Wong, T.; Campagnuolo, G.; and 9 others: Activated T cells regulate bone loss and joint destruction in adjuvant arthritis through osteoprotegerin ligand. *Nature* 402:304–309, 1999.

[30264] 3816. Lacey, D. L.; Timms, E.; Tan, H.–L.; Kelley, M. J.; Dunstan, C.R.; Burgess, T.; Elliott, R.; Colombero, A.; Elliott, G.; Scully, S.; Hsu, H.; Sullivan, J.; Hawkins, N.; Davy, E.; Capparelli, C.; Eli, A.; Qian, Y.–X.; Kaufman, S.; Sarosi, I.; Shalhoub, V.; Senaldi, G.; Guo, J.; Delaney, J.; Boyle, W. J.: Osteoprotegerin ligand is a cytokine that regulates osteoclast differentiation and activation. *Cell* 93:165–176,

1998.

- [30265] 3817. Pearse, R. N.; Sordillo, E. M.; Yaccoby, S.; Wong, B. R.; Liao, D. F.; Colman, N.; Michaeli, J.; Epstein, J.; Choi, Y.: Multiple myeloma disrupts the TRANCE/osteoprotegerin cytokine axis to trigger bone destruction and promote tumor progression. *Proc. Nat. Acad. Sci.* 98: 11581–11586, 2001.
- [30266] 3818. Wong, B. R.; Rho, J.; Arron, J.; Robinson, E.; Orlinick, J.; Chao, M.; Kalachikov, S.; Cayani, E.; Bartlett, F. S., III; Frankel, W. N.; Lee, S. Y.; Choi, Y.: TRANCE is a novel ligand of the tumor necrosis factor receptor family that activates c-Jun N-terminal kinase in T cells. *J. Biol. Chem.* 272: 25190–25194, 1997.
- [30267] 3819. Udell, C. M.; Lee, S. K.; Davey, S.: HRAD1 and MRAD1 encode mammalian homologues of the fission yeast rad1+ cell cycle checkpoint control gene. *Nucleic Acids Res.* 26: 3971–3976, 1998.
- [30268] 3820. Carvajal, J. J.; Pook, M. A.; dos Santos, M.; Doudney, K.; Hillermann, R.; Minogue, S.; Williamson, R.; Hsuan, J. J.; Chamberlain, S.: The Friedreich's ataxia gene encodes a novel phosphatidylinositol-4-phosphate 5-kinase. *Nature Genet.* 14: 157–162, 1996.
- [30269] 3821. Carvajal, J. J.; Pook, M. A.; Doudney, K.; Hillermann,

R.; Wilkes,D.; Al-Mahdawi, S.; Williamson, R.; Chamberlain, S.: Friedreich'sataxia: a defect in signal transduction? Hum. Molec. Genet. 4: 1411–1419,1995.

[30270] 3822.Pook, M. A.; Carvajal, J. J; Doudney, K.; Hillermann, R.; Chamberlain,S.: Exon–intron structure of a 2.7–kb transcript of the STM7 genewith phosphatidylinositol–4–phosphate 5–kinase activity. Genomics 42:170–172, 1997.

[30271] 3823.Carfi, A.; Willis, S. H.; Whitbeck, J. C.; Krummenacher, C.; Cohen,G. H.; Eisenberg, R. J.; Wiley, D. C.: Herpes simplex virus glycoproteinD bound to the human receptor HveA. Molec. Cell 8: 169–179, 2001.

[30272] 3824.Hsu, H.; Solovyev, I.; Colombero, A.; Elliott, R.; Kelley, M.;Boyle, W. J.: ATAR, a novel tumor necrosis factor receptor familymember, signals through TRAF2 and TRAF5. J. Biol. Chem. 272: 13471–13474,1997.

[30273] 3825.Marsters, S. A.; Ayres, T. M.; Skubatch, M.; Gray, C. L.; Rothe,M.; Ashkenazi, A.: Herpesvirus entry mediator, a member of the tumornecrosis factor receptor (TNFR) family, interacts with members ofthe TNFR–associated factor family and activates the transcriptionfactors NF–kappa–B and AP–1. J. Biol. Chem. 272: 14029–14032, 1997.

[30274] 3826.Montgomery, R. I.; Warner, M. S.; Lum, B. J.; Spear, P.

G.: Herpes simplex virus-1 entry into cells mediated by a novel member of the TNF/NGF receptor family. *Cell* 87: 427-436, 1996.

[30275] 3827. Kwon, B. S.; Tan, K. B.; Ni, J.; Oh, K.-O.; Lee, Z. H.; Kim, K. K.; Kim, Y.-J.; Wang, S.; Gentz, R.; Yu, G.-L.; Harrop, J.; Lyn, S. D.; Silverman, C.; Porter, T. G.; Truneh, A.; Young, P. R.: A newly identified member of the tumor necrosis factor receptor superfamily with a wide tissue distribution and involvement in lymphocyte activation. *J. Biol. Chem.* 272: 14272-14276, 1997.

[30276] 3828. Morel, Y.; Schiano de Colella, J.-M.; Harrop, J.; Deen, K. C.; Holmes, S. D.; Wattam, T. A.; Khandekar, S. S.; Truneh, A.; Sweet, R. W.; Gastaut, J.-A.; Olive, D.; Costello, R. T.: Reciprocal expression of the TNF family receptor herpes virus entry mediator and its ligand LIGHT on activated T cells: LIGHT down-regulates its own receptor. *J. Immun.* 165: 4397-4404, 2000.

[30277] 3829. Guan, K.-L.; Butch, E.: Isolation and characterization of a novel dual specific phosphatase, HVH2, which selectively dephosphorylates the mitogen-activated protein kinase. *J. Biol. Chem.* 270: 7197-7203, 1995.

[30278] 3830. Smith, A.; Price, C.; Cullen, M.; Muda, M.; King, A.; Ozanne, B.; Arkinstall, S.; Ashworth, A.: Chromosomal lo-

calization of three humandual specificity phosphatase genes (DUSP4, DUSP6, and DUSP7). *Genomics* 42:524–527, 1997.

- [30279] 3831.Furukawa, T.; Yatsuoka, T.; Youssef, E. M.; Abe, T.; Yokoyama,T.; Fukushige, S.; Soeda, E.; Hoshi, M.; Hayashi, Y.; Sunamura, M.;Kobari, M.; Horii, A.: Genomic analysis of DUSP6, a dual specificityMAP kinase phosphatase, in pancreatic cancer. *Cytogenet. Cell Genet.* 82:156–159, 1998.
- [30280] 3832.Groom, L. A.; Sneddon, A. A.; Alessi, D. R.; Dowd, S.; Keyse, S.M.: Differential regulation of the MAP, SAP and RK/p38 kinases byPyst1, a novel cytosolic dual–specificity phosphatase. *EMBO J.* 15:3621–3632, 1996.
- [30281] 3833.Muda, M.; Boschert, U.; Dickinson, R.; Martinou, J.–C.; Martinou,l.; Camps, M.; Schlegel, W.; Arkinstall, S.: MKP–3, a novel cytosolicprotein–tyrosine phosphatase that exemplifies a new class of mitogen–activatedprotein kinase phosphatase. *J. Biol. Chem.* 271: 4319–4326, 1996.
- [30282] 3834.Smith, A.; Price, C.; Cullen, M.; Muda, M.; King, A.; Ozanne, B.;Arkinstall, S.; Ahsworth, A.: Chromosomal localization of three humandual specificity phosphatase genes (DUSP4, DUSP6, and DUSP7). *Genomics* 42:524–527, 1997.

- [30283] 3835.Nishihira, J.; Fujinaga, M.; Kuriyama, T.; Suzuki, M.; Sugimoto,H.; Nakagawa, A.; Tanaka, I.; Sakai, M.: Molecular cloning of humanD-dopachrome tautomerase cDNA: N-terminal proline is essential forenzyme activation. Biochem. Biophys. Res. Commun. 243: 538–544,1998.
- [30284] 3836.Blouin, J.–L.; Sail, G. D.; Guipponi, M.; Rossier, C.; Pappasavas,M.–P.; Antonarakis, S. E.: Isolation of the human BACH1 transcriptionregulator gene, which maps to chromosome 21q22.1. Hum. Genet. 102:282–288, 1998.
- [30285] 3837.Conley, Y. P.; Erturk, D.; Keverline, A.; Mah, T. S.; Keravala,A.; Barnes, L. R.; Bruchis, A.; Hess, J. F.; FitzGerald, P. G.; Weeks,D. E.; Ferrell, R. E.; Gorin, M. B.: A juvenile-onset, progressivecataract locus on chromosome 3q21–q22 is associated with a missensemutation in the beaded filament structural protein–2. Am. J. Hum.Genet. 66: 1426–1431, 2000.
- [30286] 3838.Strehl, S.; Glatt, K.; Liu, Q. M.; Glatt, H.; Lalande, M.: Characterizationof two novel protocadherins (PCDH8 and PCDH9) localized on human chromosome13 and mouse chromosome 14. Genomics 53: 81–89, 1998.
- [30287] 3839.Ohbayashi, N.; Hoshikawa, M.; Kimura, S.; Yamasaki, M.; Fukui,S.; Itoh, N.: Structure and expression of the mRNA encoding a novelfibroblast growth factor, FGF–18.

J. Biol. Chem. 273: 18161–18164,1998.

- [30288] 3840.Loeffen, J.; Smeets, R.; Smeitink, J.; Triepels, R.; Sengers, R.;Trijbels, F.; van den Heuvel, L.: The human NADH:ubiquinone oxidoreductaseNDUFS5 (15 kDa) subunit: cDNA cloning, chromosomal localization, tissue distribution and the absence of mutations in isolated complex I-deficientpatients. J. Inherit. Metab. Dis. 22: 19–28, 1999.
- [30289] 3841.Jackson, A.; Panayiotidis, P.; Foroni, L.: The human homologueof the Drosophila tailless gene (TLX): characterization and mappingto a region of common deletion in human lymphoid leukemia on chromosome6q21. Genomics 50: 34–43, 1998.
- [30290] 3842.Monaghan, A. P.; Bock, D.; Gass, P.; Schwager, A.; Wolfer, D. P.;Lipp, H.–P.; Schutz, G.: Defective limbic system in mice lackingthe tailless gene. Nature 390: 515–517, 1997.
- [30291] 3843.Yu, R. T.; McKeown, M.; Evans, R. M.; Umesono, K.: Relationshipbetween Drosophila gap gene tailless and a vertebrate nuclear receptorTlx. Nature 370: 375–379, 1994.
- [30292] 3844.Plougastel, B.; Trowsdale, J.: Cloning of NKG2–F, a new memberof the NKG2 family of human natural killer

cell receptor genes. *Europ.J. Immun.* 27: 2835–2839, 1997.

[30293] 3845.Sutherland, C. L.; Chalupny, N. J.; Schooley, K.; Van-denBos, T.;Kubin, M.; Cosman, D.: UL16-binding proteins, novel MHC class I-relatedproteins, bind to NKG2D and activate multiple signaling pathways inprimary NK cells. *J. Immun.* 168: 671–679, 2002.

[30294] 3846.Wu, J.; Song, Y.; Bakker, A. B. H.; Bauer, S.; Spies, T.; Lanier,L. L.; Phillips, J. H.: An activating immunoreceptor complex formedby NKG2D and DAP10. *Science* 285: 730–732, 1999.

[30295] 3847.Chang, C.; Rodriguez, A.; Carretero, M.; Lopez-Botet, M.; Phillips,J. H.; Lanier, L. L.: Molecular characterization of human CD94: atype II membrane glycoprotein related to the C-type lectin superfamily. *Europ.J. Immun.* 25: 2433–2437, 1995.

[30296] 3848.Lazetic, S.; Chang, C.; Houchins, J. P.; Lanier, L. L.; Phillips,J. H.: Human natural killer cell receptors involved in MHC classI recognition are disulfide-linked heterodimers of CD94 and NKG2 subunits. *J.Immun.* 157: 4741–4745, 1996.

[30297] 3849.Rodriguez, A.; Carretero, M.; Glienke, J.; Bellon, T.; Ramirez,A.; Lehrach, H.; Francis, F.; Lopez-Botet, M.:

Structure of the humanCD94 C-type lectin gene. Immunogenetics 47: 305–309, 1998.

[30298] 3850.Volkmer, E.; Karnitz, L. M.: Human homologs of Schizosaccharomycespombe Rad1, Hus1, and Rad9 form a DNA damage–responsive protein complex. J.Biol. Chem. 274: 567–570, 1999.

[30299] 3851.Brandner, J. M.; Reidenbach, S.; Franke, W. W.: Evidence that 'pinin,' reportedly a differentiation–specific desmosomal protein,is actually a widespread nuclear protein. Differentiation 62: 119–127,1997.

[30300] 3852.Brandner, J. M.; Reidenbach, S.; Kuhn, C.; Franke, W. W.: Identificationand characterization of a novel kind of nuclear protein occurringfree in nucleoplasm and in ribonucleoprotein structures of the 'speckle'type. Europ. J. Cell Biol. 75: 295–308, 1998.

[30301] 3853.Degen, W. G. J.; Agterbos, M. A.; Muyrers, J. P. P.; Bloemers,H. P. J.; Swart, G. W. M.: memA/DRS, a putative mediator of multiproteincomplexes, is overexpressed in the metastasizing human melanoma celllines BLM and MV3. Biochim. Biophys. Acta 1444: 384–394, 1999.

[30302] 3854.Ouyang, P.; Sugrue, S. P.: Characterization of pinin, a novelprotein associated with the desmosome–intermediate filament complex. J.Cell Biol. 135: 1027–1042,

1996.

- [30303] 3855. Puente, X. S.; Lopez-Otin, C.: Cloning and expression analysis of a novel human serine hydrolase with sequence similarity to prokaryotic enzymes involved in the degradation of aromatic compounds. *J. Biol. Chem.* 270: 12926–12932, 1995.
- [30304] 3856. Puente, X. S.; Pendas, A. M.; Lopez-Otin, C.: Structural characterization and chromosomal localization of the gene encoding human biphenyl hydrolase-related protein (BPHL). *Genomics* 51: 459–462, 1998.
- [30305] 3857. Janssen, J. W. G.; Schleithoff, L.; Bartram, C. R.; Schulz, A. S.: An oncogenic fusion product of the phosphatidylinositol 3-kinase p85-beta subunit and HUMORF8, a putative deubiquitinating enzyme. *Oncogene* 16: 1767–1772, 1998.
- [30306] 3858. Chadwick, B. P.; Williamson, J.; Sheer, D.; Frischauf, A.-M.: cDNA cloning and chromosomal mapping of a mouse gene with homology to NTPases. *Mammalian Genome* 9: 162–164, 1998.
- [30307] 3859. Yeung, G.; Mulero, J. J.; McGowan, D. W.; Bajwa, S. S.; Ford, J. E.: CD39L2, a gene encoding a human nucleoside diphosphatase, predominantly expressed in the heart. *Biochemistry* 39: 12916–12923, 2000.

- [30308] 3860.Ji, H.; Liu, Y. E.; Jia, T.; Wang, M.; Liu, J.; Xiao, G.; Joseph,B. K.; Rosen, C.; Shi, Y. E.: Identification of a breast cancer-specificgene, BCSG1, by direct differential cDNA sequencing. *Cancer Res.* 57:759–764, 1997.
- [30309] 3861.Lavedan, C.; Leroy, E.; Dehejia, A.; Buchholtz, S.; Dutra, A.;Nussbaum, R. L.; Polymeropoulos, M. H.: Identification, localizationand characterization of the human gamma-synuclein gene. *Hum. Genet.* 103:106–112, 1998.
- [30310] 3862.Ninkina, N. N.; Alimova-Kost, M. V.; Paterson, J. W. E.; Delaney,L.; Cohen, B. B.; Imreh, S.; Gnuchev, N. V.; Davies, A. M.; Buchman,V. L.: Organization, expression and polymorphism of the human persyngene. *Hum. Molec. Genet.* 7: 1417–1424, 1998.
- [30311] 3863.Spillantini, M. G.; Crowther, R. A.; Jakes, R.; Hasegawa, M.; Goedert,M.: alpha-synuclein in filamentous inclusions of Lewy bodies fromParkinson's disease and dementia with Lewy bodies. *Proc. Nat. Acad.Sci.* 95: 6469–6473, 1998.
- [30312] 3864.Seroussi, E.; Pan, H.–Q.; Kedra, D.; Roe, B. A.; Dumanski, J. P.: Characterization of the human NIPSNAP1 gene from 22q12: a memberof a novel gene family. *Gene* 212: 13–20, 1998.
- [30313] 3865.Wang, X.–Y.; Smith, D. I.; Lui, W.; James, C. D.: GBAS,

a novel gene encoding a protein with tyrosine phosphorylation sites and a transmembrane domain, is co-amplified with EGFR. *Genomics* 49: 448–451, 1998.

[30314] 3866. Kurima, K.; Warman, M. L.; Krishnan, S.; Domowicz, M.; Krueger, R. C., Jr.; Deyrup, A.; Schwartz, N. B.: A member of a family of sulfate-activating enzymes causes murine brachymorphism. *Proc. Nat. Acad. Sci.* 95: 8681–8685, 1998.

[30315] 3867. Li, H.; Deyrup, A.; Mensch, J. R., Jr.; Domowicz, M.; Konstantinidis, A. K.; Schwartz, N. B.: The isolation and characterization of cDNA encoding the mouse bifunctional ATP sulfurylase-adenosine 5-prime-phosphosulfate kinase. *J. Biol. Chem.* 270: 29453–29459, 1995.

[30316] 3868. Hess, J. F.; Casselman, J. T.; FitzGerald, P. G.: Chromosomal locations of the genes for the beaded filament proteins CP 115 and CP 47. *Curr. Eye Res.* 14: 11–18, 1995.

[30317] 3869. Hess, J. F.; Casselman, J. T.; FitzGerald, P. G.: Gene structure and cDNA sequence identify the beaded filament protein CP49 as a highly divergent type I intermediate filament protein. *J. Biol. Chem.* 271: 6729–6735, 1996.

[30318] 3870. Jakobs, P. M.; Hess, J. F.; FitzGerald, P. G.; Kramer,

P.; Weleber, R. G.; Litt, M.: Autosomal-dominant congenital cataract associated with a deletion mutation in the human beaded filament protein gene *BFSP2*. *Am. J. Hum. Genet.* 66: 1432–1436, 2000.

[30319] 3871. Holzinger, A.; Kammerer, S.; Roscher, A. A.: Primary structure of human PMP69, a putative peroxisomal ABC-transporter. *Biochem. Biophys. Res. Commun.* 237: 152–157, 1997.

[30320] 3872. Holzinger, A.; Roscher, A. A.; Landgraf, P.; Lichtner, P.; Kammerer, S.: Genomic organization and chromosomal localization of the human peroxisomal membrane protein-1-like protein (*PXMP1-L*) gene encoding a peroxisomal ABC transporter. *FEBS Lett.* 426: 238–242, 1998.

[30321] 3873. Shani, N.; Jimenez-Sanchez, G.; Steel, G.; Dean, M.; Valle, D.: Identification of a fourth half ABC transporter in the human peroxisomal membrane. *Hum. Molec. Genet.* 6: 1925–1931, 1997.

[30322] 3874. Fink, M.; Duprat, F.; Lesage, F.; Reyes, R.; Romey, G.; Heurteaux, C.; Lazdunski, M.: Cloning, functional expression and brain localization of a novel unconventional outward rectifier K⁺ channel. *EMBO J.* 15: 6854–6862, 1996.

[30323] 3875. Lesage, F.; Lazdunski, M.: Mapping of human potassium channel genes *TREK-1* (*KCNK2*) and *TASK* (*KCNK3*) to

chromosomes 1q41 and 2p23. *Genomics* 51:478–479, 1998.

- [30324] 3876. Duprat, F.; Lesage, F.; Fink, M.; Reyes, R.; Heurteaux, C.; Lazdunski, M.: TASK, a human background K⁺ channel to sense external pH variations near physiological pH. *EMBO J.* 16: 5464–5471, 1997.
- [30325] 3877. Manjunath, N. A.; Bray-Ward, P.; Goldstein, S. A. N.; Gallagher, P. G.: Assignment of the 2P domain, acid-sensitive potassium channel OAT1 gene KCNK3 to human chromosome bands 2p24.1–p23.3 and murine 5B by in situ hybridization. *Cytogenet. Cell Genet.* 86: 242–243, 1999.
- [30326] 3878. Hacker, B. M.; Tomlinson, J. E.; Wayman, G. A.; Sultana, R.; Chan, G.; Villacres, E.; Disteché, C.; Storm, D. R.: Cloning, chromosomal mapping, and regulatory properties of the human type 9 adenylyl cyclase (ADCY9). *Genomics* 50: 97–104, 1998.
- [30327] 3879. Paterson, J. M.; Smith, S. M.; Harmar, A. J.; Antoni, F. A.: Control of a novel adenylyl cyclase by calcineurin. *Biochem. Biophys. Res. Commun.* 214: 1000–1008, 1995.
- [30328] 3880. Premont, R. T.; Matsuoka, I.; Mattei, M. G.; Pouille, Y.; Defer, N.; Hanoune, J.: Identification and characterization of a widely expressed form of adenylyl cyclase. *J. Biol.*

Chem. 271: 13900–13907,1996.

- [30329] 3881.Toyota, T.; Hattori, E.; Meerabux, J.; Yamada, K.; Saito, K.; Shibuya,H.; Nankai, M.; Yoshikawa, T.: Molecular analysis, mutation screening,and association study of adenylate cyclase type 9 gene (ADCY9) in mood disorders. Am. J. Med. Genet. (Neuropsychiat. Genet.) 114:84–92, 2002.
- [30330] 3882.Chi, N.-W.; Lodish, H. F.: Tankyrase is a Golgi-associated mitogen-activated protein kinase substrate that interacts with IRAP in GLUT4 vesicles. J.Biol. Chem. 275: 38437–38444, 2000.
- [30331] 3883.Seimiya, H.; Smith, S.: The telomeric poly(ADP-ribose) polymerase,tankyrase 1, contains multiple binding sites for telomeric repeat binding factor 1 (TRF1) and a novel acceptor, 182-kDa tankyrase-binding protein (TAB182). J. Biol. Chem. 277: 14116–14126, 2002.
- [30332] 3884.Smith, S.; Gariat, I.; Schmitt, A.; de Lange, T.: Tankyrase, a poly(ADP-ribose) polymerase at human telomeres. Science 282: 1484–1487,1998.
- [30333] 3885.Zhu, L.; Smith, S.; de Lange, T.; Seldin, M. F.: Chromosomal mapping of the tankyrase gene in human and mouse. Genomics 57: 320–321,1999.

- [30334] 3886.Occhiodoro, T.; Bernheim, L.; Liu, J.-H.; Bijlenga, P.; Sinnreich,M.; Bader, C. R.; Fischer-Lougheed, J.: Cloning of a human ether-a-go-gopotassium channel expressed in myoblasts at the onset of fusion. FEBSLett. 434: 177-182, 1998.
- [30335] 3887.Hidai, H.; Bardales, R.; Goodwin, R.; Quertermous, T.; Quertermous,E. E.: Cloning of capsulin, a basic helix-loop-helix factor expressedin progenitor cells of the pericardium and the coronary arteries. Mech.Dev. 73: 33-43, 1998.
- [30336] 3888.Lu, J.; Chang, P.; Richardson, J. A.; Gan, L.; Weiler, H.; Olson,E. N.: The basic helix-loop-helix transcription factor capsulin controlsspleen organogenesis. Proc. Nat. Acad. Sci. 97: 9525-9530, 2000.
- [30337] 3889.Lu, J.; Richardson, J. A.; Olson, E. N.: Capsulin: a novel bHLHtranscription factor expressed in epicardial progenitors and mesenchymeof visceral organs. Mech. Dev. 73: 23-32, 1998.
- [30338] 3890.Quaggin, S. E.; Vanden Heuvel, G. B.; Igarashi, P.: Pod-1, a mesoderm-specificbasic-helix-loop-helix protein expressed in mesenchymal and glomerularepithelial cells in the developing kidney. Mech. Dev. 71: 37-48,1998.

- [30339] 3891.Robb, L.; Mifsud, L.; Hartley, L.; Biben, C.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Harvey, R. P.: Epicardin: a novel basic helix–loop–helix transcription factor gene expressed in epicardium, branchial arch myoblasts, and mesenchyme of developing lung, gut, kidney, and gonads. *Dev. Dyn.* 213: 105–113, 1998.
- [30340] 3892.Stacey, P.; Rulten, S.; Dapling, A.; Phillips, S. C.: Molecular cloning and expression of human cGMP–binding cGMP–specific phosphodiesterase (PDE5). *Biochem. Biophys. Res. Commun.* 247: 249–254, 1998.
- [30341] 3893.Yanaka, N.; Kotera, J.; Ohtsuka, A.; Akatsuka, H.; Imai, Y.; Michibata, H.; Fujishige, K.; Kawai, E.; Takebayashi, S.–I.; Okumura, K.; Omori, K.: Expression, structure and chromosomal localization of the human cGMP–binding cGMP–specific phosphodiesterase PDE5A gene. *Eur. J. Biochem.* 255: 391–399, 1998.
- [30342] 3894.Hess, G. F.; Drong, R. F.; Weiland, K. L.; Slightom, J. L.; Sclafani, R. A.; Hollingsworth, R. E.: A human homolog of the yeast CDC7 gene is overexpressed in some tumors and transformed cell lines. *Gene* 211: 133–140, 1998.
- [30343] 3895.Jiang, W.; Hunter, T.: Identification and characterization of a human protein kinase related to budding yeast Cdc7p. *Proc. Nat. Acad. Sci.* 94: 14320–14325, 1997.

- [30344] 3896.Engelender, S.; Kaminsky, Z.; Guo, X.; Sharp, A. H.; Amaravi, R.K.; Kleiderlein, J. J.; Margolis, R. L.; Troncoso, J. C.; Lanahan,A. A.; Worley, P. F.; Dawson, V. L.; Dawson, T. M.; Ross, C. A.:Synphilin-1 associates with alpha-synuclein and promotes the formationof cytosolic inclusions. *Nature Genet.* 22: 110-114, 1999.
- [30345] 3897.Jakes, R.; Spillantini, M. G.; Goedert, M.: Identification oftwo distinct synucleins from human brain. *FEBS Lett.* 345: 27-32,1994.
- [30346] 3898.Tory, K.; Latif, F.; Modi, W.; Schmidt, L.; Wei, M. H.; Li, H.;Cobler, P.; Orcutt, M. L.; Delisio, J.; Geil, L.; Zbar, B.; Lerman,M. I.: A genetic linkage map of 96 loci on the short arm of humanchromosome 3. *Genomics* 13: 275-286, 1992.
- [30347] 3899.Amiel, J.; Salomon, R.; Attie, T.; Pelet, A.; Trang, H.; Mokhtari,M.; Gaultier, C.; Munnich, A.; Lyonnet, S.: Mutations of the RET-GDNFsignaling pathway in Ondine's curse. (Letter) *Am. J. Hum. Genet.* 62:715-717, 1998.
- [30348] 3900.Angrist, M.; Bolk, S.; Thiel, B.; Puffenberger, E. G.; Hofstra,R. M.; Buys, C. H. C. M.; Cass, D. T.; Chakravarti, A.: Mutationanalysis of the RET receptor tyrosine kinase in Hirschsprung disease. *Hum.Molec. Genet.* 4: 821-830, 1995.

- [30349] 3901. Antinolo, G.; Marcos, I.; Fernandez, R. M.; Romero, M.; Borrego, S.: A novel germline point mutation, c.2304G(T, in codon 768 of the RET proto-oncogene in a patient with medullary thyroid carcinoma. (Letter) Am. J. Med. Genet. 110: 85–87, 2002.
- [30350] 3902. Attie, T.; Pelet, A.; Edery, P.; Eng, C.; Mulligan, L. M.; Amiel, J.; Boutrand, L.; Beldjord, C.; Nihoul-Fekete, C.; Munnich, A.; Ponder, B. A. J.; Lyonnet, S.: Diversity of RET proto-oncogene mutations in familial and sporadic Hirschsprung disease. Hum. Molec. Genet. 4:1381–1386, 1995.
- [30351] 3903. Attie-Bitach, T.; Abitbol, M.; Gerard, M.; Delezoide, A.-L.; Auge, J.; Pelet, A.; Amiel, J.; Pachnis, V.; Munnich, A.; Lyonnet, S.; Vekemans, M.: Expression of the RET proto-oncogene in human embryos. Am. J. Med. Genet. 80: 481–486, 1998.
- [30352] 3904. Auricchio, A.; Griseri, P.; Carpentieri, M. L.; Betsos, N.; Staiano, A.; Tozzi, A.; Priolo, M.; Thompson, H.; Boccia-
rdi, R.; Romeo, G.; Ballabio, A.; Ceccherini, I.: Double heterozygosity for a RET substitution interfering with splicing and an EDNRB missense mutation in Hirschsprung disease. (Letter) Am. J. Hum. Genet. 64: 1216–1221, 1999.
- [30353] 3905. Batourina, E.; Choi, C.; Paragas, N.; Bello, N.; Hensle,

T.; Costantini, F. D.; Schuchardt, A.; Bacallao, R. L.; Mendelsohn, C. L.: Distal ureter morphogenesis depends on epithelial cell remodeling mediated by vitamin A and Ret. *Nature Genet.* 32: 109–115, 2002. Note: Erratum: *Nature Genet.* 32: 331 only, 2002.

[30354] 3906. Batourina, E.; et al; et al: Vitamin A controls epithelial/mesenchymal interactions through Ret expression. *Nature Genet.* 27: 74–78, 2001.

[30355] 3907. Berndt, I.; Reuter, M.; Saller, B.; Frank-Raue, K.; Groth, P.; Grubendorf, M.; Raue, F.; Ritter, M. M.; Hoppner, W.: A new hot spot for mutations in the ret protooncogene causing familial medullary thyroid carcinoma and multiple endocrine neoplasia type 2A. *J. Clin. Endocr. Metab.* 83: 770–774, 1998.

[30356] 3908. Boccia, L. M.; Green, J. S.; Joyce, C.; Eng, C.; Taylor, S. A. M.; Mulligan, L. M.: Mutation of RET codon 768 is associated with the FMTC phenotype. *Clin. Genet.* 51: 81–85, 1997.

[30357] 3909. Bolino, A.; Schuffenecker, I.; Luo, Y.; Seri, M.; Silengo, M.; Tocco, T.; Chabrier, G.; Houdent, C.; Murat, A.; Schlumberger, M.; Tourniaire, J.; Lenoir, G. M.; Romeo, G.: RET mutations in exons 13 and 14 of FMTC patients. *Oncogene* 10: 2415–2419, 1995.

- [30358] 3910.Bolk, S.; Angrist, M.; Schwartz, S.; Silvestri, J. M.; Weese-Mayer,D. E.; Chakravarti, A.: Congenital central hypoventilation syndrome:mutation analysis of the receptor tyrosine kinase RET. Am. J. Med.Genet. 63: 603-609, 1996.
- [30359] 3911.Bolk Gabriel, S.; Salomon, R.; Pelet, A.; Angrist, M.; Amiel,J.; Fornage, M.; Attie-Bitach, T.; Olson, J. M.; Hofstra, R.; Buys,C.; Steffann, J.; Munnich, A.; Lyonnet, S.; Chakravarti, A.: Segregationat three loci explains familial and population risk in Hirschsprungdisease. Nature Genet. 31: 89-93, 2002.
- [30360] 3912.Ceccherini, I.; Hofstra, R. M.; Luo, Y.; Stulp, R. P.; Barone,V.; Stelwagen, T.; Bocciardi, R.; Nijveen, H.; Bolino, A.; Seri, M.;Ronchetto, P.; Pasini, B.; Bozzano, M.; Buys, C. H. C. M.; Romeo,G.: DNA polymorphisms and conditions for SSCP analysis of the 20exons of the Ret proto-oncogene. Oncogene 9: 3025-3029, 1994.
- [30361] 3913.Azen, E. A.; Goodman, P. A.; Lalley, P. A.: Human salivary proline-richprotein genes on chromosome 12. Am. J. Hum. Genet. 37: 418-424,1985.
- [30362] 3914.Mamula, P. W.; Heerema, N. A.; Palmer, C. G.; Lyons, K. M.; Karn,R. C.: Localization of the human salivary protein complex (SPC) tochromosome band 12p13.2. Cyto-

genet. Cell Genet. 39: 279–284, 1985.

[30363] 3915.Azen, E. A.: Genetic protein polymorphism in human saliva: an interpretive review. Biochem. Genet. 16: 79–99, 1978.

[30364] 3916.Azen, E. A.; Denniston, C.: Genetic polymorphism of PIF (parotid isoelectric focusing variant) proteins with linkage to the PPP (parotid proline-rich protein) gene complex. Biochem. Genet. 19: 475–485, 1981.

[30365] 3917.Azen, E. A.; Denniston, C. L.: Genetic polymorphism of human salivary proline-rich proteins: further genetic analysis. Biochem. Genet. 12: 109–120, 1974.

[30366] 3918.Azen, E. A.; Kim, H.-S.; Goodman, P.; Flynn, S.; Maeda, N.: Alleles at the PRH1 locus coding for the human salivary-acidic proline-rich proteins Pa, Db, and PIF. Am. J. Hum. Genet. 41: 1035–1047, 1987.

[30367] 3919.Azen, E. A.; Lush, I. E.; Taylor, B. A.: Close linkage of mouse genes for salivary proline-rich proteins (PRPs) and taste. Trends Genet. 2: 199–200, 1986.

[30368] 3920.Azen, E. A.; Maeda, N.: Molecular genetics of human salivary proteins and their polymorphisms. Adv. Hum. Genet. 17: 141–199, 1988.

[30369] 3921.Bernstein, F.: Ergebnisse einer biostatistischen zusammenfassenden Betrachtung ueber die erblichen Blutstruk-

turen des Menschen. Klin.Wschr. 3: 1495–1497, 1924.

- [30370] 3922.Bernstein, F.: Zusammenfassende Betrachtungen ueber die erblichenBlutstrukten des Menschen. Z. indukt. Abstamm. u. VererbLehre 37:237–270, 1925. Note: Translation: Blood Transfusion Division, U.S.Army Medical Research Laboratory, Fort Knox, Kentucky 40121.
- [30371] 3923.Friedman, R. D.; Merritt, A. D.: Partial purification and characterizationof a polymorphic protein (Pa) in human parotid saliva. Am. J. Hum.Genet. 27: 304–314, 1975.
- [30372] 3924.Friedman, R. D.; Merritt, A. D.; Rivas, M. L.: Genetic studiesof human acidic salivary protein (Pa). Am. J. Hum. Genet. 27: 292–303,1975.
- [30373] 3925.Ikemoto, S.; Minaguchi, K.; Hinohara, H.: Genetic polymorphismsof human parotid salivary proteins (Pa, Pb, Pr, Db and Pm) and salivaryamylase isozyme in Japanese population. Hum. Hered. 27: 328–331,1977.
- [30374] 3926.Kim, H.–S.; Maeda, N.: Structures of two HaeIII–type genes inthe human salivary proline–rich protein multigene family. J. Biol.Chem. 261: 6712–6718, 1986.
- [30375] 3927.Maeda, N.: Inheritance of the human salivary proline–rich proteins:a reinterpretation in terms of six loci forming two subfamilies. Biochem.Genet. 23: 455–464,

1985.

- [30376] 3928. Maeda, N.; Kim, H.-S.; Azen, E. A.; Smithies, O.: Differential RNA splicing and post-translational cleavages in the human salivary proline-rich protein gene system. *J. Biol. Chem.* 260: 11123–11130, 1985.
- [30377] 3929. O'Connell, P.; Lathrop, G. M.; Law, M.; Leppert, M.; Nakamura, Y.; Hoff, M.; Kumlin, E.; Thomas, W.; Elsner, T.; Ballard, L.; Goodman, P.; Azen, E.; Sadler, J. E.; Cai, G. Y.; Lalouel, J.-M.; White, R.: A primary genetic linkage map for human chromosome 12. *Genomics* 1: 93–102, 1987.
- [30378] 3930. O'Hanlon, K.; Weissbecker, K.; Cortessis, V.; Spence, M. A.; Azen, E. A.: Genes for salivary proline-rich proteins and taste for phenylthiourea are not closely linked in humans. *Cytogenet. Cell Genet.* 49: 315–317, 1988.
- [30379] 3931. Stern, C.: *Principles of Human Genetics*. San Francisco: W.H. Freeman (pub.) (3rd ed.): 1973. Pp. 256–262.
- [30380] 3932. von Dungern, E.; Hirschfeld, L.: Ueber Vererbung gruppenspezifischer Strukturen des Blutes. II. *Z. Immunforsch.* 6: 284–292, 1910. Note: Translation: G. P. Pohlmann: *Transfusion* 2: 70–74, 1962.
- [30381] 3933. Yu, P. L.; Karn, R. C.; Merritt, A. D.: Multipoint mapping of the human parotid salivary proteins: Pr, Pa, Db. (Abstract) *Am. J. Hum. Genet.* 30: 129A only, 1978.

- [30382] 3934.Yu, P. L.; Schwartz, R. C.; Merritt, A. D.; Azen, E. A.; Rivas,M. L.; Karn, R. C.; Craft, M. A.: Linkage relationships of the proline-richsalivary proteins (Pr, Pa, Db). Cytogenet. Cell Genet. 22: 655–658,1978.
- [30383] 3935.Furlong, C. E.; Richter, R. J.; Chapline, C.; Crabb, J. W.: Purificationof rabbit and human serum paraoxonase. Biochemistry 30: 10133–10140,1991.
- [30384] 3936.Furlong, C. E.; Richter, R. J.; Seidel, S. L.; Motulsky, A. G.: Role of genetic polymorphism of human plasma paraoxonase/arylesterasein hydrolysis of the insecticide metabolites chlorpyrifos oxon andparaoxon. Am. J. Hum. Genet. 43: 230–238, 1988.
- [30385] 3937.Garin, M.–C. B.; James, R. W.; Dussoix, P.; Blanche, H.; Passa,P.; Froguel, P.; Ruiz, J.: Paraoxonase polymorphism met-leu54 isassociated with modified serum concentrations of the enzyme: a possiblelink between the paraoxonase gene and increased risk of cardiovascularisease in diabetes. J. Clin. Invest. 99: 62–66, 1997.
- [30386] 3938.Geldmacher–von Mallinckrodt, M.: Polymorphism of human serumparaoxonase. Hum. Genet. 45 (suppl. 1): 65–68, 1978.
- [30387] 3939.Geldmacher–von Mallinckrodt, M.; Lindorft, H. H.; Petenyi, M.;Flugel, M.; Fischer, T.; Hiller, T.: Genetisch de–

terminierter Polymorphismusde menschlichen Serum-
Paroxonase (E.C.3.1.1.2). Humangenetik 17:331–335,
1973.

- [30388] 3940.Hassett, C.; Richter, R. J.; Humbert, R.; Chapline, C.; Crabb,J. W.; Omiecinski, C. J.; Furlong, C. E.: Characteriza-
tion of cDNAclones encoding rabbit and human serum
paraoxonase: the mature proteinretains its signal se-
quence. Biochemistry 30: 10141–10149, 1991.
- [30389] 3941.Humbert, R.; Adler, D. A.; Disteché, C. M.; Hassett,
C.; Omiecinski,C. J.; Furlong, C. E.: The molecular basis of
the human serum paraoxonaseactivity polymorphism. Na-
ture Genet. 3: 73–76, 1993.
- [30390] 3942.Ito, T.; Yasue, H.; Yoshimura, M.; Nakamura, S.;
Nakayama, M.;Shimasaki, Y.; Harada, E.; Mizuno, Y.;
Kawano, H.; Ogawa, H.: Paraoxonasegene gln192–to–arg
(Q192R) polymorphism is associated with coronaryartery
spasm. Hum. Genet. 110: 89–94, 2002.
- [30391] 3943.Kao, Y.–L.; Donaghue, K.; Chan, A.; Knight, J.; Silink,
M.: Avariant of paraoxonase (PON1) gene is associated
with diabetic retinopathyin IDDM. J. Clin. Endocr. Metab.
83: 2589–2592, 1998.
- [30392] 3944.La Du, B. N.: The human serum paraoxonase/
arylesterase polymorphism.(Editorial) Am. J. Hum. Genet.

43: 227–229, 1988.

- [30393] 3945.Li, W.-F.; Furlong, C. E.; Costa, L. G.: Paraoxonase protects against chlorpyrifos toxicity in mice. *Toxic. Lett.* 76: 219–226, 1995.
- [30394] 3946.Li, W.-F.; Matthews, C.; Disteché, C. M.; Costa, L. G.; Furlong, C. E.: Paraoxonase (Pon1) gene in mice: sequencing, chromosomal localization and developmental expression. *Pharmacogenetics* 7: 137–144, 1997.
- [30395] 3947.Mackness, B.; Mackness, M. I.; Arrol, S.; Turkie, W.; Durrington, P. N.: Effect of the human serum paraoxonase 55 and 192 genetic polymorphisms on the protection by high density lipoprotein against low density lipoprotein oxidative modification. *FEBS Lett.* 423: 57–60, 1998.
- [30396] 3948.Mochizuki, H.; Scherer, S. W.; Xi, T.; Nickle, D. C.; Majer, M.; Huizenga, J. J.; Tsui, L.-C.; Prochazka, M.: Human PON2 gene at 7q21.3: cloning, multiple mRNA forms, and missense polymorphisms in the coding sequence. *Gene* 213: 149–157, 1998.
- [30397] 3949.Mueller, R. F.; Hornung, S.; Furlong, C. E.; Anderson, J.; Giblett, E. R.; Motulsky, A. G.: Plasma paraoxonase polymorphism: a new enzyme assay, population, family, biochemical, and linkage studies. *Am. J. Hum. Genet.* 35: 393–408, 1983.

- [30398] 3950.Navab, M.; Hama-Levy, S.; Van Lenten, B. J.; Fonarow, G. C.; Cardinez,C. J.; Castellani, L. W.; Brennan, M.-L.; Lusis, A. J.; Fogelman,A. M.: Mildly oxidized LDL induces an increased apolipoprotein J/paraoxonaseratio. J. Clin. Invest. 99: 2005-2019, 1997.
- [30399] 3951.Neel, J. V.; Tanis, R. J.; Migliazza, E. C.; Spielman, R. S.;Salzano, F. M.; Oliver, W. J.; Morrow, M.; Bachofer, S.: Geneticstudies of the Macushi and Wapishana Indians. I. Rare genetic variantsand a 'private polymorphism' of esterase A. Hum. Genet. 36: 81-108,1977.
- [30400] 3952.Nielsen, A.; Eiberg, H.; Mohr, J.: Number of loci responsiblefor the inheritance of high and low activity of paraoxonase. Clin.Genet. 29: 216-221, 1986.
- [30401] 3953.Odawara, M.; Tachi, Y.; Yamashita, K.: Paraoxonase polymorphismGln192-Arg is associated with coronary heart disease in Japanese noninsulin-dependentdiabetes mellitus. J. Clin. Endocr. Metab. 82: 2257-2260, 1997.
- [30402] 3954.Ortigoza-Ferado, J.; Richter, R. J.; Hornung, S. K.; Motulsky,A. G.; Furlong, C. E.: Paraoxon hydrolysis in human serum mediatedby a genetically variable arylesterase and albumin. Am. J. Hum. Genet. 36:295-305, 1984.
- [30403] 3955.Paolisso, G.; Manzella, D.; Tagliamonte, M. R.; Barbi-eri, M.;Marfella, R.; Zito, G.; Bonafe, M.; Giugliano, D.;

Franceschi, C.;Varricchio, M.: The BB–paraoxonase genotype is associated with impaired brachial reactivity after acute hypertriglyceridemia in healthy subjects. J.Clin. Endocr. Metab. 86: 1078–1082, 2001.

[30404] 3956.Playfer, J. R.; Eze, L. C.; Bullen, M. F.; Evans, D. A. P.: Genetic polymorphism and interethnic variability of plasma paraoxonase activity. J.Med. Genet. 13: 337–342, 1976.

[30405] 3957.Schmiegelow, K.; Eiberg, H.; Tsui, L.–C.; Buchwald, M.; Phelan, P. D.; Williamson, R.; Warwick, W.; Niebuhr, E.; Mohr, J.; Schwartz, M.; Koch, C.: Linkage between the loci for cystic fibrosis and paraoxonase. Clin.Genet. 29: 374–377, 1986.

[30406] 3958.Serrato, M.; Marian, A. J.: A variant of human paraoxonase/arylesterase(HUMPONA) gene is a risk factor for coronary artery disease. J. Clin.Invest. 96: 3005–3008, 1995.

[30407] 3959.Shih, D. M.; Gu, L.; Xia, Y.–R.; Navab, M.; Li, W.–F.; Hama, S.;Castellani, L. W.; Furlong, C. E.; Costa, L. G.; Fogelman, A. M.;Lusis, A. J.: Mice lacking serum paraoxonase are susceptible to organophosphatotoxicity and atherosclerosis. Nature 394: 284–287, 1998.

[30408] 3960.Simpson, N. E.: Serum arylesterase levels of activity in twins and their parents. Am. J. Hum. Genet. 23:

375–382, 1971.

- [30409] 3961.Sorenson, R. C.; Primo-Parmo, S. L.; Camper, S.; La Du, B. N.: The genetic mapping and gene structure of mouse paraoxonase/arylesterase. *Genomics* 30:431–438, 1995.
- [30410] 3962.Tashian, R. E.: Genetic variation and evolution of the carboxylicesterases and carbonic anhydrases of primate erythrocytes. *Am. J.Hum. Genet.* 17: 257–272, 1965.
- [30411] 3963.Tashian, R. E.; Shaw, M. W.: Inheritance of an erythrocyte acetylesterasevariant of man. *Am. J. Hum. Genet.* 14: 295–300, 1962.
- [30412] 3964.Tsui, L.–C.; Buchwald, M.; Barker, D.; Braman, J. C.; Knowlton,R.; Schumm, J. W.; Eiberg, H.; Mohr, J.; Kennedy, D.; Plavsic, N.;Zsiga, M.; Markiewicz, D.; Akots, G.; Brown, V.; Helms, C.; Gravius,T.; Parker, C.; Rediker, K.; Donis–Keller, H.: Cystic fibrosis locusdefined by a genetically linked polymorphic DNA marker. *Science* 230:1054–1057, 1985.
- [30413] 3965.Gogolin, K. J.; Wray, L. K.; Slaughter, C. A.; Harris, H.: A monoclonalantibody that reacts with nonallelic enzyme glycoproteins. *Science* 216:59–61, 1982.
- [30414] 3966.Goldstein, D. J.; Rogers, C. E.; Harris, H.: Expression of alkalinephosphatase loci in mammalian tissues. *Proc.*

Nat. Acad. Sci. 77:2857–2860, 1980.

- [30415] 3967. Gould, B. S.: Studies on the source of serum phosphatase: the nature of the increased serum phosphatase in rats after fat feeding. Arch. Biochem. 4: 175–181, 1944.
- [30416] 3968. Griffin, C. A.; Smith, M.; Henthorn, P. S.; Harris, H.; Weiss, M. J.; Raducha, M.; Emanuel, B. S.: Human placental and intestinal alkaline phosphatase genes map to 2q34–q37. Am. J. Hum. Genet. 41:1025–1034, 1987.
- [30417] 3969. Henthorn, P. S.; Raducha, M.; Edwards, Y. H.; Weiss, M. J.; Slaughter, C.; Lafferty, M. A.; Harris, H.: Nucleotide and amino acid sequences of human intestinal alkaline phosphatase: close homology to placental alkaline phosphatase. Proc. Nat. Acad. Sci. 84: 1234–1238, 1987.
- [30418] 3970. Henthorn, P. S.; Raducha, M.; Kadesch, T.; Weiss, M. J.; Harris, H.: Sequence and characterization of the human intestinal alkaline phosphatase gene. J. Biol. Chem. 263: 12011–12019, 1988.
- [30419] 3971. Langman, M. J. S.; Leuthold, E.; Robson, E. B.; Harris, J.; Luffman, J. E.; Harris, H.: Influence of diet on the 'intestinal' component of serum alkaline phosphatase in people of different ABO blood groups and secretor status. Nature 212: 41–43, 1966.
- [30420] 3972. Lehmann, F.–G.: Human alkaline phosphatases: evi–

dence of three isoenzymes (placental, intestinal and liver–bone–kidney–type) by lectin–binding affinity and immunological specificity. *Biochim. Biophys. Acta* 616:41–59, 1980.

[30421] 1973. Rajput, B.; Degen, S. F.; Reich, E.; Waller, E. K.; Axelrod, J.; Eddy, R. L.; Shows, T. B.: Chromosomal locations of human tissue plasminogen activator and urokinase genes. *Science* 230: 672–674, 1985.

[30422] 1974. Chen, I.–T.; Roufa, D. J.: The transcriptionally active human ribosomal protein S17 gene. *Gene* 70: 107–116, 1988.

[30423] 1975. Lang, F.; Klingel, K.; Wagner, C. A.; Stegen, C.; Warntges, S.; Friedrich, B.; Lanzendorfer, M.; Melzig, J.; Moschen, I.; Steuer, S.; Waldegger, S.; Sauter, M.; and 9 others: Deranged transcriptional regulation of cell–volume–sensitive kinase hSGK in diabetic nephropathy. *Proc. Nat. Acad. Sci.* 97: 8157–8162, 2000.

[30424] 1976. Ebner, R.; Chen, R.–H.; Shum, L.; Lawler, S.; Zioncheck, T. F.; Lee, A.; Lopez, A. R.; Derynck, R.: Cloning of a type I TGF– β receptor and its effect on TGF– β binding to the type II receptor. *Science* 260:1344–1348, 1993.

[30425] 1977. Franzen, P.; ten Dijke, P.; Ichijo, H.; Yamashita, H.;

Schulz,P.; Heldin, C.-H.; Miyazono, K.: Cloning of a TGF-beta type I receptorthat forms a heteromeric complex with the TGF-beta type II receptor. *Cell* 75:681-692, 1993.

[30426] 3978.Inman, G. J.; Nicolas, F. J.; Hill, C. S.: Nucleocytoplasmic shuttling of Smads 2, 3, and 4 permits sensing of TGF-beta receptor activity. *Molec.Cell* 10: 283-294, 2002.

[30427] 3979.Johnson, D. W.; Qumsiyeh, M.; Benkhalifa, M.; Marchuk, D. A.:Assignment of human transforming growth factor-beta type I and typeIII receptor genes (TGFB1 and TGFB3) to 9q33-q34 and 1p32-p33, respectively. *Genomics* 28:356-357, 1995.

[30428] 3980.Kuan, J.; Kono, D. H.: Tgfbr1 maps to chromosome 4. *MammalianGenome* 9: 95-96, 1998.

[30429] 3981.Pasche, B.; Luo, Y.; Rao, P. H.; Nimer, S. D.; Dmitrovsky, E.;Caron, P.; Luzzatto, L.; Offit, K.; Cordon-Cardo, C.; Renault, B.;Satagopan, J. M.; Murty, V. V.; Massague, J.: Type I transforminggrowth factor beta receptor maps to 9q22 and exhibits a polymorphism and a rare variant within a polyalanine tract. *Cancer Res.* 58: 2727-2732,1998.

[30430] 3982.D'Eustachio, P.; Meyuhas, O.; Ruddle, F.; Perry, R. P.: Chromosomal distribution of ribosomal protein genes in the mouse. *Cell* 24: 307-312,1981.

- [30431] 3983.Filipenko, M. L.; Iantsen, E. I.; Muravlev, A. I.; Kopantsev, E.P.; Karpova, G. G.; Mertvetsov, N. P.: Mapping the genes for ribosomal proteins S14 and S17 on human chromosomes using cDNA from a panel of hybrid cells. *Bioorg. Khim.* 21: 349–353, 1995. Note: Article in Russian.
- [30432] 3984.Woolford, J. L., Jr.; Hereford, L. M.; Rosbash, M.: Isolation of cloned DNA sequences containing ribosomal protein genes from *Saccharomyces cerevisiae*. *Cell* 18: 1247–1259, 1979.
- [30433] 3985.Nucifora, G.; Begy, C. R.; Erickson, R.; Drabkin, H. A.; Rowley, J. D.: The 3;21 translocation in myelodysplasia results in a fusion transcript between the AML1 gene and the gene for EAP, a highly conserved protein associated with the Epstein–Barr virus small RNA EBER 1. *Proc. Nat. Acad. Sci.* 90: 7784–7788, 1993.
- [30434] 3986.Chang, A.; Schalkwijk, J.; Happle, R.; van de Kerkhof, P. C. M.: Elastase-inhibiting activity in scaling skin disorders. *Acta Derm. Venerol.* 70: 147–151, 1990.
- [30435] 3987.Kuijpers, A. L. A.; Pfundt, R.; Zeeuwen, P. L. J. M.; Molhuizen, H. O. F.; Mariman, E. C. M.; van de Kerkhof, P. C. M.; Schalkwijk, J.: SKALP/elafin gene polymorphisms are not associated with pustular forms of psoriasis. *Clin.*

Genet. 54: 96–101, 1998.

- [30436] 3988. Molhuizen, H. O. F.; Alkemade, H. A. C.; Zeeuwen, P. L. J. M.; de Jongh, G. J.; Wieringa, B.; Schalkwijk, J.: SKALP/elafin: an elastase inhibitor from cultured human keratinocytes: purification, cDNA sequence, and evidence for transglutaminase cross-linking. *J. Biol. Chem.* 268:12028–12032, 1993.
- [30437] 3989. Molhuizen, H. O. F.; Zeeuwen, P. L. J. M.; Olde Weghuis, D.; Geurtsvan Kessel, A.; Schalkwijk, J.: Assignment of the human gene encoding the epidermal serine proteinase inhibitor SKALP (PI3) to chromosomal region 20q12–q13. *Cytogenet. Cell Genet.* 66: 129–131, 1994.
- [30438] 3990. Sallenave, J.-M.; Ryle, A. P.: Purification and characterization of elastase-specific inhibitor: sequence homology with mucus proteinase inhibitor. *Biol. Chem. Hoppe-Seyler* 372: 13–21, 1991.
- [30439] 3991. Schalkwijk, J.; Chang, A.; Janssen, P.; de Jongh, G. J.; Mier, P. D.: Skin-derived antileucoproteases (SKALPs): characterization of two new elastase inhibitors from psoriatic epidermis. *Brit. J. Derm.* 122: 631–641, 1990.
- [30440] 3992. Schalkwijk, J.; de Roo, C.; de Jongh, G. J.: Skin-derived antileukoproteinase (SKALP), an elastase inhibitor from human keratinocytes: purification and biochemical

properties. *Biochim. Biophys. Acta* 1096: 148–154,1991.

[30441] 1993.Wiedow, O.; Schroeder, J.–M.; Gregory, H.; Young, J. A.; Christophers,E.: Elafin: an elastase–specific inhibitor of human skin: purification,characterization, and complete amino acid sequence. *J. Biol. Chem.* 265:14791–14795, 1990.

[30442] 1994.Begley, C. G.; Visvader, J.; Green, A. R.; Aplan, P. D.; Metcalf,D.; Kirsch, I. R.; Gough, N. M.: Molecular cloning and chromosomallocalization of the murine homolog of the human helix–loop–helix geneSCL. *Proc. Nat. Acad. Sci.* 88: 869–873, 1991.

[30443] 1995.Finger, L. R.; Kagan, J.; Christopher, G.; Kurtzberg, J.; Hershfield,M. S.; Nowell, P. C.; Croce, C. M.: Involve–ment of the TCL5 geneon human chromosome 1 in T–cell leukemia and melanoma. *Proc. Nat.Acad. Sci.* 86: 5039–5043, 1989.

[30444] 1996.Gottgens, B.; Barton, L. M.; Chapman, M. A.; Sinclair, A. M.; Knudsen,B.; Grafham, D.; Gilbert, J. G. R.; Rogers, J.; Bentley, D. R.; Green,A. R.: Transcriptional regulation of the stem cell leukemia gene(SCL)--comparative analysis of five vertebrate SCL loci. *Genome Res.* 12:749–759, 2002.

[30445] 1997.Gottgens, B.; Gilbert, J. G.; Barton, L. M.; Grafham,

D.; Rogers,J.; Bentley, D. R.; Green, A. R.: Long-range comparison of human and mouse SCL loci: localized regions of sensitivity to restriction endonucleases correspond precisely with peaks of conserved noncoding sequences. *Genome Res.* 11: 87–97, 2001.

[30446] 3998.Kocher, O.; Cheresch, P.; Lee, S. W.: Identification and partial characterization of a novel membrane-associated protein (MAP17) up-regulated in human carcinomas and modulating cell replication and tumor growth. *Am.J. Path.* 149: 493–500, 1996.

[30447] 3999.Kozak, M.: An analysis of 5-prime-noncoding sequences from 699 vertebrate messenger RNAs. *Nucleic Acids Res.* 15: 8125–8148, 1987.

[30448] 4000.Kurtzberg, J.; Bigner, S. H.; Hershfield, M. S.: Establishment of the DU.528 human lymphohemopoietic stem cell line. *J. Exp. Med.* 162:1561–1578, 1985.

[30449] 4001.Robb, L.; Lyons, I.; Li, R.; Hartley, L.; Kontgen, F.; Harvey, R. P.; Metcalf, D.; Begley, C. G.: Absence of yolk sac hematopoiesis from mice with a targeted disruption of the scl gene. *Proc. Nat.Acad. Sci.* 92: 7075–7079, 1995.

[30450] 4002.Shivdasani, R. A.; Mayer, E. L.; Orkin, S. H.: Absence of blood formation in mice lacking the T-cell leukaemia oncoprotein tal-1/SCL. *Nature* 373:432–434, 1995.

- [30451] 4003.Sinclair, A. M.; Bench, A. J.; Bloor, A. J. C.; Li, J.; Gottgens,B.; Stanley, M. L.; Miller, J.; Piltz, S.; Hunter, S.; Nacheva, E.P.; Sanchez, M.-J.; Green, A. R.: Rescue of the lethal scl-/- phenotypeby the human SCL locus. *Blood* 99: 3931-3938, 2002.
- [30452] 4004.Williams, M.; Lyu, M. S.; Yang, Y. L.; Lin, E. P.; Dunbrack, R.;Birren, B.; Cunningham, J.; Hunter, K.: ler5, a novel member of theslow-kinetics immediate-early genes. *Genomics* 44: 327-334, 1999.
- [30453] 4005.Xia, Y.; Brown, L.; Yang, C. Y.-C.; Tsou Tsan, J.; Siciliano,M. J.; Espinosa, R., III; Le Beau, M. M.; Baer, R. J.: TAL2, a helix-loop-helixgene activated by the (7;9)(q34;q32) translocation in human T-cellleukemia. *Proc. Nat. Acad. Sci.* 88: 11416-11420, 1991.
- [30454] 4006.Durick, K.; Gill, G. N.; Taylor, S. S.: Shc and Enigma are bothrequired for mitogenic signaling by Ret/ptc2. *Molec. Cell. Biol.* 18:2298-2308, 1998.
- [30455] 4007.Saadat, M.; Nomoto, K.; Mizuno, Y.; Kikuchi, K.; Yoshida, M. C.: Assignment of the gene encoding type 1-gamma protein phosphatasecatalytic subunit (PPP1CC) on human, rat, and mouse chromosomes. *Jpn.J. Hum. Genet.* 41: 159-165, 1996.
- [30456] 4008.Cohen, P.; Cohen, P. T. W.: Protein phosphatases

come of age. J.Biol. Chem. 264: 21435–21438, 1989.

[30457] 4009.Groves, M. R.; Hanlon, N.; Turowski, P.; Hemmings, B. A.; Barford,D.: The structure of the protein phosphatase 2A PR65/A subunit revealsthe conformation of its 15 tandemly repeated HEAT motifs. Cell 96:99–110, 1999.

[30458] 4010.Jones, T. A.; Barker, H. M.; da Cruz e Silva, E. F.; Mayer-Jaekel,R. E.; Hemmings, B. A.; Spurr, N. K.; Sheer, D.; Cohen, P. T. W.:Localization of the genes encoding the catalytic subunits of proteinphosphatase 2A to human chromosome bands 5q23–q31 and 8p12–p11.2,respectively. Cytogenet. Cell Genet. 63: 35–41, 1993.

[30459] 4011.Stone, S. R.; Mayer, R.; Wernet, W.; Maurer, F.; Hofsteenge, J.;Hemmings, B. A.: The nucleotide sequence of the cDNA encoding thehuman lung protein phosphatase 2A alpha catalytic subunit. NucleicAcids Res. 16: 11365 only, 1988.

[30460] 4012.Hemmings, B. A.; Wernet, W.; Mayer, R.; Maurer, F.; Hofsteenge,J.; Stone, S. R.: The nucleotide sequence of the cDNA encoding thehuman lung protein phosphatase 2A–beta catalytic subunit. NucleicAcids Res. 16: 11366 only, 1988.

[30461] 4013.Imbert, A.; Chaffanet, M.; Essioux, L.; Noguchi, T.;

Adelaide,J.; Kerangueven, F.; Le Paslier, D.; Bonaiti-Pellie, C.; Sobol, H.;Birnbaum, D.; Pebusque, M.-J.: Integrated map of the chromosome 8p12-p21region, a region involved in human cancers and Werner syndrome. *Genomics* 32:29-38, 1996.

[30462] 4014.Adamkiewicz, T. V.; McSherry, C.; Bach, F. H.; Houchins, J. P.: Natural killer lectin-like receptors have divergent carboxy-termini,distinct from C-type lectins. *Immunogenetics* 39: 218 only, 1994.

[30463] 4015.Houchins, J. P.; Yabe, T.; McSherry, C.; Bach, F. H.: DNA sequenceanalysis of NKG2, a family of related cDNA clones encoding type IIintegral membrane proteins on human natural killer cells. *J. Exp.Med.* 173: 1017-1020, 1991.

[30464] 4016.Plougastel, B.; Jones, T.; Trowsdale, J.: Genomic structure, chromosomelocation, and alternative splicing of the human NKG2A gene. *Immunogenetics* 44:286-291, 1996.

[30465] 4017.Plougastel, B.; Trowsdale, J.: Sequence analysis of a 62-kb regionoverlapping the human KLRC cluster of genes. *Genomics* 49: 193-199,1998.

[30466] 4018.Renedo, M.; Arce, I.; Rodriguez, A.; Carretero, M.; Lanier, L.L.; Lopez-Botet, M.; Fernandez-Ruiz, E.: The hu-

man natural killergene complex is located on chromosome 12p12–p13. Immunogenetics 46:307–311, 1997.

[30467] 4019.Yabe, T.; McSherry, C.; Bach, F. H.; Fisch, P.; Schall, R. P.; Sondel, P. M.; Houchins, J. P.: A multigene family on human chromosome 12 encodes natural killer–cell lectins. Immunogenetics 37: 455–460, 1993.

[30468] 4020.Cooper, A. M.; Kipnis, A.; Turner, J.; Magram, J.; Ferrante, J.; Orme, I. M.: Mice lacking bioactive IL–12 can generate protective, antigen–specific cellular responses to mycobacterial infection only if the IL–12 p40 subunit is present. J. Immun. 168: 1322–1327, 2002.

[30469] 4021.Gubler, U.; Chua, A. O.; Schoenhaut, D. S.; Dwyer, C. M.; McComas, W.; Motyka, R.; Nabavi, N.; Wolitzky, A. G.; Quinn, P. M.; Familletti, P. C.; Gately, M. K.: Coexpression of two distinct genes is required to generate secreted bioactive cytotoxic lymphocyte maturation factor. Proc. Nat. Acad. Sci. 88: 4143–4147, 1991.

[30470] 4022.Hall, S. S.: IL–12 at the crossroads. Science 268: 1432–1434, 1995.

[30471] 4023.Merberg, D. M.; Wolfe, S. F.; Clark, S. C.: Sequence similarity between NKSF and the IL6/GCSF family. (Letter) Immun. Today 13: 77–78, 1992.

[30472] 4024.Schwarz, A.; Stender, S.; Berneburg, M.; Bshmi, M.;

Kulms, D.; vanSteeg, H.; Grosse-Heitmeyer, K.; Krutmann, J.; Schwarz, T.: Interleukin-12 suppresses ultraviolet radiation-induced apoptosis by inducing DNA repair. *Nature Cell Biol.* 4: 26–31, 2002.

[30473] 4025. Schweitzer, P. A.; Noben-Trauth, N.; Pelsue, S. C.; Johnson, K.R.; Wolf, S. F.; Shultz, L. D.: Genetic mapping of the IL-12 alphachain gene (Il12a) on mouse chromosome 3. *Mammalian Genome* 7: 394–395, 1996.

[30474] 4026. Sieburth, D.; Jabs, E. W.; Warrington, J. A.; Li, X.; Lasota, J.; LaForgia, S.; Kelleher, K.; Huebner, K.; Wasmuth, J. J.; Wolf, S.F.: Assignment of genes encoding a unique cytokine (IL12) composed of two unrelated subunits to chromosomes 3 and 5. *Genomics* 14: 59–62, 1992.

[30475] 4027. Wolf, S. F.; Sieburth, D.; Sypek, J.: Interleukin 12: a key modulator of immune function. *Stem Cells* 12: 154–168, 1994.

[30476] 4028. Wolf, S. F.; Temple, P. A.; Kobayashi, M.; Young, D.; Dicig, M.; Lowe, L.; Dzialo, R.; Fitz, L.; Ferenz, C.; Hewick, R. M.; Kelleher, K.; Herrmann, S. H.; Clark, S. C.; Azzoni, L.; Chan, S. H.; Trinchieri, G.; Perussia, B.: Cloning of cDNA for natural killer cell stimulatory factor, a heterodimeric cytokine with multiple biologic effects on T and natural killer cells. *J. Immun.* 146: 3074–3081, 1991.

- [30477] 4029.Mizuno, T.; Kaibuchi, K.; Yamamoto, T.; Kawamura, M.; Sakoda, T.;Fujioka, H.; Matsuura, Y.; Takai, Y.: A stimulatory GDP/GTP exchangeprotein for smg p21 is active on the post-translationally processedform of c-Ki-ras p21 and rhoA p21. *Proc. Nat. Acad. Sci.* 88: 6442–6446,1991.
- [30478] 4030.Lindner, T. H.; Njolstad, P. R.; Horikawa, Y.; Bostad, L.; Bell,G. I.; Sovik, O.: A novel syndrome of diabetes melitus, renal dysfunctionand genital malformation associated with a partial deletion of thepseudo-POU domain of hepatocyte nuclear factor-1-beta. *Hum. Molec.Genet.* 8: 2001–2008, 1999.
- [30479] 4031.Anderson, L. A.; Hall, J. M.; Lee, M. K.; Lebo, R. V.; King, M.C.: Polymorphisms near chromosome 1cen. (Abstract) *Cytogenet. CellGenet.* 51: 951, 1989.
- [30480] 4032.Gendler, S. J.; Lancaster, C. A.; Taylor-Padimitriou, J.; Duhig,T.; Peat, N.; Burchell, J.; Pemberton, L.; Lalani, E.-N.; Wilson,D.: Molecular cloning and expression of human tumor-associated polymorphicepithelial mucin. *J. Biol. Chem.* 265: 15286–15293, 1990.
- [30481] 4033.Karlsson, S.; Swallow, D. M.; Griffiths, B.; Corney, G.; Hopkinson,D. A.; Dawnay, A.; Cartron, J. P.: A genetic polymorphism of a humanurinary mucin. *Ann. Hum. Genet.* 47: 263–269, 1983.

- [30482] 4034.Kingsmore, S. F.; Spicer, A. P.; Gendler, S. J.; Seldin, M. F.: Genetic mapping of the tumor-associated mucin 1 gene on mouse chromosome3. *Mammalian Genome* 6: 378, 1995.
- [30483] 4035.Lan, M. S.; Batra, S. K.; Qi, W.-N.; Metzgar, R. S.; Hollingsworth,M. A.: Cloning and sequencing of a human pancreatic tumor mucin cDNA. *J.Biol. Chem.* 265: 15294–15299, 1990.
- [30484] 4036.Ligtenberg, M. J.; Gennissen, A. M.; Vos, H. L.; Hilkens, J.:A single nucleotide polymorphism in an exon dictates allele dependent differential splicing of episialin mRNA. *Nucleic Acids Res.* 19:297–301, 1991.
- [30485] 4037.Ligtenberg, M. J. L.; Vos, H. L.; Gennissen, A. M. C.; Hilkens,J.: Episialin, a carcinoma-associated mucin, is generated by a polymorphic gene encoding splice variants with alternative amino termini. *J.Biol. Chem.* 265: 5573–5578, 1990.
- [30486] 4038.Middleton-Price, H.; Gendler, S.; Malcolm, S.: Close linkage ofPUM and SPTA within chromosome band 1q21. *Ann. Hum. Genet.* 52: 273–278,1988.
- [30487] 4039.Pratt, W. S.; Islam, I.; Swallow, D. M.: Two additional polymorphisms within the hypervariable MUC1 gene: association of alleles either side of the VNTR region. *Ann. Hum.*

Genet. 60: 21–28, 1996.

- [30488] 4040.Silva, F.; Carvalho, F.; Peixoto, A.; Seixas, M.; Almeida, R.;Carneiro, F.; Mequita, P.; Figueiredo, C.; Nogueira, C.; Swallow,D. M.; Amorim, A.; David, L.: MUC1 gene polymorphism in the gastriccarcinogenesis pathway. Europ. J. Hum. Genet. 9: 548–552, 2001.
- [30489] 4041.Swallow, D. M.; Gendler, S.; Griffiths, B.; Corney, G.; Taylor–Papadimitriou, J.; Bramwell, M. E.: The human tumour–associated epithelial mucinsare coded by an expressed hypervariable gene locus PUM. Nature 328:82–84, 1987.
- [30490] 4042.Swallow, D. M.; Gendler, S.; Griffiths, B.; Kearney, A.; Povey,S.; Sheer, D.; Palmer, R. W.; Taylor–Papadimitriou, J.: The hypervariablegene locus PUM, which codes for the tumour associated epithelial mucins,is located on chromosome 1, within the region 1q21–24. (Abstract) Cyto–genet.Cell Genet. 46: 701, 1987.
- [30491] 4043.Swallow, D. M.; Gendler, S.; Griffiths, B.; Kearney, A.; Povey,S.; Sheer, D.; Palmer, R. W.; Taylor–Papadimitriou, J.: The hypervariablegene locus PUM, which codes for the tumour associated epithelial mucins,is located on chromosome 1, within the region 1q21–q24. Ann. Hum.Genet. 51: 289–294, 1987.

- [30492] 4044.Swallow, D. M.; Griffiths, B.; Noades, J.; Corney, G.: Linkagebetween the expressed hypervariable gene locus PUM and the gene codingfor the Duffy blood group FY. Ann. Hum. Genet. 52: 269–271, 1988.
- [30493] 4045.Cole, S. P. C.; Bhardwaj, G.; Gerlach, J. H.; Mackie, J. E.; Grant,C. E.; Almquist, K. C.; Stewart, A. J.; Kurz, E. U.; Duncan, A. M.V.; Deeley, R. G.: Overexpression of a trans–porter gene in a multidrug–resistanthuman lung cancer cell line. Science 258: 1650–1654, 1992.
- [30494] 4046.Conrad, S.; Kauffmann, H.–M.; Ito, K.; Deeley, R. G.; Cole, S.P. C.; Schrenk, D.: Identification of human mul–tidrug resistanceprotein 1 (MRP1) mutations and charac–terization of a G671V substitution. J.Hum. Genet. 46: 656–663, 2001.
- [30495] 4047.Grant, C. E.; Kurz, E. U.; Cole, S. P. C.; Deeley, R. G.: Analysisof the intron–exon organization of the human multidrug–resistanceprotein gene (MRP) and alternative splicing of its mRNA. Genomics 45:368–378, 1997.
- [30496] 4048.Lorico, A.; Bertola, A.; Baum, C.; Fodstad, O.; Rappa, G.: Roleof multidrug resistance protein 1 in protection from heavy metal oxyanions:investigations in vitro and in Mrp1–deficient mice. Biochem. Biophys.Res. Commun. 291: 617–622, 2002.

- [30497] 4049.Robbiani, D. F.; Finch, R. A.; Jager, D.; Muller, W. A.; Sartorelli, A. C.; Randolph, G. J.: The leukotriene C4 transporter MRP1 regulates CCL19 (MIP-3-beta, ELC)-dependent mobilization of dendritic cells to lymph nodes. *Cell* 103: 757-768, 2000.
- [30498] 4050.Schultz, M. J.; Wijnholds, J.; Peppelenbosch, M. P.; Vervoordeldonk, M. J. B. M.; Speelman, P.; van Deventer, S. J. H.; Borst, P.; van der Poll, T.: Mice lacking the multidrug resistance protein 1 are resistant to *Streptococcus pneumoniae*-induced pneumonia. *J. Immunol.* 166:4059-4064, 2001.
- [30499] 4051.Zaman, G. J. R.; Flens, M. J.; van Leusden, M. R.; de Haas, M.; Mulder, H. S.; Lankelma, J.; Pinedo, H. M.; Scheper, R. J.; Baas, F.; Broxterman, H. J.; Borst, P.: The human multidrug resistance-associated protein MRP is a plasma membrane drug-efflux pump. *Proc. Nat. Acad. Sci.* 91: 8822-8826, 1994.
- [30500] 4052.Polymeropoulos, M. H.; Xiao, H.; Glodek, A.; Gorski, M.; Adams, M. D.; Moreno, R. F.; Fitzgerald, M. G.; Venter, J. C.; Merrill, C. R.: Chromosomal assignment of 46 brain cDNAs. *Genomics* 12: 492-496, 1992.
- [30501] 4053.Riess, O.; Epplen, C.; Siedlaczek, I.; Epplen, J. T.: Chromosomal assignment of the human smg GDP dissoci-

ation stimulator gene to human chromosome 4q21–q25.
Hum. Genet. 92: 629–630, 1993.

- [30502] 4054.Doria, A.; Caldwell, J. S.; Ji, L.; Reynet, C.; Rich, S. S.; Weremowicz, S.; Morton, C. C.; Warram, J. H.; Kahn, C. R.; Krolewski, A. S.: Trinucleotide repeats at the rad locus: allele distributions in NIDDM and mapping to a 3–cM region on chromosome 16q. Diabetes 44: 243–247, 1995.
- [30503] 4055.Orho, M.; Carlsson, M.; Kanninen, T.; Groop, L. C.: Polymorphism at the rad gene is not associated with NIDDM in Finns. Diabetes 45: 429–433, 1996.
- [30504] 4056.Reynet, C.; Kahn, C. R.: Rad: a member of the Ras family overexpressed in muscle of type II diabetic humans. Science 262: 1441–1444, 1993.
- [30505] 4057.le Gallic, L.; Fort, P.: Structure of the human ARHG locus encoding the Rho/Rac–like RhoG GTPase. Genomics 42: 157–160, 1997.
- [30506] 4058.Taviaux, S. A.; Vincent, S.; Fort, P.; Demaille, J. G.: Localization of ARHG, a member of the RAS homolog gene family, to 11p15.5–11p15.4 by fluorescence in situ hybridization. Genomics 16: 788–790, 1993.
- [30507] 4059.Vincent, S.; Jeanteur, P.; Fort, P.: Growth–regulated expression of rhoG, a new member of the ras homolog gene family. Molec. Cell. Biol. 12: 3138–3148, 1992.

- [30508] 4060.Allan, B. B.; Moyer, B. D.; Balch, W. E.: Rab1 recruitment of p115 into a cis-SNARE complex: programming budding COPII vesicles for fusion. *Science* 289: 444–448, 2000.
- [30509] 4061.Wedemeyer, N.; Lengeling, A.; Ronsiek, M.; Korthaus, D.; Baer, K.; Wuttke, M.; Jockusch, H.: YAC contigs of the Rab1 and wobbler(wr) spinal muscular atrophy gene region on proximal mouse chromosome 11 and of the homologous region on human chromosome 2p. *Genomics* 32:447–454, 1996.
- [30510] 4062.Rousseau-Merck, M. F.; Zahraoui, A.; Touchot, N.; Tavitian, A.; Berger, R.: Chromosome assignment of four RAS-related RAB genes. *Hum.Genet.* 86: 350–354, 1991.
- [30511] 4063.Barbosa, M. D. F. S.; Johnson, S. A.; Achey, K.; Gutierrez, M.; Wakeland, E. K.; Zerial, M.; Kingsmore, S. F.: The Rab protein family: genetic mapping of six Rab genes in the mouse. *Genomics* 30: 439–444, 1995.
- [30512] 4064.Rousseau-Merck, M.-F.; Zahraoui, A.; Touchot, N.; Tavitian, A.; Berger, R.: Chromosome assignment of four RAS-related RAB genes. *Hum.Genet.* 86: 350–354, 1991.
- [30513] 4065.Bucci, C.; Parton, R. G.; Mather, I. H.; Stunnenberg, H.; Simons, K.; Hoflack, B.; Zerial, M.: The small GTPase rab5 functions as a regulatory factor in the early endocytic

pathway. Cell 70: 715–728,1992.

- [30514] 4066.Stenmark, H.; Vitale, G.; Ullrich, O.; Zerial, M.: Rabaptin-5is a direct effector of the small GTPase Rab5 in endocytic membranefusion. Cell 83: 423–432, 1995.
- [30515] 4067.Xiao, G.–H.; Shoarinejad, F.; Jin, F.; Golemis, E. A.; Yeung, R.S.: The tuberous sclerosis 2 gene product, tuberin, functions asa Rab5 GTPase activating protein (GAP) in modulating endocytosis. J.Biol. Chem. 272: 6097–6100, 1997.
- [30516] 4068.Korenberg, J. R.; Chen, X.–N.; Adams, M. D.; Venter, J. C.: Towarda cDNA map of the human genome. Genomics 29: 364–370, 1995.
- [30517] 4069.Wilson, D. B.; Wilson, M. P.: Identification and sub-cellular localizationof human rab5b, a new member of the ras-related superfamily of GTPases. J.Clin. Invest. 89: 996–1005, 1992.
- [30518] 4070.Asha, H.; de Ruiter, N. D.; Wang, M.–G.; Hariharan, I. K.: TheRap1 GTPase functions as a regulator of morphogenesis in vivo. EMBOJ. 18: 605–615, 1999.
- [30519] 4071.Boussiotis, V. A.; Freeman, G. J.; Berezovskaya, A.; Barber, D.L.; Nadler, L. M.: Maintenance of human T cell anergy: blocking ofIL–2 gene transcription by activated Rap1. Science 278: 124–128,1997.

- [30520] 4072.Kitayama, H.; Sugimoto, Y.; Matsuzaki, T.; Ikawa, Y.; Noda, M.: A ras-related gene with transformation suppressor activity. *Cell* 56:77–84, 1989.
- [30521] 4073.Knox, A. L.; Brown, N. H.: Rap1 GTPase regulation of adherensjunction positioning and cell adhesion. *Science* 295: 1285–1288,2002.
- [30522] 4074.Muller-Ladner, U.; Judex, M.; Ballhorn, W.; Kullmann, F.; Distler,O.; Schlottmann, K.; Gay, R. E.; Scholmerich, J.; Gay, S.: Activationof the IL-4 STAT pathway in rheumatoid synovium. *J. Immun.* 164:3894–3901, 2000.
- [30523] 4075.Aita, N.; Ishii, K.; Akamatsu, Y.; Ogasawara, Y.; Tanabe, S.:Cloning and expression of human liver rhodanese cDNA. *Biochem. Biophys.Res. Commun.* 231: 56–60, 1997.
- [30524] 4076.Cagianut, B.; Rhyner, K.; Furrer, W.; Schnebli, H. P.: Thiosulphate-sulphurtransferase (rhodanese) deficiency in Leber's hereditary optic atrophy.(Letter) *Lancet* II: 981–982, 1981.
- [30525] 4077.Nikoskelainen, E.: New aspects of the genetic, etiology, andclinical puzzle of Leber's disease. *Neurology* 34: 1482–1484, 1984.
- [30526] 4078.Scott, E. M.; Wright, R. C.: Genetic polymorphism of rhodanese from human erythrocytes. *Am. J. Hum. Genet.*

32: 112–114, 1980.

- [30527] 4079.Weng, L.; Heinrichson, R. L.; Westley, J.: Active site cysteinyl and arginyl residues of rhodanese. *J. Biol. Chem.* 253: 8109–8119, 1978.
- [30528] 4080.Whitehouse, D. B.; Pilz, A. J.; Porta, G.; Hopkinson, D. A.: Rhodanese isozymes in human tissues. *Ann. Hum. Genet.* 52: 1–10, 1988.
- [30529] 4081.Whitehouse, D. B.; Poole, C. J. M.; Kind, P. R. N.; Hopkinson, D. A.: Rhodanese isozymes in three subjects with Leber's optic neuropathy. *J. Med. Genet.* 26: 113–115, 1989.
- [30530] 4082.Alloway, P. G.; Howard, L.; Dolph, P. J.: The formation of stable rhodopsin–arrestin complexes induces apoptosis and photoreceptor cell degeneration. *Neuron* 28: 129–138, 2000.
- [30531] 4083.Byrne, J. A.; Smith, P. J.: The 11p15.5 ribonucleotide reductase M1 subunit locus is not imprinted in Wilms' tumour and hepatoblastoma. *Hum. Genet.* 91: 275–277, 1993.
- [30532] 4084.Bouchard, B.; Del Marmol, V.; Jackson, I. J.; Cherif, D.; Dubertret, L.: Molecular characterization of a human tyrosinase–related–protein–2 cDNA: patterns of expression in melanocytic cells. *Europ. J. Biochem.* 219: 127–134,

1994.

- [30533] 4085. Budd, P. S.; Jackson, I. J.: Structure of the mouse tyrosinase-related protein-2/dopachrome tautomerase (Tyrp2/Dct) gene and sequence of two novel slaty alleles. *Genomics* 29: 35–43, 1995.
- [30534] 4086. Cassady, J. L.; Sturm, R. A.: Sequence of the human dopachrome tautomerase-encoding TRP-2 cDNA. *Gene* 143: 295–298, 1994.
- [30535] 4087. Jackson, I. J.; Chambers, D. M.; Tsukamoto, K.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Hearing, V.: A second tyrosinase-related protein, TRP-2, maps to and is mutated at the mouse slaty locus. *EMBOJ.* 11: 527–535, 1992.
- [30536] 4088. Khong, H. T.; Rosenberg, S. A.: Pre-existing immunity to tyrosinase-related protein (TRP)-2, a new TRP-2 isoform, and the NY-ESO-1 melanoma antigen in a patient with a dramatic response to immunotherapy. *J. Immun.* 168: 951–956, 2002.
- [30537] 4089. Kwon, B. S.: Pigmentation genes: the tyrosinase gene family and the pmel 17 gene family. *J. Invest. Derm.* 100: 134S–140S, 1993.
- [30538] 4090. Sturm, R. A.; Baker, E.; Sutherland, G. R.: Assignment of the tyrosinase-related protein-2 gene (TYRP2) to

human chromosome 13q31–q32 by fluorescence in situ hybridization: extended synteny with mouse chromosome 14. *Genomics* 21: 293–296, 1994.

[30539] 4091. Sturm, R. A.; O'Sullivan B. J.; Box, N. F.; Smith, A. G.; Smit, S. E.; Puttick, E. R. J.; Parsons, P. G.; Dunn, I. S.: Chromosomal structure of the human TYRP1 and TYRP2 loci and comparison of the tyrosinase-related protein gene family. *Genomics* 29: 24–34, 1995.

[30540] 4092. Ouafik, L'H.; Stoffers, D. A.; Campbell, T. A.; Johnson, R. C.; Bloomquist, B. T.; Mains, R. E.; Eipper, B. A.: The multifunctional peptidylglycine alpha-amidating monooxygenase gene: exon/intron organization of catalytic, processing, and routing domains. *Molec. Endocr.* 6: 1571–1584, 1992.

[30541] 4093. King, C. R.; Schimke, R. N.; Arthur, T.; Davoren, B.; Collins, D.: Proximal 3p deletion in renal cell carcinoma cells from a patient with von Hippel–Lindau disease. *Cancer Genet. Cytogenet.* 27: 345–348, 1987.

[30542] 4094. Li, F. P.; Decker, H.-J. H.; Zbar, B.; Stanton, V. P., Jr.; Kovacs, G.; Seizinger, B. R.; Aburatani, H.; Sandberg, A. A.; Berg, S.; Hosoe, S.; Brown, R. S.: Clinical and genetic studies of renal cell carcinoma in a family with a constitutional chromosome 3;8 translocation: genetics of familial renal

carcinoma. *Ann. Intern. Med.* 118: 106–111, 1993.

[30543] 4095. Neel, B. G.; Jhanwar, S. C.; Chaganti, R. S. K.; Hayward, W. S.: Two human c-onc genes are located on the long arm of chromosome 8. *Proc. Nat. Acad. Sci.* 79: 7842–7846, 1982.

[30544] 4096. Ohta, M.; Inoue, H.; Cotticelli, M. G.; Kastury, K.; Baffa, R.; Palazzo, J.; Siprashvili, Z.; Mori, M.; McCue, P.; Druck, T.; Croce, C. M.; Huebner, K.: The FHIT gene, spanning the chromosome 3p14.2 fragile site and renal carcinoma-associated t(3;8) breakpoint, is abnormal in digestive tract cancers. *Cell* 84: 587–597, 1996.

[30545] 4097. Roses, A. D.: From genes to mechanisms to therapies: lessons to be learned from neurological disorders. *Nature Med.* 2: 267–269, 1996.

[30546] 4098. Wood, J. D.; Yuan, J.; Margolis, R. L.; Colomer, V.; Duan, K.; Kushi, J.; Kaminsky, Z.; Kleiderlein, J. J., Jr.; Sharp, A. H.; Ross, C. A.: Atrophin-1, the DRPLA gene product, interacts with two families of WW domain-containing proteins. *Molec. Cell. Neurosci.* 11: 149–160, 1998.

[30547] 4099. Aksoy, M.; Erdem, S.: Combination of hereditary elliptocytosis and heterozygous beta-thalassemia: a family study. *J. Med. Genet.* 5: 298–301, 1968.

- [30548] 4100.Alloisio, N.; Dorleac, E.; Delaunay, J.; Girot, R.; Ga-land, C.;Boivin, P.: A shortened variant of red cell mem-
brane protein 4.1. Blood 60:265–267, 1982.
- [30549] 4101.Alloisio, N.; Dorleac, E.; Girot, R.; Delaunay, J.: Anal-
ysis of red cell membrane in a family with hereditary ellip-
tocytes: total or partial absence of protein 4.1. Hum.
Genet. 59: 68–71, 1981.
- [30550] 4102.Alloisio, N.; Morle, L.; Dorleac, E.; Gentilhomme, O.;
Bachir,D.; Guetarni, D.; Colonna, P.; Bost, M.; Zouaoui, Z.;
Roda, L.; Roussel,D.; Delaunay, J.: The heterozygous form
of 4.1(–) hereditary elliptocytosis[the 4.1(–) trait]. Blood
65: 46–51, 1985.
- [30551] 4103.Bahary, N.; Zorich, G.; Pachter, J. E.; Leibel, R. L.;
Friedman,J. M.: Molecular genetic linkage maps of mouse
chromosomes 4 and
- [30552] 4104.Genomics 11: 33–47, 1991.6. Baklouti, F.; Huang,
S.–C.; Vulliamy, T. J.; Delaunay, J.; Benz,E. J., Jr.: Organiza-
tion of the human protein 4.1 genomic locus:new insights
into the tissue–specific alternative splicing of the pre-
mRNA. Genomics 39: 289–302, 1997.
- [30553] 4105.Bannerman, R. M.; Renwick, J. H.: The hereditary el-
liptocytoses:clinical and linkage data. Ann. Hum. Genet.
26: 23–38, 1962.

- [30554] 4106. Clarke, C. A.; Donohoe, W. T. A.; Finn, R.; McConnell, R. B.; Sheppard, P. M.; Nicol, D. S. H.: Data on linkage in man: ovalocytosis, sickling and the Rhesus blood group complex. *Ann. Hum. Genet.* 24: 283–287, 1960.
- [30555] 4107. Conboy, J.; Kan, Y. W.; Shoet, S. B.; Mohandas, N.: Molecular cloning of protein 4.1, a major structural element of the human erythrocyte membrane skeleton. *Proc. Nat. Acad. Sci.* 83: 9512–9516, 1986.
- [30556] 4108. Conboy, J.; Marchesi, S.; Kim, R.; Agre, P.; Kan, Y. W.; Mohandas, N.: Molecular analysis of insertion/deletion mutations in protein 4.1 in elliptocytosis. II. Determination of molecular genetic origin of rearrangements. *J. Clin. Invest.* 86: 524–530, 1990.
- [30557] 4109. Conboy, J.; Mohandas, N.; Tchernia, G.; Kan, Y. W.: Molecular basis of hereditary elliptocytosis due to protein 4.1 deficiency. *New Eng. J. Med.* 315: 680–685, 1986.
- [30558] 4110. Conboy, J. G.: Structure, function, and molecular genetics of erythroid membrane skeletal protein 4.1 in normal and abnormal red blood cells. *Seminars Hemat.* 30: 58–73, 1993.
- [30559] 4111. Conboy, J. G.; Chan, J.; Mohandas, N.; Kan, Y. W.: Multiple protein 4.1 isoforms produced by alternative

splicing in human erythroid cells. *Proc.Nat. Acad. Sci.* 85: 9062–9065, 1988.

[30560] 4112.Conboy, J. G.; Chasis, J. A.; Winardi, R.; Tchernia, G.; Kan,Y. W.; Mohandas, N.: An isoform–specific mutation in the protein4.1 gene results in hereditary elliptocytosis and complete deficiencyof protein 4.1 in erythrocytes but not in nonerythroid cells. *J.Clin. Invest.* 91: 77–82, 1993.

[30561] 4113.Conboy, J. G.; Mohandas, N.; Wang, C.; Tchernia, G.; Shohet, S.B.; Kan, Y. W.: Molecular cloning and characteri–zation of the genecoding for red cell membrane skeletal protein 4.1. (Abstract) *Blood* 66(suppl. 1): 31A, 1985.

[30562] 4114.Cook, P. J. L.; Noades, J. E.; Newton, M. S.; de Mey, R.: Onthe orientation of the Rh:E1–1 linkage group. *Ann. Hum. Genet.* 41:157–162, 1977.

[30563] 4115.Lane, P. W.: Association of megacolon with two re–cessive spottinggenes in the mouse. *J. Hered.* 57: 29–31, 1966.

[30564] 4116.Matsushima, Y.; Shinkai, Y.; Kobayashi, Y.; Sakamoto, M.; Kunieda,T.; Tachibana, M.: A mouse model of Waardenburg syndrome type 4 witha new spontaneous mutation of the endothelin–B receptor gene. *Mam–malianGenome* 13: 30–35, 2002.

[30565] 4117.Metallinos, D. L.; Bowling, A. T.; Rine, J.: A missense

mutation in the endothelin-B receptor gene is associated with lethal whitefoal syndrome: an equine version of Hirschsprung disease. *Mammalian Genome* 9: 426–431, 1998.

[30566] 4118. Nakamura, M.; Takayanagi, R.; Sakai, Y.; Sakamoto, S.; Hagiwara, H.; Mizuno, T.; Saito, Y.; Hirose, S.; Yamamoto, M.; Nawata, H.: Cloning and sequence analysis of a cDNA encoding human non-selective type of endothelin receptor. *Biochem. Biophys. Res. Commun.* 177:34–39, 1991.

[30567] 4119. Ogawa, Y.; Nakao, K.; Arai, H.; Nakagawa, O.; Hosoda, K.; Suga, S.; Nakanishi, S.; Imura, H.: Molecular cloning of a non-isopeptide-selective human endothelin receptor. *Biochem. Biophys. Res. Commun.* 178: 248–255, 1991.

[30568] 4120. Pao, M. M.; Tsutsumi, M.; Liang, G.; Uzvolgyi, E.; Gonzales, F. A.; Jones, P. A.: The endothelin receptor B (EDNRB) promoter displays heterogeneous, site specific methylation patterns in normal and tumor cells. *Hum. Molec. Genet.* 10: 903–910, 2001.

[30569] 4121. Puffenberger, E. G.; Hosoda, K.; Washington, S. S.; Nakao, K.; de Wit, D.; Yanagisawa, M.; Chakravarti, A.: A missense mutation of the endothelin-B receptor gene in

multigenic Hirschsprung's disease. *Cell* 79:1257–1266, 1994.

[30570] 4122.Santschi, E. M.; Purdy, A. K.; Valberg, S. J.; Vrotsos, P. D.;Kaese, H.; Mickelson, J. R.: Endothelin receptor B polymorphism associatedwith lethal white foal syndrome in horses. *Mammalian Genome* 9: 306–309,1998.

[30571] 4123.Shin, M. K.; Levorse, J. M.; Ingram, R. S.; Tilghman, S. M.:The temporal requirement for endothelin receptor–B signalling duringneural crest development. *Nature* 402: 496–501, 1999.

[30572] 4124.Shin, M. K.; Russell, L. B.; Tilghman, S. M.: Molecular characterizationof four induced alleles at the *Ednrb* locus. *Proc. Nat. Acad. Sci.* 94:13105–13110, 1997.

[30573] 4125.Svensson, P.–J.; Anvret, M.; Molander, M.–L.; Nordenskjold, A.: Phenotypic variation in a family with mutations in two Hirschsprung–relatedgenes (*RET* and endothelin receptor B). *Hum. Genet.* 103: 145–148,1998.

[30574] 4126.Syrris, P.; Carter, N. D.; Patton, M. A.: Novel non-sense mutationof the endothelin–B receptor gene in a family with Waardenburg–Hirschsprungdisease. *Am. J. Med. Genet.* 87: 69–71, 1999.

[30575] 4127.Takayanagi, R.; Ohnaka, K.; Takasaki, C.; Ohashi, M.; Nawata,H.: Multiple subtypes of endothelin receptors in

porcine tissues: characterization by ligand binding, affinity labeling and regional distribution. Regul. Pept. 32: 23–37, 1991.

- [30576] 4128. Tanaka, H.; Moroi, K.; Iwai, J.; Takahashi, H.; Ohnuma, N.; Hori, S.; Takimoto, M.; Nishiyama, M.; Masaki, T.; Yanagisawa, M.; Sekiya, S.; Kimura, S.: Novel mutations of the endothelin B receptor gene in patients with Hirschsprung's disease and their characterization. J. Biol. Chem. 273: 11378–11383, 1998.
- [30577] 4129. Vane, J. R.: Endothelins come home to roost. Nature 348: 673, 1990.
- [30578] 4130. Verheij, J. B. G. M.; Kunze, J.; Osinga, J.; van Essen, A. J.; Hofstra, R. M. W.: ABCD syndrome is caused by a homozygous mutation in the EDNRB gene. Am. J. Med. Genet. 108: 223–225, 2002.
- [30579] 4131. Yang, G. C.; Croaker, D.; Zhang, A. L.; Manglick, P.; Cartmill, T.; Cass, D.: A dinucleotide mutation in the endothelin-B receptor gene is associated with lethal white foal syndrome (LWFS); a horse variant of Hirschsprung disease (HSCR). Hum. Molec. Genet. 7: 1047–1052, 1998.
- [30580] 4132. Hanks, M.; Wurst, W.; Anson-Cartwright, L.; Auerbach, A. B.; Joyner, A. L.: Rescue of the En-1 mutant phenotype by replacement of En-1 with En-2. Science 269:

679–682, 1995.

- [30581] 4133.Johnson, R. L.; Tabin, C. J.: Molecular models for vertebrate limb development. *Cell* 90: 979–990, 1997.
- [30582] 4134.Kohler, A.; Logan, C.; Joyner, A. L.; Muenke, M.: Regional assignment of the human homeobox-containing gene EN1 to chromosome 2q13–q21. *Genomics* 15:233–235, 1993.
- [30583] 4135.Logan, C.; Hanks, M. C.; Noble-Topham, S.; Nalainathan, D.; Provart, N. J.; Joyner, A. L.: Cloning and sequence comparison of the mouse, human, and chicken engrailed genes reveal potential functional domains and regulatory regions. *Dev. Genet.* 13: 345–358, 1992.
- [30584] 4136.Logan, C.; Willard, H. F.; Rommens, J. M.; Joyner, A. L.: Chromosomal localization of the human homeo box-containing genes, EN1 and EN2. *Genomics* 4:206–209, 1989.
- [30585] 4137.Loomis, C. A.; Harris, E.; Michaud, J.; Wurst, W.; Hanks, M.; Joyner, A. L.: The mouse engrailed–1 gene and ventral limb patterning. *Nature* 382:360–363, 1996.
- [30586] 4138.Martin, G. R.; Richman, M.; Reinsch, S.; Nadeau, J. H.; Joyner, A.: Mapping of the two mouse engrailed-like genes: close linkage of En–1 to dominant hemimelia (Dh) on chromosome 1 and of En–2 to hemimelic extra-toes

(Hx) on chromosome 5. Genomics 6: 302–308,1990.

[30587] 4139.Matsui, T.; Hirai, M.; Hirano, M.; Kurosawa, Y.: The HOX complex neighbored by the EVX gene, as well as two other homeobox-containing genes, the GBX-class and the EN class, are located on the same chromosomes 2 and 7 in humans. FEBS Lett. 336: 107–110, 1993.

[30588] 4140.Wurst, W.; Auerbach, A. B.; Joyner, A. L.: Multiple developmental defects in Engrailed–1 mutant mice: an early mid–hindbrain deletion and patterning defects in forelimbs and sternum. Development 120:2065–2075, 1994.

[30589] 4141.Kohl, S.; Baumann, B.; Rosenberg, T.; Kellner, U.; Lorenz, B.; Vadala, M.; Jacobson, S. G.; Wissinger, B.: Mutations in the cone photoreceptor G–protein alpha–subunit gene GNAT2 in patients with achromatopsia. Am. J. Hum. Genet. 71: 422–425, 2002.

[30590] 4142.Morris, T. A.; Fong, S.–L.: Characterization of the gene encoding human cone transducin alpha–subunit (GNAT2). Genomics 17: 442–448,1993.

[30591] 4143.Sparkes, R. S.; Cohn, V. H.; Cire–Eversole, P.; Blatt, C.; Amatruda, T. T.; Weiner, L. P.; Nesbitt, M.; Reed, R. R.; Lochrie, M. A.; Fournier, R. E. K.; Simon, M. I.: Mapping of genes encoding the subunits of guanine nucleotide–bind–

ing proteins (G-proteins) in the mouse. (Abstract) Cytogenet. Cell Genet. 46: 696 only, 1987.

- [30592] 4144. Chipev, C. C.; Korge, B. P.; Markova, N.; Bale, S. J.; DiGiovanna, J. J.; Compton, J. G.; Steinert, P. M.: A leucine-to-proline mutation in the H1 subdomain of keratin 1 causes epidermolytic hyperkeratosis. Cell 70:821–828, 1992.
- [30593] 4145. Compton, J. G.: Epidermal disease: faulty keratin filaments take their toll. Nature Genet. 6: 6–7, 1994.
- [30594] 4146. Compton, J. G.; DiGiovanna, J. J.; Santucci, S. K.; Kearns, K. S.; Amos, C. I.; Abangan, D. L.; Korge, B. P.; McBride, O. W.; Steinert, P. M.; Bale, S. J.: Linkage of epidermolytic hyperkeratosis to the type II keratin gene cluster on chromosome 12q. Nature Genet. 1:301–305, 1992.
- [30595] 4147. Fraser, R. D. B.; MacRae, T. P.; Suzuki, E.: Structure of the α -keratin microfibril. J. Molec. Biol. 108: 435–452, 1976.
- [30596] 4148. Kimonis, V.; DiGiovanna, J. J.; Yang, J.-M.; Doyle, S. Z.; Bale, S. J.; Compton, J. G.: A mutation in the V1 end domain of keratin 1 in non-epidermolytic palmar-plantar keratoderma. J. Invest. Derm. 103:764–769, 1994.

- [30597] 4149.Lee, L. D.; Ludwig, K.; Baden, H. P.: Matrix proteins of human hair as a tool for identification of individuals. *Forensic Sci.* 11:115–121, 1978.
- [30598] 4150.Lessin, S. R.; Huebner, K.; Isobe, M.; Croce, C. M.; Steinert, P. M.: Chromosomal mapping of human keratin genes: evidence of non-linkage. *J. Invest. Derm.* 91: 572–578, 1988.
- [30599] 4151.Nadeau, J. H.: Personal Communication. Bar Harbor, Me. 7/29/1987.
- [30600] 4152.Le Marchand–Brustel, Y.; Gremeaux, T.; Ballotti, R.; van Obberghen, E.: Insulin receptor tyrosine kinase is defective in skeletal muscle of insulin-resistant obese mice. *Nature* 315: 676–679, 1985.
- [30601] 4153.Leme, C. E.; Wajchenberg, B. L.; Lerario, A. C.; Goldman, J.; Borges, J. L. C.: Acanthosis nigricans, hirsutism, insulin resistance and insulin receptor defect. *Clin. Endocr.* 17: 43–49, 1982.
- [30602] 4154.Longo, N.; Langley, S. D.; Griffin, L. D.; Elsas, L. J.: Activation of glucose transport by a natural mutation in the human insulin receptor. *Proc. Nat. Acad. Sci.* 90: 60–64, 1993.
- [30603] 4155.Longo, N.; Langley, S. D.; Griffin, L. D.; Elsas, L. J., II: Reduced mRNA and a nonsense mutation in the insulin–

receptor gene produce heritable severe insulin resistance. Am. J. Hum. Genet. 50:998–1007, 1992.

[30604] 4156. Maassen, J. A.; Klinkhamer, M. P.; Odink, R. J. H.; Sips, H.; van der Zon, G. C. M.; Wieringa, T.; Krans, H. M. J.; Moller, W.: Improper expression of insulin receptors on fibroblasts from a leprechaun patient. Europ. J. Biochem. 172: 725–729, 1988.

[30605] 4157. Mariani, S.; Pedone, A.; Meschi, F.; Di Natale, B.; Caputo, R.; Broggi, U.; Chiumello, G.: Insulin resistance in a child with acanthosis nigricans type A. Acta Paediat. Scand. 71: 667–670, 1982.

[30606] 4158. Michael, M. D.; Kulkarni, R. N.; Postic, C.; Previs, S. F.; Shulman, G. I.; Magnuson, M. A.; Kahn, C. R.: Loss of insulin signaling in hepatocytes leads to severe insulin resistance and progressive hepatic dysfunction. Molec. Cell 6: 87–97, 2000.

[30607] 4159. Moller, D. E.; Cohen, O.; Yamaguchi, Y.; Assiz, R.; Grigorescu, F.; Eberle, A.; Morrow, L. A.; Moses, A. C.; Flier, J. S.: Prevalence of mutations in the insulin receptor gene in subjects with features of the type A syndrome of insulin resistance. Diabetes 43: 247–255, 1994.

[30608] 4160. Moller, D. E.; Flier, J. S.: Insulin resistance—mechanisms, syndromes, and implications. New Eng. J.

Med. 325: 938–948, 1991.

[30609] 4161.Moller, D. E.; Flier, J. S.: Detection of an alteration in the insulin–receptor gene in a patient with insulin resistance, acanthosis nigricans, and the polycystic ovary syndrome (type A insulin resistance). *New Eng. J. Med.* 319: 1526–1529, 1988.

[30610] 4162.Moller, D. E.; Yokota, A.; Ginsberg–Fellner, F.; Flier, J. S.: Functional properties of a naturally occurring trp(1200)–to–ser(1200) mutation of the insulin receptor. *Molec. Endocr.* 4: 1183–1191, 1990.

[30611] 4163.Moller, D. E.; Yokota, A.; Pazianos, A.; Flier, J. S.: A missense mutation in one allele of the tyrosine kinase domain of the insulin receptor gene is associated with dominantly inherited insulin resistance.(Abstract) *Clin. Res.* 38: 435A only, 1990.

[30612] 4164.Moller, D. E.; Yokota, A.; White, M. F.; Pazianos, A. G.; Flier, J. S.: A naturally occurring mutation of insulin receptor alanine1134 impairs tyrosine kinase function and is associated with dominantly inherited insulin resistance. *J. Biol. Chem.* 265: 14979–14985, 1990.

[30613] 4165.Moncada, V. Y.; Hedo, J. A.; Serrano–Rios, M.; Taylor, S. I.: Insulin–receptor biosynthesis in cultured lymphocytes from an insulin–resistant patient (Rabson–Mendenhall syn–

drome): evidence for defect before insertion of receptor into plasma membrane. *Diabetes* 35: 802–807, 1986.

[30614] 4166. Norton, K. I.; Glicklich, M.; Kupchik, G.; Gray, C. E.; Ludman, M.: Leprechaunism: a case report with radiographic features. *Dysmorph. Clin. Genet.* 4: 57–62, 1990.

[30615] 4167. Odawara, M.; Kadowaki, T.; Yamamoto, R.; Shibasaki, Y.; Tobe, K.; Accili, D.; Bevins, C.; Mikami, Y.; Matsuura, N.; Akanuma, Y.; Takaku, F.; Taylor, S. I.; Kasuga, M.: Human diabetes associated with a mutation in the tyrosine kinase domain of the insulin receptor. *Science* 245:66–68, 1989.

[30616] 4168. Ojamaa, K.; Hedo, J. A.; Roberts, C. T., Jr.; Moncada, V. Y.; Gorden, P.; Ullrich, A.; Taylor, S. I.: Defects in human insulin receptor gene expression. *Molec. Endocr.* 2: 242–247, 1988.

[30617] 4169. Prince, M. J.; Smith, F. E.; Peters, E. J.; Stuart, C. A.: Functional characteristics of decreased insulin receptors on fibroblasts obtained from a subject with severe insulin resistance and acanthosis nigricans. *Diabetes* 35:148–154, 1986.

[30618] 4170. Quin, J. D.; Fisher, B. M.; Paterson, K. R.; Inoue, A.; Beastall, G. H.; MacCuish, A. C.: Acute response to recombinant insulin-like growth factor-I in a patient with

Mendenhall's syndrome. New Eng.J. Med. 323:
1425–1426, 1991.

- [30619] 4171.Quon, M. J.; Guerre–Millo, M.; Zarnowski, M. J.;
Butte, A. J.;Em, M.; Cushman, S. W.; Taylor, S. I.: Tyrosine
kinase–deficientmutant human insulin receptors
(met1153–to–ile) overexpressed in transfectedrat adipose
cells fail to mediate translocation of epitope–
taggedGLUT4. Proc. Nat. Acad. Sci. 91: 5587–5591, 1994.
- [30620] 4172.Rabson, S. M.; Mendenhall, E. N.: Familial hypertro–
phy of pinealbody, hyperplasia of adrenal cortex and dia–
betes mellitus. Am. J.Clin. Path. 26: 283–290, 1956.
- [30621] 4173.Rajala, R. V. S.; Anderson, R. E.: Interaction of the
insulinreceptor beta–subunit with phosphatidylinositol
3–kinase in bovineROS. Invest. Ophthal. Vis. Sci. 42:
3110–3117, 2001.
- [30622] 4174.Roth, R. A.; Cassell, D. J.: Insulin receptor: evidence
thatit is a protein kinase. Science 219: 299–301, 1983.
- [30623] 4175.Rubin, C. S.: Personal Communication. Bronx, N. Y.
12/8/1984.
- [30624] 4176.Rudiger, H. W.; Ahrens, P.; Dreyer, M.; Frorath, B.;
Loffel, C.;Schmidt–Preuss, U.: Impaired insulin–induced
RNA synthesis secondaryto a genetically defective insulin
receptor. Hum. Genet. 69: 76–78,1985.

- [30625] 4177. Rudiger, H. W.; Dreyer, M.; Kuhnau, J.; Bartelheimer, H.: Familial insulin-resistant diabetes secondary to an affinity defect of the insulin receptor. *Hum. Genet.* 64: 407–411, 1983.
- [30626] 4178. Salmeen, A.; Andersen, J. N.; Myers, M. P.; Tonks, N. K.; Barford, D.: Molecular basis for the dephosphorylation of the activation segment of the insulin receptor by protein tyrosine phosphatase 1B. *Molec. Cell* 6: 1401–1412, 2000.
- [30627] 4179. Scarlett, J. A.; Kolterman, O. G.; Moore, P.; Saekow, M.; Insel, J.; Griffin, J.; Mako, M.; Rubenstein, A. H.; Olefsky, J. M.: Insulin resistance and diabetes due to a genetic defect in insulin receptors. *J. Clin. Endocr. Metab.* 55: 123–132, 1982.
- [30628] 4180. Seino, S.; Seino, M.; Nishi, S.; Bell, G. I.: Structure of the human insulin receptor gene and characterization of its promoter. *Proc. Nat. Acad. Sci.* 86: 114–118, 1989.
- [30629] 4181. Polymeropoulos, M. H.; Torres, R.; Yanovski, J. A.; Chandrasekharappa, S. C.; Ledbetter, D. H.: The human corticotropin-releasing factor receptor (CRHR) gene maps to chromosome 17q12–q22. *Genomics* 28: 123–124, 1995.
- [30630] 4182. Kusafuka, T.; Wang, Y.; Puri, P.: Novel mutations of the endothelin-B receptor gene in isolated patients with

Hirschsprung's disease. Hum.Molec. Genet. 5: 347–349, 1996.

[30631] 4183.Comings, D. E.: Evidence for ancient tetraploidy and conservation of linkage groups in mammalian chromosomes. Nature 238: 455–467, 1972.

[30632] 4184.Deol, M. S.: Genetical studies on the skeleton of the mouse. XXVIII.Tail-short. Proc. Roy. Soc. Ser. B. 155: 78–95, 1961.

[30633] 4185.Hart, C. P.; Awgulewitsch, A.; Fainsod, A.; McGinnis, W.; Ruddle, F. H.: Homeo box gene complex on mouse chromosome 11: molecular cloning, expression in embryogenesis, and homology to a human homeo box locus. Cell 43:9–18, 1985.

[30634] 4186.Hauser, C. A.; Joyner, A. L.; Klein, R. D.; Learned, T. K.; Martin, G. R.; Tjian, R.: Expression of homologous homeo-box-containing genes in differentiated human teratocarcinoma cells and mouse embryos. Cell 43:19–28, 1985.

[30635] 4187.Levine, M.; Rubin, G. M.; Tjian, R.: Human DNA sequences homologous to a protein coding region conserved between homeotic genes of Drosophila. Cell 38:667–673, 1984.

[30636] 4188.Manley, J. L.; Levine, M. S.: The homeo box and

mammalian development. Cell 43:1–2, 1985.

- [30637] 4189.Meijlink, F.; de Laaf, R.; Verrijzer, P.; Destree, O.; Kroezen,V.; Hilkens, J.; Deschamps, J.: A mouse homeobox containing geneon chromosome 11: sequence and tissue-specific expression. NucleicAcids Res. 15: 6773–6786, 1987.
- [30638] 4190.Miki, T.; Murphy, P. D.; Pletcher, B. A.; Kidd, J. R.; Ferguson-Smith,A. C.; Ruddle, F. H.; Kidd, K. K.: HOX2 maps to 17q near PPY andNGFR. (Abstract) Cytogenet. Cell Genet. 46: 662 only, 1987.
- [30639] 4191.Munke, M.; Cox, D. R.; Jackson, I. J.; Hogan, B. L. M.; Francke,U.: The murine Hox–2 cluster of homeo box containing genes maps distalon chromosome 11 near the tail–short (Ts) locus. Cytogenet. CellGenet. 42: 236–240, 1986.
- [30640] 4192.Ruddle, F. H.: Personal Communication. New Haven, Conn. 1987.
- [30641] 4193.Ruddle, F. H.: Personal Communication. New Haven, Conn. 1985.
- [30642] 4194.Schughart, K.; Utset, M. F.; Awgulewitsch, A.; Ruddle, F. H.:Structure and expression of Hox–2.2, a murine homeobox–containinggene. Proc. Nat. Acad. Sci. 85: 5582–5586, 1988.

- [30643] 4195. Trainor, P. A.; Ariza-McNaughton, L.; Krumlauf, R.: Role of the isthmus and FGFs in resolving the paradox of neural crest plasticity and pre patterning. *Science* 295: 1288–1291, 2002.
- [30644] 4196. Kaur, S.; Singh, G.; Stock, J. L.; Schreiner, C. M.; Kier, A. B.; Yager, K. L.; Mucenski, M. L.; Scott, W. J., Jr.; Potter, S. S.: Dominant mutation of the murine Hox-2.2 gene results in developmental abnormalities. *J. Exp. Zool.* 264: 323–336, 1992.
- [30645] 4197. Lill, M. C.; Fuller, J. F.; Herzig, R.; Crooks, G. M.; Gasson, J. C.: The role of the homeobox gene, HOX B7, in human myelomonocytic differentiation. *Blood* 85: 692–697, 1995.
- [30646] 4198. Simeone, A.; Mavilio, F.; Acampora, D.; Giampaolo, A.; Faiella, A.; Zappavigna, V.; D'Esposito, M.; Pannese, M.; Russo, G.; Boncinelli, E.; Peschle, C.: Two human homeobox genes, c1 and c8: structure analysis and expression in embryonic development. *Proc. Nat. Acad. Sci.* 84: 4914–4918, 1987.
- [30647] 4199. Yaron, Y.; McAdara, J. K.; Lynch, M.; Hughes, E.; Gasson, J. C.: Identification of novel functional regions important for the activity of HOXB7 in mammalian cells. *J. Immun.* 166: 5058–5067, 2001.

- [30648] 4200.Sakai, K.; Yamada, M.; Horiba, N.; Wakui, M.; Demura, H.; Suda,T.: The genomic organization of the human corticotropin-releasingfactor type-1 receptor. *Gene* 219: 125-130, 1998.
- [30649] 4201.Sillaber, I.; Rammes, G.; Zimmermann, S.; Mahal, B.; Zieglgansberger,W.; Wurst, W.; Holsboer, F.; Spanagel, R.: Enhanced and delayed stress-inducedalcohol drinking in mice lacking functional CRH1 receptors. *Science* 296:931-933, 2002.
- [30650] 4202.Smith, G. W.; Aubry, J.-M.; Dellu, F.; Contarino, A.; Bilezikjian,L. M.; Gold, L. H.; Chen, R.; Marchuk, Y.; Hauser, C.; Bentley, C.A.; Sawchenko, P. E.; Koob, G. F.; Vale, W.; Lee, K.-F.: Corticotropinreleasing factor receptor 1-deficient mice display decreased anxiety,impaired stress response, and aberrant neuroendocrine development. *Neuron* 20:1093-1102, 1998.
- [30651] 4203.Timpl, P.; Spanagel, R.; Sillaber, I.; Kresse, A.; Reul, J. M.H. M.; Stalla, G. K.; Blanquet, V.; Steckler, T.; Holsboer, F.; Wurst,W.: Impaired stress response and reduced anxiety in mice lackinga functional corticotropin-releasing hormone receptor 1. *Nature Genet.* 19:162-166, 1998.
- [30652] 4204.Azim, A. C.; Knoll, J. H. M.; Beggs, A. H.; Chisti, A. H.: Isoformcloning, actin binding, and chromosomal local-

ization of human erythroiddematin, a member of the villin superfamily. J. Biol. Chem. 270:17407–17413, 1995.

- [30653] 4205. Azim, A. C.; Marfatia, S. M.; Korsgren, C.; Dotimas, E.; Cohen, C. M.; Chishti, A. H.: Human erythrocyte dematin and protein 4.2 (pallidin) are ATP binding proteins. Biochemistry 35: 3001–3006, 1996.
- [30654] 4206. Chishti, A. H.; Faquin, W.; Wu, C.-C.; Branton, D.: Purification of erythrocyte dematin (protein 4.9) reveals an endogenous protein kinase that modulates actin-bundling activity. J. Biol. Chem. 264:8985–8991, 1989.
- [30655] 4207. Gilligan, D. M.; Bennett, V.: The junctional complex of the membrane skeleton. Seminars Hemat. 30: 74–83, 1993.
- [30656] 4208. Khanna, R.; Chang, S. H.; Andrabi, S.; Azam, M.; Kim, A.; Rivera, A.; Brugnara, C.; Low, P. S.; Liu, S.-C.; Chishti, A. H.: Headpiece domain of dematin is required for the stability of the erythrocyte membrane. Proc. Nat. Acad. Sci. 99: 6637–6642, 2002.
- [30657] 4209. Peters, L. L.; Eicher, E. M.; Azim, A. C.; Chishti, A. H.: The gene encoding the erythrocyte membrane skeleton protein dematin (Epb4.9) maps to mouse chromosome 14. Genomics 26: 634–635, 1995.
- [30658] 4210. Rana, A. P.; Ruff, P.; Maalouf, G. J.; Speicher, D. W.;

Chishti,A. H.: Cloning of human erythroid dematin reveals another memberof the villin family. Proc. Nat. Acad. Sci. 90: 6651–6655, 1993.

[30659] 4211.Correas, I.; Speicher, D. W.; Marchesi, V. T.: Structure of thespectrin–actin binding site of erythrocyte protein 4.1. J. Biol.Chem. 261: 13362–13366, 1986.

[30660] 4212.Albig, W.; Drabent, B.; Kunz, J.; Kalff–Suske, M.; Grzeschik, K.–H.;Doenecke, D.: All known human H1 histone genes except the H1(0) geneare clustered on chromosome 6. Genomics 16: 649–654, 1993.

[30661] 4213.Tanguay, R. M.; Berube, D.; Gagne, R.: Localization of histonegenes to chromosomes 6, 12, and 1 by in situ hybridization. (Abstract) Cytogenet.Cell Genet. 46: 702 only, 1987.

[30662] 4214.Cheng, J.; Baumhueter, S.; Cacalano, G.; Carver–Moore, K.; Thibodeaux,H.; Thomas, R.; Broxmeyer, H. E.; Cooper, S.; Hague, N.; Moore, M.;Lasky, L. A.: Hematopoietic defects in mice lacking the sialomucinCD34. Blood 87: 479–490, 1996.

[30663] 4215.He, X.–Y.; Antao, V. P.; Basila, D.; Marx, J. C.; Davis, B. R.: Isolation and molecular characterization of the human CD34 gene. Blood 79:2296–2302, 1992.

[30664] 4216.Howell, S. M.; Molgaard, H. V.; Greaves, M. F.; Spurr,

N. K.: Localisation of the gene coding for the haemopoietic stem cell antigen CD34 to chromosome 1q32. *Hum. Genet.* 87: 625–627, 1991.

[30665] 4217. Okuno, Y.; Iwasaki, H.; Huettner, C. S.; Radomska, H. S.; Gonzalez, D. A.; Tenen, D. G.; Akashi, K.: Differential regulation of the human and murine CD34 genes in hematopoietic stem cells. *Proc. Nat. Acad. Sci.* 99: 6246–6251, 2002.

[30666] 4218. Satterthwaite, A. B.; Burn, T. C.; Le Beau, M. M.; Tenen, D. G.: Structure of the gene encoding CD34, a human hematopoietic stem cell antigen. *Genomics* 12: 788–794, 1992.

[30667] 4219. Simmons, D. L.; Satterthwaite, A. B.; Tenen, D. G.; Seed, B.: Molecular cloning of a cDNA encoding CD34, a sialomucin of human hematopoietic stem cells. *J. Immun.* 148: 267–271, 1992.

[30668] 4220. Sutherland, D. R.; Stewart, A. K.; Keating, A.: CD34 antigen: molecular features and potential clinical applications. *Stem Cells* 11(suppl. 3): 50–57, 1993.

[30669] 4221. Sutherland, D. R.; Watt, S. M.; Dowden, G.; Karhi, K.; Baker, M. A.; Greaves, M. F.; Smart, J. E.: Structural and partial amino acid sequence analysis of the human hemopoietic progenitor cell antigen CD34. *Leukemia* 2:

793–803, 1988.

- [30670] 4222.Tenen, D. G.; Satterthwaite, A. B.; Borson, R.; Simmons, D.; Eddy,R. L.; Shows, T. B.: Chromosome 1 localization of the gene for CD34,a surface antigen of human stem cells. *Cytogenet. Cell Genet.* 53:55–57, 1990.
- [30671] 4223.Clegg, J. B.: Can the product of the theta gene be a real globin? *Nature* 329:465–466, 1987.
- [30672] 4224.Fei, Y. J.; Fujita, S.; Huisman, T. H. J.: Two different thetaglobin gene deletions observed among black newborn babies. *Brit.J. Haemat.* 68: 249–253, 1988.
- [30673] 4225.Hsu, S.–L.; Marks, J.; Shaw, J.–P.; Tam, M.; Higgs, D. R.; Shen,C. C.; Shen, C.–K. J.: Structure and expression of the human theta–1globin gene. *Nature* 331: 94–96, 1988.
- [30674] 4226.Leung, S.; Proudfoot, N. J.; Whitelaw, E.: The gene for theta–globinis transcribed in human fetal erythroid tissues. *Nature* 329: 551–554,1987.
- [30675] 4227.Marks, J.; Shaw, J.–P.; Perez–Stabile, C.; Hu, W.–S.; Ayres, T.M.; Shen, C.; Shen, C.–K. J.: The primate alpha–globin gene family:a paradigm of the fluid genome. *Cold Spring Harbor Symp. Quant. Biol.* 51:499–508, 1986.
- [30676] 4228.Marks, J.; Shaw, J.–P.; Shen, C.–K. J.: Sequence organizationand genomic complexity of primary theta–1 globin gene, a novel alpha–globin–likegene. *Nature* 321:

785–788, 1986.

- [30677] 4229. Peschle, C.; Mavilio, F.; Care, A.; Migliaccio, G.; Migliaccio, A. R.; Salvo, G.; Samoggia, P.; Petti, S.; Guerrieri, R.; Marinucci, M.; Lazzaro, D.; Russo, G.; Mastroberardino, G.: Haemoglobin switching in human embryos: asynchrony of zeta-to-alpha- and epsilon-to-gamma-globin switches in primitive and definitive erythropoietic lineage. *Nature* 313:235–238, 1985.
- [30678] 4230. Utsch, B.; Albers, N.; Dame, C.; Bartmann, P.; Lentze, M. J.; Ludwig, M.: Homozygous alpha-thalassemia associated with hypospadias: SEA-type deletion does not affect expression of the -14 gene and loss of the theta-1-globin gene on 16p13.3 is compensated by its duplicate theta-2 on chromosome 10. (Letter) *Am. J. Med. Genet.* 101: 286–287, 2001.
- [30679] 4231. Feo, S.; Davies, B.; Fried, M.: The mapping of seven intron-containing ribosomal protein genes shows they are unlinked in the human genome. *Genomics* 13:201–207, 1992.
- [30680] 4232. Barton, D. E.; Crimando, C.; Hortsch, M.; Francke, U.: The genes for ribophorins I and II are on human chromosomes 3q and 20 and mouse chromosomes 6 and 12, respectively. (Abstract) *Cytogenet. Cell Genet.* 46:577 only,

1987.

- [30681] 4233.Crimaudo, C.; Hortsch, M.; Gausepohl, H.; Meyer, D. I.: Humanribophorins I and II: the primary structure and membrane topologyof two highly conserved round endoplasmic reticulum-specific glycoproteins. *EMBOJ.* 6: 75–82, 1987.
- [30682] 4234.Elsasser, S.; Gali, R. R.; Schwickart, M.; Larsen, C. N.; Leggett,D. S.; Muller, B.; Feng, M. T.; Tubing, F.; Dittmar, G. A. G.; Finley,D.: Proteasome subunit Rpn1 binds ubiquitin-like protein domains. *NatureCell Biol.* 4: 725–730, 2002.
- [30683] 4235.Kelleher, D. J.; Kreibich, G.; Gilmore, R.: Oligosaccharyltransferaseactivity is associated with a protein complex composed of ribophorinsI and II and a 48 kd protein. *Cell* 69: 55–65, 1992.
- [30684] 4236.Horbach, J. M. L. M.; Brenninkmeyer, S. J.; van de Velde, C. J.H.; Nieuwenhuyzen Kruseman, A. C.: A forme fruste of von Hippel–Lindaudisease--a combination of adrenal pheochromocytoma and ipsilateralrenal cell carcinoma: a case report. *Surgery* 105: 436–441, 1989.
- [30685] 4237.Horton, W. A.; Wong, V.; Eldridge, R.: Von Hippel–Lindau disease--clinicaland pathological manifestations in 9 families with 50 affected members. *Arch.Intern. Med.*

136: 769–777, 1976.

- [30686] 4238.Hull, M. T.; Roth, L. M.; Glover, J. L.; Walker, P. D.: Metastaticcarotid body paraganglioma in von Hippel–Lindau disease: an electronmicroscopic study. Arch. Path. Lab. Med. 106: 235–239, 1982.
- [30687] 4239.Hosoe, S.; Brauch, H.; Latif, F.; Glenn, G.; Daniel, L.; Bale,S.; Choyke, P.; Gorin, M.; Oldfield, E.; Berman, A.; Goodman, J.;Orcutt, M. L.; Hampsch, K.; Delisio, J.; Modi, W.; McBride, W.; Anglard,P.; Weiss, G.; Walther, M. M.; Linehan, W. M.; Lerman, M. I.; Zbar,B.: Localization of the von Hippel–Lindau disease gene to a smallregion of chromosome 3. Genomics 8: 634–640, 1990.
- [30688] 4240.Huson, S. M.; Harper, P. S.; Hourihan, M. D.; Cole, G.; Weeks,R. D.; Compston, D. A. S.: Cerebellar haemangioblastoma and von Hippel–Lindaudisease. Brain 109: 1297–1310, 1986.
- [30689] 4241.Iliopoulos, O.; Kibel, A.; Gray, S.; Kaelin, W. G., Jr.: Tumoursuppression by the human von Hippel–Lindau gene product. Nature Med. 1:822–826, 1995.
- [30690] 4242.Iliopoulos, O.; Levy, A. P.; Jiang, C.; Kaelin, W. G., Jr.; Goldberg,M. A.: Negative regulation of hypoxia–inducible genes by the vonHippel–Lindau protein. Proc. Nat. Acad. Sci. 93: 10595–10599, 1996.

- [30691] 4243.Iliopoulos, O.; Ohh, M.; Kaelin, W. G., Jr.: pVHL(19) is a biologically active product of the von Hippel–Lindau gene arising from internal translation initiation. *Proc. Nat. Acad. Sci.* 95: 11661–11666, 1998.
- [30692] 4244.Ivan, M.; Kondo, K.; Yang, H.; Kim, W.; Valiando, J.; Ohh, M.; Salic, A.; Asara, J. M.; Lane, W. S.; Kaelin, W. G., Jr.: HIF– α targeted for VHL–mediated destruction by proline hydroxylation: implications for $O(2)$ sensing. *Science* 292: 464–468, 2001.
- [30693] 4245.Ivanov, S. V.; Kuzmin, I.; Wei, M.–H.; Pack, S.; Geil, L.; Johnson, B. E.; Stanbridge, E. J.; Lerman, M. I.: Down–regulation of transmembrane carbonic anhydrases in renal cell carcinoma cell lines by wild–type von Hippel–Lindau transgenes. *Proc. Nat. Acad. Sci.* 95: 12596–12601, 1998.
- [30694] 4246.Jaakkola, P.; Mole, D. R.; Tian, Y.–M.; Wilson, M. I.; Gielbert, J.; Gaskell, S. J.; von Kriegsheim, A.; Hebestreit, H. F.; Mukherji, M.; Schofield, C. J.; Maxwell, P. H.; Pugh, C. W.; Ratcliffe, P. J.: Targeting of HIF– α to the von Hippel–Lindau ubiquitylation complex by $O(2)$ –regulated prolyl hydroxylation. *Science* 292: 468–472, 2001.
- [30695] 4247.James, G. P.: Personal Communication. Springfield, Ohio 8/15/1998.
- [30696] 4248.Jennings, A. M.; Smith, C.; Cole, D. R.; Jennings, C.;

Shortland, J. R.; Williams, J. L.; Brown, C. B.: Von Hippel–Lindau disease in a large British family: clinicopathological features and recommendations for screening and follow-up. *Quart. J. Med.* 66: 233–249, 1988.

[30697] 4249. Kanno, H.; Kondo, K.; Ito, S.; Yamamoto, I.; Fujii, S.; Torigoe, S.; Sakai, N.; Hosaka, M.; Shuin, T.; Yao, M.: Somatic mutations of the von Hippel–Lindau tumor suppressor gene in sporadic central nervous system hemangioblastomas. *Cancer Res.* 54: 4845–4847, 1994.

[30698] 4250. Kanno, H.; Saljooque, F.; Yamamoto, I.; Hattori, S.; Yao, M.; Shuin, T.; U, H.–S.: Role of the von Hippel–Lindau tumor suppressor protein during neuronal differentiation. *Cancer Res.* 60: 2820–2824, 2000.

[30699] 4251. Kaplan, C.; Sayre, G. P.; Greene, L. F.: Bilateral nephrogenic carcinomas in Lindau–von Hippel disease. *J. Urol.* 86: 36–42, 1961.

[30700] 4252. Karsdorp, N.; Elderson, A.; Wittebol–Post, D.; Hene, R. J.; Vos, J.; Feldberg, M. A. M.; van Gils, A. P. G.; Jansen–Schillhorn van Veen, J. M.; Vroom, T. M.; Hoppener, J. W. M.; Lips, C. J. M.: Von Hippel–Lindau disease: new strategies in early detection and treatment. *Am. J. Med.* 97: 158–168, 1994.

[30701] 4253. Keeler, L. L., III; Klauber, G. T.: von Hippel–Lindau

disease and renal cell carcinoma in a 16-year-old boy. J. Urol. 147: 1588–1591, 1992.

[30702] 4254. Kenck, C.; Wilhelm, M.; Bugert, P.; Staehler, G.; Kovacs, G.: Mutation of the VHL gene is associated exclusively with the development of non-papillary renal cell carcinomas. J. Path. 179: 157–161, 1996.

[30703] 4255. Kerr, D. J.; Scheithauer, B. W.; Miller, G. M.; Ebersold, M. J.; McPhee, T. J.: Hemangioblastoma of the optic nerve: case report. Neurosurgery 36: 573–581, 1995.

[30704] 4256. Kibel, A.; Iliopoulos, O.; DeCaprio, J. A.; Kaelin, W. G., Jr.: Binding of the von Hippel–Lindau tumor suppressor protein to elongin B and C. Science 269: 1444–1446, 1995.

[30705] 4257. Kiechle–Schwarz, M.; Neumann, H. P. H.; Decker, H.–J. H.; Dietrich, C.; Wullich, B.; Schempp, W.: Cytogenetic studies on three pheochromocytomas derived from patients with von Hippel–Lindau syndrome. Hum. Genet. 82: 127–130, 1989.

[30706] 4258. Larsen, N.; Samuelsson, T.; Swieb, C.: The Signal Recognition Particle Database (SRPDB). Nucleic Acids Res. 26: 177–178, 1998.

[30707] 4259. Pool, M. R.; Stumm, J.; Fulga, T. A.; Sinning, I.; Dobberstein, B.: Distinct modes of signal recognition particle

interaction with the ribosome. *Science* 297: 1345–1348, 2002.

[30708] 4260. Yaffe, M. B.; Farr, G. W.; Miklos, D.; Horwich, A. L.; Sternlicht, M. L.; Sternlicht, H.: TCP1 complex is a molecular chaperone in tubulin biogenesis. *Nature* 358: 245–248, 1992.

[30709] 4261. Brown, M. H.; Gorman, P. A.; Sewell, W. A.; Spurr, N. K.; Sheer, D.; Crumpton, M. J.: The gene coding for the human T-lymphocyte CD2 antigen is located on chromosome 1p. *Hum. Genet.* 76: 191–195, 1987.

[30710] 4262. Clayton, L. K.; Ramachandran, H.; Pravtcheva, D.; Chen, Y.-F.; Diamond, D. J.; Ruddle, F. H.; Reinherz, E. L.: The gene for T11 (CD2) maps to chromosome 1 in humans and to chromosome 3 in mice. *J. Immunol.* 140: 3617–3621, 1988.

[30711] 4263. Diamond, D. J.; Clayton, L. K.; Sayre, P. H.; Reinherz, E. L.: Exon-intron organization and sequence comparison of human and murine T11 (CD2) genes. *Proc. Nat. Acad. Sci.* 85: 1615–1619, 1988.

[30712] 4264. Festenstein, R.; Tolaini, M.; Corbella, P.; Mamalaki, C.; Parrington, J.; Fox, M.; Miliou, A.; Jones, M.; Kioussis, D.: Locus control region function and heterochromatin-induced position effect variegation. *Science*

271:1123–1125, 1996.

- [30713] 4265.Kingsmore, S. F.; Watson, M. L.; Moseley, W. S.; Seldin, M. F.: Physical linkage of genes encoding the lymphocyte adhesion molecules CD2 and its ligand LFA-3. *Immunogenetics* 30: 123–125, 1989.
- [30714] 4266.Lang, G.; Wotton, D.; Owen, M. J.; Sewell, W. A.; Brown, M. H.; Mason, D. Y.; Crumpton, M. J.; Kioussis, D.: The structure of the human CD2 gene and its expression in transgenic mice. *EMBO J.* 7:1675–1682, 1988.
- [30715] 4267.Richardson, N. E.; Chang, H.-C.; Brown, N. R.; Hussey, R. E.; Sayre, P. H.; Reinherz, E. L.: Adhesion domain of human T11 (CD2) is encoded by a single exon. *Proc. Nat. Acad. Sci.* 85: 5176–5180, 1988.
- [30716] 4268.Seed, B.; Aruffo, A.: Molecular cloning of the CD2 antigen, the T-cell erythrocyte receptor, by a rapid immunoselection procedure. *Proc. Nat. Acad. Sci.* 84: 3365–3369, 1987.
- [30717] 4269.Sewell, W. A.; Brown, M. H.; Dunne, J.; Owen, M. J.; Crumpton, M. J.: Molecular cloning of the human T-lymphocyte surface CD2 (T11) antigen. *Proc. Nat. Acad. Sci.* 83: 8718–8722, 1986.
- [30718] 4270.Wang, J.; Smolyar, A.; Tan, K.; Liu, J.; Kim, M.; Sun, Z. J.; Wagner, G.; Reinherz, E. L.: Structure of a heterophilic

adhesion complex between the human CD2 and CD58 (LFA-3) counterreceptors. *Cell* 97:791–803, 1999.

[30719] 4271. Brissenden, J. E.; Derynck, R.; Francke, U.: Transforming growth factor alpha gene (TGFA) maps to human chromosome 2 close to the breakpoint of the t(2;8) variant translocation in Burkitt lymphoma. (Abstract) *Cytogenet. Cell Genet.* 40: 589 only, 1985.

[30720] 4272. Collin, G. B.; Marshall, J. D.; Naggert, J. K.; Nishina, P. M.: TGFA: exon-intron structure and evaluation as a candidate gene for Alstrom syndrome. (Letter) *Clin. Genet.* 55: 61–62, 1999.

[30721] 4273. Ellis, D. L.; Kafka, S. P.; Chow, J. C.; Nanney, L. B.; Inman, W. H.; McCadden, M. E.; King, L. E., Jr.: Melanoma, growth factors, acanthosis nigricans, the sign of Leser-Trelat, and multiple acrochordons: a possible role for alpha-transforming growth factor in cutaneous paraneoplastic syndromes. *New Eng. J. Med.* 317: 1582–1587, 1987.

[30722] 4274. Fernandez-Larrea, J.; Merlos-Suarez, A.; Urena, J. M.; Baselga, J.; Arribas, J.: A role for a PDZ protein in the early secretory pathway for the targeting of proTGF-alpha to the cell surface. *Molec. Cell* 3: 423–433, 1999.

[30723] 4275. Fowler, K. J.; Mann, G. B.; Dunn, A. R.: Linkage of the

murinetransforming growth factor- α gene with Igk, Ly-2, and Fabp1 on chromosome 6. Genomics 16: 782–784, 1993.

- [30724] 4276. Tam, J. P.; Scheikh, M. A.; Solomon, D. S.; Ossowski, L.: Efficient synthesis of human type α transforming growth factor: its physical and biological characterization. Proc. Nat. Acad. Sci. 83: 8082–8086, 1986.
- [30725] 4277. Tricoli, J. V.; Nakai, H.; Byers, M. G.; Rall, L. B.; Bell, G. I.; Shows, T. B.: Assignment of the gene coding for human TGF- α to chromosome 2p13. (Abstract) Cytogenet. Cell Genet. 40: 762 only, 1985.
- [30726] 4278. Tricoli, J. V.; Nakai, H.; Byers, M. G.; Rall, L. B.; Bell, G. I.; Shows, T. B.: The gene for human transforming growth factor α is on the short arm of chromosome 2. Cytogenet. Cell Genet. 42:94–98, 1986.
- [30727] 4279. Dong, C.; Zhu, S.; Wang, T.; Yoon, W.; Li, Z.; Alvarez, R. J.; ten Dijke, P.; White, B.; Wigley, F. M.; Goldschmidt-Clermont, P. J.: Deficient Smad7 expression: a putative molecular defect in scleroderma. Proc. Nat. Acad. Sci. 99: 3908–3913, 2002.
- [30728] 4280. Dickinson, M. E.; Kobrin, M. S.; Silan, C. M.; Kingsley, D. M.; Justice, M. J.; Miller, D. A.; Ceci, J. D.; Lock, L. F.; Lee, A.; Buchberg, A. M.; Siracusa, L. D.; Lyons, K. M.;

Derynck, R.; Hogan, B. L. M.; Copeland, N. G.; Jenkins, N. A.: Chromosomal localization of seven members of the murine TGF- β superfamily suggests close linkage to several morphogenetic mutant loci. *Genomics* 6: 505–520, 1990.

[30729] 4281. Heldin, C.-H.; Miyazono, K.; ten Dijke, P.: TGF- β signalling from cell membrane to nucleus through SMAD proteins. *Nature* 390:465–471, 1997.

[30730] 4282. Thomas, S. A.; Matsumoto, A. M.; Palmiter, R. D.: Noradrenaline is essential for mouse fetal development. *Nature* 374: 643–646, 1995.

[30731] 4283. Drebin, J. A.; Hartzell, S. W.; Griffin, C.; Campbell, M. J.; Niederhuber, J. E.: Molecular cloning and chromosomal localization of the human homologue of a B-lymphocyte specific protein tyrosine kinase (blk). *Oncogene* 10:477–486, 1995.

[30732] 4284. Dymecki, S.; Niederhuber, J.; Desiderio, S.: Specific expression of a novel tyrosine kinase gene, Blk, in B lymphoid cells. *Science* 247:332–336, 1990.

[30733] 4285. Islam, K. B.; Rabbani, H.; Larsson, C.; Sanders, R.; Smith, C. I. E.: Molecular cloning, characterization, and chromosomal localization of a human lymphoid tyrosine kinase related to murine Blk. *J. Immun.* 154:1265–1272,

1995.

- [30734] 4286.Kozak, C. A.; Dymecki, S. M.; Niederhuber, J. E.; Desiderio, S.V.: Genetic mapping of the gene for a novel tyrosine kinase, Blk,to mouse chromosome 14. *Genomics* 9: 762–764, 1991.
- [30735] 4287.Kikuchi, S.; Hata, M.; Fukumoto, K.; Yamane, Y.; Matsui, T.; Tamura,A.; Yonemura, S.; Yamagishi, H.; Keppler, D.; Tsukita, S.; Tsukita,S.: Radixin deficiency causes conjugated hyperbilirubinemia withloss of Mrp2 from bile canalicular membranes. *Nature Genet.* 31:320–325, 2002.
- [30736] 4288.Wilgenbus, K. K.; Milatovich, A.; Francke, U.; Furthmayr, H.:Molecular cloning, cDNA sequence, and chromosomal assignment of thehuman radixin gene and two dispersed pseudogenes. *Genomics* 16: 199–206,1993.
- [30737] 4289.Geppert, M.; Goda, Y.; Stevens, C. F.; Sudhof, T. C.: The smallGTP-binding protein Rab3A regulates a late step in synaptic vesiclefusion. *Nature* 387: 810–814, 1997.
- [30738] 4290.Kapfhamer, D.; Valladares, O.; Sun, Y.; Nolan, P. M.; Rux, J. J.;Arnold, S. E.; Veasey, S. C.; Bucan, M.: Mutations in Rab3a altercircadian period and homeostatic response to sleep loss in the mouse. *NatureGenet.* 32: 290–295, 2002.
- [30739] 4291.Rousseau-Merck, M. F.; Zahraoui, A.; Bernheim, A.;

Touchot, N.;Miglierina, R.; Tavitian, A.; Berger, R.: Chromosome mapping of thehuman RAS related RAB3A and RAB3B genes to chromosomes 19p13.2 and1p31–p32, respectively. (Abstract) Cytogenet. Cell Genet. 51: 1070only, 1989.

[30740] 4292.Rousseau–Merck, M. F.; Zahraoui, A.; Bernheim, A.; Touchot, N.;Miglierina, R.; Tavitian, A.; Berger, R.: Chromosome mapping of thehuman RAS–related RAB3A gene to 19p13.2. Genomics 5: 694–698, 1989.

[30741] 4293.Zahraoui, A.; Touchot, N.; Chardin, P.; Tavitian, A.: The humanrab genes encode a family of GTP–binding proteins related to yeastYPT1 and SEC4 products involved in secretion. J. Biol. Chem. 264:12394–12401, 1989.

[30742] 4294.Durkin, A. S.; Maglott, D. R.; Nierman, W. C.: Chromosomal assignmentof 38 human brain expressed sequence tags (ESTs) by analyzing fluorescentlylabeled PCR products from hybrid cell panels. Genomics 14: 808–810,1992.

[30743] 4295.Hussey, D. J.; Nicola, M.; Moore, S.; Peters, G. B.; Dobrovic,A.: The (4;11)(q21;p15) translocation fuses the NUP98 and RAP1GDS1genes and is recurrent in T–cell acute lymphocytic leukemia. Blood 94:2072–2079, 1999.

[30744] 4296.Germain, P.; Iyer, J.; Zechel, C.; Gronemeyer, H.: Co–

regulator recruitment and the mechanism of retinoic acid receptor synergy. *Nature* 415:187–192, 2002.

[30745] 4297. McNamara, P.; Seo, S.; Rudic, R. D.; Sehgal, A.; Chakravarti, D.; FitzGerald, G. A.: Regulation of CLOCK and MOP4 by nuclear hormone receptors in the vasculature: a humoral mechanism to reset a peripheral clock. *Cell* 105: 877–889, 2001.

[30746] 4298. Almasan, A.; Mangelsdorf, D. J.; Ong, E. S.; Wahl, G. M.; Evans, R. M.: Chromosomal localization of the human retinoid X receptors. *Genomics* 20:397–403, 1994.

[30747] 4299. Claudel, T.; Leibowitz, M. D.; Fievet, C.; Tailleux, A.; Wagner, B.; Repa, J. J.; Torpier, G.; Lobaccaro, J.-M.; Pater-niti, J. R.; Mangelsdorf, D. J.; Heyman, R. A.; Auwerx, J.: Re-duction of atherosclerosis in apolipoprotein E knockout mice by activation of the retinoid X receptor. *Proc. Nat. Acad. Sci.* 98: 2610–2615, 2001.

[30748] 4300. de Urquiza, A. M.; Liu, S.; Sjoberg, M.; Zetterstrom, R. H.; Griffiths, W.; Sjoball, J.; Perlmann, T.: Docosa-hex-aenoic acid, a ligand for the retinoid X receptor in mouse brain. *Science* 290: 2140–2144, 2000.

[30749] 4301. Thomas, G. D.; Sander, M.; Lau, K. S.; Huang, P. L.; Stull, J. T.; Victor, R. G.: Impaired metabolic modulation of alpha-adrenergic vasoconstriction in dystrophin-deficient

skeletal muscle. Proc. Nat. Acad. Sci. 95: 15090–15095, 1998.

[30750] 4302.Xie, J.; Roddy, P.; Rife, T. K.; Murad, F.; Young, A. P.: Two closely linked but separable promoters for human neuronal nitric oxide synthase gene transcription. Proc. Nat. Acad. Sci. 92: 1242–1246, 1995.

[30751] 4303.Xu, W.; Gorman, P.; Sheer, D.; Bates, G.; Kishimoto, J.; Lizhi, L.; Emson, P.: Regional localization of the gene coding for human brain nitric oxide synthase (NOS1) to 12q24.2–24.31 by fluorescent in situ hybridization. Cytogenet. Cell Genet. 64: 62–63, 1993.

[30752] 4304.Taniyama, T.; Takai, S.; Miyazaki, E.; Fukumura, R.; Sato, J.; Kobayashi, Y.; Hirakawa, T.; Moore, K. W.; Yamada, K.: The human interleukin–10 receptor gene maps to chromosome 11q23.3. Hum. Genet. 95:99–101, 1995.

[30753] 4305.Bono, M. R.; Alcaide–Loridan, C.; Couillin, P.; Letouze, B.; Grisard, M. C.; Jouin, H.; Fellous, M.: Human chromosome 16 encodes a factor involved in induction of class II major histocompatibility antigens by interferon gamma. Proc. Nat. Acad. Sci. 88: 6077–6081, 1991.

[30754] 4306.Bono, R.; Hatat, D.; Couillin, P.; Grisard, M. C.; Van Cong, N.; Fisher, D.; Fellous, M.: Receptor for human gamma interferon is specified by human chromosomes 6

and 21. (Abstract) Cytogenet. Cell Genet. 46:584, 1987.

[30755] 4307.Dorman, S. E.; Holland, S. M.: Mutation in the signal-transducing chain of the interferon-gamma receptor and susceptibility to mycobacterial infection. J. Clin. Invest. 101: 2364–2369, 1998.

[30756] 4308.Jung, V.; Rashidbaigi, A.; Jones, C.; Tischfield, J. A.; Shows, T. B.; Pestka, S.: Human chromosomes 6 and 21 are required for sensitivity to human interferon gamma. Proc. Nat. Acad. Sci. 84: 4151–4155, 1987.

[30757] 4309.Langer, J. A.; Rashidbaigi, A.; Lai, L.-W.; Patterson, D.; Jones, C.: Sublocalization on chromosome 21 of human interferon-alpha receptor gene and the gene for an interferon-gamma response protein. Somat. Cell Molec. Genet. 16: 231–240, 1990.

[30758] 4310.Mariano, T. M.; Muthukumaran, G.; Donnelly, R. J.; Wang, N.; Adamson, M. C.; Pestka, S.; Kozak, C. A.: Genetic mapping of the gene for the mouse interferon-gamma receptor signaling subunit to the distal end of chromosome 16. Mammalian Genome 7: 321–322, 1996.

[30759] 4311.Rhee, S.; Ebensperger, C.; Dembic, Z.; Pestka, S.: The structure of the gene for the second chain of the human interferon-gamma receptor. J. Biol. Chem. 271: 28947–28952, 1996.

- [30760] 4312.Soh, J.; Donnelly, R. J.; Mariano, T. M.; Cook, J. R.; Schwartz,B.; Pestka, S.: Identification of a yeast artificial chromosome clone encoding an accessory factor for the human interferon gamma receptor:evidence for multiple accessory factors. *Proc. Nat. Acad. Sci.* 90:8737–8741, 1993.
- [30761] 4313.Badovinac, V. P.; Tvinnereim, A. R.; Harty, J. T.: Regulation of antigen-specific CD8(+) T cell homeostasis by perforin and interferon-gamma. *Science* 290:1354–1357, 2000.
- [30762] 4314.Ben-Asouli, Y.; Banai, Y.; Pel-Or, Y.; Shir, A.; Kaempfer, R.: Human interferon-gamma mRNA autoregulates its translation through a pseudoknot that activates the interferon-inducible protein kinase PKR. *Cell* 108: 221–232, 2002.
- [30763] 4315.Binder, G. K.; Griffin, D. E.: Interferon-gamma-mediated site-specific clearance of alphavirus from CNS neurons. *Science* 293: 303–306, 2001.
- [30764] 4316.Blalock, J. E.; Smith, E. M.: Human leukocyte interferon: structural and biological relatedness to adrenocorticotrophic hormone and endorphins. *Proc. Nat. Acad. Sci.* 77: 5972–5974, 1980.
- [30765] 4317.Bureau, J. F.; Bihl, F.; Brahic, M.; Le Paslier, D.: The

genecoding for interferon-gamma is linked to the D12S335 and D12S313 microsatellites and to the MDM2 gene. *Genomics* 28: 109–112, 1995.

- [30766] 4318. Burke, D. C.: The status of interferon. *Sci. Am.* 236(4): 42–50, 1977.
- [30767] 4319. Cassingena, R.; Chany, C.; Vignal, M.; Suarez, H.; Estrade, S.; Lazar, P.: Use of monkey-mouse hybrid cells for the study of the cellular regulation of interferon production and action. *Proc. Nat. Acad. Sci.* 68: 580–584, 1971.
- [30768] 4320. Creagan, R. P.; Tan, Y. H.; Chen, S.-H.; Ruddle, F. H.: Somatic cell genetic analysis of the interferon system. *Fed. Proc.* 34: 2222–2226, 1975.
- [30769] 4321. Devos, R.; Cheroutre, H.; Taya, Y.; Degraeve, W.; Van Heuverswyn, H.; Fiers, W.: Molecular cloning of human immune interferon cDNA and its expression in eukaryotic cells. *Nucleic Acids Res.* 10: 2487–2502, 1982.
- [30770] 4322. Diaz, M. O.; Bohlander, S.; Allen, G.: Nomenclature of the human interferon genes. *J. Interferon Res.* 13: 443–444, 1993.
- [30771] 4323. Diefenbach, A.; Schindler, H.; Rollinghoff, M.; Yokoyama, W. M.; Bogdan, C.: Requirement for type 2 NO synthase for IL-12 signaling in innate immunity. *Science*

284: 951–955, 1999.

- [30772] 4324.Gray, P. W.; Goeddel, D. V.: Structure of the human immune interferongene. *Nature* 298: 859–863, 1982.
- [30773] 4325.Isaacs, A.; Lindenmann, J.: Virus interference. I. The interferon. *Proc.Roy. Soc. London* 147B: 258–267, 1957.
- [30774] 4326.Isaacs, A.; Lindenmann, J.; Valentine, R. C.: Virus interference.II. Some properties of interferon. *Proc. Roy. Soc. London* 147B:268–273, 1957.
- [30775] 4327.Knight, E., Jr.: Human fibroblast interferon: amino acid analysisand amino terminal amino acid sequence. *Science* 207: 525–526, 1980.
- [30776] 4328.Lipinski, M.; Virelizier, J. L.; Tursz, T.; Griscelli, C.: Naturalkiller and killer cell activities in patients with primary immunodeficienciesor defects in immune interferon production. *Europ. J. Immun.* 10:246–249, 1980.
- [30777] 4329.Maeda, S.; McCandliss, R.; Gross, M.; Sloma, A.; Familletti, P.C.; Tabor, J. M.; Evinger, M.; Levy, W. P.; Pestka, S.: Constructionand identification of bacterial plasmids containing nucleotide sequencefor human leukocyte interferon. *Proc. Nat. Acad. Sci.* 77: 7010–7013,1980.
- [30778] 4330.Mantei, N.; Schwarzstein, M.; Streuli, M.; Panem, S.; Nagata,S.; Weissmann, C.: The nucleotide sequence of a

cloned human leukocyteinterferon cDNA. Gene 10: 1–10, 1980.

[30779] 4331.Nagata, S.; Mantei, N.; Weissmann, C.: The structure of one ofthe eight or more distinct chromosomal genes for human interferon–alpha. Nature 287:401–408, 1980.

[30780] 4332.Nagata, S.; Taira, H.; Hall, A.; Johnstrud, L.; Streuli, M.; Escodi,J.; Boll, W.; Cantell, K.; Weissmann, C.: Synthesis in E. coli ofa polypeptide with human leukocyte interferon activity. Nature 284:316–320, 1980.

[30781] 4333.Nathan, C. F.; Murray, H. W.; Wiebe, M. E.; Rubin, B. Y.: Identificationof interferon–gamma as the lymphokine that activates human macrophageoxidative metabolism and antimicrobial activity. J. Exp. Med. 158:670–689, 1983.

[30782] 4334.Naylor, S. L.; Sakaguchi, A. Y.; Shows, T. B.; Law, M. L.; Goeddel,D. V.; Gray, P. W.: Human immune interferon gene is located on chromosome12. J. Exp. Med. 57: 1020–1027, 1983.

[30783] 4335.Shimizu, A.; Sakai, Y.; Ohno, K.; Masaki, S.; Kuwano, R.; Takahashi,Y.; Miyashita, N.; Watanabe, T.: A molecular genetic linkage mapof mouse chromosome 10, including the Myb, S100b, Pah, Sl, and Ifggenes. Biochem. Genet. 30: 529–535, 1992.

- [30784] 4336. Blumenberg, M.; Savtchenko, E. S.: Linkage of human keratin genes. *Cytogenet. Cell Genet.* 42: 65–71, 1986.
- [30785] 4337. Fuchs, E.; Coppock, S.; Green, H.; Cleveland, D.: Two distinct classes of keratin genes and their evolutionary significance. *Cell* 27:75–84, 1981.
- [30786] 4338. Fuchs, E.; Green, H.: Changes in keratin gene expression during terminal differentiation of the keratinocyte. *Cell* 19: 1033–1042, 1980.
- [30787] 4339. Hanukoglu, I.; Fuchs, E.: The cDNA sequence of a human epidermal keratin: divergence of sequence but conservation of structure among intermediate filament proteins. *Cell* 31: 243–252, 1982.
- [30788] 4340. Lee, L. D.; Baden, H. P.: Organisation of the polypeptide chains in mammalian keratin. *Nature* 264: 377–379, 1976.
- [30789] 4341. Marchuk, D.; McCrohon, S.; Fuchs, E.: Complete sequence of a gene encoding a human type I keratin: sequences homologous to enhancer elements in the regulatory region of the gene. *Proc. Nat. Acad. Sci.* 82:1609–1613, 1985.
- [30790] 4342. Nadeau, J. H.; Berger, F. G.; Cox, D. R.; Crosby, J. L.; Davisson, M. T.; Ferrara, D.; Fuchs, E.; Hart, C.; Hunihan,

L.; Lalley, P. A.; Langley, S. H.; Martin, G. R.; Nichols, L.; Phillips, S. J.; Roderick, T. H.; Roop, D. R.; Ruddle, F. H.; Skow, L. C.; Compton, J. G.: A family of type I keratin genes and the homeobox-2 gene complex are closely linked to the rex locus on mouse chromosome 11. *Genomics* 5:454-462, 1989.

[30791] 4343. Ray Chaudhury, A.; Marchuk, D.; Lindhurst, M.; Fuchs, E.: Three tightly linked genes encoding human type I keratins: conservation of sequence in the 5-prime-untranslated leader and 5-prime-upstream regions of coexpressed keratin genes. *Molec. Cell. Biol.* 6: 539-548, 1986.

[30792] 4344. Romano, V.; Bosco, P.; Raimondi, E.; Feo, S.; Leube, R.; Franke, W.; Ceratto, N.: Chromosomal mapping and physical linkage analysis of human acidic cytokeratin genes. (Abstract) *Cytogenet. Cell Genet.* 58:2009-2010, 1991.

[30793] 4345. Heim, S.; Nilbert, M.; Vanni, R.; Floderus, U.-M.; Mandahl, N.; Liedgren, S.; Lecca, U.; Mitelman, F.: A specific translocation, t(12;14)(q14-15;q23-24), characterizes a subgroup of uterine leiomyomas. *Cancer Genet. Cytogenet.* 32: 13-17, 1988.

[30794] 4346. Nucifora, G.; Begy, C. R.; Kobayashi, H.; Roulston,

D.; Claxton,D.; Pedersen-Bjergaard, J.; Parganas, E.; Ihle, J. N.; Rowley, J.D.: Consistent intergenic splicing and production of multiple transcripts between AML1 at 21q22 and unrelated genes at 3q26 in (3;21)(q26;q22)translocations. Proc. Nat. Acad. Sci. 91: 4004–4008, 1994.

[30795] 4347.Nucifora, G.; Rowley, J. D.: AML1 and the 8;21 and 3;21 translocations in acute and chronic myeloid leukemia. Blood 86: 1–14, 1995.

[30796] 4348.Tan, M.; Jing, T.; Lan, K.–H.; Neal, C. L.; Li, P.; Lee, S.; Fang,D.; Nagata, Y.; Liu, J.; Arlinghaus, R.; Hung, M.–C.; Yu, D.: Phosphorylation on tyrosine–15 of p34(Cdc2) by ErbB2 inhibits p34(Cdc2) activation and is involved in resistance to taxol–induced apoptosis. Molec.Cell 9: 993–1004, 2002.

[30797] 4349.Gromeier, M.; Solecki, D.; Patel, D. D.; Wimmer, E.: Expression of the human poliovirus receptor/CD155 gene during development of the central nervous system: implications for the pathogenesis of poliomyelitis. Virology 273:248–257, 2000.

[30798] 4350.He, Y.; Bowman, V. D.; Mueller, S.; Bator, C. M.; Bella, J.; Peng,X.; Baker, T. S.; Wimmer, E.; Kuhn, R. J.; Rossmann, M. G.: Interaction of the poliovirus receptor

with poliovirus. Proc. Nat. Acad. Sci. 97:79–84, 2000.

[30799] 4351.Koike, S.; Horie, H.; Ise, I.; Okitsu, A.; Yoshida, M.; Iizuka,N.; Takeuchi, K.; Takegami, T.; Nomoto, A.: The poliovirus receptorprotein is produced both as membrane-bound and secreted forms. EMBOJ. 9: 3217–3224, 1990.

[30800] 4352.Koike, S.; Taya, C.; Kurata, T.; Abe, S.; Ise, I.; Yonekawa, H.;Nomoto, A.: Transgenic mice susceptible to poliovirus. Proc. Nat.Acad. Sci. 88: 951–955, 1991.

[30801] 4353.Mendelsohn, C.; Johnson, B.; Lionetti, K. A.; Nobis, P.; Wimmer,E.; Racaniello, V. R.: Transformation of a human poliovirus receptorgene into mouse cells. Proc. Nat. Acad. Sci. 83: 7845–7849, 1986.

[30802] 4354.Mendelsohn, C. L.; Wimmer, E.; Racaniello, V. R.: Cellular receptorfor poliovirus: molecular cloning, nucleotide sequence, and expressionof a new member of the immunoglobulin superfamily. Cell 56: 855–865,1989.

[30803] 4355.Miller, D. A.; Miller, O. J.; Dev, V. G.; Hashmi, S.; Tantravahi,R. R.; Medrano, L.; Green, H.: Human chromosome 19 carries a poliovirusreceptor gene. Cell 1: 167–174, 1974.

[30804] 4356.Larrea, F.; Escorza, A.; Granados, J.; Valencia, X.; Valero, A.;Cravioto, M. C.; Perez–Palacios, G.: Familial oc–

currence of big-bigprolactin as the predominant immunoreactive human prolactin species in blood. *Fertil. Steril.* 47: 956–963, 1987.

- [30805] 4357. Carter, R. E.; Cerosaletti, K. M.; Burkin, D. J.; Fournier, R. E. K.; Jones, C.; Greenberg, B. D.; Citron, B. A.; Festoff, B. W.: The gene for the serpin thrombin inhibitor (P17), protease nexin1, is located on human chromosome 2q33–q35 and on syntenic regions in the mouse and sheep genomes. *Genomics* 27: 196–199, 1995.
- [30806] 4358. Gloor, S.; Odink, K.; Guenther, J.; Nick, H.; Monard, D.: A glia-derived neurite promoting factor with protease inhibitory activity belongs to the protease nexins. *Cell* 47: 687–693, 1986.
- [30807] 4359. Hall, C. L.; Yang, B.; Yang, X.; Zhang, S.; Turley, M.; Samuel, S.; Lange, L. A.; Wang, C.; Curpen, G. D.; Savani, R. C.; Greenberg, A. H.; Turley, E. A.: Overexpression of the hyaluronan receptor RHAMM is transforming and is also required for H-ras transformation. *Cell* 82:19–26, 1995.
- [30808] 4360. Itano, N.; Kimata, K.: Expression cloning and molecular characterization of HAS protein, a eukaryotic hyaluronan synthase. *J. Biol. Chem.* 271:9875–9878, 1996.
- [30809] 4361. Itano, N.; Kimata, K.: Molecular cloning of human hyaluronan synthase. *Biochem. Biophys. Res. Commun.*

222: 816–820, 1996.

- [30810] 4362. Shyjan, A. M.; Heldin, P.; Bucher, E. C.; Yoshino, T.; Briskin, M. J.: Functional cloning of the cDNA for a human hyaluronan synthase. *J. Biol. Chem.* 271: 23395–23399, 1996.
- [30811] 4363. Spicer, A. P.; Seldin, M. F.; Olsen, A. S.; Brown, N.; Wells, D. E.; Doggett, N. A.; Itano, N.; Kimata, K.; Inazawa, J.; McDonald, J. A.: Chromosomal localization of the human and mouse hyaluronan synthase genes. *Genomics* 41: 493–497, 1997.
- [30812] 4364. Watanabe, K.; Yamaguchi, Y.: Molecular identification of a putative human hyaluronan synthase. *J. Biol. Chem.* 271: 22945–22948, 1996.
- [30813] 4365. Johansson, M.; Karlsson, A.: Cloning and expression of human deoxyguanosine kinase cDNA. *Proc. Nat. Acad. Sci.* 93: 7258–7262, 1996.
- [30814] 4366. Taanman, J.-W.; Kateeb, I.; Muntau, A. C.; Jaksch, M.; Cohen, N.; Mandel, H.: A novel mutation in the deoxyguanosine kinase gene causing depletion of mitochondrial DNA. *Ann. Neurol.* 52: 237–239, 2002.
- [30815] 4367. Cahill, D. P.; da Costa, L. T.; Carson-Walter, E. B.; Kinzler, K. W.; Vogelstein, B.; Lengauer, C.: Characterization of MAD2B and other mitotic spindle checkpoint genes.

Genomics 58: 181–187, 1999.

- [30816] 4368.Chen, R.–H.; Waters, J. C.; Salmon, E. D.; Murray, A. W.: Association of spindle assembly checkpoint component X MAD2 with unattached kinetochores. Science 274:242–245, 1996.
- [30817] 4369.Dobles, M.; Liberal, V.; Scott, M. L.; Benezra, R.; Sorger, P.K.: Chromosome missegregation and apoptosis in mice lacking the mitotic checkpoint protein Mad2. Cell 101: 635–645, 2000.
- [30818] 4370.Krishnan, R.; Goodman, B.; Jin, D.–Y.; Jeang, K.–T.; Collins, C.; Stetten, G.; Spencer, F.: Map location and gene structure of the Homo sapiens mitotic arrest deficient 2 (MAD2L1) gene at 4q27. Genomics 49:475–478, 1998.
- [30819] 4371.Li, X.; Nicklas, R. B.: Mitotic forces control a cell–cycle checkpoint. Nature 373:630–632, 1995.
- [30820] 4372.Li, Y.; Benezra, R.: Identification of a human mitotic checkpoint gene: hsMAD2. Science 274: 246–248, 1996.
- [30821] 4373.Luo, X.; Tang, Z.; Rizo, J.; Yu, H.: The Mad2 spindle checkpoint protein undergoes similar major conformational changes upon binding to either Mad1 or Cdc20. Molec. Cell 9: 59–71, 2002.
- [30822] 4374.Michel, L. S.; Liberal, V.; Chatterjee, A.; Kirchwegger, R.; Pasche, B.; Gerald, W.; Dobles, M.; Sorger, P. K.; Murty,

V. V. V. S.; Benezra, R.: MAD2 haplo-insufficiency causes premature anaphase and chromosome instability in mammalian cells. *Nature* 409: 355–359, 2001.

[30823] 4375. Nelson, K. K.; Schlondorff, J.; Blobel, C. P.: Evidence for an interaction of the metalloprotease-disintegrin tumour necrosis factor alpha convertase (TACE) with mitotic arrest deficient 2 (MAD2), and of the metalloprotease-disintegrin MDC9 with a novel MAD2-related protein, MAD2-beta. *Biochem. J.* 343: 673–680, 1999.

[30824] 4376. Shonn, M. A.; McCarroll, R.; Murray, A. W.: Requirement of the spindle checkpoint for proper chromosome segregation in budding yeast meiosis. *Science* 289: 300–303, 2000.

[30825] 4377. Xu, L.; Deng, H. X.; Yang, Y.; Xia, J. H.; Hung, W. Y.; Siddique, T.: Assignment of mitotic arrest deficient protein 2 (MAD2L1) to human chromosome band 5q23.3 by in situ hybridization. *Cytogenet. Cell Genet.* 78: 63–64, 1997.

[30826] 4378. De La Rosa, J.; Ostrowski, J.; Hryniewicz, M. M.; Kredich, N. M.; Kotb, M.; LeGros, H. L., Jr.; Valentine, M.; Geller, A. M.: Chromosomal localization and catalytic properties of the recombinant alpha subunit of human lymphocyte methionine adenosyltransferase. *J. Biol. Chem.* 270: 21860–21868, 1995.

- [30827] 4379.Wang, D.; Stravopodis, D.; Teglund, S.; Kitazawa, J.; Ihle, J.N.: Naturally occurring dominant negative variants of Stat5. *Molec.Cell Biol.* 16: 6141–6148, 1996.
- [30828] 4380.Hou, J.; Schindler, U.; Henzel, W. J.; Ho, T. C.; Brasseur, M.;McKnight, S. L.: An interleukin–4–induced transcription factor: IL–4Stat. *Science* 265: 1701–1706, 1994.
- [30829] 4381.Leek, J. P.; Hamlin, P. J.; Bell, S. M.; Lench, N. J.: Assignmentof the STAT6 gene (STAT6) to human chromosome band 12q13 by in situhybridization. *Cytogenet. Cell Genet.* 79: 208–209, 1997.
- [30830] 4382.Patel, B. K. R.; Keck, C. L.; O'Leary, R. S.; Popescu, N. C.;LaRochelle, W. J.: Localization of the human Stat6 gene to chromosome12q13.3–q14.1, a region implicated in multiple solid tumors. *Genomics* 52:192–200, 1998.
- [30831] 4383.Patel, B. K. R.; Pierce, J. H.; LaRochelle, W. J.: Regulationof interleukin 4–mediated signaling by naturally occurring dominantnegative and attenuated forms of human Stat6. *Proc. Nat. Acad. Sci.* 95:172–177, 1998.
- [30832] 4384.Quelle, F. W.; Shimoda, K.; Thierfelder, W.; Fischer, C.; Kim,A.; Ruben, S. M.; Cleveland, J. L.; Pierce, J. H.; Keegan, A. D.;Nelms, K.; Paul, W. E.; Ihle, J. N.: Cloning of murine Stat6 andhuman Stat6, Stat proteins that are tyro–

sine phosphorylated in response to IL-4 and IL-4 but are not required for mitogenesis. *Molec. Cell Biol.* 15: 3336–3343, 1995.

- [30833] 4385. Liu, Y.; Chiu, I.-M.: Assignment of FGF12, the human FGF homologous factor 1 gene, to chromosome 3q29–3qter by fluorescence in situ hybridization. *Cytogenet. Cell Genet.* 78: 48–49, 1997.
- [30834] 4386. Caslini, C.; Spinelli, O.; Cazzaniga, G.; Golay, J.; De Gioia, L.; Pedretti, A.; Breviario, F.; Amaru, R.; Barbui, T.; Biondi, A.; Introna, M.; Rambaldi, A.: Identification of two novel isoforms of the ZNF162 gene: a growing family of signal transduction and activator of RNA proteins. *Genomics* 42: 268–277, 1997.
- [30835] 4387. Kramer, A.; Quentin, M.; Mulhauser, F.: Diverse modes of alternative splicing of human splicing factor SF1 deduced from the exon–intron structure of the gene. *Gene* 211: 29–37, 1998.
- [30836] 4388. Reddy, U. R.; Phatak, S.; Allen, C.; Nycum, L. M.; Sulman, E. P.; White, P. S.; Biegel, J. A.: Localization of the human Ror1 gene (NTRKR1) to chromosome 1p31–p32 by fluorescence in situ hybridization and somatic cell hybrid analysis. *Genomics* 41: 283–285, 1997.
- [30837] 4389. Reddy, U. R.; Phatak, S.; Pleasure, D.: Human neural

tissues express a truncated Ror1 receptor tyrosine kinase, lacking both extracellular and transmembrane domains. *Oncogene* 13: 1555–1559, 1996.

[30838] 4390. Gong, Y.; Chitayat, D.; Kerr, B.; Chen, T.; Babul-Hirji, R.; Pal, A.; Reiss, M.; Warman, M. L.: Brachydactyly type B: clinical description, genetic mapping to chromosome 9q, and evidence for a shared ancestral mutation. *Am. J. Hum. Genet.* 64: 578–585, 1999.

[30839] 4391. Innis, J. W.; Mortlock, D. P.: Limb development: molecular dysmorphology is at hand! *Clin. Genet.* 53: 337–348, 1998.

[30840] 4392. Manouvrier-Hanu, S.; Holder-Espinasse, M.; Lyonnet, S.: Genetics of limb anomalies in humans. *Trends Genet.* 15: 409–417, 1999.

[30841] 4393. Oishi, I.; Takeuchi, S.; Hashimoto, R.; Nagabukuro, A.; Ueda, T.; Liu, Z.-J.; Hatta, T.; Akira, S.; Matsuda, Y.; Yamamura, H.; Otani, H.; Minami, Y.: Spatio-temporally regulated expression of receptor tyrosine kinases, mRor1, mRor2, during mouse development: implications in development and function of the nervous system. *Genes Cells* 4: 41–56, 1999.

[30842] 4394. Schwabe, G. C.; Tinschert, S.; Buschow, C.; Meinel, P.; Wolff, G.; Gillessen-Kaesbach, G.; Oldridge, M.;

Wilkie, A. O. M.; Komec, R.; Mundlos, S.: Distinct mutations in the receptor tyrosine kinase gene ROR2 cause brachydactyly type B. *Am. J. Hum. Genet.* 67: 822–831, 2000.

[30843] 4395. Takeuchi, S.; Takeda, K.; Oishi, I.; Nomi, M.; Ikeya, M.; Itoh, K.; Tamura, S.; Ueda, T.; Hatta, T.; Otani, H.; Terashima, T.; Takada, S.; Yamamura, H.; Akira, S.; Minami, Y.: Mouse Ror2 receptor tyrosine kinase is required for the heart development and limb formation. *Genes Cells* 5: 71–78, 2000.

[30844] 4396. van Bokhoven, H.; Brunner, H. G.: Splitting p63. *Am. J. Hum. Genet.* 71: 1–13, 2002.

[30845] 4397. Jaffrey, S. R.; Snyder, S. H.: PIN: an associated protein inhibitor of neuronal nitric oxide synthase. *Science* 274: 774–777, 1996.

[30846] 4398. Garkavtsev, I.; Demetrick, D.; Riabowol, K.: Cellular localization and chromosome mapping of a novel candidate tumor suppressor gene (ING1). *Cytogenet. Cell Genet.* 76: 176–178, 1997.

[30847] 4399. Garkavtsev, I.; Kazarov, A.; Gudkov, A.; Riabowol, K.: Suppression of the novel growth inhibitor p33(ING1) promotes neoplastic transformation. *Nature Genet.* 14: 415–420, 1996. Note: Erratum: *Nature Genet.* 23: 373 only, 1999.

- [30848] 4400.Saito, A.; Furukawa, T.; Fukushige, S.; Koyama, S.; Hoshi, M.; Hayashi, Y.; Horii, A.: p24/ING1-ALT1 and p47/ING1-ALT2, distinct alternative transcripts of p33/ING1. *J. Hum. Genet.* 45: 177-181, 2000.
- [30849] 4401.Zeremski, M.; Horrigan, S. K.; Grigorian, I. A.; Westbrook, C.A.; Gudkov, A. V.: Localization of the candidate tumor suppressor gene ING1 to human chromosome 13q34. *Somat. Cell Molec. Genet.* 23:233-236, 1997.
- [30850] 4402.Gilligan, D. M.; Lozovatsky, L.; Gwynn, B.; Brugnara, C.; Mohandas, N.; Peters, L. L.: Targeted disruption of the beta adducin gene (Add2) causes red blood cell spherocytosis in mice. *Proc. Nat. Acad. Sci.* 96:10717-10722, 1999.
- [30851] 4403.Adamson, M. C.; Dennis, C.; Delaney, S.; Christiansen, J.; Monkley, S.; Kozak, C. A.; Wainwright, B.: Isolation and genetic mapping of two novel members of the murine Wnt gene family, Wnt11 and Wnt12, and the mapping of Wnt5a and Wnt7a. *Genomics* 24: 9-13, 1994.
- [30852] 4404.Bui, T. D.; Lako, M.; Lejeune, S.; Curtis, A. R. J.; Strachan, T.; Lindsay, S.; Harris, A. L.: Isolation of a full-length human WNT7A gene implicated in limb development and cell transformation, and mapping to chromosome 3p25. *Gene* 189: 25-29, 1997.

- [30853] 4405.Ikegawa, S.; Kumano, Y.; Okui, K.; Fujiwara, T.; Takahashi, E.; Nakamura, Y.: Isolation, characterization and chromosomal assignment of the human WNT7A gene. *Cytogenet. Cell Genet.* 74: 149–152, 1996.
- [30854] 4406.Li, S.; Chiang, T.-C.; Davis, G. R.; Williams, R. M.; Wilson, V.P.; McLachlan, J. A.: Decreased expression of Wnt7a mRNA is inversely associated with the expression of estrogen receptor- α in human uterine leiomyoma. *J. Clin. Endocr. Metab.* 86: 454–457, 2001.
- [30855] 4407.Pichaud, F.; Delage-Mourroux, R.; Pidoux, E.; Jullienne, A.; Rousseau-Merck, M.-F.: Chromosomal localization of the type-I 15-PGDH gene to 4q34–q35. *Hum. Genet.* 99: 279–281, 1997.
- [30856] 4408.Walder, K.; Norman, R. A.; Hanson, R. L.; Schrauwen, P.; Neverova, M.; Jenkinson, C. P.; Easlick, J.; Warden, C. H.; Pecqueur, C.; Raimbault, S.; Ricquier, D.; Harper, M.; Silver, K.; Shuldiner, A. R.; Solanes, G.; Lowell, B. B.; Chung, W. K.; Leibel, R. L.; Pratley, R.; Ravussin, E.: Association between uncoupling protein polymorphisms (UCP2–UCP3) and energy metabolism/obesity in Pima Indians. *Hum. Molec. Genet.* 7:1431–1435, 1998.
- [30857] 4409.Lindsay, E. A.; Rizzu, P.; Antonacci, R.; Jurecic, V.; Delmas-Mata, J.; Lee, C.-C.; Kim, U.-J.; Scambler, P. J.; Bal-

dini, A.: A transcription map in the CATCH22 critical region: identification, mapping, and ordering of four novel transcripts expressed in heart. *Genomics* 32: 104–112, 1996.

[30858] 4410. Takai, S.; Hinoda, Y.; Adachi, T.; Imai, K.; Oshima, M.: A human UDP (sic)–GalNAc: polypeptide, N–acetylgalactosaminyltransferase type 1 gene is located at the chromosomal region 18q12.1. *Hum. Genet.* 99:293–294, 1997.

[30859] 4411. White, T.; Bennett, E. P.; Takio, K.; Sorensen, T.; Bonding, N.; Clausen, H.: Purification and cDNA cloning of a human UDP–N–acetyl–alpha–D–galactosamine: polypeptide N–acetylgalactosaminyltransferase. *J. Biol. Chem.* 270: 24156–24165, 1995.

[30860] 4412. Candau, R.; Moore, P. A.; Wang, L.; Barlev, N.; Ying, C. Y.; Rosen, C. A.; Berger, S. L.: Identification of human proteins functionally conserved with the yeast putative adaptors ADA2 and GCN5. *Molec. Cell. Biol.* 16: 593–602, 1996.

- [30861] 4413.Carter, K. C.; Wang, L.; Shell, B. K.; Zamir, I.; Berger, S. L.; Moore, P. A.: The human transcriptional adaptor genes TADA2L and GCN5L2 colocalize to chromosome 17q12–q21 and display a similar tissue expression pattern. *Genomics* 40: 497–500, 1997.
- [30862] 4414.Ogryzko, V. V.; Kotani, T.; Zhang, X.; Schiltz, R. L.; Howard, T.; Yang, X.-J.; Howard, B. H.; Qin, J.; Nakatani, Y.: Histone-like TAFs within the PCAF histone acetylase complex. *Cell* 94: 35–44, 1998.
- [30863] 4415.Struhl, K.; Moqtaderi, Z.: The TAFs in the HAT. *Cell* 94: 1–4, 1998.
- [30864] 4416.Imhof, M. O.; McDonnell, D. P.: Yeast RSP5 and its human homolog hRPF1 potentiate hormone-dependent activation of transcription by human progesterone and glucocorticoid receptors. *Molec. Cell. Biol.* 16:2594–2605, 1996.
- [30865] 4417.Kumar, S.; Harvey, K. F.; Kinoshita, M.; Copeland, N. G.; Noda, M.; Jenkins, N. A.: cDNA cloning, expression analysis, and mapping of the mouse Nedd4 gene. *Genomics* 40: 435–443, 1997.
- [30866] 4418.Cabin, D. E.; Gardiner, K.; Reeves, R. H.: Molecular genetic characterization and comparative mapping of the human PCP4 gene. *Somat. Cell Molec. Genet.* 22: 167–175,

1996.

- [30867] 4419.Chen, H.; Bouras, C.; Antonarakis, S. E.: Cloning of the cDNA for a human homolog of the rat PEP-19 gene and mapping to chromosome 21q22.2-q22.3. *Hum. Genet.* 98: 672-677, 1996.
- [30868] 4420.Hubert, R. S.; Korenberg, J. R.: PCP4 maps between D21S345 and P31P10SP6 on chromosome 21q22.2-q22.3. *Cytogenet. Cell Genet.* 78:44-45, 1997.
- [30869] 4421.Collum, R. G.; Fisher, P. E.; Datta, M.; Mellis, S.; Thiele, C.; Huebner, K.; Croce, C. M.; Israel, M. A.; Theil, T.; Moroy, T.; DePinho, R.; Alt, F. W.: A novel POU homeodomain gene specifically expressed in cells of the developing mammalian nervous system. *Nucleic Acids Res.* 20: 4919-4925, 1992.
- [30870] 4422.Gerrero, M. R.; McEvilly, R. J.; Turner, E.; Lin, C. R.; O'Connell, S.; Jenne, K. J.; Hobbs, M. V.; Rosenfeld, M. G.: Brn-3.0: a POU-domain protein expressed in the sensory immune and endocrine systems that functions on elements distinct from known octamer motifs. *Proc. Nat. Acad. Sci.* 90: 10841-10845, 1993.
- [30871] 4423.Still, I. H.; Cowell, J.: The Brn-3a transcription factor gene (POU4F1) maps close to the locus for the variant late infantile form of neuronal ceroid-lipofuscinosis. *Cyto-*

genet. Cell Genet. 74: 225–226,1996.

- [30872] 4424. Block, M. R.; Glick, B. S.; Wilcox, C. A.; Wieland, F. T.; Rothman, J. E.: Purification of an N-ethylmaleimide-sensitive protein catalyzing vesicular transport. Proc. Nat. Acad. Sci. 85: 7852–7856, 1988.
- [30873] 4425. Glick, B. S.; Rothman, J. E.: Possible role for fatty acyl-coenzyme A in intracellular protein transport. Nature 326: 309–312, 1987.
- [30874] 4426. Hoyle, J.; Phelan, J. P.; Bermingham, N.; Fisher, E. M. C.: Localization of human and mouse N-ethylmaleimide-sensitive factor (NSF) gene: a two-domain member of the AAA family that is involved in membrane fusion. Mammalian Genome 7: 850–852, 1996.
- [30875] 4427. Rothman, J. E.: Mechanisms of intracellular protein transport. Nature 372: 55–63, 1994.
- [30876] 4428. Wilson, D. W.; Wilcox, C. A.; Flynn, G. C.; Chen, E.; Kuang, W.-J.; Henzel, W. J.; Block, M. R.; Ullrich, A.; Rothman, J. E.: A fusion protein required for vesicle-mediated transport in both mammalian cells and yeast. Nature 339: 355–359, 1989.
- [30877] 4429. Dikstein, R.; Zhou, S.; Tjian, R.: Human TAF(II)105 is a cell type-specific TFIID subunit related to hTAF(II)130. Cell 87: 137–146, 1996.

- [30878] 4430.Freiman, R. N.; Albright, S. R.; Zheng, S.; Sha, W. C.; Hammer, R. E.; Tjian, R.: Requirement of tissue-selective TBP-associated factor TAFII105 in ovarian development. *Science* 293: 2084–2087, 2001.
- [30879] 4431.Allikmets, R.; Gerrard, B.; Hutchinson, A.; Dean, M.: Characterization of the human ABC superfamily: isolation and mapping of 21 new genes using the expressed sequence tags database. *Hum. Molec. Genet.* 5:1649–1655, 1996.
- [30880] 4432.Rizzu, P.; Lindsay, E. A.; Taylor, C.; O'Donnell, H.; Levy, A.; Scambler, P.; Baldini, A.: Cloning and comparative mapping of a gene from the commonly deleted region of DiGeorge and velocardiofacial syndromes conserved in *C. elegans*. *Mammalian Genome* 7: 639–643, 1996.
- [30881] 4433.Bennett, E. P.; Hassan, H.; Clausen, H.: cDNA cloning and expression of a novel human UDP-N-acetyl- α -D-galactosamine. *J. Biol. Chem.* 271:17006–17012, 1996.
- [30882] 4434.Bennett, E. P.; Weghuis, D. O.; Merks, G.; Geurts van Kessel, A.; Eiberg, H.; Clausen, H.: Genomic organization and chromosomal localization of three members of the UDP-N-acetylgalactosamine:polypeptide N-acetylgalactosaminyltransferase family. *Glycobiology* 8:

547–555, 1998.

- [30883] 4435.Zara, J.; Hagen, F. K.; Ten Hagen, K. G.; Van Wuyck–huyse, B. C.;Tabak, L. A.: Cloning and expression of mouse UDP–GalNAc:polypeptideN–acetylgalactosaminyltransferase–T3. *Biochem. Biophys. Res. Commun.* 228:38–44, 1996.
- [30884] 4436.Zhao, Y.–Y.; Liu, Y.; Stan, R.–V.; Fan, L.; Gu, Y.; Dalton, N.;Chu, P.–H.; Peterson, K.; Ross, J., Jr.; Chien, K. R.: Defects incaveolin–1 cause dilated cardiomyopathy and pulmonary hypertensionin knockout mice. *Proc. Nat. Acad. Sci.* 99: 11375–11380, 2002.
- [30885] 4437.Mok, S. C.; Wong, K.–K.; Chan, R. K. W.; Lau, C. C.; Tsao, S.–W.;Knapp, R. C.; Berkowitz, R. S.: Molecular cloning of differentiallyexpressed genes in human epithelial ovarian cancer. *Gynecol. Oncol.* 52:247–252, 1994.
- [30886] 4438.Xu, X.–X.; Yang, W.; Jackowski, S.; Rock, C. O.: Cloning of anovel phosphoprotein regulated by colony–stimulating factor 1 sharesa domain with the Drosophila disabled gene product. *J. Biol. Chem.* 270:14184–14191, 1995.
- [30887] 4439.Galbiati, F.; Volonte, D.; Gil, O.; Zanazzi, G.; Salzer, J. L.;Sargiacomo, M.; Scherer, P. E.; Engelman, J. A.; Schlegel, A.; Parenti,M.; Okamoto, T.; Lisanti, M. P.: Ex–

pression of caveolin-1 and -2 in differentiating PC12 cells and dorsal root ganglion neurons: caveolin-2 is up-regulated in response to cell injury. *Proc. Nat. Acad. Sci.* 95:10257-10262, 1998.

[30888] 4440. Scherer, P. E.; Okamoto, T.; Chun, M.; Nishimoto, I.; Lodish, H. F.; Lisanti, M. P.: Identification, sequence, and expression of caveolin-2 defines a caveolin gene family. *Proc. Nat. Acad. Sci.* 93: 131-135, 1996.

[30889] 4441. Campbell, H. D.; Webb, G. C.; Fountain, S.; Young, I. G.: The human PIN1 peptidyl-prolyl cis/trans isomerase gene maps to human chromosome 19p13 and the closely related PIN1L gene to 1p31. *Genomics* 44:157-162, 1997.

[30890] 4442. Liou, Y.-C.; Ryo, A.; Huang, H.-K.; Lu, P.-J.; Bronson, R.; Fujimori, F.; Uchida, T.; Hunter, T.; Lu, K. P.: Loss of Pin1 function in the mouse causes phenotypes resembling cyclin D1-null phenotypes. *Proc. Nat. Acad. Sci.* 99: 1335-1340, 2002.

[30891] 4443. Lu, K. P.; Hanes, S. D.; Hunter, T.: A human peptidyl-prolyl isomerase essential for regulation of mitosis. *Nature* 380: 544-547, 1996.

[30892] 4444. Lu, P.-J.; Wulf, G.; Zhou, X. Z.; Davies, P.; Lu, K. P.: The prolyl isomerase Pin1 restores the function of Alzheimer-associated phosphorylated tau protein. *Nature*

399: 784–788, 1999.

- [30893] 4445. Maleszka, R.; Hanes, S. D.; Hackett, R. L.; de Couet, H. G.; Gabor Miklos, G. L.: The *Drosophila melanogaster* dodo (dod) gene, conserved in humans, is functionally interchangeable with the ESS1 cell division gene of *Saccharomyces cerevisiae*. *Proc. Nat. Acad. Sci.* 93: 447–451, 1996.
- [30894] 4446. Winkler, K. E.; Swanson, K. I.; Kornbluth, S.; Means, A. R.: Requirement of the prolyl isomerase Pin1 for the replication checkpoint. *Science* 287:1644–1647, 2000.
- [30895] 4447. Takahashi, T.; Fournier, A.; Nakamura, F.; Wang, L.-H.; Murakami, Y.; Kalb, R. G.; Fujisawa, H.; Strittmatter, S. M.: Plexin-neuropilin-1 complexes form functional semaphorin-3A receptors. *Cell* 99: 59–69, 1999.
- [30896] 4448. Santagata, S.; Boggon, T. J.; Baird, C. L.; Gomez, C. A.; Zhao, J.; Shan, W. S.; Myska, D. G.; Shapiro, L.: G-protein signaling through tubby proteins. *Science* 292: 2041–2050, 2001.
- [30897] 4449. Sasaki, T.; Irie-Sasaki, J.; Horie, Y.; Bachmaier, K.; Fata, J. E.; Li, M.; Suzuki, A.; Bouchard, D.; Ho, A.; Redston, M.; Gallinger, S.; Khokha, R.; Mak, T. W.; Hawkins, P. T.; Stephens, L.; Scherer, S. W.; Tsao, M.; Penninger, J. M.: Colorectal carcinomas in mice lacking the catalytic subunit

of PI(3)K-gamma. Nature 406: 897-902,2000.

[30898] 4450.Sasaki, T.; Irie-Sasaki, J.; Jones, R. G.; Oliveira-dos-Santos,A. J.; Stanford, W. L.; Bolon, B.; Wakeham, A.; Itie, A.; Bouchard,D.; Kozieradzki, I.; Joza, N.; Mak, T. W.; Ohashi, P. S.; Suzuki,A.; Penninger, J. M.: Function of PI3K-gamma in thymocyte development,T cell activation, and neutrophil migration. Science 287: 1040-1046,2000.

[30899] 4451.Stoyanov, B.; Volinia, S.; Hanck, T.; Rubio, I.; Loubtchenkov,M.; Malek, D.; Stoyanova, S.; Vanhaesebroeck, B.; Dhand, R.; Nurnberg,B.; Gierschik, P.; Seedorf, K.; Hsuan, J. J.; Waterfield, M. D.; Wetzker,R.: Cloning and characterization of a G protein-activated human phosphoinositide-3kinase. Science 269: 690-693, 1995.

[30900] 4452.Hotten, G.; Neidhardt, H.; Schneider, C.; Pohl, J.: Cloning of a new member of the TGF-beta family: a putative new activin betaCchain. Biochem. Biophys. Res. Commun. 206: 608-613, 1995.

[30901] 4453.Ling, N.; Ying, S. Y.; Ueno, N.; Shimasaki, S.; Esch, F.; Hotta,M.; Guillemin, R.: Pituitary FSH is released by a heterodimer of the beta-subunits from the two forms of inhibin. Nature 321: 779-782,1986.

[30902] 4454.Schmitt, J.; Hotten, G.; Jenkins, N. A.; Gilbert, D. J.; Copeland,N. G.; Pohl, J.; Schrewe, H.: Structure, chromo-

somal localization, and expression analysis of the mouse inhibin/activin betaC (Inhbc) gene. *Genomics* 32: 358–366, 1996.

[30903] 4455. Savitsky, K.; Ziv, Y.; Bar-Shira, A.; Gilad, S.; Tagle, D. A.; Smith, S.; Uziel, T.; Sfez, S.; Nahmias, J.; Sartiel, A.; Eddy, R. L.; Shows, T. B.; Collins, F. S.; Shiloh, Y.; Rotman, G.: A human gene (DDX10) encoding a putative DEAD-box RNA helicase at 11q22–q23. *Genomics* 33: 199–206, 1996.

[30904] 4456. Albertsen, H. M.; Smith, S. A.; Melis, R.; Williams, B.; Holik, P.; Stevens, J.; White, R.: Sequence, genomic structure, and chromosomal assignment of human DOC-2. *Genomics* 33: 207–213, 1996.

[30905] 4457. Mok, S. C.; Chan, W. Y.; Wong, K. K.; Cheung, K. K.; Lau, C. C.; Ng, S. W.; Baldini, A.; Colitti, C. V.; Rock, C. O.; Berkowitz, R. S.: DOC-2, a candidate tumor suppressor gene in human epithelial ovarian cancer. *Oncogene* 16: 2381–2387, 1998.

[30906] 4458. Kimura, T.; Arakawa, Y.; Inoue, S.; Fukushima, Y.; Kondo, I.; Koyama, K.; Hosoi, T.; Orimo, A.; Muramatsu, M.; Nakamura, Y.; Abe, T.; Inazawa, J.: The brain finger protein gene (ZNF179), a member of the RING finger family, maps within the Smith–Magenis syndrome region at

17p11.2. Am.J. Med. Genet. 69: 320–324, 1997.

- [30907] 4459.Matsuda, Y.; Inue, S.; Seki, N.; Hosoi, T.; Orimo, A.; Muramatsu,M.; Hori, T.: Chromosome mapping of human (ZNF179), mouse, and ratgenes for brain finger protein (bfp), a member of the RING fingerfamily. Genomics 33: 325–327, 1996.
- [30908] 4460.Ambrose, H. J.; Blake, D. J.; Nawrotzki, R. A.; Davies, K. E.:Genomic organization of the mouse dystrobrevin gene: comparative analysiswith the dystrophin gene. Genomics 39: 359–369, 1997.
- [30909] 4461.Blake, D. J.; Nawrotzki, R.; Loh, N. Y.; Gorecki, D. C.; Davies,K. E.: Beta–dystrobrevin, a member of the dystrophin–related proteinfamily. Proc. Nat. Acad. Sci. 95: 241–246, 1998.
- [30910] 4462.Ichida, F.; Tsubata, S.; Bowles, K. R.; Haneda, N.; Uese, K.; Miyawaki,T.; Dreyer, W. J.; Messina, J.; Li, H.; Bowles, N. E.; Towbin, J.A.: Novel gene mutations in patients with left ventricular noncompactionor Barth syndrome. Circulation 103: 1256–1263, 2001.
- [30911] 4463.England, S. K.; Uebele, V. N.; Shear, H.; Kodali, J.; Bennett,P. B.; Tamkun, M. M.: Characterization of a voltage–gated K⁺ channelbeta subunit expressed in human heart. Proc. Nat. Acad. Sci. 92:6309–6313, 1995.

- [30912] 4464.Jones, J. M.; Bentley, E.; Meisler, M. H.; Darling, S. M.: Genetic mapping of the voltage-gated shaker potassium channel beta subunit *Kcnab1* to mouse chromosome 3. *Mammalian Genome* 9: 260 only, 1998.
- [30913] 4465.Leicher, T.; Bähring, R.; Isbrandt, D.; Pongs, O.: Co-expression of the *KCNA3B* gene product with Kv1.5 leads to a novel A-type potassium channel. *J. Biol. Chem.* 273: 35095–35101, 1998.
- [30914] 4466.Leicher, T. Roeper, J.; Weber, K.; Wang, X.; Pongs, O.: Structural and functional characterization of human potassium channel subunit beta-1 (*KCNA1B*). *Neuropharmacology* 35: 787–795, 1996.
- [30915] 4467.McCormack, K.; McCormack, T.; Tanouye, M.; Rudy, B.; Stuhmer, W.: Alternative splicing of the human Shaker K⁺ channel beta-1 gene and functional expression of the beta-2 gene product. *FEBS Lett.* 370:32–36, 1995.
- [30916] 4468.Schultz, D.; Litt, M.; Smith, L.; Thayer, M.; McCormack, K.: Localization of two potassium channel beta subunit genes, *KCNA1B* and *KCNA2B*. *Genomics* 31:389–391, 1996.
- [30917] 4469.Collin, G. B.; Nishina, P. M.; Marshall, J. D.; Naggert, J. K.: Human *DCTN1*: genomic structure and evaluation as a candidate for Alstrom syndrome. *Genomics* 53: 359–364,

1998.

- [30918] 4470. Holzbaur, E. L. F.; Tokito, M. K.: Localization of the DCTN1 gene encoding p150(Glued) to human chromosome 2p13 by fluorescence in situ hybridization. *Genomics* 31: 398–399, 1996.
- [30919] 4471. Holzbaur, E. L. F.; Vallee, R. B.: Dyneins: molecular structure and cellular function. *Ann. Rev. Cell Biol.* 10: 339–372, 1994.
- [30920] 4472. Jang, W.; Weber, J. S.; Tokito, M. K.; Holzbaur, E. L. F.; Meisler, M. H.: Mouse p150(Glued) (dynactin 1) cDNA sequence and evaluation as a candidate for the neuromuscular disease mutation mnd2. *Biochem. Biophys. Res. Commun.* 231: 344–347, 1997.
- [30921] 4473. Korthaus, D.; Wedemeyer, N.; Lengeling, A.; Ronsiek, M.; Jockusch, H.; Schmitt-John, T.: Integrated radiation hybrid map of human chromosome 2p13: possible involvement of dynactin in neuromuscular diseases. *Genomics* 43: 242–244, 1997.
- [30922] 4474. Pushkin, A.; Abuladze, N.; Newman, D.; Tatishchev, S.; Kurtz, I.: Genomic organization of the DCTN1–SLC4A5 locus encoding both NBC4 and p150(Glued). *Cytogenet. Cell Genet.* 95: 163–168, 2001.
- [30923] 4475. Miyata, A.; Hara, S.; Yokoyama, C.; Inoue, H.; Ullrich,

V.; Tanabe,T.: Molecular cloning and expression of human prostacyclin synthase. *Biochem.Biophys. Res. Commun.* 200: 1728–1734, 1994.

[30924] 4476.Inoue, H.; Ishii, H.; Alder, H.; Snyder, E.; Druck, T.; Huebner,K.; Croce, C. M.: Sequence of the FRA3B common fragile region: implicationsfor the mechanism of FHIT deletion. *Proc. Nat. Acad. Sci.* 94: 14584–14589,1997.

[30925] 4477.Morikawa, H.; Nakagawa, Y.; Hashimoto, K.; Niki, M.; Egashira,Y.; Hirata, I.; Katsu, K.; Akao, Y.: Frequent altered expressionof fragile histidine triad protein in human colorectal adenomas. *Biochem.Biophys. Res. Commun.* 278: 205–210, 2000.

[30926] 4478.Nelson, D. R.; Koymans, L.; Kamataki, T.; Stegeman, J. J.; Feyereisen,R.; Waxman, D. J.; Waterman, M. R.; Gotoh, O.; Coon, M. J.; Estabrook,R. W.; Gunsalus, I. C.; Nebert, D. W.: P450 superfamily: update onnew sequences, gene mapping, accession numbers and nomenclature. *Pharmacogenetics* 6:1–42, 1996.

[30927] 4479.Wang, L.–H.; Chen, L.: Organization of the gene encoding humanprostacyclin synthase. *Biochem. Biophys. Res. Commun.* 226: 631–637,1996.

[30928] 4480.Yokoyama, C.; Yabuki, T.; Inoue, H.; Tone, Y.; Hara, S.; Hatae,T.; Nagata, M.; Takahashi, E.–I.; Tanabe, T.: Hu–

man gene encoding prostacyclin synthase (PTGIS): genomic organization, chromosomal localization, and promoter activity. *Genomics* 36: 296–304, 1996.

- [30929] 4481. Brindle, N. P. J.; Holt, M. R.; Davies, J. E.; Price, C. J.; Critchley, D. R.: The focal-adhesion vasodilator-stimulated phosphoprotein (VASP) binds to the proline-rich domain in vinculin. *Biochem. J.* 318: 753–757, 1996.
- [30930] 4482. Haffner, C.; Jarchau, T.; Reinhard, M.; Hoppe, J.; Lohmann, S.M.; Walter, V.: Molecular cloning, structural analysis and functional expression of the proline-rich focal adhesion and microfilament-associated protein VASP. *EMBO J.* 14: 19–27, 1995.
- [30931] 4483. Zimmer, M.; Fink, T.; Fischer, L.; Hauser, W.; Scherer, K.; Lichter, P.; Walter, U.: Cloning of the VASP (vasodilator-stimulated phosphoprotein) genes in human and mouse: structure, sequence, and chromosomal localization. *Genomics* 36: 227–233, 1996.
- [30932] 4484. Brezin, A. P.; Adam, M. F.; Belmouden, A.; Lureau, M.-A.; Chaventre, A.; Copin, B.; Gomez, L.; Dupont de Dinechin, S.; Berkani, M.; Valtot, F.; Rouland, J.-F.; Dascotte, J.-C.; Bach, J.-F.; Garchon, H.-J.: Founder effect in GLC1A-linked familial open-angle glaucoma in northern-France. *Am. J. Med. Genet.* 76: 438–445, 1998.

- [30933] 4485.Colomb, E.; Nguyen, T. D.; Bechetoille, A.; Dascotte, J.-C.; Valtot,F.; Brezin, A. P.; Berkani, M.; Copin, B.; Gomez, L.; Polansky, J.R.; Garchon, H.-J.: Association of a single nucleotide polymorphismin the TIGR/MYOCILIN gene promoter with the severity of primary open-angleglaucoma. Clin. Genet. 60: 220–225, 2001.
- [30934] 4486.Escribano, J.; Ortego, J.; Coca-Prados, M.: Isolation and characterizationof cell-specific cDNA clones from a subtractive library of the ocularciliary body of a single normal human donor: transcription and synthesisof plasma proteins. J. Biochem. 118: 921–931, 1995.
- [30935] 4487.Fingert, J. H.; Ying, L.; Swiderski, R. E.; Nystuen, A. M.; Arbour,N. C.; Alward, W. L. M.; Sheffield, V. C.; Stone, E. M.: Characterizationand comparison of the human and mouse GLC1A glaucoma genes. GenomeRes. 8: 377–384, 1998.
- [30936] 4488.Fleck, B. W.; Cullen, J. F.: Autosomal dominant juvenile onsetglaucoma affecting six generations in an Edinburgh family. (Letter) Brit.J. Ophthal. 70: 715 only, 1986.
- [30937] 4489.Kubota, R.; Noda, S.; Wang, Y.; Minoshima, S.; Asakawa, S.; Kudoh,J.; Mashima, Y.; Oguchi, Y.; Shimizu, N.: A novel myosin-like protein(myocilin) expressed in the connecting cilium of the photoreceptor:molecular cloning,

tissue expression, and chromosomal mapping. *Genomics* 41:360–369, 1997.

- [30938] 4490. Fingert, J. H.; Heon, E.; Liebmann, J. M.; Yamamoto, T.; Craig, J. E.; Rait, J.; Kawase, K.; Hoh, S.-T.; Buys, Y. M.; Dickinson, J.; Hockey, R. R.; Williams-Lyn, D.; Trope, G.; Kitazawa, Y.; Ritch, R.; Mackey, D. A.; Alward, W. L. M.; Sheffield, V. C.; Stone, E. M.: Analysis of myocilin mutations in 1703 glaucoma patients from five different populations. *Hum. Molec. Genet.* 8: 899–905, 1999.
- [30939] 4491. Mansergh, F. C.; Kenna, P. F.; Ayuso, C.; Kiang, A.-S.; Humphries, P.; Farrar, G. J.: Novel mutations in the TIGR gene in early and late onset open angle glaucoma. *Hum. Mutat.* 11: 244–251, 1998.
- [30940] 4492. Michels-Rautenstrauss, K. G.; Mardin, C. Y.; Budde, W. M.; Liehr, T.; Polansky, J.; Nguyen, T.; Timmerman, V.; Van Broeckhoven, C.; Naumann, G. O. H.; Pfeiffer, R. A.; Rautenstrauss, B. W.: Juvenile open angle glaucoma: fine mapping of the TIGR gene to 1q24.3–q25.2 and mutation analysis. *Hum. Genet.* 102: 103–106, 1998.
- [30941] 4493. Morissette, J.; Clepet, C.; Moisan, S.; Dubois, S.; Winstall, E.; Vermeeren, D.; Nguyen, T. D.; Polansky, J. R.; Cote, G.; Anctil, J.-L.; Amyot, M.; Plante, M.; Falardeau, P.; Raymond, V.: Homozygotes carrying an autosomal domi-

nant TIGR mutation do not manifest glaucoma.(Letter) Nature Genet. 19: 319–321, 1998.

- [30942] 4494.Shepard, A. R.; Jacobson, N.; Fingert, J. H.; Stone, E. M.; Sheffield,V. C.; Clark, A. F.: Delayed secondary glucocorticoid responsiveness of MYOC in human trabecular meshwork cells. Invest. Ophthal. Vis.Sci. 42: 3173–3181, 2001.
- [30943] 4495.Stoilova, D.; Child, A.; Brice, G.; Crick, R. P.; Fleck, B. W.;Sarfarazi, M.: Identification of a new 'TIGR' mutation in a familywith juvenile-onset primary open angle glaucoma. Ophthalmic Genet. 18:109–118, 1997.
- [30944] 4496.Stone, E. M.: Personal Communication. Iowa City, Iowa 6/16/1999.
- [30945] 4497.Stone, E. M.: Personal Communication. Iowa City, Iowa 11/20/1997.
- [30946] 4498.Suzuki, Y.; Shirato, S.; Taniguchi, F.; Ohara, K.; Nishimaki,K.; Ohta, S.: Mutations in the TIGR gene in familial primary open-angleglaucoma in Japan. (Letter) Am. J. Hum. Genet. 1202–1204, 1997.
- [30947] 4499.Vasconcellos, J. P. C.; Melo, M. B.; Costa, V. P.; Tsukumo, D.M. L.; Basseres, D. S.; Bordin, S.; Saad, S. T. O.; Costa, F. F.:Novel mutation in the MYOC gene in primary open angle glaucoma patients. J.Med. Genet. 37:

301–303, 2000.

- [30948] 4500. Wiggs, J. L.; Allingham, R. R.; Vollrath, D.; Jones, K. H.; DeLa Paz, M.; Kern, J.; Patterson, K.; Babb, V. L.; Del Bono, E. A.; Broome, B. W.; Pericak-Vance, M. A.; Haines, J. L.: Prevalence of mutations in TIGR/myocilin in patients with adult and juvenile primary open-angle glaucoma. (Letter) *Am. J. Hum. Genet.* 63: 1549–1552, 1998.
- [30949] 4501. Zhou, Z.; Vollrath, D.: A cellular assay distinguishes normal and mutant TIGR/myocilin protein. *Hum. Molec. Genet.* 8: 2221–2228, 1999.
- [30950] 4502. Hackam, A. S.; Yassa, A. S.; Singaraja, R.; Metzler, M.; Gutekunst, C.-A.; Gan, L.; Warby, S.; Wellington, C. L.; Vaillancourt, J.; Chen, N.; Gervais, F. G.; Raymond, L.; Nicholson, D. W.; Hayden, M. R.: Huntingtin interacting protein 1 induces apoptosis via a novel caspase-dependent death effector domain. *J. Biol. Chem.* 275: 41299–41308, 2000.
- [30951] 4503. Himmelbauer, H.; Wedemeyer, N.; Haaf, T.; Wanker, E. E.; Schalkwyk, L. C.; Lehrach, H.: IRS-PCR-based genetic mapping of the huntingtin interacting protein gene (HIP1) on mouse chromosome 5. *Mammalian Genome* 9: 26–31, 1998.
- [30952] 4504. Manabe, T.; Noda, Y.; Mamiya, T.; Katagiri, H.;

Houtani, T.; Nishi, M.; Noda, T.; Takahashi, T.; Sugimoto, T.; Nabeshima, T.; Takeshima, H.: Facilitation of long-term potentiation and memory in mice lacking nociceptin receptors. *Nature* 394: 577–581, 1998.

[30953] 4505. Mollereau, C.; Simons, M.-J.; Soularue, P.; Liners, F.; Vassart, G.; Meunier, J.-C.; Parmentier, M.: Structure, tissue distribution, and chromosomal localization of the pre-nociceptin gene. *Proc. Nat. Acad. Sci.* 93: 8666–8700, 1996.

[30954] 4506. Nishi, M.; Houtani, T.; Noda, Y.; Mamiya, T.; Sato, K.; Doi, T.; Kuno, J.; Takeshima, H.; Nukada, T.; Nabeshima, T.; Yamashita, T.; Noda, T.; Sugimoto, T.: Unrestrained nociceptive response and dysregulation of hearing ability in mice lacking the nociceptin/orphanin FQ receptor. *EMBO J.* 16: 1858–1864, 1997.

[30955] 4507. Okuda-Ashitaka, E.; Minami, T.; Tachibana, S.; Yoshihara, Y.; Nishiuchi, Y.; Kimura, T.; Ito, S.: Nocistatin, a peptide that blocks nociceptin action in pain transmission. *Nature* 392: 286–289, 1998.

[30956] 4508. Pan, Z. Z.; Hirakawa, N.; Fields, H. L.: A cellular mechanism for the bidirectional pain-modulating actions of orphanin FQ/nociceptin. *Neuron* 26: 515–522, 2000.

[30957] 4509. Serhan, C. N.; Fierro, I. M.; Chiang, N.; Pouliot, M.:

Cuttingedge: nociceptin stimulates neutrophil chemotaxis and recruitment:inhibition by aspirin–triggered–15–epi–lipoxin A(4). J. Immun. 166:3650–3654, 2001.

[30958] 4510.Kanai, N.; Lu, R.; Satriano, J. A.; Bao, Y.; Wolkoff, A. W.; Schuster,V. L.: Identification and characterization of a prostaglandin transporter. Science 268:866–869, 1995.

[30959] 4511.Lu, R.; Kanai, N.; Bao, Y.; Schuster, V. L.: Cloning, in vitroexpression, and tissue distribution of a human prostaglandin transporter cDNA (hPGT). J. Clin. Invest. 98: 1142–1149, 1996.

[30960] 4512.Lu, R.; Schuster, V. L.: Molecular cloning of the gene for thehuman prostaglandin transporter hPGT: gene organization, promoteractivity, and chromosomal localization. Biochem. Biophys. Res. Commun. 246:805–812, 1998.

[30961] 4513.Frucht, D. M.; Aringer, M.; Galon, J.; Danning, C.; Brown, M.;Fan, S.; Centola, M.; Wu, C.–Y.; Yamada, N.; Galaway, H. E.; O'Shea,J. J.: Stat4 is expressed in activated peripheral blood monocytes,dendritic cells, and macrophages at sites of Th1–mediated inflammation. J.Immun. 164: 4659–4664, 2000.

[30962] 4514.Loughna, S.; Sato, T. N.: A combinatorial role of angiopoietin–1and orphan receptor TIE1 pathways in estab–

lishing vascular polarity during angiogenesis. *Molec. Cell* 7: 233–239, 2001.

- [30963] 4515. Sato, T. N.; Tozawa, Y.; Deutsch, U.; Wolburg–Buchholz, K.; Fujiwara, Y.; Gendron–Maguire, M.; Gridley, T.; Wolburg, H.; Risau, W.; Qin, Y.: Distinct roles of the receptor tyrosine kinases Tie–1 and Tie–2 in blood vessel formation. *Nature* 376: 70–74, 1995.
- [30964] 4516. Tafuri, A.; Shahinian, A.; Bladt, F.; Yoshinaga, S. K.; Jordana, M.; Wakeham, A.; Boucher, L.–M.; Bouchard, D.; Chan, V. S. F.; Duncan, G.; Odermatt, B.; Ho, A.; Itie, A.; Horan, T.; Whoriskey, J. S.; Pawson, T.; Penninger, J. M.; Ohashi, P. S.; Mak, T. W.: ICOS is essential for effective T–helper–cell responses. *Nature* 409: 105–109, 2001.
- [30965] 4517. Yoshinaga, S. K.; Whoriskey, J. S.; Khare, S. D.; Sarmiento, U.; Guo, J.; Horan, T.; Shih, G.; Zhang, M.; Coccia, M. A.; Kohno, T.; Tafuri–Bladt, A.; Brankow, D.; and 14 others: T–cell co–stimulation through B7RP–1 and ICOS. *Nature* 402: 827–832, 1999.
- [30966] 4518. Paige, A. J. W.; Taylor, K. J.; Taylor, C.; Hillier, S. G.; Farrington, S.; Scott, D.; Porteous, D. J.; Smyth, J. F.; Gabra, H.; Watson, J. E. V.: WWOX: a candidate tumor suppressor gene involved in multiple tumor types. *Proc. Nat. Acad. Sci.* 98: 11417–11422, 2001.

- [30967] 4519.Ried, K.; Finnis, M.; Hobson, L.; Mangelsdorf, M.; Dayan, S.; Nancarrow, J. K.; Woollatt, E.; Kremmidiotis, G.; Gardner, A.; Venter, D.; Baker, E.; Richards, R. I.: Common chromosomal fragile site FRA16D sequence: identification of the FOR gene spanning FRA16D and homozygous deletions and translocation breakpoints in cancer cells. *Hum. Molec. Genet.* 9:1651–1663, 2000.
- [30968] 4520.Clark, S. W.; Staub, O.; Clark, I. B.; Holzbaur, E. L. F.; Paschal, B. M.; Vallee, R. B.; Meyer, D. I.: Beta-centractin: characterization and distribution of a new member of the centractin family of actin-related proteins. *Molec. Biol. Cell* 5: 1301–1310, 1994.
- [30969] 4521.Elsea, S. H.; Clark, I. B.; Juyal, R. C.; Meyer, D. J.; Meyer, D. I.; Patel, P. I.: Assignment of beta-centractin (CTRN2) to human chromosome 2 bands q11.1–q11.2 with somatic cell hybrids and in situ hybridization. *Cytogenet. Cell Genet.* 84: 48–49, 1999.
- [30970] 4522.Ho, A. M.; Johnson, M. D.; Kingsley, D. M.: Role of the mouse ank gene in control of tissue calcification and arthritis. *Science* 289:265–270, 2000.
- [30971] 4523.Yu, H.; Peters, J.-M.; King, R. W.; Page, A. M.; Hieter, P.; Kirschner, M. W.: Identification of a cullin homology region in a subunit of the anaphase-promoting complex.

Science 279: 1219–1222, 1998.

- [30972] 4524. Zhao, N.; Lai, F.; Fernald, A. A.; Eisenbart, J. D.; Espinosa, R., III.; Wang, P. W.; Le Beau, M. M.: Human CDC23: cDNA cloning, mapping to 5q31, genomic structure, and evaluation as a candidate tumor suppressor gene in myeloid leukemias. *Genomics* 53: 184–190, 1998.
- [30973] 4525. Brambilla, R.; Draetta, G.: Molecular cloning of PISSLRE, a novel putative member of the cdk family of protein serine/threonine kinases. *Oncogene* 9:3037–3041, 1994.
- [30974] 4526. Grana, X.; Claudio, P. P.; De Luca, A.; Sang, N.; Giordano, A.: PISSLRE, a human novel CDC2–related protein kinase. *Oncogene* 9:2097–2103, 1994.
- [30975] 4527. Ma, W.-J.; Cheng, S.; Campbell, C.; Wright, A.; Furneaux, H.: Cloning and characterization of HuR, a ubiquitously expressed Elav–like protein. *J. Biol. Chem.* 271: 8144–8151, 1996.
- [30976] 4528. Ma, W.-J.; Furneaux, H.: Localization of the human HuR gene to chromosome 19p13.2. *Hum. Genet.* 99: 32–33, 1997.
- [30977] 4529. Beaudet, A. L.; O'Brien, W. E.; Bock, H.-G. O.; Freytag, S. O.; Su, T.-S.: The human argininosuccinate synthetase locus and citrullinemia. *Adv. Hum. Genet.* 15:

161–196, 1986.

- [30978] 4530. Beaudet, A. L.; Su, T.-S.; Bock, H.-G.; D'Eustachio, P.; Ruddle, F. H.; O'Brien, W. E.: Use of a cloned cDNA to study human argininosuccinatesynthetase. (Abstract) Am. J. Hum. Genet. 33: 36A only, 1981.
- [30979] 4531. Beaudet, A. L.; Su, T.-S.; O'Brien, W. E.; D'Eustachio, P.; Barker, P. E.; Ruddle, F. H.: Dispersion of argininosuccinate-synthetase-like human genes to multiple autosomes and the X chromosome. Cell 30:287–293, 1982.
- [30980] 4532. Daiger, S. P.; Wildin, R. S.; Su, T.-S.: Sequences on the human Y chromosome homologous to the autosomal gene for argininosuccinatesynthetase. Nature 298: 682–684, 1982.
- [30981] 4533. Underwood, K. W.; Song, C.; Kriz, R. W.; Chang, X. J.; Knopf, J. L.; Lin, L.-L.: A novel calcium-independent phospholipase A(2), cPLA(2)-gamma, that is prenylated and contains homology to cPLA(2). J. Biol. Chem. 273:21926–21932, 1998.
- [30982] 4534. Deak, M.; Clifton, A. D.; Lucocq, J. M.; Alessi, D. R.: Mitogen- and stress-activated protein kinase-1 (MSK1) is directly activated by MAPK and SAPK2/p38, and may mediate activation of CREB. EMBO J. 17:4426–4441, 1998.
- [30983] 4535. Jiang, C.; Yu, L.; Tu, Q.; Zhao, Y.; Zhang, H.; Zhao,

S.: Assignment of a member of the ribosomal protein S6 kinase family, RPS6KA5, to human chromosome 14q31–q32.1 by radiation hybrid mapping. *Cytogenet. Cell Genet.* 87: 261–261, 1999.

[30984] 4536. Ackerman, S. L.; Knowles, B. B.: Cloning and mapping of the UNC5C gene to human chromosome 4q21–q23. *Genomics* 52: 205–208, 1998.

[30985] 4537. Ackerman, S. L.; Kozak, L. P.; Przyborski, S. A.; Rund, L. A.; Boyer, B. B.; Knowles, B. B.: The mouse rostral cerebellar malformation gene encodes an UNC-5-like protein. *Nature* 386: 838–842, 1997.

[30986] 4538. Lane, P. W.; Bronson, R. T.; Spencer, C. A.: Rostral cerebellar malformation, (rcm): a new recessive mutation on chromosome 3 of the mouse. *J. Hered.* 83: 315–318, 1992.

[30987] 4539. Leonardo, E. D.; Hinck, L.; Masu, M.; Keino-Masu, K.; Ackerman, S. L.; Tessier-Lavigne, M.: Vertebrate homologues of *C. elegans* UNC-5 are candidate netrin receptors. *Nature* 386: 833–838, 1997.

[30988] 4540. Przyborski, S. A.; Knowles, B. B.; Ackerman, S. L.: Embryonic phenotype of *Unc5h3* mutant mice suggests chemorepulsion during the formation of the rostral cerebellar boundary. *Development* 125: 41–50, 1998.

- [30989] 4541.Marsters, S. A.; Sheridan, J. P.; Pitti, R. M.; Huang, A.; Skubatch,M.; Baldwin, D.; Yuan, J.; Gurney, A.; Goddard, A. D.; Godowski, P.;Ashkenazi, A.: A novel receptor for Apo2L/TRAIL contains a truncateddeath domain. Curr. Biol. 7: 1003–1006, 1997.
- [30990] 4542.Pan, G.; O'Rourke, K.; Chinnaiyan, A. M.; Gentz, R.; Ebner, R.;Ni, J.; Dixit, V. M.: The receptor for the cytotoxic ligand TRAIL. Science 276:111–113, 1997.
- [30991] 4543.MacFarlane, M.; Ahmad, M.; Srinivasula, S. M.; Fernandes–Alnemri,T.; Cohen, G. M.; Alnemri, E. S.: Identification and molecular cloningof two novel receptors for the cytotoxic ligand TRAIL. J. Biol. Chem. 272:25417–25420, 1997.
- [30992] 4544.Pai, S. I.; Wu, G. S.; Ozoren, N.; Wu, L.; Jen, J.; Sidransky,D.; El–Deiry, W. S.: Rare loss–of–function mutation of a death receptorgene in head and neck cancer. Cancer Res. 58: 3513–3518, 1998.
- [30993] 4545.Pan, G.; Ni, J.; Wei, Y.–F.; Yu, G.; Gentz, R.; Dixit, V. M.:An antagonist decoy receptor and a death domain–containing receptorfor TRAIL. Science 277: 815–818, 1997.
- [30994] 4546.Schneider, P.; Bodmer, J.–L.; Thome, M.; Hofmann, K.; Holler, N.;Tschopp, J.: Characterization of two recep–

tors for TRAIL. FEBS Lett. 416:329–334, 1997.

- [30995] 4547. Srean, G. R.; Mongkolsapaya, J.; Xu, X.-N.; Cowper, A. E.; McMichael, A. J.; Bell, J. I.: TRICK2, a new alternatively spliced receptor that transduces the cytotoxic signal from TRAIL. Curr. Biol. 7:693–696, 1997.
- [30996] 4548. Sheridan, J. P.; Marsters, S. A.; Pitti, R. M.; Gurney, A.; Skubatch, M.; Baldwin, D.; Ramakrishnan, L.; Gray, C. L.; Baker, K.; Wood, W. I.; Goddard, A. D.; Godowski, P.; Ashkenazi, A.: Control of TRAIL-induced apoptosis by a family of signaling and decoy receptors. Science 277:818–821, 1997.
- [30997] 4549. Boeglin, W. E.; Kim, R. B.; Brash, A. R.: A 12R-lipoxygenase in human skin: mechanistic evidence, molecular cloning, and expression. Proc. Nat. Acad. Sci. 95: 6744–6749, 1998.
- [30998] 4550. Sun, D.; McDonnell, M.; Chen, X.-S.; Lakkis, M. M.; Li, H.; Isaacs, S. N.; Elsea, S. H.; Patel, P. I.; Funk, C. D.: Human 12(R)-lipoxygenase and the mouse ortholog: molecular cloning, expression, and gene chromosomal assignment. J. Biol. Chem. 273: 33540–33547, 1998.
- [30999] 4551. Itoh, A.; Miyabayashi, T.; Ohno, M.; Sakano, S.: Cloning and expression of three mammalian homologues of Drosophila slit suggest possible roles for Slit in the for-

mation and maintenance of the nervous system.

Molec.Brain Res. 62: 175–186, 1998.

- [31000] 4552.Nakayama, M.; Nakajima, D.; Nagase, T.; Nomura, N.; Seki, N.; Ohara,O.: Identification of high molecular weight proteins with multipleEGF-like motifs by motif-trap screening. Genomics 51: 27–34, 1998.
- [31001] 4553.Duchateau, P. N.; Pullinger, C. R.; Cho, M. H.; Eng, C.; Kane,J. P.: Apolipoprotein L gene family: tissue-specific expression,splicing, promoter regions; discovery of a new gene. J. Lipid Res. 42:620–630, 2001.
- [31002] 4554.Duchateau, P. N.; Pullinger, C. R.; Orellana, R. E.; Kunitake,S. T.; Naya-Vigne, J.; O'Connor, P. M.; Malloy, M. J.; Kane, J. P.: Apolipoprotein L, a new human high density lipoprotein apolipoproteinexpressed by the pancreas: identification, cloning, characterization,and plasma distribution of apolipoprotein L. J. Biol. Chem. 272:25576–25582, 1997.
- [31003] 4555.Monajemi, H.; Fontijn, R. D.; Pannekoek, H.; Hor-revoets, A. J.G.: The apolipoprotein L gene cluster has emerged recently in evolutionand is expressed in human vascular tissue. Genomics 79: 539–546,2002.
- [31004] 4556.Page, N. M.; Butlin, D. J.; Lomthaisong, K.; Lowry, P. J.: Thehuman apolipoprotein L gene cluster: identification,

classification, and sites of distribution. *Genomics* 74: 71–78, 2001.

- [31005] 4557. Lin, C.-Y.; Huang, P.-H.; Liao, W.-L.; Cheng, H.-J.; Huang, C.-F.; Kuo, J.-C.; Patton, W. A.; Massenburg, D.; Moss, J.; Lee, F.-J. S.: ARL4, an ARF-like protein that is developmentally regulated and localized to nuclei and nucleoli. *J. Biol. Chem.* 275: 37815–37823, 2000.
- [31006] 4558. Schurmann, A.; Breiner, M.; Becker, W.; Huppertz, C.; Kainulainen, H.; Kentrup, H.; Joost, H.-G.: Cloning of two novel ADP-ribosylation factor-like proteins and characterization of their differential expression in 3T3-L1 cells. *J. Biol. Chem.* 269: 15683–15688, 1994.
- [31007] 4559. Schurmann, A.; Koling, S.; Jacobs, S.; Saftig, P.; Kraub, S.; Wennemuth, G.; Kluge, R.; Joost, H.-G.: Reduced sperm count and normal fertility in male mice with targeted disruption of the ADP-ribosylation factor-like 4 (Arl4) gene. *Molec. Cell. Biol.* 22: 2761–2768, 2002.
- [31008] 4560. Lehnert, K.; Ni, J.; Leung, E.; Gough, S. M.; Weaver, A.; Yao, W.-P.; Liu, D.; Wang, S.-X.; Morris, C. M.; Krisansen, G. W.: Cloning, sequence analysis, and chromosomal localization of the novel human integrin alpha-11 subunit (ITGA11). *Genomics* 60: 179–187, 1999.
- [31009] 4561. Velling, T.; Kusche-Gullberg, M.; Sejersen, T.; Gull-

berg, D.:cDNA cloning and chromosomal localization of human alpha-11 integrin:a collagen-binding, I domain-containing, beta-1-associated integrinalpha-chain present in muscle tissues. J. Biol. Chem. 274: 25735-25742,1999.

[31010] 4562.Mital, R.; Kobayashi, R.; Hernandez, N.: RNA polymerase III transcription from the human U6 and adenovirus type 2 VAI promoters has different requirements for human BRF, a subunit of human TFIIIB. Molec. Cell.Biol. 16: 7031-7042, 1996.

[31011] 4563.Wang, Z.; Roeder, R. G.: Structure and function of a human transcription factor TFIIIB subunit that is evolutionarily conserved and contains both TFIIIB- and high-mobility-group protein 2-related domains. Proc.Nat. Acad. Sci. 92: 7026-7030, 1995.

[31012] 4564.Comai, L.; Zomerdijs, J. C. B. M.; Beckmann, H.; Zhou, S.; Admon, A.; Tjian, R.: Reconstitution of transcription factor SL1: exclusive binding of TBP by SL1 or TFIID subunits. Science 266: 1966-1972,1994.

[31013] 4565.Di Pietro, C.; Rapisarda, A.; Amico, V.; Bonaiuto, C.; Viola, A.; Scalia, M.; Motta, S.; Amato, A.; Engel, H.; Messina, A.; Sichel, G.; Grzeschik, K.-H.; Purrello, M.: Genomic localization of the human genes TAF1A, TAF1B and

TAF1C, encoding TAFI48, TAFI63 and TAFI110 subunits of class I general transcription initiation factor SL1. Cytogenet. Cell Genet. 89: 133–136, 2000.

[31014] 4566. Nakamura, T.; Takeuchi, K.; Muraoka, S.; Takezoe, H.; Takahashi, N.; Mori, N.: A neurally enriched coronin-like protein, ClipnC, is a novel candidate for an actin cytoskeleton–cortical membrane–linking protein. J. Biol. Chem. 274: 13322–13327, 1999.

[31015] 4567. Kim, K. I.; Baek, S. H.; Jeon, Y.-J.; Nishimori, S.; Suzuki, T.; Uchida, S.; Shimbara, N.; Saitoh, H.; Tanaka, K.; Chung, C. H.: A new SUMO-1-specific protease, SUSP1, that is highly expressed in reproductive organs. J. Biol. Chem. 275: 14102–14106, 2000.

[31016] 4568. Ali, A.; Hoeflich, K. P.; Woodgett, J. R. Glycogen synthase kinase-3: properties, functions, and regulation. Chem. Rev. 101: 2527–2540, 2001.

[31017] 4569. Dajani, R.; Fraser, E.; Roe, S. M.; Young, N.; Good, V.; Dale, T. C.; Pearl, L. H.: Crystal structure of glycogen synthase kinase3-beta: structural basis for phosphate-primed substrate specificity and autoinhibition. Cell 105: 721–732, 2001.

[31018] 4570. Frame, S.; Cohen, P.; Biondi, R. M.: A common phosphate binding site explains the unique substrate specificity

of GSK3 and its inactivation by phosphorylation. *Molec. Cell* 7: 1321–1327, 2001.

- [31019] 4571. Hoeflich, K. P.; Luo, J.; Rubie, E. A.; Tsao, M.-S.; Jin, O.; Woodgett, J. R.: Requirement for glycogen synthase kinase-3-beta in cell survival and NF-kappa-B activation. *Nature* 406: 86–90, 2000.
- [31020] 4572. Lau, K. F.; Miller, C. C.; Anderton, B. H.; Shaw, P. C.: Molecular cloning and characterization of the human glycogen synthase kinase-3-beta promoter. *Genomics* 60: 121–128, 1999.
- [31021] 4573. Lau, K. F.; Miller, C. C.; Anderton, B. H.; Shaw, P. C.: Expression analysis of glycogen synthase kinase-3 in human tissues. *J. Peptide Res.* 54: 85–91, 1999.
- [31022] 4574. Lucas, J. J.; Hernandez, F.; Gomez-Ramos, P.; Moran, M. A.; Hen, R.; Avila, J.: Decreased nuclear beta-catenin, tau hyperphosphorylation and neurodegeneration in GSK-3-beta conditional transgenic mice. *EMBO J.* 20: 27–39, 2001.
- [31023] 4575. Plyte, S. E.; Hughes, K.; Nikolakaki, E.; Pulverer, B. J.; Woodgett, J. R.: Glycogen synthase kinase-3: functions in oncogenesis and development. *Biochim. Biophys. Acta* 1114: 147–162, 1992.
- [31024] 4576. Shaw, P. C.; Davies, A. F.; Lau, K. F.; Garcia-Barcelo,

M.; Waye, M. M.; Lovestone, S.; Miller, C. C.; Anderton, B. H.: Isolation and chromosomal mapping of human glycogen synthase kinase-3 alpha and 3 beta encoding genes. *Genome* 41: 720-727, 1998.

[31025] 4577. Stambolic, V.; Woodgett, J. R.: Mitogen inactivation of glycogen synthase kinase-3 beta in intact cells via serine 9 phosphorylation. *Biochem. J.* 303: 701-704, 1994.

[31026] 4578. Wang, Q. M.; Fiol, C. J.; DePaoli-Roach, A. A.; Roach, P. J.: Glycogen synthase kinase-3 beta is a dual specificity kinase differentially regulated by tyrosine and serine/threonine phosphorylation. *J. Biol. Chem.* 269: 14566-14574, 1994.

[31027] 4579. Saitoh, T.; Moriwaki, J.; Koike, J.; Takagi, A.; Miwa, T.; Shiokawa, K.; Katoh, M.: Molecular cloning and characterization of FRAT2, encoding a positive regulator of the WNT signaling pathway. *Biochem. Biophys. Res. Commun.* 281: 815-820, 2001.

[31028] 4580. Abbaszade, I.; Liu, R.-Q.; Yang, F.; Rosenfeld, S. A.; Ross, O. H.; Link, J. R.; Ellis, D. M.; Tortorella, M. D.; Pratta, M. A.; Hollis, J. M.; Wynn, R.; Duke, J. L.; and 15 others: Cloning and characterization of ADAMTS11, an aggrecanase from the ADAMTS family. *J. Biol. Chem.* 274: 23443-23450, 1999.

- [31029] 4581.Duan, H.; Wang, Y.; Aviram, M.; Swaroop, M.; Loo, J. A.; Bian,J.; Tian, Y.; Mueller, T.; Bisgaier, C. L.; Sun, Y.: SAG, a novelzinc RING finger protein that protects cells from apoptosis inducedby redox agents. *Molec. Cell. Biol.* 19: 3145–3155, 1999.
- [31030] 4582.Murakami, A.; Yajima, T.; Sakuma, H.; McLaren, M. J.; Inana, G.: X-arrestin: a new retinal arrestin mapping to the X chromosome. *FEBSLett.* 334: 203–209, 1993.
- [31031] 4583.Sakuma, H.; Murakami, A.; Fujimaki, T.; Inana, G.: Isolation andcharacterization of the human X-arrestin gene. *Gene* 224: 87–95,1998.
- [31032] 4584.Ellison, J.; Passage, M.; Yu, L.–C.; Yen, P.; Mohandas, T. K.;Shapiro, L.: Directed isolation of human genes that escape X inactivation. *Somat.Cell Molec. Genet.* 18: 259–268, 1992.
- [31033] 4585.Ellison, J. W.; Ramos, C.; Yen, P. H.; Shapiro, L. J.: Structureand expression of the human pseudoautosomal gene XE7. *Hum. Molec.Genet.* 1: 691–696, 1992.
- [31034] 4586.Amar, L. C.; Dandolo, L.; Hanauer, A.; Cook, A. R.; Arnaud, D.;Mandel, J. L.; Avner, P.: Conservation and reorganization of locion the mammalian X chromosome: a molecular framework for the identificationof homologous subchromosomal regions in man and mouse. *Genomics*

2:220–230, 1988.

- [31035] 4587.Chin, L.–S.; Li, L.; Ferreira, A.; Kosik, K. S.; Greengard, P.: Impairment of axonal development and of synaptogenesis in hippocampal neurons of synapsin I–deficient mice. *Proc. Nat. Acad. Sci.* 92:9230–9234, 1995.
- [31036] 4588.Klagges, B. R. E.; Heimbeck, G.; Godenschwege, T. A.; Hofbauer, A.; Pflugfelder, G. O.; Reifegerste, R.; Reisch, D.; Schaupp, M.; Buchner, S.; Buchner, E.: Invertebrate synapsins: a single gene codes for several isoforms in *Drosophila*. *J. Neurosci.* 16: 3154–3165, 1996.
- [31037] 4589.Li, L.; Chin, L.–S.; Shupliakov, O.; Brodin, L.; Sihra, T. S.; Hvalby, O.; Jensen, V.; Zheng, D.; McNamara, J. O.; Greengard, P.; Andersen, P.: Impairment of synaptic vesicle clustering and of synaptic transmission, and increased seizure propensity, in synapsin I–deficient mice. *Proc. Nat. Acad. Sci.* 92: 9235–9239, 1995.
- [31038] 4590.Sudhof, T. C.: The structure of the human synapsin I gene and protein. *J. Biol. Chem.* 265: 7849–7852, 1990.
- [31039] 4591.Banting, G. S.; Pym, B.; Goodfellow, P. N.: Biochemical analysis of an antigen produced by both human sex chromosomes. *EMBO J.* 4:1967–1972, 1985.
- [31040] 4592.Hagiwara, T.; Tanaka, K.; Takai, S.; Maeno–Hikichi, Y.; Mukainaka, Y.; Wada, K.: Genomic organization, pro–

moter analysis, and chromosomal localization of the gene for the mouse glial high-affinity glutamate transporter Slc1a3. *Genomics* 33: 508–515, 1996.

[31041] 4593. Harada, T.; Harada, C.; Watanabe, M.; Inoue, Y.; Sakagawa, T.; Nakayama, N.; Sasaki, S.; Okuyama, S.; Watase, K.; Wada, K.; Tanaka, K.: Functions of the two glutamate transporters GLAST and GLT-1 in the retina. *Proc. Nat. Acad. Sci.* 95: 4663–4666, 1998.

[31042] 4594. Keppen, L. D.; Gollin, S. M.; Edwards, D.; Sawyer, J.; Wilson, W.; Overhauser, J.: Clinical phenotype and molecular analysis of a three-generation family with an interstitial deletion of the short arm of chromosome 5. *Am. J. Med. Genet.* 44: 356–360, 1992.

[31043] 4595. Kirschner, M. A.; Arriza, J. L.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Magenis, E.; Amara, S. G.: The mouse and human excitatory amino acid transporter gene (EAAT1) maps to mouse chromosome 15 and a region of syntenic homology on human chromosome 5. *Genomics* 22: 631–633, 1994.

[31044] 4596. Shashidharan, P.; Huntley, G. W.; Meyer, T.; Morrison, J. H.; Plaitakis, A.: Neuron-specific human glutamate transporter: molecular cloning, characterization and expression in human brain. *Brain Res.* 662: 245–250, 1994.

- [31045] 4597.Stoffel, W.; Sasse, J.; Duker, M.; Muller, R.; Hofmann, K.; Fink,T.; Lichter, P.: Human high affinity, Na(+)-dependent L-glutamate/L-aspartatetransporter GLAST-1 (EAAT-1): gene structure and localization to chromosome5p11-p12. FEBS Lett. 386: 189-193, 1996.
- [31046] 4598.Takai, S.; Yamada, K.; Kawakami, H.; Tanaka, K.; Nakamura, S.:Localization of the gene (SLC1A3) encoding human glutamate transporter(GluT-1) to 5p13 by fluorescence in situ hybridization. Cytogenet.Cell Genet. 69: 209-210, 1995.
- [31047] 4599.Agulnik, A. I.; Mitchell, M. J.; Lerner, J. L.; Woods, D. R.; Bishop,C. E.: A mouse Y chromosome gene encoded by a region essential forspermatogenesis and expression of male-specific minor histocompatibilityantigens. Hum. Molec. Genet. 3: 873-878, 1994.
- [31048] 4600.Agulnik, A. I.; Mitchell, M. J.; Mattei, M.-G.; Borsani, G.; Avner,P. A.; Lerner, J. L.; Bishop, C. E.: A novel X gene with a widelytranscribed Y-linked homologue escapes X-inactivation in mouse andhuman. Hum. Molec. Genet. 3: 879-884, 1994.
- [31049] 4601.Lingenfelter, P. A.; Adler, D. A.; Poslinski, D.; Thomas, S.; Elliott,R. W.; Chapman, V. M.; Disteché, C. M.: Escape from X inactivationof Smcx is preceded by silenc-

ing during mouse development. NatureGenet. 18:
212–213, 1998.

- [31050] 4602.Wu, J.; Ellison, J.; Salido, E.; Yen, P.; Mohandas, T.; Shapiro, L. J.: Isolation and characterization of XE169, a novel human gene that escapes X-inactivation. Hum. Molec. Genet. 3: 153–160, 1994.
- [31051] 4603.Wu, J.; Salido, E. C.; Yen, P. H.; Mohandas, T. K.; Heng, H. H.Q.; Tsui, L.-C.; Park, J.; Chapman, V. M.; Shapiro, L. J.: The murine Xe169 gene escapes X-inactivation like its human homologue. NatureGenet. 7: 491–496, 1994.
- [31052] 4604.Evans, H. J.; Buckton, K. E.; Spowart, G.; Carothers, A. D.: Heteromorphic X chromosomes in 46,XX males: evidence for the involvement of X–Y interchange. Hum. Genet. 49: 11–31, 1979.
- [31053] 4605.Hu, X.; Burghes, A. H. M.; Ray, P. N.; Thompson, M. W.; Murphy, E. G.; Worton, R. G.: Partial gene duplication in Duchenne and Becker muscular dystrophies. J. Med. Genet. 25: 369–376, 1988.
- [31054] 4606.Berger, W.; van de Pol, D.; Warburg, M.; Gal, A.; Bleeker-Wagemakers, L.; de Silva, H.; Meindl, A.; Meitinger, T.; Cremers, F.; Ropers, H.-H.: Mutations in the candidate gene for Norrie disease. Hum. Molec. Genet. 1: 461–465,

1992.

- [31055] 4607. Bleeker–Wagemakers, L. M.; Friedrich, U.; Gal, A.; Wienker, T.F.; Warburg, M.; Ropers, H.–H.: Close linkage between Norrie disease, a cloned DNA sequence from the proximal short arm, and the centromere of the X chromosome. *Hum. Genet.* 71: 211–214, 1985.
- [31056] 4608. Chen, Z.–Y.; Battinelli, E. M.; Hendriks, R. W.; Powell, J. F.; Middleton–Price, H.; Sims, K. B.; Breakefield, X. O.; Craig, I. W.: Norrie disease gene: characterization of deletions and possible function. *Genomics* 16: 533–535, 1993.
- [31057] 4609. Chen, Z.–Y.; Battinelli, E. M.; Woodruff, G.; Young, I.; Breakefield, X. O.; Craig, I. W.: Characterization of a mutation within the NDP gene in a family with a manifesting female carrier. *Hum. Molec. Genet.* 2: 1727–1729, 1993.
- [31058] 4610. Chen, Z.–Y.; Hendriks, R. W.; Jobling, M. A.; Powell, J. F.; Breakefield, X. O.; Sims, K. B.; Craig, I. W.: Isolation and characterization of a candidate gene for Norrie disease. *Nature Genet.* 1: 204–208, 1992.
- [31059] 4611. Chen, Z.–Y.; Sims, K. B.; Coleman, M.; Donnai, D.; Monaco, A.; Breakefield, X. O.; Davies, K. E.; Craig, I. W.: Characterization of a YAC containing part or all of the Norrie disease locus. *Hum. Molec. Genet.* 1: 161–164, 1992.
- [31060] 4612. Clarke, E.: 'Pseudo–glioma' in both eyes. *Trans.*

Ophthal. Soc.U.K. 18: 136–138, 1898.

- [31061] 4613.Dahlberg–Parrow, R.: Congenital sex–linked pseudoglioma and gravitational deficiency. Acta Ophthal. 34: 250–254, 1956.
- [31062] 4614.Diergaarde, P. J.; Wieringa, B.; Bleeker–Wagemakers, E. M.; Sims,K. B.; Breakefield, X. O.; Ropers, H.–H.: Physical fine–mapping of a deletion spanning the Norrie gene. Hum. Genet. 84: 22–26, 1989.
- [31063] 4615.Donnai, D.; Mountford, R. C.; Read, A. P.: Norrie disease resulting from a gene deletion: clinical features and DNA studies. J. Med.Genet. 25: 73–78, 1988.
- [31064] 4616.Duke–Elder, J. R.: Pseudoglioma in children: aspects of clinical and pathological diagnosis. Sth. Med. J. 51: 754–759, 1958.
- [31065] 4617.Forssman, H.: Mental deficiency and pseudoglioma, a syndrome inherited as an X–linked recessive. Am. J. Ment. Defic. 64: 984–987, 1960.
- [31066] 4618.Fuchs, S.; Kellner, U.; Wedemann, H.; Gal, A.: Missense mutation(Arg121Trp) in the Norrie disease gene associated with X–linked exudative vitreoretinopathy. Hum. Mutat. 6: 257–259, 1995.
- [31067] 4619.Fuchs, S.; Xu, S. Y.; Caballero, M.; Salcedo, M.; La O, A.; Wedemann,H.; Gal, A.: A missense point mutation

(Leu13Arg) of the Norrie disease gene in a large Cuban kindred with Norrie disease. *Hum. Molec. Genet.* 3:655–656, 1994.

[31068] 4620. Collins, F. A.; Murphy, D. L.; Reiss, A. L.; Sims, K. B.; Lewis, J. G.; Freund, L.; Karoum, F.; Zhu, D.; Maumenee, I. H.; Antonarakis, S. E.: Clinical, biochemical, and neuropsychiatric evaluation of a patient with a contiguous gene syndrome due to a microdeletion Xp11.3 including the Norrie disease locus and monoamine oxidase (MAOA and-MAOB) genes. *Am. J. Med. Genet.* 42: 127–134, 1992.

[31069] 4621. Fullwood, P.; Jones, J.; Bunday, S.; Dudgeon, J.; Fielder, A. R.; Kilpatrick, M. W.: X linked exudative vitreoretinopathy: clinical features and genetic linkage analysis. *Brit. J. Ophthalmol.* 77: 168–170, 1993.

[31070] 4622. Gal, A.; Bleeker-Wagemakers, L.; Wienker, T. F.; Warburg, M.; Ropers, H.-H.: Localization of the gene for Norrie disease by linkage to the DXS7 locus. (Abstract) *Cytogenet. Cell Genet.* 40: 633, 1985.

[31071] 4623. Gal, A.; Stolzenberger, C.; Wienker, T.; Wieacker, P.; Ropers, H.-H.; Friedrich, U.; Bleeker-Wagemakers, L.; Pearson, P.; Warburg, M.: Norrie's disease: close linkage with genetic markers from the proximal short arm of the X chromosome. *Clin. Genet.* 27: 282–283, 1985.

- [31072] 4624.Gal, A.; Uhlhaas, S.; Glaser, D.; Grimm, T.: Prenatal exclusion of Norrie disease with flanking DNA markers. *Am. J. Med. Genet.* 31:449–453, 1988.
- [31073] 4625.Harendra de Silva, D. G.; de Silva, D. B. K.: Norrie's disease in an Asian family. *Brit. J. Ophthal.* 72: 62–64, 1988.
- [31074] 4626.Berminham, N. A.; Martin, J. E.; Fisher, E. M. C.: The mouse lysosomal membrane protein 1 gene as a candidate for the motor neuron degeneration (mnd) locus. *Genomics* 32: 266–271, 1996.
- [31075] 4627.Howe, C. L.; Granger, B. L.; Hull, M.; Green, S. A.; Gabel, C.A.; Helenius, A.; Mellman, I.: Derived protein sequence, oligosaccharides, and membrane insertion of the 120-kDa lysosomal membrane glycoprotein (lgp120): identification of a highly conserved family of lysosomal membrane glycoproteins. *Proc. Nat. Acad. Sci.* 85: 7577–7581, 1988.
- [31076] 4628.Mattei, M.-G.; Matterson, J.; Chen, J. W.; Williams, M. A.; Fukuda, M.: Two human lysosomal membrane glycoproteins, h-lamp-1 and h-lamp-2, are encoded by genes localized to chromosome 13q34 and chromosome Xq24–25, respectively. *J. Biol. Chem.* 265: 7548–7551, 1990.

- [31077] 4629.Schleutker, J.; Haataja, L.; Renlund, M.; Puhakka, L.; Viitala,J.; Peltonen, L.; Aula, P.: Confirmation of the chromosomal localizationof human lamp genes and their exclusion as candidate genes for Salladisease. Hum. Genet. 88: 95–97, 1991.
- [31078] 4630.Viitala, J.; Carlsson, S. R.; Siebert, P. D.; Fukuda, M.: Molecularcloning of cDNAs encoding lamp A, a human lysosomal membrane glycoproteinwith apparent M(r) about 120,000. Proc. Nat. Acad. Sci. 85: 3743–3747,1988.
- [31079] 4631.Andreae, S.; Piras, F.; Burdin, N.; Triebel, F.: Maturation andactivation of dendritic cells induced by lymphocyte activation gene–3(CD223). J. Immun. 168: 3874–3880, 2002.
- [31080] 4632.Triebel, F.; Jitsukawa, S.; Baixeras, E.; Roman–Roman, S.; Genevee,C.; Viegas–Pequignot, E.; Hercend, T.: LAG–3, a novel lymphocyteactivation gene closely related to CD4. J. Exp. Med. 171: 1393–1405,1990.
- [31081] 4633.LeClair, K. P.; Rabin, M.; Nesbitt, M. N.; Pravtcheva, D.; Ruddle,F. H.; Palfree, R. G. E.; Bothwell, A.: Murine Ly–6 multigene familyis located on chromosome 15. Proc. Nat. Acad. Sci. 84: 1638–1642,1987.
- [31082] 4634.Ravetch, J. V.; Lanier, L. L.: Immune inhibitory recep–

tors. Science 290:84–89, 2000.

- [31083] 4635.Fischer, A.; Durandy, A.; Sterkers, G.; Griscelli, C.: Role of the LFA–1 molecule in cellular interactions required for antibody production in humans. J. Immun. 136: 3198–3203, 1986.
- [31084] 4636.Marlin, S. D.; Morton, C. C.; Anderson, D. C.; Springer, T. A.: LFA–1 immunodeficiency disease: definition of the genetic defect and chromosomal mapping of alpha and beta subunits of the lymphocyte function–associated antigen 1 (LFA–1) by complementation in hybrid cells. J. Exp. Med. 164: 855–867, 1986.
- [31085] 4637.Futscher, B. W.; Oshiro, M. M.; Wozniak, R. J.; Holtan, N.; Hanigan, C. L.; Duan, H.; Domann, F. E.: Role for DNA methylation in the control of cell type–specific maspin expression. Nature Genet. 31: 175–179, 2002.
- [31086] 4638.Ngamkitidechakul, C.; Burke, J. M.; O'Brien, W. J.; Twining, S.S.: Maspin: synthesis by human cornea and regulation of in vitro stromal cell adhesion to extracellular matrix. Invest. Ophthalmol. Vis.Sci. 42: 3135–3141, 2001.
- [31087] 4639.Zou, Z.; Anisowicz, A.; Hendrix, M. J. C.; Thor, A.; Neveu, M.; Sheng, S.; Rafidi, K.; Seftor, E.; Sager, R.: Maspin, a serpin with tumor–suppressing activity in human mammary epithelial cells. Science 263:526–529, 1994.

- [31088] 4640.Blackwood, E.; Eisenman, R. N.: Max: a helix-loop-helix zipperprotein that forms a sequence-specific DNA-binding complex with Myc. *Science* 251:1211-1217, 1991.
- [31089] 4641.Eisenman, R. N.: Personal Communication. Seattle, Wash. 7/27/1994.
- [31090] 4642.Gilladoga, A. D.; Edelhoff, S.; Blackwood, E. M.; Eisenman, R.N.; Disteché, C. M.: Mapping of MAX to human chromosome 14 and mousechromosome 12 by in situ hybridization. *Oncogene* 7: 1249-1251, 1992.
- [31091] 4643.Loeffen, J. L. C. M.; Triepels, R. H.; van den Heuvel, L. P.; Schuelke,M.; Buskens, C. A. F.; Smeets, R. J. P.; Trijbels, J. M. F.; Smeitink,J. A. M.: cDNA of eight nuclear encoded subunits of NADH:ubiquinoneoxidoreductase: human complex I cDNA characterization completed. *Biochem.Biophys. Res. Commun.* 253: 415-422, 1998.
- [31092] 4644.Bulavin, D. V.; Higashimoto, Y.; Popoff, I. J.; Gaarde, W. A.;Basrur, V.; Potapova, O.; Appella, E.; Fornace, A. J., Jr.: Initiationof a G2/M checkpoint after ultraviolet radiation requires p38 kinase. *Nature* 411:102-107, 2001.
- [31093] 4645.Gould, K. L.; Moreno, S.; Tonks, N. K.; Nurse, P.: Complementationof the mitotic activator, p80(cdc25), by a human protein-tyrosinephosphatase. *Science* 250:

1573–1576, 1990.

- [31094] 4646. Moreno, S.; Nurse, P.: Clues to action of cdc25 protein. (Letter) *Nature* 351:194 only, 1991.
- [31095] 4647. Raj, K.; Ogston, P.; Beard, P.: Virus-mediated killing of cells that lack p53 activity. *Nature* 412: 914–917, 2001.
Note: Addendum: *Nature* 416: 202 only, 2002.
- [31096] 4648. Sadhu, K.; Reed, S. I.; Richardson, H.; Russell, P.: Human homolog of fission yeast cdc25 mitotic inducer is predominantly expressed in G(2). *Proc. Nat. Acad. Sci.* 87: 5139–5143, 1990.
- [31097] 4649. Strausfeld, U.; Labbe, J. C.; Fesquet, D.; Cavadore, J. C.; Picard, A.; Sadhu, K.; Russell, P.; Doree, M.: Dephosphorylation and activation of a p34(cdc2)/cyclin B complex in vitro by human CDC25 protein. *Nature* 351:242–245, 1991.
- [31098] 4650. Taviaux, S. A.; Demaille, J. G.: Localization of human cell cycle regulatory genes CDC25C to 5q31 and WEE1 to 11p15.3–11p.15.1 by fluorescence in situ hybridization. *Genomics* 15: 194–196, 1993.
- [31099] 4651. Willman, C. L.; Sever, C. E.; Pallavicini, M. G.; Harada, H.; Tanaka, N.; Slovak, M. L.; Yamamoto, H.; Harada, K.; Meeker, T. C.; List, A. F.; Taniguchi, T.: Deletion of IRF-1, mapping to chromosome 5q31.1, in human leukemia and

preleukemic myelodysplasia. *Science* 259: 968–971,1993.

- [31100] 4652. Dear, T. N.; Campbell, K.; Rabbitts, T. H.: Molecular cloning of putative odorant-binding and odorant-metabolizing proteins. *Biochemistry* 30:10376–10382, 1991.
- [31101] 4653. Lacazette, E.; Gachon, A. M.; Pitiot, G.: A novel human odorant-binding protein gene family resulting from genomic duplicons at 9q34: differential expression in the oral and genital spheres. *Hum. Molec. Genet.* 9:289–301, 2000.
- [31102] 4654. Pevsner, J.; Reed, R. R.; Feinstein, P. G.; Snyder, S. H.: Molecular cloning of odorant-binding protein: member of a ligand carrier family. *Science* 241:336–339, 1988.
- [31103] 4655. Raming, K.; Krieger, J.; Strotmann, J.; Boekhoff, I.; Kubick, S.; Baumstark, C.; Breer, H.: Cloning and expression of odorant receptors. *Nature* 361:353–356, 1993.
- [31104] 4656. Unterman, R. D.; Lynch, K. R.; Nakhasi, H. L.; Dolan, K. P.; Hamilton, J. W.; Cohn, D. V.; Feigelson, P.: Cloning and sequence of several alpha-2-microglobulin cDNAs. *Proc. Nat. Acad. Sci.* 78: 3478–3482, 1981.
- [31105] 4657. Miles, S. A.; Martinez-Maza, O.; Rezai, A.; Magpantay, L.; Kishimoto, T.; Nakamura, S.; Radka, S. F.; Linsley, P. S.: Oncostatin M as a potent mitogen for AIDS-Kaposi's

sarcoma-derived cells. *Science* 255:1432–1434, 1992.

- [31106] 4658. Modur, V.; Feldhaus, M. J.; Weyrich, A. S.; Jicha, D. L.; Prescott, S. M.; Zimmerman, G. A.; McIntyre, T. M.: Oncostatin M is a proinflammatory mediator: in vivo effects correlate with endothelial cell expression of inflammatory cytokines and adhesion molecules. *J. Clin. Invest.* 100:158–168, 1997.
- [31107] 4659. Nair, B. C.; DeVico, A. L.; Nakamura, S.; Copeland, T. D.; Chen, Y.; Patel, A.; O'Neil, T.; Oroszlan, S.; Gallo, R. C.; Sarngadharan, M. G.: Identification of a major growth factor for AIDS-Kaposi's sarcoma cells as oncostatin M. *Science* 255: 1430–1432, 1992.
- [31108] 4660. Rose, T. M.; Bruce, A. G.: Oncostatin M is a member of a cytokine family that includes leukemia-inhibitory factor, granulocyte colony-stimulating factor, and interleukin 6. *Proc. Nat. Acad. Sci.* 88: 8641–8645, 1991.
- [31109] 4661. Rose, T. M.; Lagrou, M. J.; Fransson, I.; Werelius, B.; Delattre, O.; Thomas, G.; de Jong, P. J.; Todaro, G. J.; Dumaniski, J. P.: The genes for oncostatin M (OSM) and leukemia inhibitory factor (LIF) are tightly linked on human chromosome 22. *Genomics* 17: 136–140, 1993.
- [31110] 4662. Zarling, J. M.; Shoyab, M.; Marquardt, H.; Hanson, M. B.; Lioubin, M. N.; Todaro, G. J.: Oncostatin M: a growth

regulator produced by differentiated histiocytic lymphoma cells. *Proc. Nat. Acad. Sci.* 83:9739–9743, 1986.

- [31111] 4663. Harder, K. W.; Parsons, L. M.; Armes, J.; Evans, N.; Kountouri, N.; Clark, R.; Quillici, C.; Grail, D.; Hodgson, G. S.; Dunn, A. R.; Hibbs, M. L.: Gain- and loss-of-function Lyn mutant mice define a critical inhibitory role for Lyn in the myeloid lineage. *Immunity* 15:603–615, 2001.
- [31112] 4664. Fujimoto, M.; Kantaputra, P. N.; Ikegawa, S.; Fukushima, Y.; Sonta, S.; Matsuo, M.; Ishida, T.; Matsumoto, T.; Kondo, S.; Tomita, H.; Deng, H.-X.; D'urso, M.; Rinaldi, M. M.; Ventruto, V.; Takagi, T.; Nakamura, Y.; Niiikawa, N.: The gene for mesomelic dysplasia Kantaputratype is mapped to chromosome 2q24–q32. *J. Hum. Genet.* 43: 32–36, 1998.
- [31113] 4665. Ventruto, V.; Pisciotta, R.; Renda, S.; Fosta, B.; Rinaldi, M. M.; Stabile, M.; Cavaliere, M. L.; Esposito, M.: Multiple skeletal familial abnormalities associated with balanced reciprocal translocation 2;8(q32;p13). *Am. J. Med. Genet.* 16: 589–594, 1983.
- [31114] 4666. Joyner, A. L.; Herrup, K.; Auerbach, B. A.; Davis, C. A.; Rossant, J.: Subtle cerebellar phenotype in mice homozygous for a targeted deletion of the *En-2* homeobox. *Science* 251: 1239–1243, 1991.

- [31115] 4667.Joyner, A. L.; Skarnes, W. C.; Rossant, J.: Production of a mutation in mouse En-2 gene by homologous recombination in embryonic stem cells. *Nature* 338:153–156, 1989.
- [31116] 4668.Poole, S. J.; Law, M. L.; Kao, F.-T.; Lau, Y.-F.: Isolation and chromosomal localization of the human En-2 gene. *Genomics* 4: 225–231, 1989.
- [31117] 4669.Kiss, C.; Li, J.; Szeles, A.; Gizatullin, R. Z.; Kashuba, V. I.; Lushnikova, T.; Protopopov, A. I.; Kelve, M.; Kiss, H.; Kholodnyuk, I. D.; Imreh, S.; Klein, G.; Zabarovsky, E. R.: Assignment of the ARHA and GPX1 genes to human chromosome bands 3p21.3 by in situ hybridization and with somatic cell hybrids. *Cytogenet. Cell Genet.* 79: 228–230, 1997.
- [31118] 4670.Ma, J. J.; Nishimura, M.; Mine, H.; Kuroki, S.; Nukina, M.; Ohta, M.; Saji, H.; Obayashi, H.; Kawakami, H.; Saida, T.; Uchiyama, T.: Genetic contribution of the tumor necrosis factor region in Guillain-Barre syndrome. *Ann. Neurol.* 44: 815–818, 1998.
- [31119] 4671.Giuli, G.; Roechel, N.; Scholl, U.; Mattei, M.-G.; Guellaen, G.: Colocalization of the genes coding for the alpha-3 and beta-3 subunits of soluble guanylyl cyclase to human chromosome 4 at q31.3–q33. *Hum. Genet.* 91:

257–260, 1993.

- [31120] 4672.Cameron, H. S.; Szczepaniak, D.; Weston, B. W.: Expression of human chromosome 19p alpha-(1,3)-fucosyltransferase genes in normal tissues: alternative splicing, polyadenylation, and isoforms. *J.Biol. Chem.* 270: 20112–20122, 1995.
- [31121] 4673.Koszdin, K. L.; Bowen, B. R.: The cloning and expression of a human alpha-1,3 fucosyltransferase capable of forming the E-selectin ligand. *Biochem. Biophys. Res. Commun.* 187: 152–157, 1992.
- [31122] 4674.Mollicone, R.; Reguigne, I.; Fletcher, A.; Aziz, A.; Rustam, M.; Weston, B. W.; Kelly, R. J.; Lowe, J. B.; Oriol, R.: Molecular basis for plasma alpha(1,3)-fucosyltransferase gene deficiency (FUT6). *J.Biol. Chem.* 269: 12662–12671, 1994.
- [31123] 4675.Pang, H.; Koda, Y.; Soejima, M.; Schlaphoff, T.; Du Toit, E. D.; Kimura, H.: Allelic diversity of the human plasma alpha(1,3)fucosyltransferase gene (FUT6). *Ann. Hum. Genet.* 63: 277–284, 1999.
- [31124] 4676.Schnyder-Candrian, S.; Borsig, L.; Moser, R.; Berger, E. G.: Localization of alpha-1,3-fucosyltransferase VI in Weibel-Palade bodies of human endothelial cells. *Proc. Nat. Acad. Sci.* 97: 8369–8374, 2000.

- [31125] 4677. Doonan, S.; Barra, D.; Bossa, F.: Structural and genetic relationships between cytosolic and mitochondrial isoenzymes. *Int. J. Biochem.* 16:1193–1199, 1984.
- [31126] 4678. Allen, S. J.; O'Donnell, A.; Alexander, N. D. E.; Alpers, M. P.; Peto, T. E. A.; Clegg, J. B.; Weatherall, D. J.: Alpha(+)-thalassaemia protects children against disease caused by other infections as well as malaria. *Proc. Nat. Acad. Sci.* 94: 14736–14741, 1997.
- [31127] 4679. Keats, B. J. B.; Morton, N. E.; Rao, D. C.: Possible linkages (lod score over 1.5) and a tentative map of the Jk–Km linkage group. *Cytogenet. Cell Genet.* 22: 304–308, 1978.
- [31128] 4680. Keats, B. J. B.; Morton, N. E.; Rao, D. C.: Likely linkage: In V with Jk. *Hum. Genet.* 39: 157–159, 1977.
- [31129] 4681. Davenne, M.; Maconochie, M. K.; Neun, R.; Pattyn, A.; Chambon, P.; Krumlauf, R.; Rijli, F. M.: Hoxa2 and Hoxb2 control dorsoventral patterns of neuronal development in the rostral hindbrain. *Neuron* 22:677–691, 1999.
- [31130] 4682. Pollock, R. A.; Sreenath, T.; Ngo, L.; Bieberich, C. J.: Gain of function mutations for paralogous Hox genes: implications for the evolution of Hox gene function. *Proc. Nat. Acad. Sci.* 92: 4492–4496, 1995.
- [31131] 4683. Vieille-Grosjean, I.; Huber, P.: Transcription factor

GATA-1 regulates human HOXB2 gene expression in erythroid cells. *J. Biol. Chem.* 270:4544–4550, 1995.

[31132] 4684. Cannizzaro, L. A.; Croce, C. M.; Griffin, C. A.; Simone, A.; Boncinelli, E.; Huebner, K.: Human homeobox-containing genes located at chromosomal regions 2q31–2q37 and 12q12–12q13. *Am. J. Hum. Genet.* 41: 1–15, 1987.

[31133] 4685. Chariot, A.; Castronovo, V.; Le, P.; Gillet, C.; Sobel, M. E.; Gielen, J.: Cloning and expression of a new HOXC6 transcript encoding a repressing protein. *Biochem. J.* 319: 91–97, 1996.

[31134] 4686. Gibbs, S.; Fijneman, R.; Wiegant, J.; Geurts van Kessel, A.; vande Putte, P.; Backendorf, C.: Molecular characterization and evolution of the SPRR family of keratinocyte differentiation markers encoding small proline-rich proteins. *Genomics* 16: 630–637, 1993.

[31135] 4687. Marvin, K. W.; George, M. D.; Fujimoto, W.; Saunders, N. A.; Bernacki, S. H.; Jetten, A. M.: Cornifin, a cross-linked envelope precursor in keratinocytes that is down-regulated by retinoids. *Proc. Nat. Acad. Sci.* 89: 11026–11030, 1992.

[31136] 4688. Bressler, J.; Tsai, T.-F.; Wu, M.-Y.; Tsai, S.-F.; Ramirez, M. A.; Armstrong, D.; Beaudet, A. L.: The SNRPN

promoter is not required for genomic imprinting of the Prader–Willi/Angelman domain in mice. *Nature Genet.* 28: 232–240, 2001.

- [31137] 4689. Buiting, K.; Saitoh, S.; Gross, S.; Dittrich, B.; Schwartz, S.; Nicholls, R. D.; Horsthemke, B.: Inherited microdeletions in the Angelman and Prader–Willi syndromes define an imprinting centre on human chromosome 15. *Nature Genet.* 9: 395–400, 1995.
- [31138] 4690. Dittrich, B.; Buiting, K.; Korn, B.; Rickard, S.; Buxton, J.; Saitoh, S.; Nicholls, R. D.; Poustka, A.; Winterpacht, A.; Zabel, B.; Horsthemke, B.: Imprint switching on human chromosome 15 may involve alternative transcripts of the SNRPN gene. *Nature Genet.* 14: 163–170, 1996.
- [31139] 4691. Gallagher, R. C.; Pils, B.; Albalwi, M.; Francke, U.: Evidence for the role of PWCR1/HBII–85 C/D box small nucleolar RNAs in Prader–Willi syndrome. *Am. J. Hum. Genet.* 71: 669–678, 2002.
- [31140] 4692. Glenn, C. C.; Porter, K. A.; Jong, M. T. C.; Nicholls, R. D.; Driscoll, D. J.: Functional imprinting and epigenetic modification of the human SNRPN gene. *Hum. Molec. Genet.* 2: 2001–2005, 1993.
- [31141] 4693. Gray, T. A.; Saitoh, S.; Nicholls, R. D.: An imprinted, mammalian bicistronic transcript encodes two independent

proteins. Proc. Nat.Acad. Sci. 96: 5616–5621, 1999.

[31142] 4694.Kuslich, C. D.; Kobori, J. A.; Mohapatra, G.; Gregorio-King, C.;Donlon, T. A.: Prader–Willi syndrome is caused by disruption of theSNRPN gene. Am. J. Hum. Genet. 64: 70–76, 1999.

[31143] 4695.Leff, S. E.; Brannan, C. I.; Reed, M. L.; Ozcelik, T.; Francke,U.; Copeland, N. G.; Jenkins, N. A.: Maternal imprinting of the mouseSnrpn gene and conserved linkage homology with the human Prader–Willisyndrome region. Nature Genet. 2: 259–264, 1992.

[31144] 4696.Li, S.; Klein, E. S.; Russo, A. F.; Simmons, D. M.; Rosenfeld,M. G.: Isolation of cDNA clones encoding small nuclear ribonucleoparticle–associatedproteins with different tissue specificities. Proc. Nat. Acad. Sci. 86:9778–9782, 1989.

[31145] 4697.Lyko, F.; Buiting, K.; Horsthemke, B.; Paro, R.: Identificationof a silencing element in the human 15q11–q13 imprinting center byusing transgenic Drosophila. Proc. Nat. Acad. Sci. 95: 1698–1702,1998.

[31146] 4698.McAllister, G.; Amara, S. G.; Lerner, M. R.: Tissue-specificexpression and cDNA cloning of small nuclear ribonucleoprotein–associatedpolypeptide N. Proc. Nat. Acad. Sci. 85: 5296–5300, 1988.

- [31147] 4699.Mutirangura, A.; Jayakumar, A.; Sutcliffe, J. S.; Nakao, M.; McKinney,M. J.; Buiting, K.; Horsthemke, B.; Beaudet, A. L.; Chinault, A. C.;Ledbetter, D. H.: A complete YAC contig of the Prader–Willi/Angelmanchromosome region (15q11–q13) and refined localization of the SNRPN–gene. *Genomics* 18: 546–552, 1993.
- [31148] 4700.Ozcelik, T.; Leff, S.; Robinson, W.; Donlon, T.; Lalande, M.;Sanjines, E.; Schinzel, A.; Francke, U.: Small nuclear ribonucleoproteinpolypeptide N (SNRPN), an expressed gene in the Prader–Willi syndromecritical region. *Nature Genet.* 2: 265–269, 1992.
- [31149] 4701.Reed, M. L.; Leff, S. E.: Maternal imprinting of human SNRPN,a gene deleted in Prader–Willi syndrome. *Nature Genet.* 6: 163–167,1994.
- [31150] 4702.Saitoh, S.; Wada, T.: Parent–of–origin specific histone acetylationand reactivation of a key imprinted gene locus in Prader–Willisyndrome. *Am.J. Hum. Genet.* 66: 1958–1962, 2000.
- [31151] 4703.Kantaputra, P. N.; Gorlin, R. J.; Langer, L. O., Jr.: Dominantmesomelic dysplasia, ankle, carpal, and tarsal synostosis type: anew autosomal dominant bone disorder. *Am. J. Med. Genet.* 44: 730–737,1992.
- [31152] 4704.Karin, M.; Eddy, R. L.; Henry, W. M.; Haley, L. L.; By–

ers, M. G.;Shows, T. B.: Human metallothionein genes are clustered on chromosome16. Proc. Nat. Acad. Sci. 81: 5494–5498, 1984.

[31153] 4705.West, A. K.; Stallings, R.; Hildebrand, C. E.; Chiu, R.; Karin,M.; Richards, R. I.: Human metallothionein genes: structure of thefunctional locus at 16q13. Genomics 8: 513–518, 1990.

[31154] 4706.Wen, L.; Huang, J.–K.; Johnson, B. H.; Reeck, G. R.: A human placentalcDNA clone that encodes nonhistone chromosomal protein HMG–1. NucleicAcids Res. 17: 1197–1214, 1989.

[31155] 4707.Laing, N. G.; Wilton, S. D.; Akkari, P. A.; Dorosz, S.; Boundy,K.; Kneebone, C.; Blumbergs, P.; White, S.; Watkins, H.; Love, D.R.; Haan, E.: A mutation in the alpha tropomyosin gene TPM3 associatedwith autosomal dominant nemaline myopathy. Nature Genet. 9: 75–79,1995.

[31156] 4708.Shirakawa, H.; Yoshida, M.: Structure of a gene coding for humanHMG2 protein. J. Biol. Chem. 267: 6641–6645, 1992.

[31157] 4709.Wanschura, S.; Schoenmakers, E. F. P. M.; Huysmans, C.; Bartnitzke,S.; Van de Ven, W. J. M.; Bullerdiek, J.: Mapping of the human HMG2gene to 4q31. Genomics 31: 264–265, 1996.

- [31158] 4710.Bustin, M.: Regulation of DNA-dependent activities by the functional motifs of the high-mobility-group chromosomal proteins. *Molec. Cell.Biol.* 19: 5237–5246, 1999.
- [31159] 4711.Landsman, D.; Bustin, M.: Chromosomal proteins HMG-14 and HMG-17: distinct multigene families coding for similar types of transcripts. *J.Biol. Chem.* 261: 16087–16091, 1986.
- [31160] 4712.Landsman, D.; Soares, N.; Gonzalez, F. J.; Bustin, M.: Chromosomal protein HMG-17: complete human cDNA sequence and evidence for a multigene family. *J. Biol. Chem.* 261: 7479–7484, 1986.
- [31161] 4713.Mitchell, A.; McBride, W.; Landsman, D.; Bustin, M.: Chromosomal mapping of HMG-17 gene to human chromosome 1p. (Abstract) *Am. J.Hum. Genet.* 43: A152 only, 1988.
- [31162] 4714.Mitchell, A. L.; Bale, A. E.; Bustin, M.; Landsman, D.; Popescu, N.; McBride, O. W.: Localization of HMG17 gene to chromosome 1p35–36.1. (Abstract) *Cytogenet. Cell Genet.* 51: 1045 only, 1989.
- [31163] 4715.Popescu, N.; Landsman, D.; Bustin, M.: Mapping the human gene coding for chromosomal protein HMG-17. *Hum. Genet.* 85: 376–378, 1990.
- [31164] 4716.Porkka, K.; Laakkonen, P.; Hoffman, J. A.;

Bernasconi, M.; Ruoslahti, E.: A fragment of the HMGN2 protein homes to the nuclei of tumor cells and tumor endothelial cells in vivo. *Proc. Nat. Acad. Sci.* 99:7444–7449, 2002.

[31165] 4717. Srikantha, J.; Landsman, D.; Bustin, M.: Retrosequences for human chromosomal protein HMG-17. *J. Molec. Biol.* 197: 405–413, 1987.

[31166] 4718. Ding, H.-F.; Rimsky, S.; Batson, S. C.; Bustin, M.; Hansen, U.: Stimulation of RNA polymerase II elongation by chromosomal protein HMG-14. *Science* 265: 796–799, 1994.

[31167] 4719. Landsman, D.; McBride, O. W.; Soares, N.; Crippa, M. P.; Srikantha, T.; Bustin, M.: Chromosomal protein HMG-14: identification, characterization, and chromosome localization of a functional gene from the large human-multigene family. *J. Biol. Chem.* 264: 3421–3427, 1989.

[31168] 4720. Pash, J.; Popescu, N.; Matocha, M.; Rapoport, S.; Bustin, M.: Chromosomal protein HMG-14 gene maps to the Down syndrome region of human chromosome 21 and is overexpressed in mouse trisomy 16. *Proc. Nat. Acad. Sci.* 87: 3836–3840, 1990.

[31169] 4721. Petersen, M. B.; Economou, E. P.; Slaugenaupt, S. A.; Chakravarti, A.; Antonarakis, S. E.: Linkage analysis of

the human HMG14 gene on chromosome 21 using a GT dinucleotide repeat as polymorphic marker. *Genomics* 7:136–138, 1990.

[31170] 4722. Bajalica, S.; Allander, S. V.; Ehrenborg, E.; Brondum-Nielsen, K.; Luthman, H.; Larsson, C.: Localization of the human insulin-like growth factor-binding protein 4 gene to chromosomal region 17q12–21.1. *Hum. Genet.* 89: 234–236, 1992.

[31171] 4723. Kiefer, M. C.; Schmid, C.; Waldvogel, M.; Schlapfer, I.; Futo, E.; Masiarz, F. R.; Green, K.; Barr, P. J.; Zapf, J.: Characterization of recombinant human insulin-like growth factor binding proteins 4, 5, and 6 produced in yeast. *J. Biol. Chem.* 267: 12692–12699, 1992.

[31172] 4724. Shimasaki, S.; Uchiyama, F.; Shimonaka, M.; Ling, N.: Molecular cloning of the cDNAs encoding a novel insulin-like growth factor-binding protein from rat and human. *Molec. Endocr.* 4: 1451–1458, 1990.

[31173] 4725. Tonin, P.; Ehrenborg, E.; Lenoir, G.; Feunteun, J.; Lynch, H.; Morgan, K.; Zazzi, H.; Vivier, A.; Pollak, M.; Huynh, H.; Luthman, H.; Larsson, C.; Narod, S.: The human insulin-like growth factor-binding protein 4 gene maps to chromosome region 17q12–q21.1 and is close to the gene for hereditary breast–ovarian cancer. *Genomics* 18:

414–417,1993.

- [31174] 4726.Zazzi, H.; Nikoshkov, A.; Hall, K.; Luthman, H.: Structure andtranscription regulation of the human in–sulin–like growth factor bindingprotein 4 gene (IGFBP4). Genomics 49: 401–410, 1998.
- [31175] 4727.Kou, K.; James, P. L.; Clemmons, D. R.; Copeland, N. G.; Gilbert,D. J.; Jenkins, N. A.; Rotwein, P.: Identification of two clustersof mouse insulin–like growth factor binding protein genes on chromosomes1 and 11. Genomics 21: 653–655, 1994.
- [31176] 4728.Brink, P. A.; Steyn, L. T.; Coetzee, G. A.; van der Westhuyzen,D. R.: Familial hypercholesterolemia in South African Afrikaners:PvuII and StuI DNA polymorphisms in the LDL–receptor gene consistentwith a predominating founder gene effect. Hum. Genet. 77: 32–35,1987.
- [31177] 4729.Brown, M. S.; Goldstein, J. L.: Analysis of a mutant strain ofhuman fibroblasts with a defect in the internal–ization of receptor–boundlow density lipoproteins. Cell 9: 663–674, 1976.
- [31178] 4730.Brown, M. S.; Goldstein, J. L.: Familial hypercholes–terolemia:defective binding of lipoproteins to cultured fi–broblasts associatedwith impaired regulation of 3–hydroxy–3–methylglutaryl coenzyme atreductase activ–

ity. Proc. Nat. Acad. Sci. 71: 788–792, 1974.

- [31179] 4731.Davis, C. G.; Lehrman, M. A.; Russell, D. W.; Anderson, R. G.W.; Brown, M. S.; Goldstein, J. L.: The J.D. mutation in familialhypercholesterolemia: amino acid substitution in cytoplasmic domainimpedes internalization of LDL receptors. Cell 45: 15–24, 1986.
- [31180] 4732.De Braekeleer, M.: Hereditary disorders in Saguenay–Lac–St–Jean(Quebec, Canada). Hum. Hered. 41: 141–146, 1991.
- [31181] 4733.Defesche, J. C.; Kastelein, J. J. P.: Molecular epidemiologyof familial hypercholesterolaemia. (Letter) Lancet 352: 1643–1644,1998.
- [31182] 4734.Durst, R.; Colombo, R.; Shpitzen, S.; Ben Avi, L.; Friedlander,Y.; Wexler, R.; Raal, F. J.; Marais, D. A.; Defesche, J. C.; Mandelshtam,M. Y.; Kotze, M. J.; Leitersdorf, E.; Meiner, V.: Recent origin andspread of a common Lithuanian mutation, G197del LDLR, causing familialhypercholesterolemia: positive selection is not always necessary toaccount for disease incidence among Ashkenazi Jews. Am. J. Hum. Genet. 68:1172–1188, 2001.
- [31183] 4735.Ekstrom, U.; Abrahamson, M.; Floren, C.–H.; Tollig, H.; Wettrell,G.; Nilsson, G.; Sun, X.–M.; Soutar, A. K.; Nilsson–Ehle, P.: Anindividual with a healthy phenotype in

spite of a pathogenic LDL receptormutation (C240F). Clin. Genet. 55: 332–339, 1999.

[31184] 4736.Elston, R. C.; Namboodiri, K. K.; Go, R. C. P.; Siervogel, R.M.; Glueck, C. J.: Probable linkage between essential familial hypercholesterolemiaand third complement component (C3). Cytogenet. Cell Genet. 16:294–297, 1976.

[31185] 4737.Feussner, G.; Dobmeyer, J.; Nissen, H.; Hansen, T. S.: UnusualXanthomas in a young patient with heterozygous familial hypercholesterolemiaand type III hyperlipoproteinemia. Am. J. Med. Genet. 65: 149–154,1996.

[31186] 4738.Francke, U.; Brown, M. S.; Goldstein, J. L.: Assignment of thehuman gene for the low density lipoprotein receptor to chromosome19: synteny of a receptor, a ligand, and a genetic disease. Proc.Nat. Acad. Sci. 81: 2826–2830, 1984.

[31187] 4739.Frank, S. L.; Taylor, B. A.; Lusis, A. J.: Linkage of the mouseLDL receptor gene on chromosome 9. Genomics 5: 646–648, 1989.

[31188] 4740.Copeland, N. G.; Silan, C. M.; Kingsley, D. M.; Jenkins, N. A.;Cannizzaro, L. A.; Croce, C. M.; Huebner, K.; Sims, J. E.: Chromosomallocation of murine and human IL-1 receptor genes. Genomics 9: 44–50,1991.

[31189] 4741.Dale, M.; Nicklin, M. J.: Interleukin-1 receptor clus-

ter: geneorganization of IL1R2, IL1R1, IL1RL2 (IL-1Rrp2), IL1RL1 (T1/ST2),and IL18R1 (IL-1Rrp) on human chromosome 2q. Genomics 57: 177-179,1999.

- [31190] 4742.Dower, S. K.; Kronheim, S. R.; Hopp, T. P.; Cantrell, M.; Deeley,M.; Gillis, S.; Henney, C. S.; Urdal, D. L.: The cell surface receptorsfor interleukin-1(alpha) and interleukin-1(beta) are identical. Nature 324:266-268, 1986.
- [31191] 4743.Sims, J. E.; Acres, R. B.; Grubin, C. E.; McMahan, C. J.; Wignall,J. M.; March, C. J.; Dower, S. K.: Cloning the interleukin 1 receptorfrom human T cells. Proc. Nat. Acad. Sci. 86: 8946-8950, 1989.
- [31192] 4744.Habets, G. G. M.; van der Kammen, R. A.; Willemsen, V.; Balemans,M.; Wiegant, J.; Collard, J. G.: Sublocalization of an invasion-inducinglocus and other genes on human chromosome 7. Cytogenet. Cell Genet. 60:200-205, 1992.
- [31193] 4745.Ballantyne, C. M.; Kozak, C. A.; O'Brien, W. E.; Beaudet, A. L.: Assignment of the gene for intercellular adhesion molecule-1 (Icam-1)to proximal mouse chromosome 9. Genomics 9: 547-550, 1991.
- [31194] 4746.Bella, J.; Kolatkar, P. R.; Marlbor, C. W.; Greve, J. M.; Rossmann,M. G.: The structure of the two amino-terminal domains of human ICAM-1suggests how it functions as a rhinovirus receptor and as an LFA-1integrin ligand. Proc.

Nat. Acad. Sci. 95: 4140–4145, 1998.

[31195] 4747. Bellamy, R.; Kwiatkowski, D.; and Hill, A. V. S.: Absence of an association between intercellular adhesion molecule 1, complement receptor 1 and interleukin 1 receptor antagonist gene polymorphisms and severe malaria in a West African population. *Trans. R. Soc. Trop. Med. Hyg.* 92: 312–316, 1998.

[31196] 4748. Craig, A.; Fernandez-Reyes, D.; Mesri, M.; McDowall, A.; Altieri, D. C.; Hogg, N.; Newbold, C.: A functional analysis of a natural variant of intercellular adhesion molecule-1 (ICAM-1-Kilifi). *Hum. Molec. Genet.* 9: 525–530, 2000.

[31197] 4749. Fernandez-Reyes, D.; Craig, A. G.; Kyes, S. A.; Peshu, N.; Snow, R. W.; Berendt, A. R.; Marsh, K.; Newbold, C. I.: A high frequency African coding polymorphism in the N-terminal domain of ICAM-1 predisposing to cerebral malaria in Kenya. *Hum. Molec. Genet.* 6: 1357–1360, 1997.

[31198] 4750. Greve, J. M.; Davis, G.; Meyer, A. M.; Forte, C. P.; Yost, S. C.; Marlor, C. W.; Kamarck, M. E.; McClelland, A.: The major human rhinovirus receptor is ICAM-1. *Cell* 56: 839–847, 1989.

[31199] 4751. Hill, A. V. S.: The immunogenetics of resistance to malaria. *Proc. Assoc. Am. Phys.* 111: 272–277, 1999.

- [31200] 4752.Katz, F. E.; Parkar, M.; Stanley, K.; Murray, L. J.; Clark, E.A.; Greaves, M. F.: Chromosome mapping of cell membrane antigensexpressed on activated B cells. *Europ. J. Immun.* 15: 103–106, 1985.
- [31201] 4753.Le Beau, M. M.; Ryan, D., Jr.; Pericak–Vance, M. A.: Report ofthe committee on the genetic constitution of chromosomes 18 and 19. *Cytogenet.Cell Genet.* 51: 338–357, 1989.
- [31202] 4754.Lu, T. T.; Cyster, J. G.: Integrin–mediated long–term B cellretention in the splenic marginal zone. *Science* 297: 409–412, 2002.
- [31203] 4755.Prieto, J.; Takei, F.; Gendelman, R.; Christenson, B.; Biberfeld,P.; Patarroyo, M.: MALA–2, mouse homologue of human adhesion moleculeICAM–1 (CD54). *Europ. J. Immun.* 19: 1551–1557, 1989.
- [31204] 4756.Simmons, D.; Makgoba, M. W.; Seed, B.: ICAM, an adhesion ligandof LFA–1, is homologous to the neural cell adhesion molecule NCAM. *Nature* 331:624–627, 1988.
- [31205] 4757.Gedde–Dahl, T., Jr.; Dupuy, B. M.; Jonassen, R.; Winberg, J.–O.;Anton–Lamprecht, I.; Olaisen, B.: Junctional epidermolysis bullosainversa (locus EBR2A) assigned to 1q31 by linkage and associationof LAMC1. *Hum. Molec. Genet.* 3: 1387–1391, 1994.

- [31206] 4758.Kallunki, T.; Ikonen, J.; Chow, L. T.; Kallunki, P.; Tryggvason,K.: Structure of the human laminin B2 chain gene reveals extensive divergence from the laminin B1 chain gene. *J. Biol. Chem.* 266: 221–228,1991.
- [31207] 4759.Kallunki, T.; Pikkarainen, T.; Tryggvason, K.; Savolainen, E.–R.: A Pst I polymorphism in the human laminin B2 chain gene on 1q25–q31. *NucleicAcids Res.* 17: 4423 only, 1989.
- [31208] 4760.Mattei, M.–G.; Weil, D.; Passage, E.; Van Cong, N.; Pribula–Conway,D.; Timpl, R.; Chu, M.–L.: Human gene for laminin B2 chain (LAMB2)maps to the long arm of chromosome 1. (Abstract) *Cytogenet. CellGenet.* 46: 659 only, 1987.
- [31209] 4761.Mattei, M.–G.; Weil, D.; Pribula–Conway, D.; Bernard, M. P.; Passage,E.; Van Cong, N.; Timpl, R.; Chu, M.–L.: cDNA cloning, expressionand mapping of human laminin B2 gene to chromosome 1q31. *Hum. Genet.* 79:235–241, 1988.
- [31210] 4762.Miner, J. H.; Patton, B. L.; Lentz, S. I.; Gilbert, D. J.; Jenkins,N. A.; Copeland, N. G.; Sanes, J. R.: The laminin alpha chains: expression,developmental transitions, and chromosomal locations of alpha1–5,identification of heterodimeric laminins 8–11, and cloning of a novelalpha3

isoform. *J. Cell Biol.* 137: 685–701, 1997.

[31211] 4763. Crozat, A.; Aman, P.; Mandahl, N.; Ron, D.: Fusion of CHOP to a novel RNA-binding protein in human myxoid liposarcoma. *Nature* 363:640–644, 1993.

[31212] 4764. Mrozek, K.; Karakousis, C. P.; Bloomfield, C. D.: Chromosome 12 breakpoints are cytogenetically different in benign and malignant lipogenic tumors: localization of breakpoints in lipoma to 12q15 and in myxoid liposarcoma to 12q13.3. *Cancer Res.* 53: 1670–1675, 1993.

[31213] 4765. Panagopoulos, I.; Aman, P.; Mertens, F.; Mandahl, N.; Rydholm, A.; Bauer, H. F. C.; Mitelman, F.: Genomic PCR detects tumor cells in peripheral blood from patients with myxoid liposarcoma. *Genes Chromosomes Cancer* 17: 102–107, 1996.

[31214] 4766. Park, J. S.; Luethy, J. D.; Wang, M. G.; Fargnoli, J.; Fornace, A. J., Jr.; McBride, O. W.; Holbrook, N. J.: Isolation, characterization and chromosomal localization of the human GADD153 gene. *Gene* 116:259–267, 1992.

[31215] 4767. Rabbitts, T. H.; Forster, A.; Larson, R.; Nathan, P.: Fusion of the dominant negative transcription regulator CHOP with a novel gene FUS by translocation t(12;16) in malignant liposarcoma. *Nature Genet.* 4:175–180, 1993.

[31216] 4768. Ron, D.; Habener, J. F.: CHOP, a novel developmen-

tally regulated nuclear protein that dimerizes with transcription factors C/EBP and LAP and functions as a dominant-negative inhibitor of gene transcription. *Genes Dev.* 6: 439–453, 1992.

- [31217] 4769. Kaneda, Y.; Yoshida, M. C.; Kohno, K.; Uchida, T.; Okada, Y.: Chromosomal assignment of the gene for human elongation factor-2. *Proc. Nat. Acad. Sci.* 81: 3158–3162, 1984.
- [31218] 4770. Rapp, G.; Klaudiny, J.; Hagendorff, G.; Luck, M. R.; Scheit, K.H.: Complete sequence of the coding region of human elongation factor 2 (EF-2) by enzymatic amplification of cDNA from human ovarian granulosa cells. *Biol. Chem. Hoppe-Seyler* 370: 1071–1075, 1989.
- [31219] 4771. Chen, I.-T.; Dixit, A.; Rhoads, D. D.; Roufa, D. J.: Homologous ribosomal proteins in bacteria, yeast, and humans. *Proc. Nat. Acad. Sci.* 83: 6907–6911, 1986.
- [31220] 4772. Kenmochi, N.; Kawaguchi, T.; Rozen, S.; Davis, E.; Goodman, N.; Hudson, T. J.; Tanaka, T.; Page, D. C.: A map of 75 human ribosomal protein genes. *Genome Res.* 8: 509–523, 1998.
- [31221] 4773. Nakamichi, N. N.; Kao, F.-T.; Wasmuth, J.; Roufa, D. J.: Ribosomal protein gene sequences map to human chromosomes 5, 8, and 17. *Somat. Cell Molec. Genet.* 12:

225–236, 1986.

- [31222] 4774. Justice, M. J.; Siracusa, L. D.; Gilbert, D. J.; Heisterkamp, N.; Groffen, J.; Chada, K.; Silan, C. M.; Copeland, N. G.; Jenkins, N. A.: A genetic linkage map of mouse chromosome 10: localization of eighteen molecular markers using a single interspecific backcross. *Genetics* 125:855–866, 1990.
- [31223] 4775. King, M. C.: Personal Communication. Berkeley, Calif. 5/1996.
- [31224] 4776. Korach, K. S.: Insights from the study of animals lacking functional estrogen receptor. *Science* 266: 1524–1527, 1994.
- [31225] 4777. Kos, M.; Reid, G.; Denger, S.; Gannon, F.: Minireview: genomic organization of the human ER- α gene promoter region. *Molec. Endocr.* 15:2057–2063, 2001.
- [31226] 4778. Kumar, R.; Wang, R.-A.; Mazumdar, A.; Talukder, A. H.; Mandal, M.; Yang, Z.; Bagheri-Yarmand, R.; Sahin, A.; Hortobagyi, G.; Adam, L.; Barnes, C. J.; Vadlamudi, R. K.: A naturally occurring MTA1 variant sequesters oestrogen receptor- α in the cytoplasm. *Nature* 418:654–657, 2002.
- [31227] 4779. Lawson, J. S.; Field, A. S.; Champion, S.; Tran, D.; Ishikura, H.; Trichopoulos, D.: Low oestrogen receptor α -

pha expression in normal breast tissue underlies low breast cancer incidence in Japan. (Letter) Lancet 354: 1787–1788, 1999.

- [31228] 4780. Lonard, D. M.; Nawaz, Z.; Smith, C. L.; O'Malley, B. W.: The 26S proteasome is required for estrogen receptor- α and coactivator turnover and for efficient estrogen receptor- α transactivation. *Molec. Cell* 5: 939–948, 2000.
- [31229] 4781. Lorentzon, M.; Lorentzon, R.; Backstrom, T.; Nordstrom, P.: Estrogen receptor gene polymorphism, but not estradiol levels, is related to bone density in healthy adolescent boys: a cross-sectional and longitudinal study. *J. Clin. Endocr. Metab.* 84: 4597–4601, 1999.
- [31230] 4782. Mader, S.; Kumar, V.; de Verneuil, H.; Chambon, P.: Three amino acids of the oestrogen receptor are essential to its ability to distinguish oestrogen from a glucocorticoid-responsive element. *Nature* 338: 271–274, 1989.
- [31231] 4783. McGuire, W. L.; Chamness, G. C.; Fuqua, S. A. W.: Estrogen receptor variants in clinical breast cancer. *Molec. Endocr.* 5: 1571–1577, 1991.
- [31232] 4784. McGuire, W. L.; Chamness, G. C.; Fuqua, S. A. W.: Abnormal estrogen receptor in clinical breast cancer. *J. Steroid Biochem. Molec. Biol.* 43: 243–247, 1992.

- [31233] 4785. McInerney, E. M.; Ince, B. A.; Shapiro, D. J.; Katzenellenbogen, B. S.: A transcriptionally active estrogen receptor mutant is a novel type of dominant negative inhibitor of estrogen action. *Molec. Endocr.* 10:1519–1526, 1996.
- [31234] 4786. Menasce, L. P.; White, G. R. M.; Harrison, C. J.; Boyle, J. M.: Localization of the estrogen receptor locus (ESR) to chromosome 6q25.1 by FISH and a simple post-FISH banding technique. *Genomics* 17:263–265, 1993.
- [31235] 4787. Metzger, D.; White, J. H.; Chambon, P.: The human oestrogen receptor functions in yeast. *Nature* 334: 31–36, 1988.
- [31236] 4788. Murphy, L. C.; Wang, M.; Coutt, A.; Dotzlaw, H.: Novel mutations in the estrogen receptor messenger RNA in human breast cancers. *J. Clin. Endocr. Metab.* 81: 1420–1427, 1996.
- [31237] 4789. Pelletier, G.; El-Alfy, M.: Immunocytochemical localization of estrogen receptors alpha and beta in the human reproductive organs. *J. Clin. Endocr. Metab.* 85: 4835–4840, 2000.
- [31238] 4790. Ponglikitmongkol, M.; Green, S.; Chambon, P.: Genomic organization of the human oestrogen receptor gene. *EMBO J.* 7: 3385–3388, 1988.

- [31239] 4791. Reese, J. C.; Katzenellenbogen, B. S.: Characterization of a temperature-sensitive mutation in the hormone binding domain of the human estrogen receptor: studies in cell extracts and intact cells and their implications for hormone-dependent transcriptional activation. *J. Biol. Chem.* 267: 9868–9873, 1992.
- [31240] 4792. Reese, J. C.; Katzenellenbogen, B. S.: Mutagenesis of cysteines in the hormone binding domain of the human estrogen receptor: alterations in binding and transcriptional activation by covalently and reversibly attaching ligands. *J. Biol. Chem.* 266: 10880–10887, 1991.
- [31241] 4793. Scott, G. K.; Kushner, P.; Vigne, J.-L.; Benz, C. C.: Truncated forms of DNA-binding estrogen receptors in human breast cancer. *J. Clin. Invest.* 88: 700–706, 1991.
- [31242] 4794. Shang, Y.; Brown, M.: Molecular determinants for the tissue specificity of SERMs. *Science* 295: 2465–2468, 2002.
- [31243] 4795. Shiau, A. K.; Barstad, D.; Loria, P. M.; Cheng, L.; Kushner, P. J.; Agard, D. A.; Greene, G. L.: The structural basis of estrogen receptor/coactivator recognition and the antagonism of this interaction by tamoxifen. *Cell* 95: 927–937, 1998.
- [31244] 4796. Shupnik, M. A.; Pitt, L. K.; Soh, A. Y.; Anderson, A.;

Lopes, M. B.; Laws, E. R., Jr.: Selective expression of estrogen receptor alpha and beta isoforms in human pituitary tumors. *J. Clin. Endocrinol. Metab.* 83: 3965–3972, 1998.

[31245] 4797. Simoncini, T.; Hafezi-Moghadam, A.; Brazil, D. P.; Ley, K.; Chin, W. W.; Liao, J. K.: Interaction of oestrogen receptor with the regulatory subunit of phosphatidylinositol-3-OH kinase. *Nature* 407: 538–541, 2000.

[31246] 4798. Sluyser, M.: Mutations in the estrogen receptor gene. *Hum. Mutat.* 6: 97–103, 1995.

[31247] 4799. Sluyser, M.; Mester, J.: Oncogenes homologous to steroid receptors? (Letter) *Nature* 315: 546 only, 1985.

[31248] 4800. Smith, E. P.; Boyd, J.; Frank, G. R.; Takahashi, H.; Cohen, R. M.; Specker, B.; Williams, T. C.; Lubahn, D. B.; Korach, K. S.: Estrogen resistance caused by a mutation in the estrogen-receptor gene in a man. *New Eng. J. Med.* 331: 1056–1061, 1994.

[31249] 4801. Sudhir, K.; Chou, T. M.; Chatterjee, K.; Smith, E. P.; Williams, T. C.; Kane, J. P.; Malloy, M. J.; Korach, K. S.; Rubanyi, G. M.: Premature coronary artery disease associated with a disruptive mutation in the estrogen receptor gene in a man. *Circulation* 96: 3774–3777, 1997.

[31250] 4802. Takeyama, J.; Suzuki, T.; Inoue, S.; Kaneko, C.; Nagura, H.; Harada, N.; Sasano, H.: Expression and cellular

localization of estrogen receptors alpha and beta in the human fetus. *J. Clin. Endocr. Metab.* 86:2258–2262, 2001.

[31251] 4803. Walter, P.; Green, S.; Greene, G.; Krust, A.; Bornert, J.-M.; Jeltsch, J.-M.; Staub, A.; Jensen, E.; Scrace, G.; Waterfield, M.; Chambon, P.: Cloning of the human estrogen receptor cDNA. *Proc. Nat. Acad. Sci.* 82: 7889–7893, 1985.

[31252] 4804. Weel, A. E. A. M.; Uitterlinden, A. G.; Westendorp, I. C. D.; Burger, H.; Schuit, S. C. E.; Hofman, A.; Helmerhorst, T. J. M.; van Leeuwen, J. P. T. M.; Pols, H. A. P.: Estrogen receptor polymorphism predicts the onset of natural and surgical menopause. *J. Clin. Endocr. Metab.* 84: 3146–3150, 1999.

[31253] 4805. Weis, K. E.; Ekena, K.; Thomas, J. A.; Lazennec, G.; Katzenellenbogen, B. S.: Constitutively active human estrogen receptors containing amino acid substitutions for tyrosine 537 in the receptor protein. *Molec. Endocr.* 10: 1388–1398, 1996.

[31254] 4806. Liu, B. P.; Fournier, A.; GrandPre, T.; Strittmatter, S. M.: Myelin-associated glycoprotein as a functional ligand for the Nogo-66 receptor. *Science* 297:1190–1193, 2002.

[31255] 4807. Costa, R. M.; Federov, N. B.; Kogan, J. H.; Murphy, G. G.; Stern, J.; Ohno, M.; Kucherlapati, R.; Jacks, T.; Silva, A. J.: Mechanism for the learning deficits in a mouse model of

neurofibromatosis type1. Nature 415: 526–530, 2002.

[31256] 4808.van de Vijver, M. J.; Peterse, J. L.; Mooi, W. J.; Wisman, P.; Lomans, J.; Dalesio, O.; Nusse, R.: NEU-protein overexpression in breast cancer: association with comedo-type ductal carcinoma in situ and limited prognostic value in stage II breast cancer. New Eng. J. Med. 319:

1239–1245, 1988.

[31257] 4809.Xie, D.; Shu, X. O.; Deng, Z.; Wen, W.-Q.; Creek, K. E.; Dai, Q., Gao, Y.-T.; Jin, F.; Zheng, W.: Population-based, case-control study of HER2 genetic polymorphism and breast cancer risk. J. Nat. Cancer Inst. 92: 412–417, 2000.

[31258] 4810.Yamamoto, T.; Ikawa, S.; Akiyama, T.; Semba, K.; Nomura, N.; Miyajima, N.; Saito, T.; Toyoshima, K.: Similarity of protein encoded by the human c-erb-B-2 gene to epidermal growth factor receptor. Nature 319:230–234, 1986.

[31259] 4811.Yang-Feng, T. L.; Schechter, A. L.; Weinberg, R. A.; Francke, U.: Oncogene from rat neuro/glioblastomas (human gene symbol NGL) is located on the proximal long arm of human chromosome 17 and EGFR is confirmed at 7p13–q11.2. (Abstract) Cytogenet. Cell Genet. 40:784 only, 1985.

[31260] 4812.Yu, D.; Jing, T.; Liu, B.; Yao, J.; Tan, M.; McDonnell,

T. J.;Hung, M.-C.: Overexpression of ErbB2 blocks Taxol-induced apoptosisby upregulation of p21Cip1, which inhibits p34Cdc2 kinase. *Molec.Cell* 2: 581–591, 1998.

[31261] 4813.Klappacher, G. W.; Lunyak, V. V.; Sykes, D. B.; Sawka-Verhelle,D.; Sage, J.; Brard, G.; Ngo, S. D.; Gangadharan, D.; Jacks, T.; Kamps,M. P.; Rose, D. W.; Rosenfeld, M. G.: An induced Ets repressor complexregulates growth arrest during terminal macrophage differentiation. *Cell* 109:169–180, 2002.

[31262] 4814.Klemsz, M.; Hromas, R.; Raskind, W.; Bruno, E.; Hoffman, R.: PE-1,a novel ETS oncogene family member, localizes to chromosome 1q21–q23. *Genomics* 20:291–294, 1994.

[31263] 4815.Kastury, K.; Li, J.; Druck, T.; Su, H.; Vogt, P. K.; Croce, C.M.; Huebner, K.: The human homologue of the retroviral oncogene qinmaps to chromosome 14q13. *Proc. Nat. Acad. Sci.* 91: 3616–3618, 1994.

[31264] 4816.Li, J.; Chang, H. W.; Lai, E.; Parker, E. J.; Vogt, P. K.: Theoncogene qin codes for a transcriptional repressor. *Cancer Res.* 55:5540–5544, 1995.

[31265] 4817.Li, J.; Vogt, P. K.: The retroviral oncogene qin belongs to thetranscription factor family that includes the homeotic gene fork head. *Proc.Nat. Acad. Sci.* 90:

4490–4494, 1993.

- [31266] 4818. Murphy, D. B.; Wiese, S.; Burfeind, P.; Schmundt, D.; Mattei, M.–G.; Schulz–Schaeffer, W.; Thies, U.: Human brain factor 1, a new member of the fork head gene family. *Genomics* 21: 551–557, 1994.
- [31267] 4819. Wiese, S.; Murphy, D. B.; Schlung, A.; Burfeind, P.; Schmundt, D.; Schnulle, V.; Mattei, M.–G.; Thies, U.: The genes for human brainfactor 1 and 2, members of the fork head gene family, are clustered on chromosome 14q. *Biochim. Biophys. Acta* 1262: 105–112, 1995.
- [31268] 4820. Bustelo, X. R.: Regulatory and signaling properties of the Vav family. *Molec. Cell. Biol.* 20: 1461–1477, 2000.
- [31269] 4821. Bustelo, X. R.; Barbacid, M.: Tyrosine phosphorylation of the VAV proto–oncogene product in activated B cells. *Science* 256: 1196–1199, 1992.
- [31270] 4822. Denninger, D. J.; Borges, C. R.; Butler, C. L.; Cushman, A. M.; Kawahara, R. S.: Genomic organization and regulation of the vav proto–oncogene. *Biochim. Biophys. Acta* 1491: 253–262, 2000.
- [31271] 4823. Fackler, O. T.; Luo, W.; Geyer, M.; Alberts, A. S.; Peterlin, B. M.: Activation of Vav by Nef induces cytoskeletal rearrangements and downstream effector functions. *Molec. Cell* 3: 729–739, 1999.

- [31272] 4824.Fischer, K.-D.; Zmuidzinas, A.; Gardner, S.; Barbacid, M.; Bernstein,A.; Guidos, C.: Defective T-cell receptor signalling and positiveselection of Vav-deficient CD4(+) CD8(+) thymocytes. *Nature* 374:474-477, 1995.
- [31273] 4825.Katzav, S.; Martin-Zanca, D.; Barbacid, M.: VAV, a novel humanoncogene derived from a locus ubiquitously expressed in hematopoieticcells. *EMBO J.* 8: 2283-2290, 1989.
- [31274] 4826.Martinerie, C.; Cannizzaro, L. A.; Croce, C. M.; Huebner, K.; Katzav,S.; Barbacid, M.: The human VAV proto-oncogene maps to chromosomeregion 19p12-19p13.2. *Hum. Genet.* 86: 65-68, 1990.
- [31275] 4827.Moores, S. L.; Selfors, L. M.; Fredericks, J.; Breit, T.; Fujikawa,K.; Alt, F. W.; Brugge, J. S.; Swat, W.: Vav family proteins coupleto diverse cell surface receptors. *Molec. Cell. Biol.* 20: 6364-6373,2000.
- [31276] 4828.Tarakhovsky, A.; Turner, M.; Schaal, S.; Mee, P. J.; Duddy, L.P.; Rajewsky, K.; Tybulewicz, V. L. J.: Defective antigen receptor-mediatedproliferation of B and T cells in the absence of Vav. *Nature* 374:467-470, 1995.
- [31277] 4829.Zhang, R.; Alt, F. W.; Davidson, L.; Orkin, S. H.; Swat, W.:Defective signalling through the T- and B-cell antigen receptors inlymphoid cells lacking the vav proto-

oncogene. Nature 374: 470–473,1995.

[31278] 4830.Fukuhara, S.; Rowley, J. D.; Variakojis, D.; Sweet, D. L.: Chromosomeabnormalities in poorly differentiated lymphocytic lymphoma. CancerRes. 39: 3119–3128, 1979.

[31279] 4831.Ohno, H.; Fukuhara, S.; Takahashi, R.; Mihara, K.; Sugiyama, T.;Doi, S.; Uchino, H.; Toyoshima, K.: c–yes and bcl–2 genes locatedon 18q21.3 in a follicular lymphoma cell line carrying a t(14;18)chromosomal translocation. Int. J. Cancer 39: 785–788, 1987.

[31280] 4832.Semba, K.; Nishizawa, M.; Satoh, H.; Fukushige, S.; Yoshida, M.C.; Sasaki, M.; Matsubara, K.; Yamamoto, T.; Toyoshima, K.: Nucleotidesequence and chromosomal mapping of the human c–yes–2 gene. Jpn.J. Cancer Res. 79: 710–717, 1988.

[31281] 4833.Semba, K.; Yamanashi, Y.; Nishizawa, M.; Sukegawa, J.; Yoshida,M.; Sasaki, M.; Yamamoto, T.; Toyoshima, K.: Location of the c–yesgene on the human chromosome and its expression in various tissues. Science 227:1038–1040, 1985.

[31282] 4834.Silverman, G. A.; Kuo, W.–L.; Taillon–Miller, P.; Gray, J. W.:Chromosomal reassignment: YACs containing both YES1 and thymidylatesynthase map to the short arm of

chromosome 18. *Genomics* 15: 442–445, 1993.

- [31283] 4835. Sukegawa, J.; Semba, K.; Yamanashi, Y.; Nishizawa, M.; Miyajima, N.; Yamamoto, T.; Toyoshima, K.: Characterization of cDNA clones for the human c-yes gene. *Molec. Cell. Biol.* 7: 41–47, 1987.
- [31284] 4836. Alves, S. E.; Lopez, V.; McEwen, B. S.; Weiland, N. G.: Differential colocalization of estrogen receptor beta (ER-beta) with oxytocin and vasopressin in the paraventricular and supraoptic nuclei of the female rat brain: an immunocytochemical study. *Proc. Nat. Acad. Sci.* 95: 3281–3286, 1998.
- [31285] 4837. Inoue, T.; Kimura, T.; Azuma, C.; Inazawa, J.; Takemura, M.; Kikuchi, T.; Kubota, Y.; Ogita, K.; Saji, F.: Structural organization of the human oxytocin receptor gene. *J. Biol. Chem.* 269: 32451–32456, 1994.
- [31286] 4838. Kimura, T.; Tanizawa, O.; Mori, K.; Brownstein, M. J.; Okayama, H.: Structure and expression of a human oxytocin receptor. *Nature* 356: 526–529, 1992.
- [31287] 4839. Simmons, C. F., Jr.; Clancy, T. E.; Quan, R.; Knoll, J. H. M.: The oxytocin receptor gene (OXTR) localizes to human chromosome 3p25 by fluorescence in situ hybridization and PCR analysis of somatic cell hybrids. *Genomics* 26: 623–625, 1995.

- [31288] 4840. Yang, M.; Wang, W.; Zhong, M.; Philippi, A.; Lichtarge, O.; Sanborn, B. M.: Lysine 270 in the third intracellular domain of the oxytocin receptor is an important determinant for G- α -q coupling specificity. *Molec. Endocr.* 16: 814–823, 2002.
- [31289] 4841. Owerbach, D.; Rutter, W. J.; Cooke, N. E.; Martial, J. A.; Shows, T. B.: The prolactin gene is located on chromosome 6 in humans. *Science* 212: 815–816, 1981.
- [31290] 4842. Shome, B.; Parlow, A. F.: Human pituitary prolactin (hPRL): the entire linear amino acid sequence. *J. Clin. Endocrin. Metab.* 45: 1112–1115, 1977.
- [31291] 4843. Sun, Z.; Lee, M. S.; Rhee, H. K.; Arrandale, J. M.; Dannies, P. S.: Inefficient secretion of human H27A-prolactin, a mutant that does not bind Zn(2+). *Molec. Endocr.* 11: 1544–1551, 1997.
- [31292] 4844. Taggart, R. T.; Mohandas, T. K.; Bell, G. I.: Assignment of the human preprogastrin (PGC) to chromosome 6 and regional localization of PGC (6pter–p21.1), prolactin PRL (6pter–p21.1). (Abstract) *Cytogenet. Cell Genet.* 46: 701–702, 1987.
- [31293] 4845. Truong, A. T.; Duez, C.; Belayew, A.; Renard, A.; Pictet, R.; Bell, G. I.; Martial, J. A.: Isolation and characterization of the human prolactin gene. *EMBO J.* 3: 429–437,

1984.

- [31294] 4846.Slate, D. L.; Ruddle, F. H.: Fibroblast interferon in man is coded by two loci on separate chromosomes. *Cell* 16: 171–180, 1979.
- [31295] 4847.Rosenberg, M.; RayChaudhury, A.; Shows, T. B.; Le Beau, M. M.;Fuchs, E.: A group of type I keratin genes on human chromosome 17:characterization and expression. *Molec. Cell. Biol.* 8: 722–736,1988.
- [31296] 4848.Gross, J. A.; Johnston, J.; Mudri, S.; Enselman, R.; Dillon, S.R.; Madden, K.; Xu, W.; Parrish–Novak, J.; Foster, D.; Lofton–Day,C.; Moore, M.; Littau, A.; Grossman, A.; Haugen, H.; Foley, K.; Blumberg,H.; Harrison, K.; Kindsvo–gel, W.; Clegg, C. H.: TACI and BCMA are receptors for a TNF homologue implicated in B–cell autoimmune disease. *Nature* 404:995–999, 2000.
- [31297] 4849.de Wit, T. P. M.; Morton, H. C.; Capel, P. J. A.; van de Winkel,J. G. J.: Structure of the gene for the human myeloid IgA Fc receptor(CD89). *J. Immun.* 155: 1203–1209, 1995.
- [31298] 4850.Kremer, E. J.; Kalatzis, V.; Baker, E.; Callen, D. F.; Sutherland,G. R.; Maliszewski, C. R.: The gene for the human IgA Fc receptormaps to 19q13.4. *Hum. Genet.* 89: 107–108, 1992.

- [31299] 4851.Maliszewski, C. R.; March, C. J.; Schoenborn, M. A.; Gimpel, S.;Shen, L.: Expression cloning of a human Fc receptor for IgA. *J.Exp. Med.* 172: 1665–1672, 1990.
- [31300] 4852.Monteiro, R. C.; Hostoffer, R. W.; Cooper, M. D.; Bonner, J. R.;Gartland, G. L.; Kubagawa, H.: Definition of immunoglobulin A receptorson eosinophils and their enhanced expression in allergic individuals. *J.Clin. Invest.* 92: 1681–1685, 1993.
- [31301] 4853.Narita, I.; Goto, S.; Saito, N.; Sakatsume, M.; Jin, S.; Omori,K.; Gejyo, F.: Genetic polymorphisms in the promoter and 5–primeUTR region of the Fc alpha receptor (CD89) are not associated witha risk of IgA nephropathy. *J. Hum. Genet.* 46: 694–698, 2001.
- [31302] 4854.Pleass, R. J.; Andrews, P. D.; Kerr, M. A.; Woof, J. M.: Alternativesplicing of the human IgA Fc receptor CD89 in neutrophils and eosinophils. *Biochem.J.* 318: 771–777, 1996.
- [31303] 4855.Shimokawa, T.; Tsuge, T.; Okumura, K.; Ra, C.: Identificationand characterization of the promoter for the gene encoding the humanmyeloid IgA Fc receptor (Fc–alpha–R, CD89). *Immunogenetics* 51: 945–954,2000.
- [31304] 4856.Tsuge, T.; Shimokawa, T.; Horikoshi, S.; Tomino, Y.; Ra, C.: Polymorphismin promoter region of Fc–alpha re–

ceptor gene in patients with IgA nephropathy. *Hum. Genet.* 108: 128–133, 2001.

- [31305] 4857. Lalley, P. A.; Sakaguchi, A. Y.; Eddy, R. L.; Honey, N. H.; Bell, G. I.; Shen, L.-P.; Rutter, W. J.; Jacobs, J. W.; Heinrich, G.; Chin, W. W.; Naylor, S. L.: Mapping polypeptide hormone genes in the mouse: somatostatin, glucagon, calcitonin, and parathyroid hormone. *Cytogenet. Cell Genet.* 44: 92–97, 1987.
- [31306] 4858. Dillon, J. S.; Tanizawa, Y.; Wheeler, M. B.; Leng, X.-H.; Ligon, B. B.; Rabin, D. U.; Yoo-Warren, H.; Permutt, M. A.; Boyd, A. E., III: Cloning and functional expression of the human glucagon-like peptide-1 (GLP-1) receptor. *Endocrinology* 133: 1907–1910, 1993.
- [31307] 4859. Kershaw, E. E.; Chua, S. C., Jr.; Leibel, R. L.: Localization of a (CA)_n repeat in glucagon-like peptide-1 receptor gene (*Glp1r*) to proximal mouse chromosome 17 and its linkage to other markers. *Mammalian Genome* 6: 301–303, 1995.
- [31308] 4860. Stoffel, M.; Espinosa, R., III; Le Beau, M. M.; Bell, G. I.: Human glucagon-like peptide-1 receptor gene: localization to chromosome band 6p21 by fluorescence in situ hybridization and linkage of a highly polymorphic simple tandem repeat DNA polymorphism to other markers on

chromosome 6. Diabetes 42: 1215–1218, 1993.

- [31309] 4861.Thorens, B.: Expression cloning of the pancreatic beta cell receptor for the gluco–incretin hormone glucagon–like peptide 1. Proc. Nat.Acad. Sci. 89: 8641–8645, 1992.
- [31310] 4862.Cohen, P.; Miyazaki, M.; Socci, N. D.; Hagge–Greenberg, A. Liedtke,W.; Soukas, A. A.; Sharma, R.; Hudgins, L. C.; Ntambi, J. M.; Friedman,J. M.: Role for stearoyl–CoA desaturase–1 in leptin–mediated weightloss. Science 297: 240–243, 2002.
- [31311] 4863.Comuzzie, A. G.; Hixson, J. E.; Almasy, L.; Mitchell, B. D.; Mahaney,M. C.; Dyer, T. D.; Stern, M. P.; MacCluer, J. W.; Blangero, J.:A major quantitative trait locus determining serum leptin levels and fat mass is located on human chromosome 2. Nature Genet. 15: 273–276,1997.
- [31312] 4864.Dahms, N. M.; Lobel, P.; Breitmeyer, J.; Chirgwin, J. M.; Kornfeld,S.: 46 kd mannose 6–phosphate receptor: cloning, expression, andhomology to the 215 kd mannose 6–phosphate receptor. Cell 50: 181–192,1987.
- [31313] 4865.Ludwig, T.; Ruther, U.; Metzger, R.; Copeland, N. G.; Jenkins,N. A.; Lobel, P.; Hoflack, B.: Gene and pseudogene of the mouse cation–dependentmannose 6–phosphate receptor: genomic organization, expression, andchromoso–

- mal localization. J. Biol. Chem. 267: 12211–12219, 1992.
- [31314] 4866. Pohlmann, R.; Boeker, M. W. C.; von Figura, K.: The two mannose 6-phosphate receptors transport distinct complements of lysosomal proteins. J. Biol. Chem. 270: 27311–27318, 1995.
- [31315] 4867. Pohlmann, R.; Nagel, G.; Schmidt, B.; Stein, M.; Lorkowski, G.; Krentler, C.; Cully, J.; Meyer, H. E.; Grzeschik, K.-H.; Mersmann, G.; Hasilik, A.; von Figura, K.: Cloning of a cDNA encoding the human cation-dependent mannose 6-phosphate-specific receptor. Proc. Nat. Acad. Sci. 84: 5575–5579, 1987.
- [31316] 4868. Roberts, D. L.; Weix, D. J.; Dahms, N. M.; Kim, J.-J. P.: Molecular basis of lysosomal enzyme recognition: three-dimensional structure of the cation-dependent mannose 6-phosphate receptor. Cell 93: 639–648, 1998.
- [31317] 4869. Watanabe, H.; Grubb, J. H.; Sly, W. S.: The overexpressed human 46-kDa mannose 6-phosphate receptor mediates endocytosis and sorting of β -glucuronidase. Proc. Nat. Acad. Sci. 87: 8036–8040, 1990.
- [31318] 4870. Vigon, I.; Mornon, J.-P.; Cocault, L.; Mitjavila, M.-T.; Tambourin, P.; Gisselbrecht, S.; Souyri, M.: Molecular cloning and characterization of MPL, the human homolog of the v-mpl oncogene: identification of a member of the

hematopoietic growth factor receptor superfamily.

Proc.Nat. Acad. Sci. 89: 5640–5644, 1992.

- [31319] 4871.Bucan, M.; Gatalica, B.; Nolan, P.; Chung, A.; Leroux, A.; Grossman,M. H.; Nadeau, J. H.; Emanuel, B. S.; Budarf, M.: Comparative mapping of 9 human chromosome 22q loci in the laboratory mouse. Hum. Molec.Genet. 2: 1245–1252, 1993.
- [31320] 4872.Griffiths, D. F. R.; Williams, G. T.; Williams, E. D.: Duodenalcarcinoid tumours, phaeochromocytoma and neurofibromatosis: isletcell tumour, phaeochromocytoma and the von Hippel–Lindau complex:two distinctive neuroendocrine syndromes. Quart. J. Med. 245: 769–782,1987.
- [31321] 4873.Mackman, N.; Morrissey, J. H.; Fowler, B.; Edgington, T. S.: Complete sequence of the human tissue factor gene, a highly regulated cellularreceptor that initiates the coagulation protease cascade. Biochemistry 28:1755–1762, 1989.
- [31322] 4874.Popescu, N. C.; Bowden, P. E.; DiPaolo, J. A.: Two type II keratingenes are localized on human chromosome 12. Hum. Genet. 82: 109–112,1989.
- [31323] 4875.Pulkkinen, L.; Christiano, A. M.; Knowlton, R. G.; Uitto, J.:Epidermolytic hyperkeratosis (bullous congenital

ichthyosiform erythroderma):genetic linkage to chromosome 12q in the region of the type II keratingene cluster. J. Clin. Invest. 91: 357–361, 1993.

- [31324] 4876.Rothnagel, J. A.; Dominey, A. M.; Dempsey, L. D.; Longley, M.A.; Greenhalgh, D. A.; Gagne, T. A.; Huber, M.; Frenk, E.; Hohl, D.;Roop, D. R.: Mutations in the rod domains of keratins 1 and 10 in epidermolytic hyperkeratosis. Science 257: 1128–1130, 1992.
- [31325] 4877.Schimkat, M.; Baur, M. P.; Henke, J.: Inheritance of some electrophoretic phenotypes of human hair. Hum. Genet. 85: 311–314, 1990.
- [31326] 4878.Sybert, V. P.; Francis, J. S.; Corden, L. D.; Smith, L. T.; Weaver, M.; Stephens, K.; McLean, W. H. I.: Cyclic ichthyosis with epidermolytic hyperkeratosis: a phenotype conferred by mutations in the 2B domain of keratin K1. Am. J. Hum. Genet. 64: 732–738, 1999.
- [31327] 4879.Syder, A. J.; Yu, Q.-C.; Paller, A. S.; Giudice, G.; Pearson, R.; Fuchs, E.: Genetic mutations in the K1 and K10 genes of patients with epidermolytic hyperkeratosis: correlation between location and disease severity. J. Clin. Invest. 93: 1533–1542, 1994.
- [31328] 4880.Yoon, S.-J.; LeBlanc-Straceski, J.; Ward, D.; Krauter, K.; Kucherlapati, R.: Organization of the human keratin

type II gene cluster at 12q13. Genomics 24:502–508, 1994.

- [31329] 4881.Hermouet, S.; Merendino, J. J., Jr.; Gutkind, J. S.; Spiegel, A.M.: Activating and inactivating mutations of the alpha subunit ofG(i2) protein have opposite effects on proliferation of NIH 3T3 cells. Proc.Nat. Acad. Sci. 88: 10455–10459, 1991.
- [31330] 4882.Lerman, B. B.; Dong, B.; Stein, K. M.; Markowitz, S. M.; Linden,J.; Catanzaro, D. F.: Right ventricular outflow tract tachycardiadue to a somatic cell mutation in G protein subunit–alpha–i2. J.Clin. Invest. 101: 2862–2868, 1998.
- [31331] 4883.Lyons, J.; Landis, C. A.; Harsh, G.; Vallar, L.; Grunewald, K.;Feichtinger, H.; Duh, Q.–Y.; Clark, O. H.; Kawasaki, E.; Bourne, H.R.; McCormick, F.: Two G protein oncogenes in human endocrine tumors. Science 249:655–659, 1990.
- [31332] 4884.Rudolph, U.; Finegold, M. J.; Rich, S. S.; Harriman, G. R.; Srinivasan,Y.; Brabet, P.; Boulay, G.; Bradley, A.; Birnbaumer, L.: Ulcerativecolitis and adenocarcinoma of the colon in G alpha(i2)–deficient mice. NatureGenet. 10: 143–150, 1995.
- [31333] 4885.Baron, B.; Fernandez, M. A.; Toledo, F.; Le Roscouet,

D.; Mayau,V.; Martin, N.; Buttin, G.; Debatisse, M.: The highly conserved Chinesehamster GNAI3 gene maps less than 60 kb from the AMPD2 gene and lacksthe intronic U6 snRNA present in its human counterpart. *Genomics* 24:288–294, 1994.

[31334] 4886.Goodman, F. R.: Limb malformations and the human HOX genes. *Am.J. Med. Genet.* 112: 256–265, 2002.

[31335] 4887.Magli, M. C.; Barba, P.; Celetti, A.; De Vita, G.; Cillo, C.; Boncinelli,E.: Coordinate regulation of HOX genes in human hematopoietic cells. *Proc.Nat. Acad. Sci.* 88: 6348–6352, 1991.

[31336] 4888.Pravtcheva, D.; Newman, M.; Hunihan, L.; Lonai, P.; Ruddle, F.H.: Chromosome assignment of the murine Hox–4.1 gene. *Genomics* 5:541–545, 1989.

[31337] 4889.Taniguchi, Y.; Komatsu, N.; Moriuchi, T.: Overexpression of theHOX4A (HOXD3) homeobox gene in human erythroleukemia HEL cells resultsin altered adhesive properties. *Blood* 85: 2786–2794, 1995.

[31338] 4890.Zakany, J.; Duboule, D.: Hox genes and the making of sphincters. *Nature* 401:761–762, 1999.

[31339] 4891.Mavilio, F.; Simeone, A.; Giampaolo, A.; Faiella, A.; Zappavigna,V.; Acampora, D.; Poiana, G.; Russo, G.; Peschle, C.; Boncinelli,E.: Differential and stage–related

expression in embryonic tissues of a new human homeobox gene. *Nature* 324: 664–668, 1986.

[31340] 4892. Oliver, G.; Sidell, N.; Fiske, W.; Heinzmann, C.; Mohandas, T.; Sparkes, R. S.; De Robertis, E. M.: Complementary homeo protein gradients in developing limb buds. *Genes Dev.* 3: 641–650, 1989.

[31341] 4893. Shaw, D. J.; Meredith, A. L.; Brook, J. D.; Sarfarazi, M.; Harley, H. G.; Huson, S. M.; Bell, G. I.; Harper, P. S.: Linkage relationships of the insulin receptor gene with the complement component 3, LDL receptor, apolipoprotein C2 and myotonic dystrophy loci on chromosome 19. *Hum. Genet.* 74: 267–269, 1986.

[31342] 4894. Blangy, A.; Lane, H. A.; d'Herin, P.; Harper, M.; Kress, M.; Nigg, E. A.: Phosphorylation by p34(cdc2) regulates spindle association of human Eg5, a kinesin-related motor essential for bipolar spindle formation in vivo. *Cell* 83: 1159–1169, 1995.

[31343] 4895. Shimada, F.; Suzuki, Y.; Taira, M.; Hashimoto, N.; Nozaki, O.; Makino, H.; Yoshida, S.: Abnormal messenger ribonucleic acid (mRNA) transcribed from a mutant insulin receptor gene in a patient with type A insulin resistance. *Diabetologia* 35: 639–644, 1992.

[31344] 4896. Shimada, F.; Taira, M.; Suzuki, Y.; Hashimoto, N.;

Nozaki, O.;Taira, M.; Tatibana, M.; Ebina, Y.; Tawata, M.; Onaya, T.; Makino,H.; Yoshida, S.: Insulin-resistant diabetes associated with partialdeletion of insulin-receptor gene. Lancet 335: 1179–1181, 1990.

[31345] 4897.Taira, M.; Taira, M.; Hashimoto, N.; Shimada, F.; Suzuki, Y.;Kanatsuka, A.; Nakamura, F.; Ebina, Y.; Tati-bana, M.; Makino, H.;Yoshida, S.: Human diabetes associ-ated with a deletion of the tyrosinekinase domain of the insulin receptor. Science 245: 63–66, 1989.

[31346] 4898.Takahashi, Y.; Kadowaki, H.; Ando, A.; Quin, J. D.; MacCuish,A. C.; Yazaki, Y.; Akanuma, Y.; Kadowaki, T.: Two aberrant splicingscaused by mutations in the insulin receptor gene in cultured lymphocytesfrom a patient with Rabson–Mendenhall's syndrome. J. Clin. Invest. 101:588–594, 1998.

[31347] 4899.Taylor, S. I.; Cama, A.; Accili, D.; Barbetti, F.; Imano, E.;Kadowaki, H.; Kadowaki, T.: Genetic basis of endocrine disease 1:molecular genetics of insulin resistant diabetes mellitus. J. Clin.Endocr. Metab. 73: 1158–1163, 1991.

[31348] 4900.Taylor, S. I.; Marcus–Samuels, B.; Ryan–Young, J.; Leventhal,S.; Elders, M. J.: Genetics of the insulin receptor defect in a patientwith extreme insulin resistance. J. Clin. Endocr. Metab. 62: 1130–1135,1986.

- [31349] 4901. Taylor, S. I.; Underhill, L. H.; Hedo, J. A.; Roth, J.; Serrano Rios, M.; Blizzard, R. M.: Decreased insulin binding to cultured cells from a patient with the Rabson-Mendenhall syndrome: dichotomy between studies with cultured lymphocytes and cultured fibroblasts. *J. Clin. Endocr. Metab.* 56: 856–861, 1983.
- [31350] 4902. Tevaarwerk, G. J. M.; Strickland, K. P.; Lin, C. H.; Hudson, A. J.: Studies on insulin resistance and insulin receptor binding in myotonia dystrophica. *J. Clin. Endocr. Metab.* 49: 216–222, 1979.
- [31351] 4903. Ullrich, A.; Bell, J. R.; Chen, E. Y.; Herrera, R.; Petruzzelli, L. M.; Dull, T. J.; Gray, A.; Coussens, L.; Liao, Y.-C.; Tsubokawa, M.; Mason, A.; Seeburg, P. H.; Grunfeld, C.; Rosen, O. M.; Ramachandran, J.: Human insulin receptor and its relationship to the tyrosine kinase family of oncogenes. *Nature* 313: 756–761, 1985.
- [31352] 4904. van der Vorm, E. R.; Kuipers, A.; Kielkopf-Renner, S.; Krans, H. M. J.; Moller, W.; Maassen, J. A.: A mutation in the insulin receptor that impairs proreceptor processing but not insulin binding. *J. Biol. Chem.* 269: 14297–14302, 1994.
- [31353] 4905. van der Vorm, E. R.; van der Zon, G. C. M.; Moller, W.; Krans, H. M. J.; Lindhout, D.; Maassen, J. A.: An arg for

gly substitution at position 31 in the insulin receptor, linked to insulin resistance, inhibits receptor processing and transport. *J. Biol. Chem.* 267:66–71, 1992.

[31354] 4906. Ward, G. M.; Harrison, L. C.: Structure of the human erythrocyte insulin receptor. *Diabetes* 35: 101–105, 1986.

[31355] 4907. Wertheimer, E.; Lu, S.-P.; Backeljauw, P. F.; Davenport, M. L.; Taylor, S. I.: Homozygous deletion of the human insulin receptor gene results in leprechaunism. *Nature Genet.* 5: 71–73, 1993.

[31356] 4908. Williams, D. L.; Look, A. T.; Melvin, S. L.; Roberson, P. K.; Dahl, G.; Flake, T.; Stass, S.: New chromosomal translocations correlate with specific immunophenotypes of childhood acute lymphoblastic leukemia. *Cell* 36:101–109, 1984.

[31357] 4909. Williams, J. F.; McClain, D. A.; Dull, T. J.; Ullrich, A.; Olefsky, J. M.: Characterization of an insulin receptor mutant lacking the subunit processing site. *J. Biol. Chem.* 265: 8463–8469, 1990.

[31358] 4910. Yamamoto-Honda, R.; Koshio, O.; Tobe, K.; Shibasaki, Y.; Momomura, K.; Odawara, M.; Kadowaki, T.; Takaku, F.; Akanuma, Y.; Kasuga, M.: Phosphorylation state and biological function of a mutant human insulin receptor val(996). *J. Biol. Chem.* 265: 14777–14783,

1990.

- [31359] 4911. Yang-Feng, T. L.; Francke, U.; Ullrich, A.: Gene for human insulin receptor: localization to site on chromosome 19 involved in pre-B-cell leukemia. *Science* 228: 728–731, 1985.
- [31360] 4912. Yoshimasa, Y.; Seino, S.; Whittaker, J.; Kakehi, T.; Kosaki, A.; Kuzuya, H.; Imura, H.; Bell, G. I.; Steiner, D. F.: Insulin-resistant diabetes due to a point mutation that prevents insulin proreceptor processing. *Science* 240: 784–787, 1988.
- [31361] 4913. Kurachi, H.; Jobo, K.; Ohta, M.; Kawasaki, T.; Itoh, N.: A new member of the insulin receptor family, insulin receptor-related receptor, is expressed preferentially in the kidney. *Biochem. Biophys. Res. Commun.* 187: 934–939, 1992.
- [31362] 4914. Shier, P.; Watt, V. M.: Primary structure of a putative receptor for a ligand of the insulin family. *J. Biol. Chem.* 264: 14605–14608, 1989.
- [31363] 4915. Shier, P.; Willard, H. F.; Watt, V. M.: Localization of the insulin receptor-related receptor gene to human chromosome 1. *Cytogenet. Cell Genet.* 54: 80–81, 1990.
- [31364] 4916. Whitmore, T. E.; Maurer, M. F.; Day, H. L.; Jelmberg, A. C.; Dasovich, M. M.; Sundborg, L. M.; Burkhead, S. K.;

Heipel, M. D.; Madden, K.L.; Kramer, J. M.; Kuijper, J. L.; Xu, W. F.; Jaspers, S. R.; Holly, R. D.; Lok, S.: The assignment of the human insulin receptor-related receptor gene (INSRR) to chromosome 1q21-q23 by the use of radiation hybrid mapping. *Cytogenet. Cell Genet.* 87: 93-94, 1999.

[31365] 4917. Alnemri, E. S.; Livingston, D. J.; Nicholson, D. W.; Salvesen, G.; Thornberry, N. A.; Wong, W. W.; Yuan, J.: Human ICE/CED-3 protease nomenclature. (Letter) *Cell* 87: 171 only, 1996.

[31366] 4918. Casano, F. J.; Rolando, A. M.; Mudgett, J. S.; Moliniaux, S. M.: The structure and complete nucleotide sequence of the murine gene encoding interleukin-1-beta converting enzyme (ICE). *Genomics* 20:474-481, 1994.

[31367] 4919. Cerretti, D. P.; Hollingsworth, L. T.; Kozlosky, C. J.; Valentine, M. B.; Shapiro, D. N.; Morris, S. W.; Nelson, N.: Molecular characterization of the gene for human interleukin-1-beta converting enzyme (IL1BC). *Genomics* 20:468-473, 1994.

[31368] 4920. Cerretti, D. P.; Kozlosky, C. J.; Mosley, B.; Nelson, N.; Van Ness, K.; Greenstreet, T. A.; March, C. J.; Kronheim, S. R.; Druck, T.; Cannizzaro, L. A.; Huebner, K.; Black, R. A.: Molecular cloning of the interleukin-1-beta converting en-

zyme. Science 256: 97–100, 1992.

- [31369] 4921.Humke, E. W.; Ni, J.; Dixit, V. M.: ERICE, a novel FLICE–activatablecaspase. J. Biol. Chem. 273: 15702–15707, 1998.
- [31370] 4922.Koenig, U.; Eckhart, L.; Tschachler, E.: Evidence that caspase–13is not a human but a bovine gene. Biochem. Biophys. Res. Commun. 285:1150–1154, 2001.
- [31371] 4923.Baker, E.; Hort, Y. J.; Ball, H.; Sutherland, G. R.; Shine, J.;Herzog, H.: Assignment of the human neuropeptide Y gene to chromosome7p15.1 by nonisotopic in situ hybridization. Genomics 26: 163–164,1995.
- [31372] 4924.Carr, L. G.; Foroud, T.; Bice, P.; Gobbett, T.; Ivashina, J.; Edenberg,H.; Lumeng, L.; Li, T. K.: A quantitative trait locus for alcoholconsumption in selectively bred rat lines. Alcohol Clin. Exp. Res. 22:884–887, 1998.
- [31373] 4925.Dockray, G. J.: Neuropeptide Y: in search of a function. Neurochem.Int. 8: 9–11, 1986.
- [31374] 4926.Erickson, J. C.; Clegg, K. E.; Palmiter, R. D.: Sensitivity toleptin and susceptibility to seizures of mice lacking neuropeptideY. Nature 381: 415–421, 1996.
- [31375] 4927.Erickson, J. C.; Hollopeter, G.; Palmiter, R. D.: Attenuationof the obesity syndrome of ob/ob mice by the loss of neuropeptideY. Science 274: 1704–1706, 1996.

- [31376] 4928.Hansel, D. E.; Eipper, B. A.; Ronnett, G. V.: Neuropeptide Y functions as a neuroproliferative factor. *Nature* 410: 940–943, 2001.
- [31377] 4929.Karvonen, M. K.; Koulu, M.; Pesonen, U.; Uusitupa, M. I. J.; Tammi, A.; Viikari, J.; Simell, O.; Ronnema, T.: Leucine 7 to proline 7 polymorphism in the prepro-neuropeptide Y is associated with birthweight and serum triglyceride concentration in preschool-aged children. *J.Clin. Endocr. Metab.* 85: 1455–1460, 2000.
- [31378] 4930.Karvonen, M. K.; Pesonen, U.; Koulu, M.; Niskanen, L.; Laakso, M.; Rissanen, A.; Dekker, J. M.; 't Hart, L. M.; Valve, R.; Uusitupa, M. I.: Association of a leucine(7)–to–proline(7) polymorphism in the signal peptide of neuropeptide Y with high serum cholesterol and LDL cholesterol levels. *Nature Med.* 4: 1434–1437, 1998.
- [31379] 4931.Kauhanen, J.; Karvonen, M. K.; Pesonen, U.; Koulu, M.; Tuomainen, T.–P.; Uusitupa, M. I. J.; Salonen, J. T.: Neuropeptide Y polymorphism and alcohol consumption in middle-aged men. *Am. J. Med. Genet.* 93:117–121, 2000.
- [31380] 4932.Maccarrone, C.; Jarrott, B.: Neuropeptide Y: a putative neurotransmitter. *Neurochem.Int.* 8: 13–22, 1986.
- [31381] 4933.Meisler, M. H.; Spence, J. E.; Dixon, J. E.; Caldwell, R. M.; Minth, C. D.; Beaudet, A. L.: Exclusion of close linkage

between the loci for cystic fibrosis and neuropeptide Y on human chromosome 7. *Cytogenet. Cell Genet.* 44: 175–176, 1987.

[31382] 4934. Minth, C. D.; Andrews, P. C.; Dixon, J. E.: Characterization, sequence, and expression of the cloned human neuropeptide Y gene. *J. Biol. Chem.* 261: 11974–11979, 1986.

[31383] 4935. Minth, C. D.; Bloom, S. R.; Polak, J. M.; Dixon, J. E.: Cloning, characterization, and DNA sequence of a human cDNA encoding neuropeptide tyrosine. *Proc. Nat. Acad. Sci.* 81: 4577–4581, 1984.

[31384] 4936. Takeuchi, T.; Gumucio, D.; Eddy, R.; Meisler, M.; Minth, C.; Dixon, J.; Yamada, T.; Shows, T.: Assignment of the related pancreatic polypeptide (PPY) and neuropeptide Y (NPY) genes to regions on human chromosomes 17 and 7. (Abstract) *Cytogenet. Cell Genet.* 40: 759 only, 1985.

[31385] 4937. Takeuchi, T.; Gumucio, D. L.; Yamada, T.; Meisler, M. H.; Minth, C. D.; Dixon, J. E.; Eddy, R. E.; Shows, T. B.: Genes encoding pancreatic polypeptide and neuropeptide Y are on human chromosomes 17 and 7. *J. Clin. Invest.* 77: 1038–1041, 1986.

[31386] 4938. Terenghi, G.; Polak, J. M.; Hamid, Q.; O'Brien, E.; Denny, P.; Legon, S.; Dixon, J.; Minth, C. D.; Palay, S. L.;

Yasargil, G.; Chan-Palay, V.: Localization of neuropeptide Y mRNA in neurons of human cerebral cortex by means of in situ hybridization with a complementary RNA probe. *Proc. Nat. Acad. Sci.* 84: 7315–7318, 1987.

[31387] 4939. Thiele, T. E.; Marsh, D. J.; Ste. Marie, L.; Bernstein, I. L.; Palmiter, R. D.: Ethanol consumption and resistance are inversely related to neuropeptide Y levels. *Nature* 396: 366–369, 1998.

[31388] 4940. Niskanen, L.; Karvonen, M. K.; Valve, R.; Koulu, M.; Pesonen, U.; Mercuri, M.; Rauramaa, R.; Toyry, J.; Laakso, M.; Uusitupa, M. I. J.: Leucine 7 to proline 7 polymorphism in the neuropeptide Y gene is associated with enhanced carotid atherosclerosis in elderly patients with type 2 diabetes and control subjects. *J. Clin. Endocr. Metab.* 85: 2266–2269, 2000.

[31389] 4941. Uusitupa, M. I. J.; Karvonen, M. K.; Pesonen, U.; Koulu, M.: Neuropeptide Y: a novel link between the neuroendocrine system and cholesterol metabolism. *Ann. Med.* 30: 508–510, 1998.

[31390] 4942. Herzog, H.; Baumgartner, M.; Vivero, C.; Selbie, L. A.; Auer, B.; Shine, J.: Genomic organization, localization, and allelic differences in the gene for the human neuropeptide Y Y1 receptor. *J. Biol. Chem.* 268: 6703–6707,

1993.

- [31391] 4943. Herzog, H.; Darby, K.; Ball, H.; Hort, Y.; Beck-Sickinger, A.; Shine, J.: Overlapping gene structure of the human neuropeptide Y receptor subtypes Y1 and Y5 suggests coordinate transcriptional regulation. *Genomics* 41:315–319, 1997.
- [31392] 4944. Herzog, H.; Hort, Y. J.; Ball, H. J.; Hayes, G.; Shine, J.; Selbie, L. A.: Cloned human neuropeptide Y receptor couples to two different second messenger systems. *Proc. Nat. Acad. Sci.* 89: 5794–5798, 1992.
- [31393] 4945. Larhammar, D.; Blomqvist, A. G.; Yee, F.; Jazin, E.; Yoo, H.; Wahlestedt, C.: Cloning and functional expression of a human neuropeptide Y/peptide YY receptor of the Y1 type. *J. Biol. Chem.* 267: 10935–10938, 1992.
- [31394] 4946. Lutz, C. M.; Frankel, W. N.; Richards, J. E.; Thompson, D. A.: Neuropeptide Y receptor genes on human chromosome 4q31–q32 map to conserved linkage groups on mouse chromosomes 3 and 8. *Genomics* 41:498–500, 1997.
- [31395] 4947. Naveilhan, P.; Hassani, H.; Lucas, G.; Blakeman, K. H.; Hao, J.-X.; Xu, X.-J.; Wiesenfeld-Hallin, Z.; Thoren, P.; Ernfor, P.: Reduced antinociception and plasma extravasation in mice lacking a neuropeptide Y receptor. *Nature*

409: 513–517, 2001.

- [31396] 4948.Ammar, D. A.; Eadie, D. M.; Wong, D. J.; Ma, Y.–Y.; Kolakowski, L. F., Jr.; Yang–Feng, T. L.; Thompson, D. A.: Characterization of the human type 2 neuropeptide Y receptor gene (NPY2R) and localization to the chromosome 4q region containing the type 1 neuropeptide Y receptor–gene. *Genomics* 38: 392–398, 1996.
- [31397] 4949.Gerald, C.; Walker, M. W.; Vaysse, P. J.–J.; He, C.; Branchek, T. A.; Weinshank, R. L.: Expression cloning and pharmacological characterization of a human hippocampal neuropeptide Y/peptide YY Y2 receptor subtype. *J.Biol. Chem.* 270: 26758–26761, 1995.
- [31398] 4950.Weydert, A.; Daubas, P.; Lazaridis, I.; Barton, P.; Garner, I.; Leader, D. P.; Bonhomme, F.; Catalan, J.; Simon, D.; Guenet, J. L.; Gros, F.; Buckingham, M. E.: Genes for skeletal muscle myosin heavy chains are clustered and are not located on the same mouse chromosome as a cardiac myosin heavy chain gene. *Proc. Nat. Acad. Sci.* 82:7183–7187, 1985.
- [31399] 4951.Jandreski, M. A.; Sole, M. J.; Liew, C.–C.: Two different forms of beta myosin heavy chain are expressed in human striated muscle. *Hum.Genet.* 77: 127–131, 1987.
- [31400] 4952.Leinwand, L. A.; Fournier, R. E. K.; Nadal–Ginard, B.;

Shows, T.B.: Multigene family for sarcomeric myosin heavy chain in mouse and human DNA: localization on a single chromosome. *Science* 221: 766–769, 1983.

[31401] 4953. Leinwand, L. A.; Fournier, R. E. K.; Nadal-Ginard, B.; Shows, T.B.: Assignment of the sarcomeric myosin heavy chain multigene family to chromosome 17 in humans and chromosome 11 in the mouse. (Abstract) *Cytogenet. Cell Genet.* 37: 521–522, 1984.

[31402] 4954. Leinwand, L. A.; Saez, L.; McNally, E.; Bernardo, N.-G.: Isolation and characterization of human myosin heavy chain genes. *Proc. Nat. Acad. Sci.* 80: 3716–3720, 1983.

[31403] 4955. Soussi-Yanicostas, N.; Whalen, R. G.; Petit, C.: Five skeletal myosin heavy chain genes are organized as a multigene complex in the human genome. *Hum. Molec. Genet.* 2: 563–569, 1993.

[31404] 4956. Weiss, A.; McDonough, D.; Wertman, B.; Acakpo-Satchivi, L.; Montgomery, K.; Kucherlapati, R.; Leinwand, L.; Krauter, K.: Organization of human and mouse skeletal myosin heavy chain gene clusters is highly conserved. *Proc. Nat. Acad. Sci.* 96: 2958–2963, 1999.

[31405] 4957. Yoon, S.-J.; Seiler, S. H.; Kucherlapati, R.; Leinwand, L.: Organization of the human skeletal myosin heavy chain

gene cluster. Proc. Nat.Acad. Sci. 89: 12078–12082, 1992.

- [31406] 4958.Karsch–Mizrachi, I.; Feghali, R.; Shows, T. B.; Leinwand, L. A.: Generation of a full–length human perinatal myosin heavy chain cDNA. Gene 89:289–294, 1990.
- [31407] 4959.Weiss, A.; Schiaffino, S.; Leinwand, L. A.: Comparative sequenceanalysis of the complete human sarcomeric myosin heavy chain family:implications for functional diversity. J. Molec. Biol. 290: 61–75,1999.
- [31408] 4960.Castilla, L. H.; Garrett, L.; Adya, N.; Orlic, D.; Dutra, A.; Anderson,S.; Owens, J.; Eckhaus, M.; Bodine, D.; Liu, P. P.: The fusion geneCbfb–MYH11 blocks myeloid differentiation and predisposes mice toacute myelomonocytic leukaemia. (Letter) Nature Genet. 23: 144–146,1999.
- [31409] 4961.Deng, Z.; Liu, P.; Marlton, P.; Claxton, D. F.; Lane, S.; Callen,D. F.; Collins, F. S.; Siciliano, M. J.: Smooth muscle myosin heavychain locus (MYH11) maps to 16p13.13–p13.12 and establishes a newregion of conserved synteny between human 16p and mouse 16. Genomics 18:156–159, 1993.
- [31410] 4962.Stewart, R. J.; Pesavento, P. A.; Woerpel, D. N.; Goldstein, L.S. B.: Identification and partial characterization of six membersof the kinesin superfamily in Drosophila.

Proc. Nat. Acad. Sci. 88:8470–8474, 1991.

- [31411] 4963.Tihy, F.; Kress, M.; Harper, M.; Dutrillaux, B.; Lemieux, N.:Localization of the human kinesin-related gene to band 10q24 by fluorescencein situ hybridization. Genomics 13: 1371–1372, 1992.
- [31412] 4964.Shimura, H.; Schlossmacher, M. G.; Hattori, N.; Frosch, M. P.;Trockenbacher, A.; Schneider, R.; Mizuno, Y.; Kosik, K. S.; Selkoe,D. J.: Ubiquitination of a new form of alpha-synuclein by parkinfrom human brain: implications for Parkinson's disease. Science 293:263–269, 2001.
- [31413] 4965.Spillantini, M. G.; Divane, A.; Goedert, M.: Assignment of humanalpha-synuclein (SNCA) and beta-synuclein (SNCB) genes to chromosomes4q21 and 5q35. Genomics 27: 379–381, 1995.
- [31414] 4966.Spillantini, M. G.; Schmidt, M. L.; Lee, V. M.–Y.; Trojanowski,J. Q.; Jakes, R.; Goedert, M.: Alpha-synuclein in Lewy bodies. Nature 388:839–840, 1997.
- [31415] 4967.Calogero, S.; Grassi, F.; Aguzzi, A.; Voigtlander, T.; Ferrier,P.; Ferrari, S.; Bianchi, M. E.: The lack of chromosomal proteinHmg1 does not disrupt cell growth but causes lethal hypoglycaemiain newborn mice. Nature Genet. 22: 276–280, 1999.
- [31416] 4968.Ferrari, S.; Finelli, P.; Rocchi, M.; Bianchi, M. E.: The

activegene that encodes human high mobility group 1 protein (HMG1) containsintrons and maps to chromosome 13. Genomics 35: 367–371, 1996.

[31417] 4969.Scaffidi, P.; Misteli, T.; Bianchi, M. E.: Release of chromatinprotein HMGB1 by necrotic cells triggers inflammation. Nature 418:191–195, 2002.

[31418] 4970.Taguchi, A.; Blood, D. C.; del Toro, G.; Canet, A.; Lee, D. C.;Qu, W.; Tanji, N.; Lu, Y.; Lalla, E.; Fu, C.; Hofmann, M. A.; Kislinger,T.; Ingram, M.; Lu, A.; Tanaka, H.; Hori, O.; Ogawa, S.; Stern, D.M.; Schmidt, A. M.: Blockade of RAGE–amphoterin signalling suppresses tumour growth and metastases. Nature 405: 354–360, 2000.

[31419] 4971.Wang, H.; Bloom, O.; Zhang, M.; Vishnubhakat, J. M.; Ombrellino,M.; Che, J.; Frazier, A.; Yang, H.; Ivanova, S.; Borovikova, L.; Manogue,K. R.; Faist, E.; Abraham, E.; Andersson, J.; Andersson, U.; Molina,P. E.; Abumrad, N. N.; Sama, A.; Tracey, K. J.: HMG–1 as a late mediatorof endotoxin lethality in mice. Science 285: 248–251, 1999.

[31420] 4972.Cetta, F.; Chiappetta, G.; Melillo, R. M.; Petracci, M.; Montalto,G.; Santoro, M.; Fusco, A.: The ret/ptc1 oncogene is activated infamilial adenomatous polyposis–associated thyroid papillary carcinomas. J.Clin. Endocr. Metab. 83: 1003–1006, 1998.

- [31421] 4973.Decker, R. A.; Peacock, M. L.; Watson, P.:
Hirschsprung disease in MEN 2A: increased spectrum of
RET exon 10 genotypes and strong genotype–phenotype
correlation. *Hum. Molec. Genet.* 7: 129–134, 1998.
- [31422] 4974.Donis–Keller, H.; Dou, S.; Chi, D.; Carlson, K. M.;
Toshima, K.; Lairmore, T. C.; Howe, J. R.; Moley, J. F.;
Goodfellow, P.; Wells, S. A., Jr.: Mutations in the RET proto-
oncogene are associated with MEN 2A and FMTC. *Hum.*
Molec. Genet. 2: 851–856, 1993.
- [31423] 4975.Doray, B.; Salomon, R.; Amiel, J.; Pelet, A.; Touraine,
R.; Billaud, M.; Attie, T.; Bachy, B.; Munnich, A.; Lyonnet, S.:
Mutation of the RET ligand, neurturin, supports multigenic
inheritance in Hirschsprung disease. *Hum. Molec. Genet.* 7:
1449–1452, 1998.
- [31424] 4976.Edery, P.; Lyonnet, S.; Mulligan, L. M.; Pelet, A.; Dow,
E.; Abel, L.; Holder, S.; Nihoul–Fekete, C.; Ponder, B. A. J.;
Munnich, A.: Mutations of the RET proto–oncogene in
Hirschsprung's disease. *Nature* 367:378–380, 1994.
- [31425] 4977.Eng, C.: The RET proto–oncogene in multiple en-
docrine neoplasia type 2 and Hirschsprung's disease. *New*
Eng. J. Med. 335: 943–951, 1996.
- [31426] 4978.Eng, C.; Crossey, P. A.; Mulligan, L. M.; Healey, C. S.;
Houghton, C.; Prowse, A.; Chew, S. L.; Dahia, P. L. M.;

O'Riordan, J. L. H.; Toledo, S. P. A.; Smith, D. P.; Maher, E. R.; Ponder, B. A. J.: Mutations in the RET proto-oncogene and the von Hippel-Lindau disease tumour suppressor gene in sporadic and syndromic pheochromocytomas. *J. Clin. Genet.* 32: 934-937, 1995.

[31427] 4979. Eng, C.; Mulligan, L. M.: Mutations of the RET proto-oncogene in the multiple endocrine neoplasia type 2 syndromes, related sporadic tumours, and Hirschsprung disease. *Hum. Mutat.* 9: 97-109, 1997.

[31428] 4980. Eng, C.; Mulligan, L. M.; Smith, D. P.; Healey, C. S.; Frilling, A.; Raue, F.; Neumann, H. P. H.; Pfragner, R.; Behmel, A.; Lorenzo, M. J.; Stonehouse, T. J.; Ponder, M. A.; Ponder, B. A. J.: Mutation of the RET proto-oncogene in sporadic medullary thyroid carcinoma. *Genes Chromosomes Cancer* 12: 209-212, 1995.

[31429] 4981. Eng, C.; Smith, D. P.; Mulligan, L. M.; Healey, C. S.; Zvelebil, M. J.; Stonehouse, T. J.; Ponder, M. A.; Jackson, C. E.; Waterfield, M. D.; Ponder, B. A. J.: A novel point mutation in the tyrosine kinase domain of the RET proto-oncogene in sporadic medullary thyroid carcinoma and in a family with FMTC. *Oncogene* 10: 509-513, 1995.

[31430] 4982. Eng, C.; Smith, D. P.; Mulligan, L. M.; Nagai, M. A.; Healey, C. S.; Ponder, M. A.; Gardner, E.; Scheumann, G. F.

W.; Jackson, C.E.; Tunnacliffe, A.; Ponder, B. A. J.: Point mutation within the tyrosine kinase domain of the RET proto-oncogene in multiple endocrine neoplasia type 2B and related sporadic tumors. *Hum. Molec. Genet.* 3:237–241, 1994.

[31431] 4983. Fearon, E. R.: Human cancer syndromes: clues to the origin and nature of cancer. *Science* 278: 1043–1050, 1997.

[31432] 4984. Fitze, G.; Schreiber, M.; Kuhlisch, E.; Schackert, H. K.; Roesner, D.: Association of RET protooncogene codon 45 polymorphism with Hirschsprung disease. (Letter) *Am. J. Hum. Genet.* 65: 1469–1473, 1999.

[31433] 4985. Gardner, E.; Mulligan, L. M.; Eng, C.; Healey, C. S.; Kwok, J. B. J.; Ponder, M. A.; Ponder, B. A. J.: Haplotype analysis of MEN2 mutations. *Hum. Molec. Genet.* 3: 1771–1774, 1994.

[31434] 4986. Grieco, M.; Santoro, M.; Berlingieri, M. T.; Melillo, R. M.; Donghi, R.; Bongarzone, I.; Pierotti, M. A.; Della Porta, G.; Fusco, A.; Vecchio, G.: PTC is a novel rearranged form of the ret proto-oncogene and is frequently detected in vivo in human thyroid papillary carcinomas. *Cell* 60:557–563, 1990.

[31435] 4987. Hofstra, R. M. W.; Landsvater, R. M.; Ceccherini, I.;

Stulp, R.P.; Stelwagen, T.; Luo, Y.; Pasini, B.; Hoppener, J. W. M.; Ploosvan Amstel, H. K.; Romeo, G.; Lips, C. J. M.; Buys, C. H. C. M.:A mutation in the RET proto-oncogene associated with multiple endocrineneoplasia type 2B and sporadic medullary thyroid carcinoma. *Nature* 367:375–376, 1994.

[31436] 4988.Hoppener, J. W. M.; Lips, C. J. M.: RET receptor tyrosine kinase gene mutations: molecular biological, physiological and clinical aspects. *Europ.J. Clin. Invest.* 26: 613–624, 1996.

[31437] 4989.Hoppner, W.; Ritter, M. M.: A duplication of 12 bp in the critical cysteine rich domain of the RET proto-oncogene results in a distinct phenotype of multiple endocrine neoplasia type 2A. *Hum. Molec. Genet.* 6:587–590, 1997.

[31438] 4990.Ikeda, I.; Ishizaka, Y.; Tahira, T.; Suzuki, T.; Onda, M.; Sugimura, T.; Nagao, M.: Specific expression of the ret proto-oncogene in human neuroblastoma cell lines. *Oncogene* 5: 1291–1296, 1990.

[31439] 4991.Ishizaka, Y.; Itoh, F.; Tahira, T.; Ikeda, I.; Sugimura, T.; Tucker, J.; Fertitta, A.; Carrano, A. V.; Nagao, M.: Human ret proto-oncogene mapped to chromosome 10q11.2. *Oncogene* 4: 1519–1521, 1989.

- [31440] 4992.Japon, M. A.; Urbano, A. G.; Saez, C.; Segura, D. I.; Cerro, A.L.; Dieguez, C.; Alvarez, C. V.: Glial-derived neurotrophic factor and RET gene expression in normal human anterior pituitary cell types and in pituitary tumors. *J. Clin. Endocr. Metab.* 87: 1879–1884, 2002.
- [31441] 4993.Julies, M. G.; Moore, S. W.; Kotze, M. J.; du Plessis, L.: Novel RET mutations in Hirschsprung's disease patients from the diverse South African population. *Europ. J. Hum. Genet.* 9: 419–423, 2001.
- [31442] 4994.Klugbauer, S.; Demidchik, E. P.; Lengfelder, E.; Rabes, H. M.: Detection of a novel type of RET rearrangement (PTC5) in thyroid carcinomas after Chernobyl and analysis of the involved RET-fused gene RFG5. *Cancer Res.* 58: 198–203, 1998.
- [31443] 4995.de Oca Luna, R. M.; Tabor, A. D.; Eberspaecher, H.; Hulboy, D.L.; Worth, L. L.; Colman, M. S.; Finlay, C. A.; Lozano, G.: The organization and expression of the mdm2 gene. *Genomics* 33: 352–357, 1996.
- [31444] 4996.Watson, C. E.; Draganov, D. I.; Billecke, S. S.; Bisgaier, C.L.; La Du, B. N.: Rabbits possess a serum paraoxonase polymorphism similar to the human Q192R. *Pharmacogenetics* 11: 123–134, 2001.
- [31445] 4997.Rajput, B.; Marshall, A.; Killary, A. M.; Lalley, P. A.;

Naylor, S. L.; Belin, D.; Rickles, R. J.; Strickland, S.: Chromosomal assignments of genes for tissue plasminogen activator and urokinase in mouse. *Somat. Cell Molec. Genet.* 13: 581–586, 1987.

- [31446] 4998. Yamasaki, Y.; Sakamoto, K.; Watada, H.; Kajimoto, Y.; Hori, M.: The arg-192 isoform of paraoxonase with low sarin-hydrolyzing activity is dominant in the Japanese. *Hum. Genet.* 101: 67–68, 1997.
- [31447] 4999. Fong, L.; Brockstedt, D.; Benike, C.; Breen, J. K.; Strang, G.; Ruegg, C. L.; Engleman, E. G.: Dendritic cell-based xenoantigen vaccination for prostate cancer immunotherapy. *J. Immun.* 167: 7150–7156, 2001.
- [31448] 5000. Li, S. S.-L.; Sharief, F. S.: The prostatic acid phosphatase (ACPP) gene is localized to human chromosome 3q21–q23. *Genomics* 17: 765–766, 1993.
- [31449] 5001. Sharief, F. S.; Lee, H.; Leuderman, M. M.; Lundwall, A.; Deaven, L. L.; Lee, C.; Li, S. S.-L.: Human prostatic acid phosphatase: cDNA cloning, gene mapping and protein sequence homology with lysosomal acid phosphatase. *Biochem. Biophys. Res. Commun.* 160: 79–86, 1989.
- [31450] 5002. Sharief, F. S.; Li, S. S.-L.: Nucleotide sequence of human prostatic acid phosphatase ACPP gene, including 7 Alu repeats. *Biochem. Molec. Biol. Int.* 33: 561–565, 1994.

- [31451] 5003.Sharief, F. S.; Li, S. S.-L.: Structure of human prostatic acidphosphatase gene. *Biochem. Biophys. Res. Commun.* 184: 1468–1476,1992.
- [31452] 5004.Tailor, P. G.; Govindan, M. V.; Patel, P. C.: Nucleotide sequence of human prostatic acid phosphatase determined from a full-length cDNA clone. *Nucleic Acids Res.* 18: 4928 only, 1990.
- [31453] 5005.Winqvist, R.; Virkkunen, P.; Grzeschik, K.-H.; Vihko, P.: Chromosomal localization to 3q21-qter and two TaqI RFLPs of the human prostate-specific acid phosphatase gene (ACPP). *Cytogenet. Cell Genet.* 52: 68–71,1989.
- [31454] 5006.Winqvist, R.; Virkkunen, P.; Grzeschik, K.-H.; Vihko, P.: The gene for prostate-specific acid phosphatase is located in segment q21-qter of human chromosome 3 and shows two RFLPs with TaqI. (Abstract) *Cytogenet. Cell Genet.* 51: 1108 only, 1989.
- [31455] 5007.Yeh, L.-C. C.; Lee, A. J.; Lee, N. E.; Lam, K.-W.; Lee, J. C.: Molecular cloning of cDNA for human prostatic acid phosphatase. *Gene* 60:191–196, 1987.
- [31456] 5008.Badger, K. S.; Sussman, H. H.: Structural evidence that human liver and placental alkaline phosphatase isoenzymes are coded by different genes. *Proc. Nat. Acad. Sci.* 73: 2201–2205, 1976.

- [31457] 5009.Beckman, L.; Beckman, G.; Christodoulou, C.; Ifekwunigwe, A.:Variations in human placental alkaline phosphatase. *Acta Genet. Statist.Med.* 17: 406–412, 1967.
- [31458] 5010.Beckman, L.; Bjorling, G.; Christodoulou, C.: Pregnancy enzymesand placental polymorphism: alkaline phosphatase. *Acta Genet. Statist.Med.* 16: 59–73, 1966.
- [31459] 5011.Boyer, S. H.: Alkaline phosphatase in human sera and placenta. *Science* 134:1002–1004, 1961.
- [31460] 5012.Antalis, T. M.; Clark, M. A.; Barnes, T.; Lehrbach, P. R.; Devine,P. L.; Schevzov, G.; Goss, N. H.; Stephens, R. W.; Tolstoshev, P.: Cloning and expression of a cDNA coding for a human monocyte–derivedplasminogen activator inhibitor. *Proc. Nat. Acad. Sci.* 85: 985–989,1988.
- [31461] 5013.Bartuski, A. J.; Kamachi, Y.; Schick, C.; Overhauser, J.; Silverman,G. A.: Cytoplasmic antiproteinase 2 (PI8) and bomapin (PI10) mapto the serpin cluster at 18q21.3. *Genomics* 43: 321–328, 1997.
- [31462] 5014.Dickinson, J. L.; Bates, E. J.; Ferrante, A.; Antalis, T. M.:Plasminogen activator inhibitor type 2 inhibits tumor necrosis factoralpha–induced apoptosis: evidence for an alternate biological function. *J.Biol. Chem.* 270: 27894–27904, 1995.
- [31463] 5015.Dougherty, K. M.; Pearson, J. M.; Yang, A. Y.;

Westrick, R. J.; Baker, M. S.; Ginsburg, D.: The plasminogen activator inhibitor-2 gene is not required for normal murine development or survival. *Proc. Nat. Acad. Sci.* 96: 686–691, 1999.

[31464] 5016. Kruithof, E. K. O.; Baker, M. S.; Bunn, C. L.: Biological and clinical aspects of plasminogen activator inhibitor type 2. *Blood* 86:4007–4024, 1995.

[31465] 5017. Oldenburg, M.; Wijnen, J. T.; van den Berg, E. A.; le Clercq, E.; Kooistra, T.; Meera Khan, P.: Assignment of plasminogen activator-inhibitor type 2 (PAI2) to chromosome 18. (Abstract) *Cytogenet. Cell Genet.* 51:1055, 1989.

[31466] 5018. Samia, J. A.; Alexander, S. J.; Horton, K. W.; Auron, P. E.; Byers, M. G.; Shows, T. B.; Webb, A. C.: Chromosomal organization and localization of the human urokinase inhibitor gene: perfect structural conservation with ovalbumin. *Genomics* 6: 159–167, 1990.

[31467] 5019. Silverman, G. A.; Jockel, J. I.; Domer, P. H.; Mohr, R. M.; Taillon-Miller, P.; Korsmeyer, S. J.: Yeast artificial chromosome cloning of a two-megabase-size contig within chromosomal band 18q21 establishes physical linkage between BCL2 and plasminogen activator inhibitor type-2. *Genomics* 9:219–228, 1991.

- [31468] 5020. Webb, A. C.; Alexander, S. J.; Samia, J. A.; Auron, P. E.; Byers, M. G.; Shows, T. B.: Localization of the urokinase-type plasminogen activator inhibitor (PLANH2) gene to the long arm of chromosome 18 at 18q21.2-q22. (Abstract) Cytogenet. Cell Genet. 51: 1103, 1989.
- [31469] 5021. Webb, A. C.; Collins, K. L.; Snyder, S. E.; Alexander, S. J.; Rosenwasser, L. J.; Eddy, R. L.; Shows, T. B.; Auron, P. E.: Human monocyte arg-serpin cDNA: sequence, chromosomal assignment, and homology to plasminogen activator-inhibitor. J. Exp. Med. 166: 77-94, 1987.
- [31470] 5022. Borlum, A.; Anette, B.; Roldan, A. L.; Blasi, F.; Bolund, L.; Kruse, T. A.: Assignment of the gene for urokinase-type plasminogen activator receptor to chromosome 19q13. (Abstract) Cytogenet. Cell Genet. 58: 2016-2017, 1991.
- [31471] 5023. Borlum, A. D.; Byskov, A.; Ragno, P.; Roldan, A. L.; Tripputi, P.; Cassani, G.; Dano, K.; Blasi, F.; Bolund, L.; Kruse, T. A.: Assignment of the urokinase-type plasminogen activator receptor gene (PLAUR) to chromosome 19q13.1-q13.2. Am. J. Hum. Genet. 50: 492-497, 1992.
- [31472] 5024. Dewerchin, M.; Van Nuffelen, A.; Wallays, G.; Bouche, A.; Moons, L.; Carmeliet, P.; Mulligan, R. C.;

Collen, D.: Generation and characterization of urokinase receptor-deficient mice. *J. Clin. Invest.* 97: 870–878, 1996.

[31473] 5025. Foca, C.; Moses, E. K.; Quinn, M. A.; Rice, G. E.: Differential mRNA expression of urokinase-type plasminogen activator, plasminogen activator receptor and plasminogen activator inhibitor type-2 in normal human endometria and endometrial carcinomas. *Gynec. Oncol.* 79: 244–250, 2000.

[31474] 5026. Memarzadeh, S.; Kozak, K. R.; Chang, L.; Natarajan, S.; Shintaku, P.; Reddy, S. T.; Farias-Eisner, R.: Urokinase plasminogen activator receptor: prognostic biomarker for endometrial cancer. *Proc. Nat. Acad. Sci.* 99: 10647–10652, 2002. Note: Erratum: *Proc. Nat. Acad. Sci.* 99: 12501 only, 2002.

[31475] 5027. Min, H. Y.; Semnani, R.; Mizukami, I. F.; Watt, K.; Todd, R. F., III; Liu, D. Y.: cDNA for Mo3, a monocyte activation antigen, encodes the human receptor for urokinase plasminogen activator. *J. Immun.* 148: 3636–3642, 1992.

[31476] 5028. Rijneveld, A. W.; Levi, M.; Florquin, S.; Speelman, P.; Carmeliet, P.; van der Poll, T.: Urokinase receptor is necessary for adequate host defense against pneumococcal pneumonia. *J. Immun.* 168: 3507–3511, 2002.

[31477] 5029. Amagai, M.; Nishikawa, T.; Nousari, H. C.; Anhalt, G.

J.; Hashimoto, T.: Antibodies against desmoglein 3 (pemphigus vulgaris antigen) are present in sera from patients with paraneoplastic pemphigus and cause acantholysis in vivo in neonatal mice. *J. Clin. Invest.* 102:775–782, 1998.

[31478] 5030. Arnemann, J.; Spurr, N. K.; Buxton, R. S.: The human gene (DSG3) coding for the pemphigus vulgaris antigen is, like the genes coding for the other two known desmogleins, assigned to chromosome 18. *Hum. Genet.* 89: 347–350, 1992.

[31479] 5031. Ishikawa, H.; Silos, S. A.; Tamai, K.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Uitto, J.: cDNA cloning and chromosomal assignment of the mouse gene for desmoglein 3 (*Dsg3*), the pemphigus vulgaris antigen. *Mammalian Genome* 5: 803–804, 1994.

[31480] 5032. Silos, S. A.; Tamai, K.; Li, K.; Kivirikko, S.; Kouba, D.; Christiano, A. M.; Uitto, J.: Cloning of the gene for human pemphigus vulgaris antigen (desmoglein 3), a desmosomal cadherin. *J. Biol. Chem.* 271:17504–17511, 1996.

[31481] 5033. Allikmets, R.; Seddon, J. M.; Bernstein, P. S.; Hutchinson, A.; Atkinson, A.; Sharma, S.; Gerrard, B.; Li, W.; Metzker, M. L.; Wadelius, C.; Caskey, C. T.; Dean, M.; Petrukhin, K.: Evaluation of the Best disease gene in pa-

tients with age-related macular degeneration and other maculopathies. Hum. Genet. 104: 449–453, 1999.

- [31482] 5034. Bascom, R. A.; Liu, L.; Chen, J.; Duncan, A.; Kimberling, W. J.; Moller, C. G.; Humphries, P.; Nathans, J.; McInnes, R. R.: ROM1: a candidate gene for autosomal dominant retinitis pigmentosa (ADRP), Usher syndrome type 1, and Best vitelliform macular dystrophy. (Abstract) Am. J. Hum. Genet. 51 (suppl.): A6, 1992.
- [31483] 5035. Best, F.: Ueber eine hereditaere Maculaaffektion. Z. Augenheilk. 13: 199–212, 1905.
- [31484] 5036. Braley, A. E.: Dystrophy of the macula. Am. J. Ophthal. 61: 1–24, 1966.
- [31485] 5037. Braley, A. E.; Spivey, B. E.: Hereditary vitelline macular degeneration: a clinical and functional evaluation of a new pedigree with variable expressivity and dominant inheritance. Arch. Ophthal. 72: 743–762, 1964.
- [31486] 5038. Brecher, R.; Bird, A. C.: Adult vitelliform macular dystrophy. Eye 4: 210–215, 1990.
- [31487] 5039. Davis, C. T.; Hollenhorst, R. W.: Hereditary degeneration of the macula: occurring in five generations. Am. J. Ophthal. 39: 637–643, 1955.
- [31488] 5040. Deutman, A. F.: Electro-oculography in families with vitelliform dystrophy of the fovea: detection of the carrier

state. Arch. Ophthal. 81:305–316, 1969.

[31489] 5041.Falls, H. F.: Hereditary congenital macular degeneration. Am.J. Hum. Genet. 1: 96–104, 1949.

[31490] 5042.Forsman, K.; Graff, C.; Nordstrom, S.; Johansson, K.; Westermarck,E.; Lundgren, E.; Gustavson, K.–H.; Wadelius, C.; Holmgren, G.: The gene for Best's macular dystrophy is located at 11q13 in a Swedish family. Clin. Genet. 42: 156–159, 1992.

[31491] 5043.Francois, J.: Vitelliform degeneration of the macula. Bull.N.Y. Acad. Med. 44: 18–27, 1968.

[31492] 5044.Friedenwald, J. S.; Maumenee, A. E.: Peculiar macular lesions with unaccountably good vision. Arch. Ophthal. 45: 567–570, 1951.

[31493] 5045.Goodstadt, L.; Ponting, C. P.: Sequence variation and disease in the wake of the draft human genome. Hum. Molec. Genet. 10: 2209–2214, 2001.

[31494] 5046.Graff, C.; Eriksson, A.; Forsman, K.; Sandgren, O.; Holmgren, G.; Wadelius, C.: Refined genetic localization of the Best disease gene in 11q13 and physical mapping of linked markers on radiation hybrids. Hum. Genet. 101: 263–270, 1997.

[31495] 5047.Graff, C.; Forsman, K.; Larsson, C.; Nordstrom, S.; Lind, L.; Johansson, K.; Sandgren, O.; Weissenbach, J.;

Holmgren, G.; Gustavson, K.-H.; Wadelius, C.: Fine mapping of Best's macular dystrophy localizes the gene in close proximity to but distinct from the D11S480/ROM1 loci. *Genomics* 24: 425–434, 1994.

[31496] 5048. Hagemeijer, A.; Hoovers, J.; Smit, E. M. E.; Bootsma, D.: Replication pattern of the X chromosomes in three X/autosomal translocations. *Cytogenet. Cell Genet.* 18: 333–348, 1977.

[31497] 5049. Hou, Y.-C.; Richards, J. E.; Bingham, E. L.; Pawar, H.; Scott, K.; Segal, M.; Lunetta, K. L.; Boehnke, M.; Sieving, P. A.: Linkage study of Best's vitelliform macular dystrophy (VMD2) in a large North American family. *Hum. Hered.* 46: 211–220, 1996.

[31498] 5050. Jung, E. E.: Ueber eine Sippe mit angeborener Maculadegeneration. Giessen: Seibert (pub.) 1936.

[31499] 5051. Cullen, T. S.: Embryology, Anatomy, and Diseases of the Umbilicus Together with Diseases of the Urachus. Philadelphia: W. B. Saunders (pub.) 1916.

[31500] 5052. Friedman, J. M.: Umbilical dysmorphology: the importance of contemplating the belly button. *Clin. Genet.* 28: 343–347, 1985.

[31501] 5053. Legius, E.; de Die-Smulders, C. E. M.; Verbraak, F.; Habex, H.; Decorte, R.; Marynen, P.; Fryns, J. P.; Cassiman,

J. J.: Geneticheterogeneity in Rieger eye malformation. J. Med. Genet. 31: 340–341,1994.

[31502] 5054.Phillips, J. C.; Del Bono, E. A.; Haines, J. L.; Pralea, A. M.;Cohen, J. S.; Greff, L. J.; Wiggs, J. L.: A second locus for Riegersyndrome maps to chromosome 13q14. Am. J. Hum. Genet. 59: 613–619,1996.

[31503] 5055.Chu, X.; Thompson, D.; Yee, L. J.; Sung, L. A.: Genomic organizationof mouse and human erythrocyte tropomodulin genes encoding the pointedend capping protein for the actin filaments. Gene 256: 271–281,2000.

[31504] 5056.Fowler, V. M.; Sussmann, M. A.; Miller, P. G.; Flucher, B. E.;Daniels, M. P.: Tropomodulin is associated with the free (pointed)ends of the thin filaments in rat skeletal muscle. J. Cell Biol. 120:411–420, 1993.

[31505] 5057.Gilligan, D. M.; Bennett, V.: The junctional complex of the membraneskeleton. Semin. Hemat. 30: 74–83, 1993.

[31506] 5058.Lench, N. J.; Telford, E. A.; Andersen, S. E.; Moynihan, T. P.;Robinson, P. A.; Markham, A. F.: An EST and STS–based YAC contigmap of human chromosome 9q22.3 Genomics 38: 199–205, 1996.

[31507] 5059.Sung, L. A.; Fan, Y.–S.; Lin, C. C.: Gene assignment, expression,and homology of human tropomodulin. Ge–

nomics 34: 92–96, 1996.

- [31508] 5060.Sung, L. A.; Fan, Y. S.; Lambert, K.; Fowler, V.; Chien, S.; Lin,C.: Assignment of human erythrocyte tropomodulin gene to q22 of chromosome9. (Abstract) Cytogenet. Cell Genet. 58: 1944, 1991.
- [31509] 5061.Sung, L. A.; Fowler, V. M.; Lambert, K.; Chien, S.: Molecularcloning of human erythroid tropomodulin. (Abstract) FASEB J. 5:A1625, 1991.
- [31510] 5062.Sung, L. A.; Fowler, V. M.; Lambert, K.; Sussman, M. A.; Karr,D.; Chien, S.: Molecular cloning and characteriza- tion of human fetalliver tropomodulin: a tropomyosin-bind- ing protein. J. Biol. Chem. 267:2616–2621, 1992.
- [31511] 5063.White, R. A.; Dowler, L. L.; Woo, M.; Adkison, L. R.; Pal, S.;Gershon, D.; Fowler, V. M.: The tropomodulin (Tmod) gene maps tochromosome 4, closely linked to Mup1. Mammalian Genome 6: 332–333,1995.
- [31512] 5064.Helander, H. M.; Koivuranta, K. T.; Horelli-Kuitunen, N.; Palvimo,J. J.; Palotie, A.; Hiltunen, J. K.: Molecular cloning and characterizationof the human mitochondrial 2,4-dienoyl-CoA reductase gene (DECR). Genomics 46:112–119, 1997.
- [31513] 5065.Roe, C. R.; Millington, D. S.; Norwood, D. L.; Kodo, N.; Sprecher,H.; Mohammed, B. S.; Nada, M.; Schulz, H.;

McVie, R.: 2,4-Dienoyl-coenzymeA reductase deficiency: a possible new disorder of fatty acid oxidation. *J.Clin. Invest.* 85: 1703–1707, 1990.

[31514] 5066.Tauchi, H.; Matsuura, S.; Isomura, M.; Kinjo, T.; Nakamura, A.;Sakamoto, S.; Kondo, N.; Endo, S.; Komatsu, K.; Nakamura, Y.: Sequenceanalysis of an 800-kb genomic DNA region on chromosome 8q21 that containsthe Nijmegen breakage syndrome gene, NBS1. *Genomics* 55: 242–247,1999.

[31515] 5067.Hamajima, N.; Matsuda, K.; Sakata, S.; Tamaki, N.; Sasaki, M.;Nonaka, M.: A novel gene family defined by human dihydropyrimidinaseand three related proteins with differential tissue distribution. *Gene* 180:157–163, 1996.

[31516] 5068.Wheeler, J. M. D.; Warren, B. F.; Mortensen, N. J. M.; Kim, H.C.; Biddolph, S. C.; Elia, G.; Beck, N. E.; Williams, G. T.; Shepherd,N. A.; Bateman, A. C.; Bodmer, W. F.: An insight into the geneticpathway of adenocarcinoma of the small intestine. *Gut* 50: 218–223,2002.

[31517] 5069.Bardwick, P. A.; Zvaifler, N. J.; Gill, G. N.; Newman, D.; Greenway,G. D.; Resnick, D. L.: Plasma cell dyscrasia with polyneuropathy,organomegaly, endocrinopathy, M protein, and skin changes: the POEMSsyndrome. Report on two cases and a review of the literature. *Medicine*

59:311–322, 1980.

- [31518] 5070. Alhonen–Hongisto, L.; Leinonen, P.; Sinervirta, R.; Laine, R.; Winqvist, R.; Alitalo, K.; Janne, O. A.; Janne, J.: Mouse and human ornithine decarboxylase genes: methylation polymorphism and amplification. *Biochem.J.* 242: 205–210, 1987.
- [31519] 5071. Brabant, M.; McConlogue, L.; van Daalen Wetters, T.; Coffino, P.: Mouse ornithine decarboxylase gene: cloning, structure, and expression. *Proc.Nat. Acad. Sci.* 85: 2200–2204, 1988.
- [31520] 5072. Cox, D. R.; Trouillot, T.; Ashley, P. L.; Brabant, M.; Coffino, P.: A functional mouse ornithine decarboxylase gene (*Odc*) maps to chromosome 12: further evidence of homoeology between mouse chromosome 12 and the short arm of human chromosome 2. *Cytogenet. Cell Genet.* 48:92–94, 1988.
- [31521] 5073. Bandmann, O.; Davis, M. B.; Marsden, C. D.; Wood, N. W.: The human homologue of the weaver mouse gene in familial and sporadic Parkinson's disease. *Neuroscience* 72: 877–879, 1996.
- [31522] 5074. Domer, P. H.; Fakharzadeh, S. S.; Chen, C.–S.; Jockel, J.; Johansen, L.; Silverman, G. A.; Kersey, J. H.; Korsmeyer, S. J.: Acute mixed–lineage leukemia t(4;11)(q21;q23) gen–

erates an MLL–AF4 fusion product. *Proc.Nat. Acad. Sci.* 90: 7884–7888, 1993.

[31523] 5075.Gu, Y.; Nakamura, T.; Alder, H.; Prasad, R.; Canaani, O.; Cimino,G.; Croce, C. M.; Canaani, E.: The t(4;11) chromosome translocationof human acute leukemias fuses the ALL–1 gene, related to *Drosophilatrithorax*, to the AF–4 gene. *Cell* 71: 701–708, 1992.

[31524] 5076.Nakamura, T.; Alder, H.; Gu, Y.; Prasad, R.; Canaani, O.; Kamada,N.; Gale, R. P.; Lange, B.; Crist, W. M.; Nowell, P. C.; Croce, C.M.; Canaani, E.: Genes on chromosomes 4, 9, and 19 involved in 11q23abnormalities in acute leukemia share sequence homology and/or commonmotifs. *Proc. Nat. Acad. Sci.* 90: 4631–4635, 1993.

[31525] 5077.Raffini, L. J.; Slater, D. J.; Rappaport, E. F.; Lo Nigro, L.;Cheung, N.–K. V.; Biegel, J. A.; Nowell, P. C.; Lange, B. J.; Felix,C. A.: Panhandle and reverse–panhandle PCR enable cloning of der(11)and der(other) genomic breakpoint junctions of MLL translocationsand identify complex translocation of MLL, AF–4, and CDK6. *Proc.Nat. Acad. Sci.* 99: 4568–4573, 2002.

[31526] 5078.Hickok, N. J.; Seppanen, P. J.; Gunsalus, G. L.; Janne, O. A.:Complete amino acid sequence of human ornithine decarboxylase deducedfrom complementary DNA. *DNA* 6:

179–187, 1987.

- [31527] 5079. Michalak, M.; Quackenbush, E. J.; Letarte, M.: Inhibition of $\text{Na}^+/\text{Ca}^{2+}$ exchanger activity in cardiac and skeletal muscle sarcolemmal vesicles by monoclonal antibody 44D7. *J. Biol. Chem.* 261: 92–95, 1986.
- [31528] 5080. Posillico, J. T.; Srikanta, S.; Brown, E. M.; Eisenbarth, G. S.: The 4F2 cell surface protein modulates intracellular calcium. (Abstract) *Clin. Res.* 33: 385A only, 1985.
- [31529] 5081. Posillico, J. T.; Srikanta, S.; Eisenbarth, G.; Quaranta, V.; Kajiji, S.; Brown, E. M.: Binding of monoclonal antibody (4F2) to its cell surface antigen on dispersed adenomatous parathyroid cells raises cytosolic calcium and inhibits parathyroid hormone secretion. *J. Clin. Endocr. Metab.* 64: 43–50, 1987.
- [31530] 5082. Quackenbush, E.; Clabby, M.; Gottesdiener, K. M.; Barbosa, J.; Jones, N. H.; Strominger, J. L.; Speck, S.; Leiden, J. M.: Molecular cloning of complementary DNAs encoding the heavy chain of the human 4F2 cell-surface antigen: a type II membrane glycoprotein involved in normal and neoplastic cell growth. *Proc. Nat. Acad. Sci.* 84: 6526–6530, 1987. Note: Correction: *Proc. Nat. Acad. Sci.* 84: 6526–6530, 1987.
- [31531] 5083. Aljada, A.; Ghanim, H.; Saadeh, R.; Dandona, P.: In-

sulin inhibits NF- κ B and MCP-1 expression in human aortic endothelial cells. *J. Clin. Endocr. Metab.* 86: 450–453, 2001.

[31532] 5084. Harroch, S.; Furtado, G. C.; Brueck, W.; Rosenbluth, J.; Lafaille, J.; Chao, M.; Buxbaum, J. D.; Schlessinger, J.: A critical role for the protein tyrosine phosphatase receptor type Z in functional recovery from demyelinating lesions. *Nature Genet.* 30 Sept, 2002. Note: Advance Electronic Publication.

[31533] 5085. Marsden, P. A.; Heng, H. H. Q.; Scherer, S. W.; Stewart, R. J.; Hall, A. V.; Shi, X.-M.; Tsui, L.-C.; Schappert, K. T.: Structure and chromosomal localization of the human constitutive endothelial nitric oxide synthase gene. *J. Biol. Chem.* 268: 17478–17488, 1993.

[31534] 5086. Miyamoto, Y.; Saito, Y.; Nakayama, M.; Shimasaki, Y.; Yoshimura, T.; Yoshimura, M.; Harada, M.; Kajiyama, N.; Kishimoto, I.; Kuwahara, K.; Hino, J.; Ogawa, E.; Hamanaka, I.; Kamitani, S.; Takahashi, N.; Kawakami, R.; Kangawa, K.; Yasue, H.; Nakao, K.: Replication protein A1 reduces transcription of the endothelial nitric oxide synthase gene containing a -786T-C mutation associated with coronary spastic angina. *Hum. Molec. Genet.* 9: 2629–2637, 2000.

[31535] 5087. Nakayama, M.; Yasue, H.; Yoshimura, M.; Shimasaki,

Y.; Kugiyama, K.; Ogawa, H.; Motoyama, T.; Saito, Y.;
Ogawa, Y.; Miyamoto, Y.; Nakao, K.: T(−786)–C mutation in
the 5–prime–flanking region of the endothelial nitric oxide
synthase gene is associated with coronary spasm. *Circulation* 99:2864–2870, 1999.

[31536] 5088. Bloch, K. D.; Wolfram, J. R.; Brown, D. M.; Roberts, J. D., Jr.; Zapol, D. G.; Lepore, J. J.; Filippov, G.; Thomas, J. E.; Jacob, H. J.; Bloch, D. B.: Three members of the nitric oxide synthase II gene family (NOS2A, NOS2B, and NOS2C) colocalize to human chromosome 17. *Genomics* 27:526–530, 1995.

[31537] 5089. Chartrain, N. A.; Geller, D. A.; Koty, P. P.; Sitrin, N. F.; Nussler, A. K.; Hoffman, E. P.; Billiar, T. R.; Hutchinson, N. I.; Mudgett, J. S.: Molecular cloning, structure, and chromosomal localization of the human inducible nitric oxide synthase gene. *J. Biol. Chem.* 269:6765–6772, 1994.

[31538] 5090. Choi, H.–S.; Rai, P. R.; Chu, H. W.; Cool, C.; Chan, E. D.: Analysis of nitric oxide synthase and nitrotyrosine expression in human pulmonary tuberculosis. *Am. J. Resp. Crit. Care Med.* 166: 178–186, 2002.

[31539] 5091. Facchetti, F.; Vermi, W.; Fiorentini, S.; Chilosi, M.; Caruso, A.; Duse, M.; Notarangelo, L. D.; Badolato, R.: Expression of inducible nitric oxide synthase in human gran–

ulomas and histiocytic reactions. *Am.J. Path.* 154:
145–152, 1999.

- [31540] 5092. Pyronnet, S.; Pradayrol, L.; Sonenberg, N.: A cell cycle-dependent internal ribosome entry site. *Molec. Cell* 5: 607–616, 2000.
- [31541] 5093. Radford, D. M.; Nakai, H.; Byers, M. G.; Eddy, R. L.; Haley, L. L.; Henry, W. M.; Shows, T. B.: Mapping the ornithine decarboxylase gene (ODC1 and ODC2) to 2p25 and 7q31-qter, respectively. (Abstract) *Cytogenet. Cell Genet.* 46: 678 only, 1987.
- [31542] 5094. Villani, V.; Coffino, P.; D'Eustachio, P.: Linkage genetics of mouse ornithine decarboxylase (Odc). *Genomics* 5: 636–638, 1989.
- [31543] 5095. Winqvist, R.; Makela, T. P.; Seppanen, P.; Janne, O. A.; Alhonen-Hongisto, L.; Janne, J.; Grzeschik, K.-H.; Altitalo, K.: Human ornithine decarboxylase sequences map to chromosome regions 2pter-p23 and 7cen-qter but are not coamplified with the NMYC oncogene. *Cytogenet. Cell Genet.* 42:133–140, 1986.
- [31544] 5096. Yang-Feng, T. L.; Barton, D. E.; Thelander, L.; Lewis, W. H.; Srinivasan, P. R.; Francke, U.: Ribonucleotide reductase M2 subunit sequences mapped to four different chromosomal sites in humans and mice: functional locus iden-

tified by its amplification in hydroxyurea-resistant cell lines. *Genomics* 1: 77–86, 1987.

- [31545] 5097. Clark, C. C.; Cohen, I.; Eichstetter, I.; Cannizzaro, L. A.; McPherson, J. D.; Wasmuth, J. J.; Iozzo, R. V.: Molecular cloning of the human proto-oncogene Wnt-5A and mapping of the gene (WNT5A) to chromosome 3p14–p21. *Genomics* 18: 249–260, 1993.
- [31546] 5098. He, X.; Saint-Jeannet, J.-P.; Wang, Y.; Nathans, J.; Dawid, I.; Varmus, H.: A member of the frizzled protein family mediating axis induction by Wnt-5A. *Science* 275: 1652–1654, 1997.
- [31547] 5099. Adelaide, J.; Mattei, M.-G.; Marics, I.; Raybaud, F.; Planche, J.; De Lapeyriere, O.; Birnbaum, D.: Chromosomal localization of the hst oncogene and its co-amplification with the int.2 oncogene in a human melanoma. *Oncogene* 2: 413–416, 1988.
- [31548] 5100. Dudley, A. T.; Ros, M. A.; Tabin, C. J.: A re-examination of proximodistal patterning during vertebrate limb development. *Nature* 418: 539–544, 2002.
- [31549] 5101. Feldman, B.; Poueymirou, W.; Papaionnou, V. E.; DeChiara, T. M.; Goldfarb, M.: Requirement of FGF-4 for postimplantation mouse development. *Science* 267: 246–249, 1995.

- [31550] 5102.Huebner, K.; Ferrari, A. C.; Delli Bovi, P.; Croce, C. M.; Basilico,C.: The FGF-related oncogene, K-FGF, maps to human chromosome region11q13, possibly near int-2. *Oncogene Res.* 3: 263–270, 1988.
- [31551] 5103.Sakamoto, H.; Mori, M.; Taira, M.; Yoshida, T.; Matsukawa, S.;Shimizu, K.; Sekiguchi, M.; Terada, M.; Sugimura, T.: Transforminggene from human stomach cancers and a noncancerous portion of stomachmucosa. *Proc. Nat. Acad. Sci.* 83: 3997–4001, 1986.
- [31552] 5104.Sun, X.; Lewandoski, M.; Meyers, E. N.; Liu, Y.–H.; Maxson, R.E., Jr.; Martin, G. R.: Conditional inactivation of *Fgf4* revealscomplexity of signalling during limb bud development. *Nature Genet.* 25:83–86, 2000.
- [31553] 5105.Sun, X.; Mariani, F. V.; Martin, G. R.: Functions of FGF signallingfrom the apical ectodermal ridge in limb development. *Nature* 418:501–508, 2002.
- [31554] 5106.Lesch, K.–P.; Bengel, D.; Heils, A.; Sabol, S. Z.; Greenberg, B.D.; Petri, S.; Benjamin, J.; Muller, C. R.; Hamer, D. H.; Murphy,D. L.: Association of anxiety-related traits with a polymorphism in the serotonin transporter gene regulatory region. *Science* 274:1527–1530, 1996.
- [31555] 5107.Sharp, D.; Blinderman, L.; Combs, K. A.; Kienzle, B.; Ricci, B.;Wager–Smith, K.; Gil, C. M.; Turck, C. W.; Bouma,

M.-E.; Rader, D.J.; Aggerbeck, L. P.; Gregg, R. E.; Gordon, D. A.; Wetterau, J. R.: Cloning and gene defects in microsomal triglyceride transfer protein associated with abetalipoproteinaemia. *Nature* 365: 65-69, 1993.

[31556] 5108.Julier, C.; Lathrop, M.; Lalouel, J. M.; Kaplan, J. C.: Use of multilocus tests of gene order: example for chromosome 22. (Abstract) *Cytogenet.Cell Genet.* 40: 663-664, 1985.

[31557] 5109.Julier, C.; Lathrop, M.; Lalouel, J. M.; Reghis, A.; Szajnert, M. F.; Kaplan, J. C.: New restriction fragment length polymorphism on human chromosome 22 at loci SIS, MB and IGLV. (Abstract) *Cytogenet.Cell Genet.* 40: 664 only, 1985.

[31558] 5110.Julier, C.; Reghis, A.; Szajnert, M. F.; Kaplan, J. C.; Lathrop, G. M.; Lalouel, J. M.: A preliminary linkage map of human chromosome 22. (Abstract) *Cytogenet. Cell Genet.* 40: 665 only, 1985.

[31559] 5111.Nakamura, Y.; Leppert, M.; O'Connell, P.; Wolff, R.; Holm, T.; Culver, M.; Martin, C.; Fujimoto, E.; Hoff, M.; Kumlin, E.; White, R.: Variable number of tandem repeat (VNTR) markers for human genemapping. *Science* 235: 1616-1622, 1987.

[31560] 5112.Romero-Herrera, A. E.; Lehmann, H.: The amino acid

sequence of human myoglobin and its minor fractions.

Proc. Roy. Soc. London 186B:249–279, 1974.

- [31561] 5113. Romero–Herrera, A. E.; Lehmann, H.: Primary structure of human myoglobin. *Nature* N.B. 232: 149–152, 1971.
- [31562] 5114. Weller, P.; Jeffreys, A. J.; Wilson, V.; Blanchetot, A.: Organization of the human myoglobin gene. *EMBO J.* 3: 439–446, 1984.
- [31563] 5115. Wyman, A. R.; White, R.: A highly polymorphic locus in human DNA. *Proc. Nat. Acad. Sci.* 77: 6754–6758, 1980.
- [31564] 5116. Almarri, A.; Batchelor, J. R.: HLA and hepatitis B infection. *Lancet* 344:1194–1195, 1994.
- [31565] 5117. Ayala, F. J.; Escalante, A.; O'hUigin, C.; Klein, J.: Molecular genetics of speciation and human origins. *Proc. Nat. Acad. Sci.* 91:6787–6794, 1994.
- [31566] 5118. Bell, J. I.; Estess, P.; St. John, T.; Saiki, R.; Watling, D. L.; Erlich, H. A.; McDevitt, H. O.: DNA sequence and characterization of human class II major histocompatibility complex beta chains from the DR1 haplotype. *Proc. Nat. Acad. Sci.* 82: 3405–3409, 1985.
- [31567] 5119. Bodmer, J. G.; Marsh, S. G. E.; Albert, E.: Nomenclature for factors of the HLA system, 1989. *Immun. Today* 11: 3–10, 1990.

- [31568] 5120. Bodmer, J. G.; Marsh, S. G. E.; Albert, E. D.; Bodmer, W. F.; Bontrop, R. E.; Charron, D.; Dupont, B.; Erlich, H. A.; Fauchet, R.; Mach, B.; Mayr, W. R.; Parham, P.; Sasazuki, T.; Schreuder, G. M. T.; Strominger, J. L.; Svejgaard, A.; Terasaki, P. I.: Nomenclature for factors of the HLA system, 1996. *Europ. J. Immunogenet.* 24: 105–151, 1997.
- [31569] 5121. Bodmer, J. G.; Marsh, S. G. E.; Albert, E. D.; Bodmer, W. F.; Dupont, B.; Erlich, H. A.; Mach, B.; Mayr, W. R.; Parham, P.; Sasazuki, T.; Schreuder, G. M. T.; Strominger, J. L.; Svejgaard, A.; Terasaki, P. I.: Nomenclature for factors of the HLA system, 1994. *Tissue Antigens* 44:1–18, 1994.
- [31570] 5122. Bodmer, W. F.; Bodmer, J. G.; Batchelor, J. R.; Festenstein, H.; Morris, P. J.: *Histocompatibility Testing* 1977. Copenhagen: Munksgaard(pub.) 1978.
- [31571] 5123. Boss, J. M.; Strominger, J. L.: Cloning and sequence analysis of the human major histocompatibility complex gene DC-3-beta. *Proc. Nat. Acad. Sci.* 81: 5199–5203, 1984.
- [31572] 5124. Charron, D. J.; McDevitt, H. O.: Analysis of HLA-D region-associated molecules with monoclonal antibody. *Proc. Nat. Acad. Sci.* 76: 6567–6571, 1979.
- [31573] 5125. Chelladurai, M.; Honn, K. V.; Walz, D. A.: HLA-DR is a procoagulant. *Biochem. Biophys. Res. Commun.* 178:

467–473, 1991.

- [31574] 5126.Corte, G.; Damiani, G.; Calabi, F.; Fabbi, M.; Bargellesi, A.: Analysis of HLA–DR polymorphism by two-dimensional peptide mapping. *Proc.Nat. Acad. Sci.* 78: 534–538, 1981.
- [31575] 5127.Das, H. K.; Lawrance, S. K.; Weissman, S. M.: Structure and nucleotidesequence of the heavy chain gene of HLA–DR. *Proc. Nat. Acad. Sci.* 80:3543–3547, 1983.
- [31576] 5128.Delovitch, T. L.; Falk, J. A.: Evidence for structural homologybetween murine and human Ia antigens. *Immunogenetics* 8: 405–418,1979.
- [31577] 5129.Dunham, I.; Sargent, C. A.; Dawkins, R. L.; Campbell, R. D.:An analysis of variation in the long-range genomic organization ofthe human major histocompatibility complex class II region by pulsed–fieldgel electrophoresis. *Genomics* 5: 787–796, 1989.
- [31578] 5130.Erlich, H. A.; Stetler, D.; Saiki, R.; Gladstone, P.; Pious, D.: Mapping of the genes encoding the HLA–DR alpha chain and the HLA–relatedantigens to a chromosome 6 deletion by using genomic blotting. *Proc.Nat. Acad. Sci.* 80: 2300–2304, 1983.
- [31579] 5131.Ferber, K. M.; Keller, E.; Albert, E. D.; Ziegler, A.–G.: Predictivevalue of human leukocyte antigen class II typing

for the development of islet autoantibodies and insulin-dependent diabetes postpartum in women with gestational diabetes. *J. Clin. Endocr. Metab.* 84:2342–2348, 1999.

[31580] 5132. Fuller, T. C.; Einarson, M.; Pinto, C.; Ahern, A.; Yunis, E. J.: Genetic evidence that HLA-DR (Ia) specifications include multiple HLA-D determinants on a single haplotype. *Transplant. Proc.* 10:781–784, 1978.

[31581] 5133. Hardy, D. A.; Bell, J. I.; Long, E. O.; Lindsten, T.; McDevitt, H. O.: Mapping of the class II region of the human major histocompatibility complex by pulsed-field gel electrophoresis. *Nature* 323: 453–455, 1986.

[31582] 5134. Hui, K.; Festenstein, H.; de Klein, A.; Grosveld, G.; Grosveld, F.: HLA-DR genotyping by restriction fragment length polymorphism analyses. *Immunogenetics* 22: 231–239, 1985.

[31583] 5135. Kaufman, J. F.; Strominger, J. L.: HLA-DR light chain has a polymorphic N-terminal region and a conserved immunoglobulin-like C-terminal region. *Nature* 297:694–697, 1982.

[31584] 5136. Korman, A. J.; Auffray, C.; Schamboeck, A.; Strominger, J. L.: The amino acid sequence and gene organization of the heavy chain of the HLA-DR antigen: homology to immunoglobulins. *Proc. Nat. Acad. Sci.* 79:

6013-6017, 1982.

- [31585] 5137.Korman, A. J.; Knudsen, P. J.; Kaufman, J. F.; Strominger, J.L.: cDNA clones for the heavy chain of HLA-DR antigens obtained after immunopurification of polysomes by monoclonal antibody. Proc. Nat.Acad. Sci. 79: 1844-1848, 1982.
- [31586] 5138.Kratzin, H.; Yang, C.; Gotz, H.; Pauly, E.; Kolbel, S.; Egert, G.; Thinnes, F. P.; Wernet, P.; Altevogt, P.; Hilschmann, N.: Primärstruktur menschlicher Histokompatibilitätsantigene der Klasse II. 1. Mitteilung: Aminosäuresequenz der N-terminalen 198 Reste der beta-Kette des HLA-Dw2,2;DR2,2-Alloantigens. Hoppe-Seyler's Z. Physiol. Chem. 362: 1665-1669, 1981.
- [31587] 5139.Lamm, L. U.: Another segregant series, DR, in HLA. Cytogenet. Cell Genet. 22: 309-312, 1978.
- [31588] 5140.Larhammar, D.; Schenning, L.; Gustafsson, K.; Wiman, K.; Claesson, L.; Rask, L.; Peterson, P. A.: Complete amino acid sequence of an HLA-DR antigen-like beta chain as predicted from the nucleotide sequence: similarities with immunoglobulins and HLA-A, -B, and -C antigens. Proc. Nat. Acad. Sci. 79: 3687-3691, 1982.
- [31589] 5141.Lee, J. S.; Trowsdale, J.; Bodmer, W. F.: cDNA clones coding for the heavy chain of human HLA-DR antigen.

Proc. Nat. Acad. Sci. 79:545–549, 1982.

- [31590] 5142. Levine, F.; Mach, B.; Long, E.; Erlich, H.; Pious, D.: Mapping in the HLA–D region with deletion variants and cloned genes. (Abstract) Cytogenet. Cell Genet. 37: 523 only, 1984.
- [31591] 5143. Mann, D. L.; Abelson, L.; Harris, S. D.; Amos, D. B.: Second genetic locus in the HLA region for human B–cell alloantigens. Nature 259:145–146, 1976.
- [31592] 5144. Mann, D. L.; Abelson, L.; Henkart, P.; Harris, S. D.; Amos, D. B.: Specific B–lymphocyte alloantigens linked to HLA. Proc. Nat. Acad. Sci. 72: 5103–5106, 1975.
- [31593] 5145. Markert, M. L.; Cresswell, P.: Polymorphism of human B–cell alloantigens: evidence for three loci within the HLA system. Proc. Nat. Acad. Sci. 77:6101–6104, 1980.
- [31594] 5146. Mayer, W. E.; O'hUigin, C.; Klein, J.: Resolution of the HLA–DRB6 puzzle: a case of grafting a de novo–generated exon on an existing gene. Proc. Nat. Acad. Sci. 90: 10720–10724, 1993.
- [31595] 5147. McDevitt, H. O.; Bodmer, W. F.: HLA, immune–response genes, and disease. Lancet I: 1269–1275, 1974.
- [31596] 5148. Disteche, C. M.; Adler, D. A.; Tedder, T. F.; Saito, H.: Mapping of the genes for LYAM1, a new lymphocyte adhesion molecule, and for LAR, a new receptor–linked protein

tyrosine phosphatase, to human chromosome 1. (Abstract)
Cytogenet. Cell Genet. 51: 990 only, 1989.

- [31597] 5149. Wijmenga, C.; Hansen, R. S.; Gimelli, G.; Bjorck, E. J.; Davies, E. G.; Valentine, D.; Belohradsky, B. H.; van Dongen, J. J.; Smeets, D. F. C. M.; van den Heuvel, L. P. W. J.; Luyten, J. A. F. M.; Strengman, E.; Weemaes, C.; Pearson, P. L.: Genetic variation in ICF syndrome: evidence for genetic heterogeneity. Hum. Mutat. 16: 509–517, 2000.
- [31598] 5150. Ehrlich, M.; Buchanan, K. L.; Tsien, F.; Jiang, G.; Sun, B.; Uicker, W.; Weemaes, C. M. R.; Smeets, D.; Sperling, K.; Belohradsky, B. H.; Tommerup, N.; Misek, D. E.; Rouillard, J.-M.; Kuick, R.; Hanash, S. M.: DNA methyltransferase 3B mutations linked to the ICF syndrome cause dysregulation of lymphogenesis genes. Hum. Molec. Genet. 10: 2917–2931, 2001.
- [31599] 5151. Fredrickson, D. S.; Levy, R. I.: Familial hyperlipoproteinemia. In: Stanbury, J. B.; Wyngaarden, J. B.; Fredrickson, D. S.: The Metabolic Basis of Inherited Disease. New York: McGraw-Hill (pub.) (3rd ed.): 1972. Pp. 545–614.
- [31600] 5152. Ameis, D.; Kobayashi, J.; Davis, R. C.; Ben-Zeev, O.; Malloy, M. J.; Kane, J. P.; Lee, G.; Wong, H.; Havel, R. J.; Schotz, M. C.: Familial chylomicronemia (type I hyperlipoproteinemia) due to a single missense mutation in the

lipoprotein lipase gene. J. Clin. Invest. 87:1165–1170, 1991.

[31601] 5153. Auwerx, J. H.; Babirak, S. P.; Fujimoto, W. Y.; Iverius, P. H.; Brunzell, J. D.: Defective enzyme protein in lipoprotein lipase deficiency. Europ. J. Clin. Invest. 19: 433–437, 1989.

[31602] 5154. Beg, O. U.; Meng, M. S.; Skarlatos, S. I.; Previato, L.; Brunzell, J. D.; Brewer, H. B., Jr.; Fojo, S. S.: Lipoprotein lipase (Bethesda): a single amino acid substitution (ala176-to-thr) leads to abnormal heparin binding and loss of enzymic activity. Proc. Nat. Acad. Sci. 87:3474–3478, 1990.

[31603] 5155. Benlian, P.; De Gennes, J. L.; Foubert, L.; Zhang, H.; Gagne, S. E.; Hayden, M.: Premature atherosclerosis in patients with familial chylomicronemia caused by mutations in the lipoprotein lipase gene. New Eng. J. Med. 335: 848–854, 1996.

[31604] 5156. Berger, G. M. B.: An incomplete form of familial lipoprotein lipase deficiency presenting with type I hyperlipoproteinemia. Am. J. Clin. Path. 88: 369–373, 1987.

[31605] 5157. Berger, H.; Richter, A.; Gilardi, A.; Wagner, H.: Essential familial hyperlipaemia in a 2-year-old child. Ann. Paediat. 199: 445–466, 1962.

- [31606] 5158. Bergeron, J.; Normand, T.; Bharucha, A.; Ven Murthy, M. R.; Julien, P.; Gagne, C.; Dionne, C.; De Braekeleer, M.; Brun, D.; Hayden, M.R.; Lupien, P. J.: Prevalence, geographical distribution and genealogical investigations of mutation 188 of lipoprotein lipase gene in the French-Canadian population of Quebec. *Clin. Genet.* 41: 206–210, 1992.
- [31607] 5159. Bertolini, S.; Simone, M. L.; Pes, G. M.; Ghisellini, M.; Rolleri, M.; Bellocchio, A.; Elicio, N.; Masturzo, P.; Calandra, S.: Pseudodominance of lipoprotein lipase (LPL) deficiency due to a nonsense mutation (tyr302-to-term) in exon 6 of LPL gene in an Italian family from Sardinia (LPL-Olbia). *Clin. Genet.* 57: 140–147, 2000.
- [31608] 5160. Boer, J. M. A.; Kuivenhoven, J. A.; Feskens, E. J. M.; Schouten, E. G.; Havekes, L. M.; Seidell, J. C.; Kastelein, J. J. P.; Kromhout, D.: Physical activity modulates the effect of a lipoprotein lipase mutation (D9N) on plasma lipids and lipoproteins. *Clin. Genet.* 56: 158–163, 1999.
- [31609] 5161. Boggs, J. D.; Hsia, D. Y.-Y.; Mais, R. F.; Bigler, J. A.: The genetic mechanism of idiopathic hyperlipemia. *New Eng. J. Med.* 257: 1101–1108, 1957.
- [31610] 5162. Breckenridge, W. C.; Little, A. C.; Steiner, G.; Chow, A.; Poapst, M.: Hypertriglyceridemia associated with defi-

ciency of C-II apoprotein in plasma lipoproteins. *New Eng. J. Med.* 298: 1265–1273, 1978.

- [31611] 5163. Brunzell, J. D.; Chait, A.; Nikkila, E. A.; Ehnholm, C.; Huttunen, J. K.; Steiner, G.: Heterogeneity of primary lipoprotein lipase deficiency. *Metabolism* 29:624–629, 1980.
- [31612] 5164. Busca, R.; Martinez, M.; Vilella, E.; Pognonec, P.; Deeb, S.; Auwerx, J.; Reina, M.; Vilaro, S.: The mutation gly142-to-glu in human lipoprotein lipase produces a missorted protein that is diverted to lysosomes. *J. Biol. Chem.* 271: 2139–2146, 1996.
- [31613] 5165. Cantin, B.; Boudriau, S.; Bertrand, M.; Brun, L.-D.; Gagne, C.; Rogers, P. A.; Ven Murthy, M. R.; Lupien, P.-J.; Julien, P.: Hemolysis in primary lipoprotein lipase deficiency. *Metabolism* 44: 652–658, 1995.
- [31614] 5166. Chimienti, G.; Capurso, A.; Resta, F.; Pepe, G.: A G-to-C change at the donor splice site of intron 1 causes lipoprotein lipase deficiency in a Southern-Italian family. *Biochem. Biophys. Res. Commun.* 187:620–627, 1992.
- [31615] 5167. Clark, A. G.; Weiss, K. M.; Nickerson, D. A.; Taylor, S. L.; Buchanan, A.; Stengard, J.; Salomaa, V.; Vartiainen, E.; Perola, M.; Boerwinkle, E.; Sing, C. F.: Haplotype structure and population genetic inferences from nucleotide-se-

quence variation in human lipoprotein lipase. *Am.J. Hum. Genet.* 63: 595–612, 1998.

- [31616] 5168.Clee, S. M.; Loubser, O.; Collins, J.; Kastelein, J. J. P.; Hayden, M. R.: The LPL S447X cSNP is associated with decreased blood pressure and plasma triglycerides, and reduced risk of coronary artery disease. *Clin. Genet.* 60: 293–300, 2001.
- [31617] 5169.De Braekeleer, M.; Dionne, C.; Gagne, C.; Julien, P.; Brun, D.; Ven Murthy, M. R.; Lupien, P.-J.: Founder effect in familial hyperchylomicronemia among French Canadians of Quebec. *Hum. Hered.* 41: 168–173, 1991.
- [31618] 5170.De Bruin, T. W. A.; Maily, F.; Van Barlingen, H. H. J. J.; Fisher, R.; Castro Cabezas, M.; Talmud, P.; Dallinga-Thie, G. M.; Humphries, S. E.: Lipoprotein lipase gene mutations D9N and N291S in four pedigrees with familial combined hyperlipidaemia. *Europ. J. Clin. Invest.* 26: 631–639, 1996.
- [31619] 5171.Deeb, S. S.; Peng, R.: Structure of the human lipoprotein lipase gene. *Biochemistry* 28: 4131–4135, 1989.
- [31620] 5172.Henderson, H. E.; Hassan, F.; Berger, G. M. B.; Hayden, M. R.: The lipoprotein lipase gly188-to-glu mutation in South Africans of Indian descent: evidence suggesting common origins and an increased frequency. *J. Med. Genet.* 29: 119–122, 1992.

- [31621] 5173.Henderson, H. E.; Hassan, F.; Marais, D.; Hayden, M. R.: A newmutation destroying disulphide bridging in the C-terminal domain oflipoprotein lipase. *Biochem. Biophys. Res. Commun.* 227: 189–194,1996.
- [31622] 5174.Henderson, H. E.; Ma, Y.; Hassan, M. F.; Monsalve, M. V.; Marais,A. D.; Winkler, F.; Gubernator, K.; Peterson, J.; Brunzell, J. D.;Hayden, M. R.: Amino acid substitution (ile194-to-thr) in exon 5of the lipoprotein lipase gene causes lipoprotein lipase deficiencyin three unrelated probands: support for a multicentric origin. *J.Clin. Invest.* 87: 2005–2011, 1991.
- [31623] 5175.Sekiguchi, K.; Klos, A. M.; Kurachi, K.; Yoshitake, S.; Hakomori,S.: Human liver fibronectin complementary DNAs: identification oftwo different messenger RNAs possibly encoding the alpha and betasubunits of plasma fi-bronectin. *Biochemistry* 25: 4936–4941, 1986.
- [31624] 5176.Shirakami, A.; Shigekiyo, T.; Hirai, Y.; Takeichi, T.; Kawauchi,S.; Saito, S.; Miyoshi, K.: Plasma fibronectin deficiency in eightmembers of one family. *Lancet I*: 473–474, 1986.
- [31625] 5177.Shows, T. B.: Personal Communication. Buffalo, N. Y. 1982.
- [31626] 5178.Skow, L. C.; Adkison, L.; Womack, J. E.; Beamer, W.

G.; Taylor, B. A.: Mapping of the mouse fibronectin gene (Fn-1) to chromosome 1: conservation of the Idh-1--Cryg--Fn-1 syntenic group in mammals. *Genomics* 1:283-286, 1987.

[31627] 5179. Smith, M.; Gold, L. I.; Pearlstein, E.; Krinsky, A.: Expression of mouse and human fibronectin in hybrid cells. (Abstract) *Cytogenet. Cell Genet.* 25: 205 only, 1979.

[31628] 5180. Smith, M.; Krinsky, A. M.; Arredondo-Vega, F. X.; Pearlstein, E.: Production of soluble fibronectin by RAG x human fibroblast hybrids. (Abstract) *Cytogenet. Cell Genet.* 32: 318 only, 1982.

[31629] 5181. Wu, B.-L.; Milunsky, A.; Wyandt, H.; Hoth, C.; Baldwin, C.; Skare, J.: In situ hybridization applied to Waardenburg syndrome. *Cytogenet. Cell Genet.* 63: 29-32, 1993.

[31630] 5182. Zardi, L.; Cianfriglia, M.; Balza, E.; Carnemolla, B.; Siri, A.; Croce, C. M.: Species-specific monoclonal antibodies in the assignment of the gene for human fibronectin to chromosome 2. *EMBO J.* 1: 929-933, 1982.

[31631] 5183. Zardi, L.; Siri, A.; Carnemolla, B.; Santi, L.; Gardner, W. D.; Hoch, S. O.: Fibronectin: a chromatin-associated protein? *Cell* 18:649-657, 1979.

[31632] 5184. Adkison, L. R.; White, R. A.; Haney, D. M.; Lee, J. C.; Pusey, K. T.; Gardner, J.: The fibronectin receptor, alpha

subunit (Itga5) maps to murine chromosome 15, distal to D15Mit16. *Mammalian Genome* 5:456–457, 1994.

[31633] 5185. Argraves, W. S.; Pytela, R.; Suzuki, S.; Millan, J. L.; Pierschbacher, M. D.; Ruoslahti, E.: cDNA sequences from the alpha subunit of the fibronectin receptor predict a transmembrane domain and a short cytoplasmic peptide. *J. Biol. Chem.* 261: 12922–12924, 1986.

[31634] 5186. Argraves, W. S.; Suzuki, S.; Arai, H.; Thompson, K.; Pierschbacher, M. D.; Ruoslahti, E.: Amino acid sequence of the human fibronectin receptor. *J. Cell Biol.* 105: 1183–1190, 1987.

[31635] 5187. Fitzgerald, L. A.; Poncz, M.; Steiner, B.; Rall, S. C., Jr.; Bennett, J. S.; Phillips, D. R.: Comparison of cDNA-derived protein sequences of the human fibronectin and vitronectin receptor alpha-subunits and platelet glycoprotein IIb. *Biochemistry* 26: 8158–8165, 1987.

[31636] 5188. Krissansen, G. W.; Yuan, Q.; Jenkins, D.; Jiang, W.-M.; Rooke, L.; Spurr, N. K.; Eccles, M.; Leung, E.; Watson, J. D.: Chromosomal locations of the genes coding for the integrin beta-6 and beta-7 subunits. *Immunogenetics* 35:58–61, 1992.

[31637] 5189. Sosnoski, D.; Emanuel, B. S.; Hawkins, A. L.; van Tuinen, P.; Ledbetter, D. H.; Nussbaum, R. L.; Kaos, F.-T.;

Schwartz, E.; Phillips, D.; Bennett, J. S.; Fitzgerald, L. A.; Poncz, M.: Chromosomal localization of the genes for the vitronectin and fibronectin receptors alpha-subunits and for platelet glycoproteins IIb and IIIa. *J. Clin. Invest.* 81:1993–1998, 1988.

[31638] 5190. Spurr, N. K.; Rooke, L.: Confirmation of the assignment of the vitronectin (VNRA) and fibronectin (FNRA) receptor alpha-subunits. *Ann. Hum. Genet.* 55: 217–223, 1991.

[31639] 5191. Akula, S. M.; Pramod, N. P.; Wang, F.-Z.; Chandran, B.: Integrin alpha-3/beta-1 (CD 49c/29) is a cellular receptor for Kaposi's sarcoma-associated herpesvirus (KSHV/HHV-8) entry into the target cells. *Cell* 108:407–419, 2002.

[31640] 5192. Arregui, C.; Pathre, P.; Lilien, J.; Balsamo, J.: The nonreceptor tyrosine kinase Fer mediates cross-talk between N-cadherin and beta-1-integrins. *J. Cell Biol.* 149: 1263–1273, 2000.

[31641] 5193. Giuffra, L. A.; Lichter, P.; Wu, J.; Kennedy, J. L.; Pakstis, A. J.; Rogers, J.; Kidd, J. R.; Harley, H.; Jenkins, T.; Ward, D. C.; Kidd, K. K.: Genetic and physical mapping and population studies of a fibronectin receptor beta-subunit-like sequence on human chromosome 19. *Genomics*

8: 340–346, 1990.

- [31642] 5194. Giuffra, L. A.; Wu, J.; Lichter, P.; Kennedy, J. L.; Castiglione, C.; Pakstis, A. J.; Ward, D.; Kidd, K. K.: Mapping of a fibronectin receptor beta subunit-like sequence to chromosome 19. (Abstract) *Am. J. Hum. Genet.* 45 (suppl.): A141 only, 1989.
- [31643] 5195. Goodfellow, P. J.; Nevanlinna, H. A.; Gorman, P.; Sheer, D.; Lam, G.; Goodfellow, P. N.: Assignment of the gene encoding the beta-subunit of the human fibronectin receptor (beta-FNR) to chromosome 10p11.2. *Ann. Hum. Genet.* 53: 15–22, 1989.
- [31644] 5196. Graus-Porta, D.; Blaess, S.; Senften, M.; Littlewood-Evans, A.; Damsky, C.; Huang, Z.; Orban, P.; Klein, R.; Schittny, J. C.; Muller, U.: Beta-1-class integrins regulate the development of laminae and folia in the cerebral and cerebellar cortex. *Neuron* 31: 367–379, 2001.
- [31645] 5197. Hynes, R. O.: Integrins: a family of cell surface receptors. *Cell* 48: 549–554, 1987.
- [31646] 5198. Johansson, S.; Forsberg, E.; Lundgren, B.: Comparison of fibronectin receptors from rat hepatocytes and fibroblasts. *J. Biol. Chem.* 262: 7819–7824, 1987.
- [31647] 5199. Lu, T. T.; Cyster, J. G.: Integrin-mediated long-term B cell retention in the splenic marginal zone. *Science* 297:

409–412, 2002.

- [31648] 5200.Messer Peters, P.; Kamarck, M. E.; Hemler, M. E.; Strominger, J. L.; Ruddle, F. H.: Genetic and biochemical characterization of human lymphocyte cell surface antigens: the A-1A5 and A-3A4 determinants. *J. Exp. Med.* 159: 1441–1454, 1984.
- [31649] 5201.Pytela, R.; Pierschbacher, M. D.; Ginsberg, M. H.; Plow, E. F.; Ruoslahti, E.: Platelet membrane glycoprotein IIb/IIIa: member of a family of arg-gly-asp-specific adhesion receptors. *Science* 231:1159–1162, 1986.
- [31650] 5202.Woods, V. L., Jr.; Pischel, K. D.; Avery, E. D.; Bluestein, H.G.: Antigenic polymorphism of human very late activation protein-2(platelet glycoprotein Ia-IIa): platelet alloantigen Hc(a). *J. Clin. Invest.* 83: 978–985, 1989.
- [31651] 5203.Wu, J. S.; Giuffra, L. A.; Goodfellow, P. J.; Myers, S.; Carson, N. L.; Anderson, L.; Hoyle, L. S.; Simpson, N. E.; Kidd, K. K.: The beta subunit locus of the human fibronectin receptor: DNA restriction fragment length polymorphism and linkage mapping studies. *Hum. Genet.* 83:383–390, 1989.
- [31652] 5204.Wilcox, A. S.; Warrington, J. A.; Gardiner, K.; Berger, R.; Whiting, P.; Altherr, M. R.; Wasmuth, J. J.; Patterson, D.;

Sikela, J. M.: Human chromosomal localization of genes encoding the gamma-1 and gamma-2 subunits of the gamma-aminobutyric acid receptor indicates members of this gene family are often clustered in the genome. *Proc. Nat. Acad. Sci.* 89: 5857–5861, 1992.

- [31653] 5205. Huang, F.; Shi, L. J.; Heng, H. H. Q.; Fei, J.; Guo, L.-H.: Assignment of the human GABA transporter gene (GABATHG) locus to chromosome 3p24–p25. *Genomics* 29:302–304, 1995.
- [31654] 5206. Lam, D. M.-K.; Fei, J.; Zhang, X.-Y.; Tam, A. C. W.; Zhu, L.-H.; Huang, F.; King, S. C.; Guo, L.-H.: Molecular cloning and structure of the human (GABATHG) GABA transporter gene. *Molec. Brain Res.* 19:227–232, 1993.
- [31655] 5207. Nelson, H.; Mandiyan, S.; Nelson, N.: Cloning of the human brain GABA transporter. *FEBS Lett.* 269: 181–184, 1990.
- [31656] 5208. Figlewicz, D. A.; Delattre, O.; Guellaen, G.; Krizus, A.; Thomas, G.; Zucman, J.; Rouleau, G. A.: Mapping of human gamma-glutamyl transpeptidase genes on chromosome 22 and other human autosomes. *Genomics* 17: 299–305, 1993.
- [31657] 5209. Pawlak, A.; Wu, S.-J.; Bulle, F.; Suzuki, A.; Chikhi, N.; Ferry, N.; Baik, J.-H.; Siegrist, S.; Guellaen, G.: Different

gamma-glutamyltranspeptidase mRNAs are expressed in human liver and kidney. *Biochem.Biophys. Res. Commun.* 164: 912–918, 1989.

- [31658] 5210.Rajpert-De Meyts, E.; Heisterkamp, N.; Groffen, J.: Cloning and nucleotide sequence of human gamma-glutamyl transpeptidase. *Proc.Nat. Acad. Sci.* 85: 8840–8844, 1988.
- [31659] 5211.Lopez de Castro, J. A.; Strominger, J. L.; Strong, D. M.; Orr, H. T.: Structure of crossreactive human histocompatibility antigens HLA-A28 and HLA-A2: possible implications for the generation of HLA polymorphism. *Proc. Nat. Acad. Sci.* 79: 3813–3817, 1982.
- [31660] 5212.Malissen, M.; Malissen, B.; Jordan, B. R.: Exon/intron organization and complete nucleotide sequence of an HLA gene. *Proc. Nat. Acad.Sci.* 79: 893–897, 1982.
- [31661] 5213.Mann, D. L.; Rogentine, G. N., Jr.; Fahey, J. L.; Nathenson, S.G.: Molecular heterogeneity of human lymphoid (HL-A) alloantigens. *Science* 163:1460–1462, 1969.
- [31662] 5214.MacDonald, K. S.; Fowke, K. R.; Kimani, J.; Dunand, V. A.; Nagelkerke, N. J. D.; Ball, T. B.; Oyugi, J.; Njagi, E.; Gaur, L. K.; Brunham, R. C.; Wade, J.; Luscher, M. A.; Krausa, P.; Rowland-Jones, S.; Ngugi, E.; Bwayo, J. J.; Plummer, F. A.: Influence of HLA supertypes on susceptibility

and resistance to human immunodeficiency virus type 1 infection. *J. Infect. Dis.* 181: 1581–1589, 2000.

[31663] 5215. Mayr, W. R.; Mayr, D.: Analysis of the linkage between the HL-A loci and the genes of other markers. *Humangenetik* 24: 129–133, 1974.

[31664] 5216. Messer, G.; Zemmour, J.; Orr, H. T.; Parham, P.; Weiss, E. H.; Girdlestone, J.: HLA-J, a second inactivated class I HLA gene related to HLA-G and HLA-A. *J. Immun.* 148: 4043–4053, 1992.

[31665] 5217. Morton, C. C.; Kirsch, I. R.; Nance, W. E.; Evans, G. A.; Korman, A. J.; Strominger, J. L.: Orientation of loci within the human major histocompatibility complex by chromosomal in situ hybridization. *Proc. Nat. Acad. Sci.* 81: 2816–2820, 1984.

[31666] 5218. Morton, N. E.; Rao, D. C.; Lindsten, J.; Hulten, M.; Yee, S.: A chiasma map of man. *Hum. Hered.* 27: 38–51, 1977.

[31667] 5219. Mulley, J. C.; Hay, J.; Sheffield, L. J.; Sutherland, G. R.: Regional localization for HLA by recombination with a fragile site at 6p23. *Am. J. Hum. Genet.* 35: 1284–1288, 1983.

[31668] 5220. Newell, W. R.; Trowsdale, J.; Beck, S.: MHCDB—database of the human MHC. *Immunogenetics* 40:

109–115, 1994.

- [31669] 5221.Orr, H. T.; Bach, F. H.; Ploegh, H. L.; Strominger, J. L.; Kavathas,P.; DeMars, R.: Use of HLA loss mutants to analyse the structureof the human major histocompatibil-ity complex. *Nature* 296: 454–456,1982.
- [31670] 5222.Paabo, S.: Molecular cloning of ancient Egyptian mummy DNA. *Nature* 314:644–645, 1985.
- [31671] 5223.Patel, R.; Mickey, M. R.; Terasaki, P. I.: Serotyping for homotransplantationof kidneys from unrelated donors. *New Eng. J. Med.* 279: 501–506,1968.
- [31672] 5224.Payne, R.; Tripp, M.; Weigle, J.; Bodmer, W. F.; Bodmer, J. G.: A new leukocyte isoantigen system in man. *Cold Spring Harbor Symp.Quant. Biol.* 29: 285–295, 1964.
- [31673] 5225.Polacek, L. A.; Phillips, R. B.; Hackbarth, S. A.; Duquesnoy,R. J.: A linkage study of the HLA region using C-band heteromorphisms. *Clin.Genet.* 23: 177–185, 1983.
- [31674] 5226.Ragoussis, J.; Bloemer, K.; Pohla, H.; Messer, G.; Weiss, E. H.;Ziegler, A.: A physical map including a new class I gene (cda12)of the human major histocompatibility complex (A2/B13 haplotype) derivedfrom a monosomy 6 mutant cell line. *Genomics* 4: 301–308, 1989.
- [31675] 5227.Ragoussis, J.; van der Bliek, A.; Trowsdale, J.;

Ziegler, A.: Mapping of HLA genes using pulsed field gradient electrophoreses. FEBS Lett. 204: 1–4, 1986.

[31676] 5228. Salter, R. D.; Norment, A. M.; Chen, B. P.; Clayberger, C.; Krensky, A. M.; Littman, D. R.; Parham, P.: Polymorphism in the alpha(3) domain of HLA-A molecules affects binding to CD8. Nature 338: 345–347, 1989.

[31677] 5229. Schunter, F.; Wernet, P.; Kompf, J.; Bissbort, S.; Gohler, F.: Mapping of the linkage group GLO-Bf-HLA-B, C, A-PGM. II. Segregation analysis. Hum. Genet. 44: 321–331, 1978.

[31678] 5230. Shivdasani, R. A.; Haluska, F. G.; Dock, N. L.; Dover, J. S.; Kineke, E. J.; Anderson, K. C.: Graft-versus-host disease associated with transfusion of blood from unrelated HLA-homozygous donors. New Eng. J. Med. 328: 766–770, 1993.

[31679] 5231. Shukla, H.; Gillespie, G. A.; Srivastava, R.; Collins, F.; Chorney, M. J.: A class I jumping clone places the HLA-G gene approximately 100 kilobases from HLA-H within the HLA-A subregion of the human MHC. Genomics 10: 905–914, 1991.

[31680] 5232. Snell, G. D.: Studies in histocompatibility. Science 213: 172–178, 1981.

[31681] 5233. Solheim, B. G.; Bratlie, A.; Sandberg, L.; Staub-

Nielsen, L.;Thorsby, E.: Further evidence of a third HL-A locus. *Tissue Antigens* 3:439–453, 1973.

[31682] 5234.Strachan, T.: Molecular genetics and polymorphism of class IHLA antigens. *Brit. Med. Bull.* 43: 1–14, 1987.

[31683] 5235.Szpak, Y.; Vieville, J.-C.; Tabary, T.; Naud, M.-C.; Chopin, M.;Edelson, C.; Cohen, J. H. M.; Dausset, J.; de Kozak, Y.; Pla, M.:Spontaneous retinopathy in HLA-A29 transgenic mice. *Proc. Nat. Acad.Sci.* 98: 2572–2576, 2001.

[31684] 5236.Teshima, T.; Ordemann, R.; Reddy, P.; Gagin, S.; Liu, C.; Cooke,K. R.; Ferrara, J. L. M.: Acute graft-versus-host disease does notrequire alloantigen expression on host epithelium. *Nature Med.* 8:575–581, 2002.

[31685] 5237.Thorsby, E.; Sandberg, L.; Lindholm, A.; Kissmeyer-Nielsen, F.: The HL-A system: evidence of a third sub-locus. *Scand. J. Haemat.* 7:195–200, 1970.

[31686] 5238.Tragardh, L.; Rask, L.; Wiman, K.; Fohlman, J.; Peterson, P. A.: Amino acid sequence of an immunoglobulin HLA antigen heavy chaindomain. *Proc. Nat. Acad. Sci.* 76: 5839–5842, 1979.

[31687] 5239.Trowsdale, J.: Personal Communication. London, England 1/12/1983.

[31688] 5240.Trowsdale, J.; Ragoussis, J.; Campbell, R. D.: Map of

the humanMHC. *Immun. Today* 12: 443–446, 1991.

[31689] 5241. Van Leeuwen, A.; Eernisse, J. G.; Van Rood, J. J.: A new leucocytegroup with two alleles: leucocyte group five. *Vox Sang.* 9: 431–446, 1964.

[31690] 5242. Van Rood, J. J.: Leucocyte grouping and organ transplantation. *Brit.J. Haemat.* 16: 211–220, 1969.100. Van Rood, J. J.: Tissue typing and organ transplantation. *Lancet* I:1142–1146, 1969.101. Van Rood, J. J.; Amos, D. B.: In memoriam: Ruggero Ceppellini, 1917–1988. *Hum. Immun.* 23: 1–3, 1988.102. Van Rood, J. J.; Van Leeuwen, A.: Leukocyte grouping: a method and its application. *J. Clin. Invest.* 42: 1382–1390, 1963.103. Van Someren, H.; Westerveld, A.; Hagemeijer, A.; Mees, J. R.; Meera Khan, P.; Zaalberg, O. B.: Human antigen and enzyme markers in man–Chinese hamster somatic cell hybrids: evidence for synteny between the HL–A, PGM–3, ME–1, and IPO–B loci. *Proc. Nat. Acad. Sci.* 71:962–965, 1974.104. Walford, R. L.; Finkelstein, S.; Hanna, C.; Collins, Z.: Third sublocus in the HL–A human transplantation system. *Nature* 224: 74–75, 1969.105. Weitkamp, L. R.; Van Rood, J. J.; Thorsby, E.; Bias, W. B.; Fotino, M.; Lawler, S. D.; Dausset, J.; Mayr, W. R.; Bodmer, J.; Ward, F.S.; Seignalet, J.; Payne, R.; Kissmeyer–Nielsen, F.; Gatti, R. A.; Sachs, J. A.; Lamm,

L. U.: The relation of parental sex and age to recombination in the HL-A system. *Hum. Hered.* 23: 197–205, 1973.106. Wolski, K. P.; Schmid, F. R.; Mittal, K. K.: Genetic linkage between the HL-A system and a deficit of the second component (C2) of complement. *Science* 188: 1020–1022, 1975.107. Ziegler, A.; Ragoussis, J.; Fonatsch, C.; Weiss, E.: A physical map of the human major histocompatibility (HLA) complex derived from a monosomy 6 mutant cell line. (Abstract) *Cytogenet. Cell Genet.* 51:1116 only, 1989.

[31691] 5243. Wildhage, I.; Trusheim, H.; Goke, B.; Lankat-Buttgereit, B.: Gene expression of the human glucagon-like peptide-1 receptor is regulated by Sp1 and Sp3. *Endocrinology* 140: 624–631, 1999.

[31692] 5244. Chambers, S. M.; Morris, B. J.: Glucagon receptor gene mutation in essential hypertension. (Letter) *Nature Genet.* 12: 122, 1996.

[31693] 5245. Hager, J.; Hansen, L.; Vaisse, C.; Vionnet, N.; Philippi, A.; Poller, W.; Velho, G.; Carcassi, C.; Contu, L.; Julier, C.; Cambien, F.; Passa, P.; Lathrop, M.; Kindsvogel, W.; Demenais, F.; Nishimura, E.; Froguel, P.: A missense mutation in the glucagon receptor gene is associated with non-insulin-dependent diabetes mellitus. *Nature Genet.*

9: 299–304,1995.

- [31694] 5246.Lok, S.; Kuijper, J. L.; Jelinek, L. J.; Kramer, J. M.; Whitmore,T. E.; Sprecher, C. A.; Mathewes, S.; Grant, F. J.; Biggs, S. H.;Rosenberg, G. B.; Sheppard, P. O.; O'Hara, P. J.; Foster, D. C.; Kindsvogel,W.: The human glucagon receptor encoding gene: structure, cDNA sequenceand chromosomal localization. *Gene* 140: 203–209, 1994.
- [31695] 5247.Menzel, S.; Stoffel, M.; Espinosa, R., III; Fernald, A. A.; LeBeau, M. M.; Bell, G. I.: Localization of the glucagon receptor geneto human chromosome band 17q25. *Genomics* 20: 327–328, 1994.
- [31696] 5248.Weitkamp, L. R.: Concerning the linkage relationships of theGc and MNSs loci. *Hum. Genet.* 43: 215–220, 1978.
- [31697] 5249.Boguszewski, C. L.; Svensson, P.–A.; Jansson, T.; Clark, R.; Carlsson,L. M. S.; Carlsson, B.: Cloning of two novel growth hormone transcriptsexpressed in human placenta. *J. Clin. Endocr. Metab.* 83: 2878–2885,1998.
- [31698] 5250.Chen, E. Y.; Liao, Y.–C.; Smith, D. H.; Barrera–Saldana, H. A.;Gelinas, R. E.; Seeburg, P. H.: The human growth hormone locus: nucleotidesequence, biology, and evolution. *Genomics* 4: 479–497, 1989.
- [31699] 5251.Frankenne, F.; Rentier–Delrue, F.; Scippo, M.–L.;

Martial, J.;Hennen, G.: Expression of the growth hormone variant gene in humanplacenta. J. Clin. Endocr. Metab. 64: 635–637, 1987.

- [31700] 5252.Lewis, U. J.; Dunn, J. T.; Bonewald, L. F.; Seavey, B. K.; VanderLaan,W. P.: A naturally occurring structural variant of human growth hormone. J.Biol. Chem. 253: 2679–2687, 1978.
- [31701] 5253.Liebhaber, S. A.; Urbanek, M.; Ray, J.; Tuan, R. S.; Cooke, N.E.: Characterization and histologic localization of human growthhormone–variant gene expression in the placenta. J. Clin. Invest. 83:1985–1991, 1989.
- [31702] 5254.MacLeod, J. N.; Liebhaber, S. A.; MacGillivray, M. H.; Cooke, N.E.: Identification of a splice–site mutation in the human growthhormone–variant gene. Am. J. Hum. Genet. 48: 1168–1174, 1991.
- [31703] 5255.MacLeod, J. N.; Worsley, I.; Ray, J.; Friesen, H. G.; Liebhaber,S. A.; Cooke, N. E.: Human growth hormone variant is a biologicallyactive somatogen and lactogen. Endocrinology 128: 1298–1302, 1991.
- [31704] 5256.Owerbach, D.; Rutter, W. J.; Martial, J. A.; Baxter, J. D.; Shows,T. B.: Genes for growth hormone, chorionic somatomammotropin andgrowth hormone–like genes on chromosome 17 in humans. Science 209:289–292, 1980.

- [31705] 5257. Harada, H.; Fujita, T.; Miyamoto, M.; Kimura, Y.; Maruyama, M.; Furia, A.; Miyata, T.; Taniguchi, T.: Structurally similar but functionally distinct factors, IRF-1 and IRF-2, bind to the same regulatory elements of IFN and IFN-inducible genes. *Cell* 58: 729–739, 1989.
- [31706] 5258. Wong, P.; Colucci-Guyon, E.; Takahashi, K.; Gu, C.; Babinet, C.; Coulombe, P. A.: Introducing a null mutation in the mouse K6-alpha and K6-beta genes reveals their essential structural role in the oral mucosa. *J. Cell Biol.* 150: 921–928, 2000.
- [31707] 5259. Nakashima, K.; Yanagisawa, M.; Arakawa, H.; Kimura, N.; Hisatsune, T.; Kawabata, M.; Miyazono, K.; Taga, T.: Synergistic signaling in fetal brain by STAT3-Smad1 complex bridged by p300. *Science* 284: 479–482, 1999.
- [31708] 5260. Bae, J.; Leo, C. P.; Hsu, S. Y.; Hsueh, A. J. W.: MCL-1S, a splicing variant of the antiapoptotic BCL-2 family member MCL-1, encodes a proapoptotic protein possessing only the BH3 domain. *J. Biol. Chem.* 275: 25255–25261, 2000.
- [31709] 5261. Craig, R. W.; Jabs, E. W.; Zhou, P.; Kozopas, K. M.; Hawkins, A. L.; Rochelle, J. M.; Seldin, M. F.; Griffin, C. A.: Human and mouse chromosomal mapping of the myeloid

cell leukemia-1 gene: MCL1 maps to human chromosome 1q21, a region that is frequently altered in preneoplastic and neoplastic disease. *Genomics* 23: 457-463, 1994.

[31710] 5262. Kozopas, K. M.; Yang, T.; Buchan, H. L.; Zhou, P.; Craig, R. W.: MCL1, a gene expressed in programmed myeloid cell differentiation, has sequence similarity to BCL2. *Proc. Nat. Acad. Sci.* 90: 3516-3520, 1993.

[31711] 5263. Rinkenberger, J. L.; Horning, S.; Klocke, B.; Roth, K.; Korsmeyer, S. J.: Mcl-1 deficiency results in peri-implantation embryonic lethality. *Genes Dev.* 14: 23-27, 2000.

[31712] 5264. Hizawa, N.; Yamaguchi, E.; Furuya, K.; Ohnuma, N.; Kodama, N.; Kojima, J.; Ohe, M.; Kawakami, Y.: Association between high serum total IgE levels and D11S97 on chromosome 11q13 in Japanese subjects. *J. Med. Genet.* 32: 363-369, 1995.

[31713] 5265. Sandford, A. J.; Shirakawa, T.; Moffatt, M. F.; Daniels, S. E.; Ra, C.; Faux, J. A.; Young, R. P.; Nakamura, Y.; Lathrop, G. M.; Cookson, W. O. C. M.; Hopkin, J. M.: Localisation of atopy and beta subunit of high-affinity IgE receptor (FCER1) on chromosome 11q. *Lancet* 341: 332-334, 1993.

[31714] 5266. Harada, H.; Kondo, T.; Ogawa, S.; Tamura, T.; Kitagawa, M.; Tanaka, N.; Lamphier, M. S.; Hirai, H.; Taniguchi,

T.: Accelerated exon skipping of IRF-1 mRNA in human myelodysplasia/leukemia: a possible mechanism of tumor suppressor inactivation *Oncogene* 9: 3313–3320, 1994.

- [31715] 5267. Harada, H.; Willison, K.; Sakakibara, J.; Miyamoto, M.; Fujita, T.; Taniguchi, T.: Absence of the type I IFN system in EC cells: transcriptional activator (IRF-1) and repressor (IRF-2) genes are developmentally regulated. *Cell* 63: 303–312, 1990.
- [31716] 5268. Itoh, S.; Harada, H.; Nakamura, Y.; White, R.; Taniguchi, T.: Assignment of the human interferon regulatory factor-1 (IRF1) gene to chromosome 5q23–q31. *Genomics* 10: 1097–1099, 1991.
- [31717] 5269. Ko, J.; Gendron-Fitzpatrick, A.; Splitter, G. A.: Susceptibility of IFN regulatory factor-1 and IFN consensus sequence binding protein-deficient mice to brucellosis. *J. Immun.* 168: 2433–2440, 2002.
- [31718] 5270. Miyamoto, M.; Fujita, T.; Kimura, Y.; Maruyama, M.; Harada, H.; Sudo, Y.; Miyata, T.; Taniguchi, T.: Regulated expression of a gene encoding a nuclear factor, IRF-1, that specifically binds to IFN- β gene regulatory elements. *Cell* 54: 903–913, 1988.
- [31719] 5271. Nozawa, H.; Oda, E.; Ueda, S.; Tamura, G.; Maesawa, C.; Muto, T.; Taniguchi, T.; Tanaka, N.: Functionally inacti-

vating point mutation in the tumor-suppressor IRF-1 gene identified in human gastric cancer. *Int.J. Cancer* 77: 522-527, 1998.

- [31720] 5272. Tamura, G.; Sakata, K.; Nishizuka, S.; Maesawa, C.; Suzuki, Y.; Terashima, M.; Eda, Y.; Satodate, R.: Allelotype of adenoma and differentiated adenocarcinoma of the stomach. *J. Path.* 180: 371-377, 1996.
- [31721] 5273. Willman, C. L.; Sever, C. E.; Pallavicini, M. G.; Harada, H.; Tanaka, N.; Slovak, M. L.; Yamamoto, H.; Harada, K.; Meeker, T. C.; List, A. F.; Taniguchi, T.: Deletion of IRF-1, mapping to chromosome 5q31.1, in human leukemia and preleukemic myelodysplasia. *Science* 259: 968-971, 1993.
- [31722] 5274. Yamada, G.; Ogawa, M.; Akagi, K.; Miyamoto, H.; Nakano, N.; Itoh, S.; Miyazaki, J.; Nishikawa, S.; Yamamura, K.; Taniguchi, T.: Specific depletion of the B-cell population induced by aberrant expression of human interferon regulatory factor 1 gene in transgenic mice. *Proc. Nat. Acad. Sci.* 88: 532-536, 1991.
- [31723] 5275. Harada, H.; Kitagawa, M.; Tanaka, N.; Yamamoto, H.; Harada, K.; Ishihara, M.; Taniguchi, T.: Anti-oncogenic and oncogenic potential of interferon regulatory factors-1 and -2. *Science* 259: 971-974, 1993.
- [31724] 5276. Harada, H.; Takahashi, E.-I.; Itoh, S.; Harada, K.;

Hori, T.-A.; Taniguchi, T.: Structure and regulation of the human interferon regulatory factor 1 (IRF-1) and IRF-2 genes: implications for a gene network in the interferon system. *Molec. Cell. Biol.* 14: 1500–1509, 1994.

[31725] 5277. Hida, S.; Ogasawara, K.; Sato, K.; Abe, M.; Takayanagi, H.; Yokochi, T.; Sato, T.; Hirose, S.; Shirai, T.; Taki, S.; Taniguchi, T.: CD8⁺T cell-mediated skin disease in mice lacking IRF-2, the transcriptional attenuator of interferon- α /beta signaling. *Immunity* 13: 643–655, 2000.

[31726] 5278. Nishio, Y.; Noguchi, E.; Ito, S.; Ichikawa, E.; Umebayashi, Y.; Otsuka, F.; Arinami, T.: Mutation and association analysis of the interferon regulatory factor 2 gene (IRF2) with atopic dermatitis. *J. Hum. Genet.* 46: 664–667, 2001.

[31727] 5279. Guo, B.; Yu, Y.; Leibold, E. A.: Iron regulates cytoplasmic levels of a novel iron-responsive element-binding protein without aconitase activity. *J. Biol. Chem.* 269: 24252–24260, 1994.

[31728] 5280. Henderson, B. R.; Seiser, C.; Kuhn, L. C.: Characterization of a second RNA-binding protein in rodents with specificity for iron-responsive elements. *J. Biol. Chem.* 268: 27327–27334, 1993.

- [31729] 5281.Hentze, M. W.; Kuhn, L. C.: Molecular control of vertebrate ironmetabolism: mRNA-based regulatory circuits operated by iron, nitricoxide, and oxidative stress. Proc. Nat. Acad. Sci. 93: 8175–8182,1996.
- [31730] 5282.Iwai, K.; Klausner, R. D.; Rouault, T. A.: Requirements for iron-regulateddegradation of the RNA binding protein, iron regulatory protein 2. EMBOJ. 14: 5350–5357, 1995.
- [31731] 5283.Rouault, T. A.; Tang, C. K.; Kaptain, S.; Burgess, W. H.; Haile,D. J.; Samaniego, F.; McBride, O. W.; Harford, J. B.; Klausner, R.D.: Cloning of the cDNA encoding an RNA regulatory protein: the humaniron-responsive element-binding protein. Proc. Nat. Acad. Sci. 87:7958–7962, 1990.
- [31732] 5284.LaVaute, T.; Smith, S.; Cooperman, S.; Iwai, K.; Land, W.; Meyron-Holtz,E.; Drake, S. K.; Miller, G.; Abu-Asab, M.; Tsokos, M.; Switzer, R.,III; Grinberg, A.; Love, P.; Tresser, N.; Rouault, T. A.: Targeteddeletion of the gene encoding iron regulatory protein-2 causes misregulationof iron metabolism and neurodegenerative disease in mice. NatureGenet. 27: 209–214, 2001.
- [31733] 5285.Samaniego, F.; Chin, J.; Iwai, K.; Rouault, T. A.; Klausner, R.D.: Molecular characterization of a second

iron-responsive element-binding protein, iron regulatory protein 2: structure, function, and post-translational regulation. *J. Biol. Chem.* 269: 30904–30910, 1994.

- [31734] 5286. Smith, F. J. D.; Jonkman, M. F.; van Goor, H.; Coleman, C. M.; Covello, S. P.; Uitto, J.; McLean, W. H. I.: A mutation in human keratin K6b produces a phenocopy of the K17 disorder pachyonychia congenita type 2. *Hum. Molec. Genet.* 7: 1143–1148, 1998.
- [31735] 5287. Moll, R.; Franke, W. W.; Schiller, D. L.; Geiger, B.; Krepler, R.: The catalog of human cytokeratins: patterns of expression in normal epithelia, tumors and cultured cells. *Cell* 31: 11–24, 1982.
- [31736] 5288. Casanova, M. L.; Bravo, A.; Ramirez, A.; Morreale de Escobar, G.; Were, F.; Merlino, G.; Vidal, M.; Jorcano, J. L.: Exocrine pancreatic disorders in transgenic (sic) mice expressing human keratin 8. *J. Clin. Invest.* 103: 1587–1595, 1999.
- [31737] 5289. Jackson, B. W.; Grund, C.; Schmid, E.; Burke, K.; Franke, W.; Illmensee, K.: Formation of cytoskeletal elements during mouse embryogenesis: intermediate filaments of the cytokeratin type and desmosomes in preimplantation embryos. *Differentiation* 17: 161–179, 1980.
- [31738] 5290. Ku, N.-O.; Gish, R.; Wright, T. L.; Omary, M. B.: Ker-

atin 8 mutations in patients with cryptogenic liver disease.
New Eng. J. Med. 344:1580–1587, 2001.

- [31739] 5291. Ku, N.-O.; Wright, T. L.; Terrault, N. A.; Gish, R.; Omary, M.B.: Mutation of human keratin 18 in association with cryptogenic cirrhosis. *J. Clin. Invest.* 99: 19–23, 1997.
- [31740] 5292. Waseem, A.; Alexander, C. M.; Steel, J. B.; Lane, E. B.: Embryonic simple epithelial keratins 8 and 18: chromosomal location emphasizes difference from other keratin pairs. *New Biologist* 2: 464–478, 1990.
- [31741] 5293. Yamamoto, R.; Kao, L.-C.; McKnight, C. E.; Strauss, J. F., III: Cloning and sequence of cDNA for human placental cytokeratin 8: regulation of the mRNA in trophoblastic cells by cAMP. *Molec. Endocr.* 4:370–374, 1990.
- [31742] 5294. Fan, Q. R.; Long, E. O.; Wiley, D. C.: Crystal structure of the human natural killer cell inhibitory receptor KIR2DL1–HLA–Cw4 complex. *Nature Immun.* 2: 452–460, 2001.
- [31743] 5295. Yen, J.-H.; Moore, B. E.; Nakajima, T.; Scholl, D.; Schaid, D.J.; Weyand, C. M.; Goronzy, J. J.: Major histocompatibility complex class I–recognizing receptors are disease risk genes in rheumatoid arthritis. *J. Exp. Med.* 193: 1159–1167, 2001.
- [31744] 5296. Kogan, S. C.; Lagasse, E.; Atwater, S.; Bae, S.; Weiss–

man, I.; Ito, Y.; Bishop, J. M.: The PEBP2-beta-MYH11 fusion created by inv(16)(p13;q22) in myeloid leukemia impairs neutrophil maturation and contributes to granulocytic dysplasia. Proc. Nat. Acad. Sci. 95: 11863-11868, 1998.

[31745] 5297. Liu, P.; Tarle, S. A.; Hajra, A.; Claxton, D. F.; Marlton, P.; Freedman, M.; Siciliano, M. J.; Collins, F. S.: Fusion between transcription factor CBF-beta/PEBP2-beta and a myosin heavy chain in acute myeloid leukemia. Science 261: 1041-1044, 1993.

[31746] 5298. Liu, P. P.; Hajra, A.; Wijmenga, C.; Collins, F. S.: Molecular pathogenesis of the chromosome 16 inversion in the M4Eo subtype of acute myeloid leukemia. Blood 85: 2289-2302, 1995.

[31747] 5299. Lutterbach, B.; Hou, Y.; Durst, K. L.; Hiebert, S. W.: The inv(16) encodes an acute myeloid leukemia 1 transcriptional corepressor. Proc. Nat. Acad. Sci. 96: 12822-12827, 1999.

[31748] 5300. O'Reilly, J.; Chipper, L.; Springall, F.; Herrmann, R.: A unique structural abnormality of chromosome 16 resulting in a CBF-beta-MYH11 fusion transcript in a patient with acute myeloid leukemia, FAB M4. Cancer Genet. Cytogenet. 121: 52-55, 2000.

- [31749] 5301.Ogawa, E.; Inuzuka, M.; Maruyama, M.; Satake, M.; Naito-Fujimoto,M.; Ito, Y.; Shigesada, K.: Molecular cloning and characterization of PEBP2-beta, the heterodimeric partner of a novel Drosophila runt-related DNA binding protein PEBP2-alpha. *Virology* 194: 314-331, 1993.
- [31750] 5302.Wang, S.; Wang, Q.; Crute, B. E.; Melnikova, I. N.; Keller, S.R.; Speck, N. A.: Cloning and characterization of subunits of the T-cell receptor and murine leukemia virus enhancer core-binding factor. *Molec.Cell. Biol.* 13: 3324-3339, 1993.
- [31751] 5303.Pellegata, N. S.; Dieguez-Lucena, J. L.; Joensuu, T.; Lau, S.;Montgomery, K. T.; Krahe, R.; Kivela, T.; Kucherlapati, R.; Forsius,H.; de la Chapelle, A.: Mutations in KERA, encoding keratocan, cause cornea plana. *Nature Genet.* 25: 91-95, 2000.
- [31752] 5304.Fabrizi, G. M.; Rizzuto, R.; Nakase, H.; Mita, S.; Lomax, M. I.;Grossman, L. I.; Schon, E. A.: Sequence of a cDNA specifying subunit VIIa of human cytochrome c oxidase. *Nucleic Acids Res.* 17: 7107only, 1989.
- [31753] 5305.Wolz, W.; Kress, W.; Mueller, C. R.: Genomic sequence structure and organization of the human gene for cytochrome c oxidase subunit (COX7A1) VIIa-M. *Genomics*

45: 438–442, 1997.

- [31754] 5306. Brooks, B. A.; McBride, O. W.; Dolphin, C. T.; Farrall, M.; Scambler, P. J.; Gonzalez, F. J.; Idle, J. R.: The gene CYP3 encoding P450PCN1 (nifedipine oxidase) is tightly linked to the gene COL1A2 encoding collagen type 1 alpha on 7q21–q22.1. *Am. J. Hum. Genet.* 43: 280–284, 1988.
- [31755] 5307. Chen, H.; Sandler, D. P.; Taylor, J. A.; Shore, D. L.; Liu, E.; Bloomfield, C. D.; Bell, D. A.: Increased risk for myelodysplastic syndromes in individuals with glutathione transferase theta 1 (GSTT1) gene defect. *Lancet* 347: 295–297, 1996.
- [31756] 5308. Daly, A. K.; Salh, B. S.; Bilton, D.; Allen, J.; Knight, A. D.; Webb, A. K.; Braganza, J. M.; Idle, J. R.: Deficient nifedipine oxidation: a rare inherited trait associated with cystic fibrosis kindreds. *Pharmacogenetics* 2: 19–24, 1992.
- [31757] 5309. Elshourbagy, N. A.; Guzelian, P. S.: Separation, purification, and characterization of a novel form of hepatic cytochrome P-450 from rats treated with pregnenolone-16- α -carbonitrile. *J. Biol. Chem.* 255: 1279–1285, 1980.
- [31758] 5310. Felix, C. A.; Walker, A. H.; Lange, B. J.; Williams, T. M.; Winick, N. J.; Cheung, N.-K. V.; Lovett, B. D.; Nowell, P. C.; Blair, I. A.; Rebbeck, T. R.: Association of CYP3A4 geno-

type with treatment-related leukemia. Proc. Nat. Acad. Sci. 95: 13176–13181, 1998.

[31759] 5311. Forrester, L. M.; Neal, G. E.; Judah, D. J.; Glancey, M. J.; Wolf, C. R.: Evidence for involvement of multiple forms of cytochrome P-450 in aflatoxin B(1) metabolism in human liver. Proc. Nat. Acad. Sci. 87: 8306–8310, 1990.

[31760] 5312. Gonzalez, F. J.; Schmid, B. J.; Umeno, M.; McBride, O. W.; Hardwick, J. P.; Meyer, U. A.; Gelboin, H. V.; Idle, J. R.: Human P450PCN1: sequence, chromosome localization, and direct evidence through cDNA expression that P450PCN1 is nifedipine oxidase. DNA 7: 79–86, 1988.

[31761] 5313. Hoyo-Vadillo, C.; Castaneda-Hernandez, G.; Herrera, J. E.; Vidal-Garate, J.; Moreno-Ramos, A.; Chavez, F.; Hong, E.: Pharmacokinetics of nifedipine slow release tablet in Mexican subjects: further evidence for an oxidation polymorphism. J. Clin. Pharm. 29: 816–820, 1989.

[31762] 5314. Inoue, K.; Inazawa, J.; Nakagawa, H.; Shimada, T.; Yamazaki, H.; Guengerich, F. P.; Abe, T.: Assignment of the human cytochrome P-450 nifedipine oxidase gene (CYP3A4) to chromosome 7 at band q22.1 by fluorescence in situ hybridization. Jpn. J. Hum. Genet. 37: 133–138, 1992.

[31763] 5315. Kittles, R. A.; Chen, W.; Panguluri, R. K.; Ahaghotu,

C.; Jackson,A.; Adebamowo, C. A.; Griffin, R.; Williams, T.; Ukoli, F.; Adams–Campbell,L.; Kwagyan, J.; Isaacs, W.; Freeman, V.; Dunston, G. M.: CYP3A4–Vand prostate cancer in African Americans: causal or confounding associationbecause of population stratification? Hum. Genet. 110: 553–560,2002.

[31764] 5316.Kleinbloesem, C. H.; van Brummelen, P.; Faber, H.; Danhof, M.;Vermeulen, N. P. E.; Breimer, D. D.: Variability in nifedipine pharmacokineticsand dynamics: a new oxidation polymorphism in man. Biochem. Pharm. 33:3721–3724, 1984.

[31765] 5317.Lehmann, J. M.; McKee, D. D.; Watson, M. A.; Willson, T. M.; Moore,J. T.; Klierwer, S. A.: The human orphan nuclear receptor PXR is activatedby compounds that regulate CYP3A4 gene expression and cause drug interactions. J.Clin. Invest. 102: 1016–1023, 1998.

[31766] 5318.Lown, K. S.; Bailey, D. G.; Fontana, R. J.; Janardan, S. K.; Adair,C. H.; Fortlage, L. A.; Brown, M. B.; Guo, W.; Watkins, P. B.: Grapefruitjuice increases felodipine oral availability in humans by decreasingintestinal CYP3A protein expression. J. Clin. Invest. 99: 2545–2553,1997.

[31767] 5319.Molowa, D. T.; Schuetz, E. G.; Wrighton, S. A.; Watkins, P. B.;Kremers, P.; Mendez–Picon, G.; Parker, G.

A.; Guzelian, P. S.: Complete cDNA sequence of a cytochrome P-450 inducible by glucocorticoids in human liver. *Proc. Nat. Acad. Sci.* 83: 5311–5315, 1986.

[31768] 5320. Paris, P. L.; Kupelian, P. A.; Hall, J. M.; Williams, T. L.; Levin, H.; Klein, E. A.; Casey, G.; Witte, J. S.: Association between a CYP3A4 genetic variant and clinical presentation in African-American prostate cancer patients. *Cancer Epidemiol. Biomarkers Prev.* 8:901–905, 1999.

[31769] 5321. Rebbeck, T. R.; Jaffe, J. M.; Walker, A. H.; Wein, A. J.; Malkowicz, S. B.: Modification of clinical presentation of prostate tumors by a novel genetic variant in CYP3A4. *J. Nat. Cancer Inst.* 90: 1225–1229, 1998.

[31770] 5322. Renwick, A. G.; Robertson, D. R. C.; Macklin, B.; Challenor, V.; Waller, D. G.; George, C. F.: The pharmacokinetics of oral nifedipine—a population study. *Brit. J. Clin. Pharm.* 25: 701–708, 1988.

[31771] 5323. Nebert, D. W.; Adesnik, M.; Coon, M. J.; Estabrook, R. W.; Gonzalez, F. J.; Guengerich, F. P.; Gunsalus, I. C.; Johnson, E. F.; Kemper, B.; Levin, W.; Phillips, I. R.; Sato, R.; Waterman, M. R.: The P450 gene superfamily: recommended nomenclature. *DNA* 6: 1–11, 1987.

[31772] 5324. Spielberg, S. P.: Personal Communication. Toronto, Ontario, Canada 2/26/1988.

- [31773] 5325.Thum, T.; Borlak, J.: Gene expression in distinct regions of the heart. *Lancet* 355: 979–983, 2000.
- [31774] 5326.Chen, Y.; Faraco, J.; Yin, W.; Germiller, J.; Francke, U.; Bonadio, J.: Structure, chromosomal localization, and expression pattern of the murine Magp gene. *J. Biol. Chem.* 268: 27381–27389, 1993.
- [31775] 5327.Faraco, J.; Bashir, M.; Rosenbloom, J.; Francke, U.: Characterization of the human gene for microfibril-associated glycoprotein (MFAP2), assignment to chromosome 1p36.1–p35, and linkage to D1S170. *Genomics* 25:630–637, 1995.
- [31776] 5328.Gibson, M. A.; Hughes, J. L.; Fanning, J. C.; Cleary, E. G.: The major antigen of elastin-associated microfibrils is a 31-kDa glycoprotein. *J. Biol. Chem.* 261: 11429–11436, 1986.
- [31777] 5329.Low, F. N.: Microfibrils: fine filamentous components of the tissue space. *Anat. Rec.* 142: 131–137, 1962.
- [31778] 5330.Aberdam, E.; Bertolotto, C.; Sviderskaya, E. V.; de Thillot, V.; Hemesath, T. J.; Fisher, D. E.; Bennett, D. C.; Ortonne, J.-P.; Ballotti, R.: Involvement of microphthalmia in the inhibition of melanocyte lineage differentiation and of melanogenesis by agouti signal protein. *J. Biol. Chem.* 273: 19560–19565, 1998.

- [31779] 5331.Kelavkar, U. P.; Badr, K. F.: Effects of mutant p53 expressionon human 15-lipoxygenase-promoter activity and murine 12/15-lipoxygenasegene expression: evidence that 15-lipoxygenase is a mutator gene. *Proc.Nat. Acad. Sci.* 96: 4378–4383, 1999.
- [31780] 5332.Sigal, E.; Craik, C. S.; Highland, E.; Grunberger, D.; Costello,L. L.; Dixon, R. A. F.; Nadel, J. A.: Molecular cloning and primarystructure of human 15-lipoxygenase. *Biochem. Biophys. Res. Commun.* 157:457–464, 1988.
- [31781] 5333.Yoshimoto, T.; Suzuki, H.; Yamamoto, S.; Takai, T.; Yokoyama, C.;Tanabe, T.: Cloning and sequence analysis of the cDNA for arachidonate12-lipoxygenase of porcine leukocytes. *Proc. Nat. Acad. Sci.* 87:2142–2146, 1990.
- [31782] 5334.Minoshima, S.; Fukuyama, R.; Yamamoto, T.; Shimizu, N.: Mappingof human long-chain acyl-CoA synthetase to chromosome 4. (Abstract) *Cytogenet.Cell Genet.* 58: 1888 only, 1991.
- [31783] 5335.Cantu, E. S.; Sprinkle, T. J.; Ghosh, B.; Singh, I.: The humanpalmitoyl-CoA ligase (FACL2) gene maps to the chromosome 4q34–q35region by fluorescence in situ hybridization (FISH) and somatic cellhybrid panels. *Genomics* 28: 600–602, 1995.
- [31784] 5336.Benson, D. W.; MacRae, C. A.; Vesely, M. R.; Walsh, E.

P.; Seidman, J. G.; Seidman, C. E.; Satler, C. A.: Missense mutation in the poreregion of HERG causes familial long QT syndrome. *Circulation* 93:1791–1795, 1996.

[31785] 5337. Brook, J. D.; Shaw, D. J.; Meredith, A. L.; Bruns, G. A. P.; Harper, P. S.: Localisation of genetic markers and orientation of the linkage group on chromosome 19. *Hum. Genet.* 68: 282–285, 1984.

[31786] 5338. Bruns, G. A. P.; Regina, V. M.; Gerald, P. S.: Lysosomal DNase and chromosome 19. (Abstract) *J. Cell Biol.* 83: 444a only, 1979.

[31787] 5339. Kawane, K.; Fukuyama, H.; Kondoh, G.; Takeda, J.; Ohsawa, Y.; Uchiyama, Y.; Nagata, S.: Requirement of DNase II for definitive erythropoiesis in the mouse fetal liver. *Science* 292: 1546–1549, 2001.

[31788] 5340. Yasuda, T.; Nadano, D.; Sawazaki, K.; Kishi, K.: Genetic polymorphism of human deoxyribonuclease II (DNase II): low activity levels in urine and leukocytes are due to an autosomal recessive allele. *Ann. Hum. Genet.* 56: 1–10, 1992.

[31789] 5341. Yasuda, T.; Takeshita, H.; Iida, R.; Nakajima, T.; Hosomi, O.; Nakashima, Y.; Kishi, K.: Molecular cloning of the cDNA encoding human deoxyribonuclease II. *J. Biol. Chem.* 273: 2610–2616, 1998.

- [31790] 5342.Yasuda, T.; Takeshita, H.; Iida, R.; Nakajima, T.; Hosomi, O.; Nakashima, Y.; Mogi, K.; Kishi, K.: Chromosomal localization of a human deoxyribonuclease II gene (DNASE2) to 19p13.2–p13.1 using both the polymerase chain reaction and fluorescence in situ hybridization analysis. *Biochem. Biophys. Res. Commun.* 244: 815–818, 1998.
- [31791] 5343.Baylin, S. B.: Tying it all together: epigenetics, genetics, cell cycle, and cancer. *Science* 277: 1948–1949, 1997.
- [31792] 5344.Bestor, T.; Laudano, A.; Mattaliano, R.; Ingram, V.: Cloning and sequencing of a cDNA encoding DNA methyltransferase of mouse cells: the carboxyl-terminal domain of the mammalian enzymes is related to bacterial restriction methyltransferases. *J. Molec. Biol.* 203: 971–983, 1988.
- [31793] 5345.Bestor, T. H.: The DNA methyltransferases of mammals. *Hum. Molec. Genet.* 9: 2395–2402, 2000.
- [31794] 5346.Bestor, T. H.: DNA methylation: evolution of a bacterial immune function into a regulator of gene expression and genome structure in higher eukaryotes. *Phil. Trans. Roy. Soc. London B* 326: 179–187, 1990.
- [31795] 5347.Chuang, L. S.–H.; Ian, H.–I.; Koh, T.–W.; Ng, H.–H.;

Xu, G.; Li, B. F. L.: Human DNA-(cytosine-5) methyltransferase-PCNA complex as a target for p21(WAF1). *Science* 277: 1996-2000, 1997.

- [31796] 5348. El-Deiry, W. S.; Nelkin, B. D.; Celano, P.; Chiu Yen, R.-W.; Falco, J. P.; Hamilton, S. R.; Baylin, S. B.: High expression of the DNA methyltransferase gene characterizes human neoplastic cells and progression stages of colon cancer. *Proc. Nat. Acad. Sci.* 88: 3470-3474, 1991.
- [31797] 5349. Fuks, F.; Burgers, W. A.; Brehm, A.; Hughes-Davies, L.; Kouzarides, T.: DNA methyltransferase Dnmt1 associates with histone deacetylase activity. *Nature Genet.* 24: 88-91, 2000.
- [31798] 5350. Howell, C. Y.; Bestor, T. H.; Ding, F.; Latham, K. E.; Mertineit, C.; Trasler, J. M.; Chaillet, J. R.: Genomic imprinting disrupted by a maternal effect mutation in the Dnmt1 gene. *Cell* 104: 829-838, 2001.
- [31799] 5351. Hsu, D.-W.; Lin, M.-J.; Lee, T.-L.; Wen, S.-C.; Chen, X.; Shen, C.-K. J.: Two major forms of DNA (cytosine-5) methyltransferase in human somatic tissues. *Proc. Nat. Acad. Sci.* 96: 9751-9756, 1999.
- [31800] 5352. Lee, P. P.; Fitzpatrick, D. R.; Beard, C.; Jessup, H. K.; Lehar, S.; Makar, K. W.; Perez-Melgosa, M.; Sweetser, M. T.; Schlissel, M. S.; Nguyen, S.; Cherry, S. R.; Tsai, J. H.;

Tucker, S. M.; Weaver, W. M.; Kelso, A.; Jaenisch, R.; Wilson, C. B.: A critical role for Dnmt1 and DNA methylation in T cell development, function, and survival. *Immunity* 15:763–774, 2001.

[31801] 5353. Li, E.; Bestor, T. H.; Jaenisch, R.: Targeted mutation of the DNA methyltransferase gene results in embryonic lethality. *Cell* 69:915–926, 1992.

[31802] 5354. Lyko, F.; Ramsahoye, B. H.; Kashevsky, H.; Tudor, M.; Mastrangelo, M.-A.; Orr-Weaver, T. L.; Jaenisch, R.: Mammalian (cytosine-5) methyltransferases cause genomic DNA methylation and lethality in *Drosophila*. *Nature Genet.* 23: 363–366, 1999.

[31803] 5355. Rhee, I.; Bachman, K. E.; Park, B. H.; Jair, K.-W.; Yen, R.-W. C.; Schuebel, K. E.; Cui, H.; Feinberg, A. P.; Lengauer, C.; Kinzler, K. W.; Baylin, S. B.; Vogelstein, B.: DNMT1 and DNMT3b cooperate to silence genes in human cancer cells. *Nature* 416: 552–556, 2002.

[31804] 5356. Rhee, I.; Jair, K.-W.; Yen, R.-W. C.; Lengauer, C.; Herman, J. G.; Kinzler, K. W.; Vogelstein, B.; Baylin, S. B.; Schuebel, K. E.: CpG methylation is maintained in human cancer cells lacking DNMT1. *Nature* 404:1003–1007, 2000.

[31805] 5357. Robertson, K. D.; Ait-Si-Ali, S.; Yokochi, T.; Wade, P.

A.; Jones, P. L.; Wolffe, A. P.: DNMT1 forms a complex with Rb, E2F1 and HDAC1 and represses transcription from E2F-responsive promoters. *Nature Genet.* 25: 338–342, 2000.

[31806] 5358. Rouleau, J.; Tanigawa, G.; Szyf, M.: The mouse DNA methyltransferase 5-prime region: a unique housekeeping gene promoter. *J. Biol. Chem.* 267: 7368–7377, 1992.

[31807] 5359. Rountree, M. R.; Bachman, K. E.; Baylin, S. B.: DNMT1 binds HDAC2 and a new co-repressor, DMAP1, to form a complex at replication foci. *Nature Genet.* 25: 269–277, 2000.

[31808] 5360. Tucker, K. L.; Talbot, D.; Lee, M. A.; Leonhardt, H.; Jaenisch, R.: Complementation of methylation deficiency in embryonic stem cells by a DNA methyltransferase mini-gene. *Proc. Nat. Acad. Sci.* 93: 12920–12925, 1996.

[31809] 5361. Yen, R.-W. C.; Vertino, P. M.; Nelkin, B. D.; Yu, J. J.; El-Deiry, W.; Cumaraswamy, A.; Lennon, G. G.; Trask, B. J.; Celano, P.; Baylin, S. B.: Isolation and characterization of the cDNA encoding human DNA methyltransferase. *Nucleic Acids Res.* 20: 2287–2291, 1992.

[31810] 5362. Ye, Q.; Chung, L. W. K.; Li, S.; Zhau, H. E.: Identification of a novel FAS/ER- α fusion transcript expressed in human cancer cells. *Biochim. Biophys. Acta* 1493:

373–377, 2000.

- [31811] 5363.Zuppan, P.; Hall, J. M.; Lee, M. K.; Ponglikitmongkol, M.; King, M.-C.: Possible linkage of the estrogen receptor gene to breast cancer in a family with late-onset disease. *Am. J. Hum. Genet.* 48: 1065–1068, 1991.
- [31812] 5364.Zuppan, P. J.; Hall, J. M.; Ponglikitmongkol, M.; Spielman, R.; King, M. C.: Polymorphisms at the estrogen receptor (ESR) locus and linkage relationships on chromosome 6q. (Abstract) *Cytogenet. Cell Genet.* 51: 1116 only, 1989.
- [31813] 5365.Calabi, F.; Cilli, V.: CBFA2T1, a gene rearranged in human leukemia, is a member of a multigene family. *Genomics* 52: 332–341, 1998.
- [31814] 5366.Erickson, P.; Gao, J.; Chang, K.-S.; Look, T.; Whisenant, E.; Raimondo, S.; Lasher, R.; Trujillo, J.; Rowley, J. D.; Drabkin, H. A.: Identification of breakpoints in t(8;21) acute myelogenous leukemia and isolation of a fusion transcript, AML1/ETO, with similarity to *Drosophila* segmentation gene, runt. *Blood* 80: 1825–1831, 1992.
- [31815] 5367.Miyamoto, T.; Weissman, I. L.; Akashi, K.: AML1/ETO-expressing nonleukemic stem cells in acute myelogenous leukemia with 8;21 chromosomal translocation. *Proc. Nat. Acad. Sci.* 97: 7521–7526, 2000.

- [31816] 5368.Miyoshi, H.; Kozu, T.; Shimizu, K.; Enomoto, K.; Maseki, N.; Kaneko,Y.; Kamada, N.; Ohki, M.: The t(8;21) translocation in acute myeloidleukemia results in production of an AML1–MTG8 fusion transcript. EMBOJ. 12: 2715–2721, 1993.
- [31817] 5369.Miyoshi, H.; Shimizu, K.; Kozu, T.; Maseki, N.; Kaneko, Y.; Ohki,M.: t(8;21) breakpoints on chromosome 21 in acute myeloid leukemiaare clustered within a limited region of a single gene, AML1. Proc.Nat. Acad. Sci. 88: 10431–10434, 1991.
- [31818] 5370.Linggi, B.; Muller–Tidow, C.; van de Locht, L.; Hu, M.; Nip, J.;Serve, H.; Berdel, W. E.; van der Reijden, B.; Quelle, D. E.; Rowley,J. D.; Cleveland, J.; Jansen, J. H.; Pandolfi, P. P.; Hiebert, S.W.: The t(8;21) fusion protein, AML1–ETO, specifically represses the transcription of the p14(ARF) tumor suppressor in acute myeloidleukemia. Nature Med. 8: 743–750, 2002.
- [31819] 5371.Minucci, S.; Maccarana, M.; Cioce, M.; De Luca, P.; Gelmetti, V.;Segalla, S.; Di Croce, L.; Giavara, S.; Matteucci, C.; Gobbi, A.;Bianchini, A.; Colombo, E.; Schiavoni, I.; Badaracco, G.; Hu, X.;Lazar, M. A.; Landsberger, N.; Nervi, C.; Pelicci, P. G.: Oligomerizationof RAR and AML1 transcription factors as a novel mechanism of oncogenicacti–

vation. *Molec. Cell* 5: 811–820, 2000.

- [31820] 5372.Niwa–Kawakita, M.; Miyoshi, H.; Gotoh, O.; Matsushima, Y.; Nishimura,M.; Shisa, H.; Ohki, M.: Cloning and gene mapping of the mouse homologueof the CBFA2T1 gene associated with human acute myeloid leukemia. *Genomics* 29:755–759, 1995.
- [31821] 5373.Nucifora, G.; Rowley, J. D.: The AML1 and ETO genes in acute myeloidleukemia with a t(8;21). *Leukemia Lym–phoma* 14: 353–362, 1994.
- [31822] 5374.Schoch, C.; Kohlmann, A.; Schnittger, S.; Brors, B.; Dugas, M.;Mergenthaler, S.; Kern, W.; Hiddemann, S.; Eils, R.; Haferlach, T.: Acute myeloid leukemias with reciprocal rearrangements can be distinguishedby specific gene expression profiles. *Proc. Nat. Acad. Sci.* 99:10008–10013, 2002.
- [31823] 5375.Wolford, J. K.; Bogardus, C.; Prochazka, M.: Polymorphism inthe 3–prime untranslated region of MTG8 is associated with obesityin Pima Indian males. *Biochem. Bio–phys. Res. Commun.* 246: 624–626,1998.
- [31824] 5376.Wolford, J. K.; Prochazka, M.: Structure and expression of thehuman MTG8/ETO gene. *Gene* 212: 103–109, 1998.
- [31825] 5377.Yergeau, D. A.; Hetherington, C. J.; Wang, Q.; Zhang,

P.; Sharpe, A. H.; Binder, M.; Marin-Padilla, M.; Tenen, D. G.; Speck, N. A.; Zhang, D.-E.: Embryonic lethality and impairment of haematopoiesis in mice heterozygous for an AML1-ETO fusion gene. *Nature Genet.* 15:303-306, 1997.

[31826] 5378. Dorfman, J.; Lazaris-Karatzas, A.; Malo, D.; Sonenberg, N.; Gros, P.: Chromosomal assignment of one of the mammalian translation initiation factor eIF-4E genes. *Genomics* 9: 785-788, 1991.

[31827] 5379. Jones, R. M.; Branda, J.; Johnston, K. A.; Polymenis, M.; Gadd, M.; Rustgi, A.; Callanan, L.; Schmidt, E. V.: An essential E box in the promoter of the gene encoding the mRNA cap-binding protein (eukaryotic initiation factor 4E) is a target for activation by c-myc. *Molec. Cell. Biol.* 16: 4754-4764, 1996.

[31828] 5380. Jones, R. M.; MacDonald, M. E.; Branda, J.; Altherr, M. R.; Louis, D. N.; Schmidt, E. V.: Assignment of the human gene encoding eukaryotic initiation factor 4E (EIF4E) to the region q21-25 on chromosome 4. *Somat. Cell Molec. Genet.* 23: 221-223, 1997.

[31829] 5381. Pause, A.; Belsham, G. J.; Gingras, A.-C.; Donze, O.; Lin, T.-A.; Lawrence, J. C., Jr.; Sonenberg, N.: Insulin-dependent stimulation of protein synthesis by phosphorylation of a regulator of 5-prime-cap function. *Nature* 371:

762–767, 1994.

- [31830] 5382. Pelletier, J.; Brook, J. D.; Housman, D. E.: Assignment of two of the translation initiation factor-4E (EIF4EL1 and EIF4EL2) genes to human chromosomes 4 and 20. *Genomics* 10: 1079–1082, 1991.
- [31831] 5383. Pyronnet, S.; Imataka, H.; Gingras, A.-C.; Fukunaga, R.; Hunter, T.; Sonenberg, N.: Human eukaryotic translation initiation factor 4G (eIF4G) recruits Mnk1 to phosphorylate eIF4E. *EMBO J.* 18: 270–279, 1999.
- [31832] 5384. Rychlik, W.; Domier, L. L.; Gardner, P. R.; Hellmann, G. M.; Rhoads, R. E.: Amino acid sequence of the mRNA cap-binding protein from human tissues. *Proc. Nat. Acad. Sci.* 84: 945–949, 1987.
- [31833] 5385. Waskiewicz, A. J.; Flynn, A.; Proud, C. G.; Cooper, J. A.: Mitogen-activated protein kinases activate the serine/threonine kinases Mnk1 and Mnk2. *EMBO J.* 16: 1909–1920, 1997.
- [31834] 5386. Aman, P.; Panagopoulos, I.; Lassen, C.; Fioretos, T.; Mencinger, M.; Toresson, H.; Hoglund, M.; Forster, A.; Rabbitts, T. H.; Ron, D.; Mandahl, N.; Mitelman, F.: Expression patterns of the human sarcoma-associated genes FUS and EWS and the genomic structure of FUS. *Genomics* 37:1–8, 1996.

- [31835] 5387. Aurias, A.; Rimbaut, C.; Buffe, D.; Zucker, J.-M.; Mazabraud, A.: Translocation involving chromosome 22 in Ewing's sarcoma: a cytogenetic study of four fresh tumors. *Cancer Genet. Cytogenet.* 12: 21-25, 1984.
- [31836] 5388. Ben-David, Y.; Giddens, E. B.; Letwin, K.; Bernstein, A.: Erythroleukemia induction by Friend murine leukemia virus: insertional activation of a new member of the *ets* gene family, *Fli-1*, closely linked to *c-ets-1*. *Genes Dev.* 5: 908-918, 1991.
- [31837] 5389. Budarf, M.; Sellinger, B.; Griffin, C.; Emanuel, B. S.: Comparative mapping of the constitutional and tumor-associated 11;22 translocations. *Am. J. Hum. Genet.* 45: 128-139, 1989.
- [31838] 5390. Zhang, Y.; Saison, M.; Spaepen, M.; De Strooper, B.; Van Leuven, F.; David, G.; Van den Berghe, H.; Cassiman, J.-J.: Mapping of human fibronectin receptor beta subunit gene to chromosome 10. *Somat. Cell Molec. Genet.* 14: 99-104, 1988.
- [31839] 5391. Argraves, W. S.; Dickerson, K.; Burgess, W. H.; Ruoslahti, E.: Fibulin, a novel protein that interacts with the fibronectin receptor-beta subunit cytoplasmic domain. *Cell* 58: 623-629, 1989.
- [31840] 5392. Korenberg, J. R.; Chen, X.-N.; Tran, H.; Argraves, W.

S.: Localization of the human gene for fibulin-1 (FBLN1) to chromosome band 22q13.3. *Cytogenet. Cell Genet.* 68: 192-193, 1995.

- [31841] 5393. Mattei, M.-G.; Pan, T.-C.; Zhang, R.-Z.; Timpl, R.; Chu, M.-L.: The fibulin-1 gene (FBLN1) is located on human chromosome 22 and on mouse chromosome 15. *Genomics* 22: 437-438, 1994.
- [31842] 5394. Collod, G.; Chu, M.-L.; Sasaki, T.; Coulon, M.; Timpl, R.; Renkart, L.; Weissenbach, J.; Jondeau, G.; Bourdarias, J. P.; Junien, C.; Boileau, C.: Fibulin-2: genetic mapping and exclusion as a candidate gene in Marfan syndrome type 2. *Europ. J. Hum. Genet.* 4: 292-295, 1996.
- [31843] 5395. Kuivaniemi, H.; Marshall, A.; Ganguly, A.; Chu, M.-L.; Abbott, W. M.; Tromp, G.: Fibulin-2 exhibits high degree of variability, but no structural changes concordant with abdominal aortic aneurysms. *Europ. J. Hum. Genet.* 6: 642-646, 1998.
- [31844] 5396. Pavlakis, G. N.; Hizuka, N.; Gorden, P.; Seeburg, P. H.; Hamber, D. H.: Expression of two human growth hormone genes in monkey cells infected by simian virus 40 recombinants. *Proc. Nat. Acad. Sci.* 78: 7398-7402, 1981.
- [31845] 5397. Leone, A.; McBride, O. W.; Weston, A.; Wang, M. G.; Anglard, P.; Cropp, C. S.; Goepel, J. R.; Lidereau, R.; Calla-

han, R.; Marston Linehan, W.; Rees, R. C.; Harris, C. C.; Liotta, L. A.; Steeg, P. S.: Somatic allelic deletion of nm23 in human cancer. *Cancer Res.* 51: 2490–2493, 1991.

[31846] 5398. Broide, D. H.; Hoffman, H.; Sriramaraio, P.: Genes that regulate eosinophilic inflammation. *Am. J. Hum. Genet.* 65: 302–307, 1999.

[31847] 5399. Campbell, H. D.; Tucker, W. Q. J.; Hort, Y.; Martinson, M. E.; Mayo, G.; Clutterbuck, E. J.; Sanderson, C. J.; Young, I. G.: Molecular cloning, nucleotide sequence, and expression of the gene encoding human eosinophil differentiation factor (interleukin–5). *Proc. Nat. Acad. Sci.* 84: 6629–6633, 1987.

[31848] 5400. Chandrasekharappa, S. C.; Rebelsky, M. S.; Firak, T. A.; Le Beau, M. M.; Westbrook, C. A.: A long-range restriction map of the interleukin–4 and interleukin–5 linkage group on chromosome 5. *Genomics* 6: 94–99, 1990.

[31849] 5401. Coffman, R. L.; Seymour, B. W. P.; Hudak, S.; Jackson, J.; Rennick, D.: Antibody to interleukin–5 inhibits helminth–induced eosinophilia in mice. *Science* 245: 308–310, 1989.

[31850] 5402. Cogan, E.; Schandene, L.; Crusiaux, A.; Cochaux, P.; Velu, T.; Goldman, M.: Clonal proliferation of type 2 helper T cells in a man with the hypereosinophilic syndrome. *New*

Eng. J. Med. 330: 535–538,1994.

[31851] 5403.Kozak, C.: Personal Communication. Bethesda, Md. 6/9/1988.

[31852] 5404.Pereira, E.; Goldblatt, J.; Rye, P.; Sanderson, C.; Le Souef, P.: Mutation analysis of interleukin–5 in an asthmatic cohort. Hum.Mutat. 11: 51–54, 1998.

[31853] 5405.Rodrigues, V., Jr.; Abel, L.; Piper, K.; Dessein, A. J.: Segregationanalysis indicates a major gene in the control of interleukin–5 productionin humans infected with Schistosoma mansoni. Am. J. Hum. Genet. 59:453–461, 1996.

[31854] 5406.Simon, H. U.; Plotz, S. G.; Dummer, R.; Blaser, K.: Abnormalclones of T cells producing interleukin–5 in idiopathic eosinophilia. NewEng. J. Med 341: 1112–1120, 1999.

[31855] 5407.Sutherland, G. R.; Baker, E.; Callen, D. F.; Campbell, H. D.;Young, Y. G.; Sanderson, C. J.; Garson, O. M.; Lopez, A. F.; Vadas,M. A.: Interleukin–5 is at 5q31 and is deleted in the 5q– syndrome. Blood 71:1150–1152, 1988.

[31856] 5408.Tanabe, T.; Konishi, M.; Mizuta, T.; Noma, T.; Honjo, T.: Molecularcloning and structure of the human interleukin–5 gene. J. Biol. Chem. 262:16580–16584, 1987.

[31857] 5409.Yokota, T.; Coffman, R. L.; Hagiwara, H.; Rennick, D. M.; Takebe,Y.; Yokota, K.; Gemmell, L.; Shrader, B.; Yang,

G.; Meyerson, P.; Luh, J.; Hoy, P.; Pene, J.; Briere, F.; Spits, H.; Banchereau, J.; de Vries, J.; Lee, F. D.; Arai, N.; Arai, K.: Isolation and characterization of lymphokine cDNA clones encoding mouse and human IgA-enhancing factor and eosinophil colony-stimulating factor activities: relationship to interleukin 5. *Proc. Nat. Acad. Sci.* 84: 7388–7392, 1987.

[31858] 5410. Geijsen, N.; Uings, I. J.; Pals, C.; Armstrong, J.; McKinnon, M.; Raaijmakers, J. A. M.; Lammers, J.-W. J.; Koenderman, L.; Coffey, P. J.: Cytokine-specific transcriptional regulation through an IL-5R- α interacting protein. *Science* 293: 1136–1138, 2001.

[31859] 5411. Gough, N. M.; Rakar, S.: Localization of the IL-5 receptor gene to the distal half of murine chromosome 6 using recombinant inbred strains of mice. *Genomics* 12: 855–856, 1992.

[31860] 5412. Isobe, M.; Kumura, Y.; Murata, Y.; Takaki, S.; Tomimaga, A.; Takatsu, K.; Ogita, Z.: Localization of the gene encoding the α subunit of human interleukin-5 receptor (IL5RA) to chromosome region 3p24–3p26. *Genomics* 14: 755–758, 1992.

[31861] 5413. Jacob, C. O.; Myktyyn, K.; Varcony, T.; Drabkin, H. A.: Mapping of the interleukin 5 receptor gene to human

chromosome 3p25–p26 and to mouse chromosome 6 close to the Raf–1 locus with polymorphic tandem repeat sequences. *Mammalian Genome* 4: 435–439, 1993.

- [31862] 5414. Sun, Z.; Yergeau, D. A.; Tuypens, T.; Tavernier, J.; Paul, C. C.; Baumann, M. A.; Tenen, D. G.; Ackerman, S. J.: Identification and characterization of a functional promoter region in the human eosinophil IL–5 receptor alpha subunit gene. *J. Biol. Chem.* 270: 1462–1471, 1995.
- [31863] 5415. Tavernier, J.; Tuypens, T.; Plaetinck, G.; Verhee, A.; Fiers, W.; Devos, R.: Molecular basis of the membrane–anchored and two soluble isoforms of the human interleukin 5 receptor alpha subunit. *Proc. Nat. Acad. Sci.* 89: 7041–7045, 1992.
- [31864] 5416. Zhang, J.; Kuvelkar, R.; Cheewatrakoolpong, B.; Williams, S.; Egan, R. W.; Billah, M. M.: Evidence for multiple promoters of the human IL–5 receptor alpha subunit gene: a novel 6–base pair element determines cell–specific promoter function. *J. Immun.* 15: 5412–5421, 1997.
- [31865] 5417. Povey, S.; Morton, N. E.; Sherman, S. L.: Report of the committee on the genetic constitution of chromosomes 1 and 2. *Cytogenet. Cell Genet.* 40: 67–106, 1985.
- [31866] 5418. Bird, A.; Taggart, M.; Frommer, M.; Miller, O. J.; Macleod, D.: A fraction of the mouse genome that is de–

rived from islands of nonmethylated,CpG-rich DNA. Cell 40: 91–99, 1985.

- [31867] 5419.Pikkarainen, T.; Kallunki, T.; Tryggvason, K.: Human laminin B2chain: comparison of the complete amino acid sequence with the B1chain reveals variability in sequence homology between different structuraldomains. J. Biol. Chem. 263: 6751–6758, 1988.
- [31868] 5420.Sasaki, M.; Yamada, Y.: The laminin B2 chain has a multidomainstructure homologous to the B1 chain. J. Biol. Chem. 262: 17111–17117,1987.
- [31869] 5421.Smyth, N.; Vatansever, H. S.; Murray, P.; Meyer, M.; Frie, C.;Paulsson, M.; Edgar, D.: Absence of basement membranes after targetingthe LAMC1 gene results in embryonic lethality due to failure of endodermdifferentiation. J. Cell Biol. 144: 151–160, 1999.
- [31870] 5422.Benlian, P.; Foubert, L.; Gagne, E.; Bernard, L.; De Gennes, J.L.; Langlois, S.; Robinson, W.; Hayden, M.: Complete paternal isodisomyfor chromosome 8 unmasked by lipoprotein lipase deficiency. Am. J.Hum. Genet. 59: 431–436, 1996.
- [31871] 5423.Schoenmakers, E. P. P. M.; Wanschura, S.; Mols, R.; Bullerdiek,J.; Van den Berghe, H.; Van de Ven, W. J. M.: Recurrent rearrangementsin the high mobility group protein

gene, HMGI-C, in benign mesenchymaltumours. *Nature Genet.* 10: 436–444, 1995.

[31872] 5424.Turc-Carel, C.; Pietrzak, E.; Kakati, S.; Kinniburgh, A. J.; Sandberg, A. A.: The human int-1 gene is located at chromosome region 12q12–12q13 and is not rearranged in myxoid liposarcoma with t(12;16)(q13;p11). *Onco-geneRes.* 1: 397–405, 1987.

[31873] 5425.Beck, Y.; Oren, R.; Amit, B.; Levanon, A.; Gorecki, M.; Hartman, J. R.: Human Mn superoxide dismutase cDNA sequence. *Nucleic AcidsRes.* 15: 9076, 1987.

[31874] 5426.Church, S. L.; Grant, J. W.; Meese, E. U.; Trent, J. M.: Sublocalization of the gene encoding manganese superoxide dismutase (MnSOD/SOD2) to 6q25 by fluorescence in situ hybridization and somatic cell hybrid mapping. *Genomics* 14: 823–825, 1992.

[31875] 5427.Creagan, R.; Tischfield, J.; Ricciuti, F.; Ruddle, F. H.: Chromosome assignments of genes in man using mouse-human somatic cell hybrids: mitochondrial superoxide dismutase (indophenol oxidase-B, tetrameric) to chromosome 6. *Humangenetik* 20: 203–209, 1973.

[31876] 5428.Figueroa, F.; Vincek, V.; Kasahara, M.; Bell, G. I.; Klein, J.: Mapping of the Sod-2 locus into the t complex on mouse chromosome 17. *Immunogenetics* 28: 260–264,

1988.

- [31877] 5429. Heckl, K.: Isolation of cDNAs encoding human manganese superoxidedismutase. *Nucleic Acids Res.* 16: 6224, 1988.
- [31878] 5430. Li, Y.; Huang, T.-T.; Carlson, E. J.; Melov, S.; Ursell, P. C.; Olson, J. L.; Noble, L. J.; Yoshimura, M. P.; Berger, C.; Chan, P. H.; Wallace, D. C.; Epstein, C. J.: Dilated cardiomyopathy and neonatal lethality in mutant mice lacking manganese superoxide dismutase. *Nature Genet.* 11: 376–381, 1995.
- [31879] 5431. Melov, S.; Coskun, P.; Patel, M.; Tuinstra, R.; Cottrell, B.; Jun, A. S.; Zastawny, T. H.; Dizdaroglu, M.; Goodman, S. I.; Huang, T.-T.; Mizziorko, H.; Epstein, C. J.; Wallace, D. C.: Mitochondrial disease in superoxide dismutase 2 mutant mice. *Proc. Nat. Acad. Sci.* 96:846–851, 1999.
- [31880] 5432. Melov, S.; Schneider, J. A.; Day, B. J.; Hinerfeld, D.; Coskun, P.; Mirra, S. S.; Crapo, J. D.; Wallace, D. C.: A novel neurological phenotype in mice lacking mitochondrial manganese superoxide dismutase. *Nature Genet.* 18: 159–163, 1998.
- [31881] 5433. Michelson, A. M.; McCord, J. M.; Fridovich, I.: *Superoxide and Superoxide Dismutases*. New York: Academic Press, 1977.

- [31882] 5434.Hassett, C.; Robinson, K. B.; Beck, N. B.; Omiecinski, C. J.:The human microsomal epoxide hydrolase gene (EPHX1): complete nucleotidesequence and structural characterization. *Genomics* 23: 433–442,1994.
- [31883] 5435.Jackson, M. R.; Craft, J. A.; Burchell, B.: Nucleotide and deducedamino acid sequence of human liver microsomal epoxide hydrolase. *NucleicAcids Res.* 15: 7188 only, 1987.
- [31884] 5436.Kutt, H.; Brennan, R.; Dehejia, H.; Verebely, K.: Diphenylhydantoinintoxication: a complication of isoni-azid therapy. *Am. Rev. Resp.Dis.* 101: 377–383, 1970.
- [31885] 5437.Kutt, H.; Verebely, K.; McDowell, F.: Inhibition of diphenylhydantoinmetabolism in rat and in rat liver microsome by antitubercular drugs. *Neurology* 18:706–710, 1968.
- [31886] 5438.Kutt, H.; Wolk, M.; Scherman, R.; McDowell, F.: Insufficientparahydroxylation as a cause of diphenylhydantoin toxicity. *Neurology* 14:542–548, 1964.
- [31887] 5439.McGlynn, K. A.; Rosvold, E. A.; Lustbader, E. D.; Hu, Y.; Clapper,M. L.; Zhou, T.; Wild, C. P.; Xia, X.–L.; Baffoe–Bonnie, A.; Ofori–Adjei,D.; Chen, G.–C.; London, W. T.; Shen, F.–M.; Buetow, K. H.: Susceptibilityto hepatocellular carcinoma is associated with genetic variation inthe enzy–

matic detoxification of aflatoxin B1. Proc. Nat. Acad. Sci. 92:2384–2387, 1995.

[31888] 5440.Nadeau, J. H.: Personal Communication. Bar Harbor, Me. 6/22/1988.

[31889] 5441.Phelan, M. C.; Pellock, J. M.; Nance, W. E.: Discordant expression of fetal hydantoin syndrome in a pair of dizygotic twins with different fathers. (Abstract) Am. J. Hum. Genet. 33: 67A only, 1981.

[31890] 5442.Sabry, M. A.; Farag, T. I.: Hand anomalies in fetal-hydantoin syndrome: from nail/phalangeal hypoplasia to unilateral acheiria.(Letter) Am. J. Med. Genet. 62: 410–412, 1996.

[31891] 5443.Skoda, R. C.; Demierre, A.; McBride, O. W.; Gonzalez, F. J.; Meyer, U. A.: Human microsomal xenobiotic epoxide hydrolase: complementary DNA sequence, complementary DNA-directed expression in COS-1 cells, and chromosomal localization. J. Biol. Chem. 263: 1549–1554, 1988.

[31892] 5444.Spielberg, S. P.; Gordon, G. B.; Blake, D. A.; Goldstein, D. A.; Herlong, H. F.: Predisposition to phenytoin hepatotoxicity assessed in vitro. New Eng. J. Med. 305: 722–727, 1981.

[31893] 5445.Strickler, S. M.; Dansky, L. V.; Miller, M. A.; Seni, M. H.; Andermann, E.; Spielberg, S. P.: Genetic predisposition

to phenytoin-induced birth defects. Lancet II: 746–749, 1985.

- [31894] 5446. Vasko, M. R.; Bell, R. D.; Daly, D. D.: Inheritance of diphenylhydantoin hypometabolism: a pharmacokinetic study of one family. (Abstract) Clin. Pharm. Therap. 21: 120 only, 1977.
- [31895] 5447. Vasko, M. R.; Bell, R. D.; Daly, D. D.; Pippenger, C. E.: Inheritance of phenytoin hypometabolism: a kinetic study of one family. Clin. Pharm. Therap. 27: 96–103, 1980.
- [31896] 5448. Vermeij, P.; Ferrari, M. D.; Buruma, O. J. S.; Veenema, H.; de Wolff, F. A.: Inheritance of poor phenytoin parahydroxylation capacity in a Dutch family. Clin. Pharm. Therap. 44: 588–593, 1988.
- [31897] 5449. Vesell, E. S.: Pharmacogenetics: multiple interactions between genes and environment as determinants of drug response. (Editorial) Am. J. Med. 66: 183–187, 1979.
- [31898] 5450. Zusterzeel, P. L. M.; Peters, W. H. M.; Visser, W.; Hermsen, K. J. M.; Roelofs, H. M. J.; Steegers, E. A. P.: A polymorphism in the gene for microsomal epoxide hydrolase is associated with pre-eclampsia. J. Med. Genet. 38: 234–237, 2001.
- [31899] 5451. Grant, D. F.; Spearow, J. L.; Storms, D. H.; Edelhoff,

S.; Adler, D. A.; Disteché, C. M.; Taylor, B. A.; Hammock, B. D.: Chromosomal mapping and expression levels of a mouse soluble epoxide hydrolase gene. *Pharmacogenetics* 4: 64–72, 1994.

[31900] 5452. Larsson, C.; White, I.; Johansson, C.; Stark, A.; Meijer, J.: Localization of the human soluble epoxide hydrolase gene (EPHX2) to chromosomal region 8p21–p12. *Hum. Genet.* 95: 356–358, 1995.

[31901] 5453. Norris, K. K.; DeAngelo, T. M.; Vesell, E. S.: Genetic and environmental factors that regulate cytosolic epoxide hydrolase activity in normal human lymphocytes. *J. Clin. Invest.* 84: 1749–1756, 1989.

[31902] 5454. Vesell, E. S.: Genetic factors that regulate cytosolic epoxide hydrolase activity in normal human lymphocytes. *Ann. Genet.* 34: 167–172, 1991.

[31903] 5455. Fonatsch, C.; Latza, U.; Durkop, H.; Rieder, H.; Stein, H.: Assignment of the human CD30 (Ki-1) gene to 1p36. *Genomics* 14: 825–826, 1992.

[31904] 5456. Pauling, L.: The discovery of the superoxide radical. *Trends Biochem. Sci.* 4(11): 270–271, 1979.

[31905] 5457. Ma, T.; Yang, B.; Umenishi, F.; Verkman, A. S.: Closely spaced tandem arrangement of AQP2, AQP5, and AQP6 genes in a 27-kilobase segment at chromosome lo-

cus 12q13. Genomics 43: 387–389, 1997.

- [31906] 5458.Benn, P.; Chern, C. J.; Bruns, G.; Craig, I. W.; Croce, C. M.:Assignment of the genes for human beta-glucuronidase and mitochondrial malate dehydrogenase to the region pter–q22 of chromosome 7. Cytogenet.Cell Genet. 19: 273–280, 1977.
- [31907] 5459.Blake, N. M.: Malate dehydrogenase types in the Asian–Pacific area, and a description of new phenotypes. Hum. Genet. 43: 69–80, 1978.
- [31908] 5460.Davidson, R. G.; Cortner, J. A.: Mitochondrial malate dehydrogenase: a new genetic polymorphism in man. Science 157: 1569–1571, 1967.
- [31909] 5461.Shimizu, N.; Shimizu, Y.; Ruddle, F. H.: Assignment of the human mitochondrial NAD–linked malate dehydrogenase gene to the p22–qter region of chromosome 7. Cytogenet. Cell Genet. 22: 441–445, 1978.
- [31910] 5462.Van Heyningen, V.; Bobrow, M.; Bodmer, W. F.; Gardiner, S. E.; Povey, S.; Hopkinson, D. A.: Chromosome assignment of some human enzyme loci: mitochondrial malate dehydrogenase to 7, mannose phosphate isomerase and pyruvate kinase to 15 and probably, esterase D to 13. Ann.Hum. Genet. 38: 295–303, 1975.
- [31911] 5463.Larson, L. M.; Bruce, A. W.; Saumur, J. H.; Wasdahl,

W. A.: Further evidence by gene dosage for the regional assignment of erythrocyte acid phosphatase (ACP1) and malate dehydrogenase (MDH1) loci on chromosome 2p. Clin. Genet. 22: 220–225, 1982.

[31912] 5464. D'Andrea, R.; Harrison-Findik, D.; Butcher, C. M.; Finnie, J.; Blumbergs, P.; Bartley, P.; McCormack, M.; Jones, K.; Rowland, R.; Gonda, T. J.; Vadas, M. A.: Dysregulated hematopoiesis and a progressive neurological disorder induced by expression of an activated form of the human common beta chain in transgenic mice. J. Clin. Invest. 102:1951–1960, 1998.

[31913] 5465. Rosenblum, J. S.; Gilula, N. B.; Lerner, R. A.: On signal sequence polymorphisms and diseases of distribution. Proc. Nat. Acad. Sci. 93:4471–4473, 1996.

[31914] 5466. Yoshimitsu, K.; Nichi, Y.; Kobayashi, Y.; Yoshimura, O.; Ohama, K.; Oguma, N.; Usui, T.: Decreased superoxide dismutase-2 activity in a patient with ring chromosome 6. Am. J. Med. Genet. 28: 211–214, 1987.

[31915] 5467. Klugbauer, S.; Rabes, H. M.: The transcription coactivator HTIF1 and a related protein are fused to the RET receptor tyrosine kinase in childhood papillary thyroid carcinomas. Oncogene 18: 4388–4393, 1999.

[31916] 5468. Lairmore, T. C.; Dou, S.; Howe, J. R.; Chi, D.; Carl-

son, K.; Veile, R.; Mishra, S. K.; Wells, S. A., Jr.; Donis-Keller, H.: A 1.5-megabase yeast artificial chromosome contig from human chromosome 10q11.2 connecting three genetic loci (RET, D10S94, and D10S102) closely linked to the MEN2A locus. *Proc. Nat. Acad. Sci.* 90: 492–496, 1993.

- [31917] 5469. Lombardo, F.; Baudin, E.; Chiefari, E.; Arturi, F.; Bardet, S.; Caillou, B.; Conte, C.; Dallapiccola, B.; Giuffrida, D.; Bidart, J.-M.; Schlumberger, M.; Filetti, S.: Familial medullary thyroid carcinoma: clinical variability and low aggressiveness associated with RET mutation at codon 804. *J. Clin. Endocr. Metab.* 87: 1674–1680, 2002.
- [31918] 5470. Lore, F.; Di Cairano, G.; Talidis, F.: Unilateral renal agenesis in a family with medullary thyroid carcinoma. (Letter) *New Eng. J. Med.* 342: 1218–1219, 2000.
- [31919] 5471. Machens, A.; Gimm, O.; Hinze, R.; Hoppner, W.; Boehm, B. O.; Dralle, H.: Genotype–phenotype correlations in hereditary medullary thyroid carcinoma: oncological features and biochemical properties. *J. Clin. Endocr. Metab.* 86: 1104–1109, 2001.
- [31920] 5472. Mendelsohn, C.; et al; et al: Function of the retinoic acid receptors (RARs) during development (II). Multiple abnormalities at various stages of organogenesis in RAR

double mutants. *Development* 120:2749–2771, 1994.

- [31921] 5473. Menko, F. H.; van der Luijt, R. B.; de Valk, I. A. J.; Toorians, A. W. F. T.; Sepers, J. M.; van Diest, P. J.; Lips, C. J. M.: Atypical MEN type 2B associated with two germline RET mutations on the same allele not involving codon 918. *J. Clin. Endocr. Metab.* 87: 393–397, 2002.
- [31922] 5474. Mulligan, L. M.; Kwok, J. B. J.; Healey, C. S.; Elsdon, M. J.; Eng, C.; Gardner, E.; Love, D. R.; Mole, S. E.; Moore, J. K.; Papi, L.; Ponder, M. A.; Telenius, H.; Tunnacliffe, A.; Ponder, B. A. J.: Germ-line mutations of the RET proto-oncogene in multiple endocrine neoplasia type 2A. *Nature* 363: 458–460, 1993.
- [31923] 5475. Munnes, M.; Fanaei, S.; Schmitz, B.; Muiznieks, I.; Holschneider, A. M.; Doerfler, W.: Familial form of Hirschsprung disease: nucleotide sequence studies reveal point mutations in the RET proto-oncogene in two of six families but not in other candidate genes. *Am. J. Med. Genet.* 94: 19–27, 2000.
- [31924] 5476. Nakata, T.; Kitamura, Y.; Shimizu, K.; Tanaka, S.; Fujimori, M.; Yokoyama, S.; Ito, K.; Emi, M.: Fusion of a novel gene, ELKS, to RET due to translocation t(10;12)(q11;p13) in a papillary thyroid carcinoma. *Genes Chromosomes Cancer* 25: 97–103, 1999.

- [31925] 5477.Niccoli-Sire, P.; Murat, A.; Rohmer, V.; Franc, S.; Chabrier,G.; Baldet, L.; Maes, B.; Savagner, F.; Giraud, S.; Bezieau, S.; Kottler,M.-L.; Morange, S.; Conte-Devolx, B.: The French Calcitonin TumorsStudy Group (GETC).: Familial medullary thyroid carcinoma with noncysteineRET mutations: phenotype-genotype relationship in a large series ofpatients. J. Clin. Endocr. Metab. 86: 3746-3753, 2001.
- [31926] 5478.Pachnis, V.; Mankoo, B.; Costantini, F.: Expression of the c-retproto-oncogene during mouse embryogenesis. Development 119: 1005-1017,1993.
- [31927] 5479.Pasini, B.; Hofstra, R. M. W.; Yin, L.; Bocciardi, R.; Santamaria,G.; Grootsholten, P. M.; Ceccherini, I.; Patrone, G.; Priolo, M.;Buys, C. H. C. M.; Romeo, G.: The physical map of the human RET proto-oncogene. Oncogene 11:1737-1743, 1995.
- [31928] 5480.Pelet, A.; Geneste, O.; Edery, P.; Pasini, A.; Chappuis, S.; Attie,T.; Munnich, A.; Lenoir, G.; Lyonnet, S.; Billaud, M.: Various mechanismscause RET-mediated signaling defects in Hirschsprung's disease. J.Clin. Invest. 101: 1415-1423, 1998.
- [31929] 5481.Pigny, P.; Bauters, C.; Wemeau, J.-L.; Houcke, M. L.; Crepin,M.; Caron, P.; Giraud, S.; Calender, A.; Buisine, M.-P.; Kerckaert,J.-P.; Porchet, N.: A novel 9-base pair

duplication in RET exon 8 in familial medullary thyroid carcinoma. *J. Clin. Endocr. Metab.* 84:1700–1704, 1999.

[31930] 5482. Pierotti, M. A.; Santoro, M.; Jenkins, R. B.; Sozzi, G.; Bongarzone, I.; Grieco, M.; Monzini, N.; Miozzo, M.; Hermann, M. A.; Fusco, A.; Hay, I. D.; Della Porta, G.; Vecchio, G.: Characterization of an inversion on the long arm of chromosome 10 juxtaposing D10S170 and RET and creating the oncogenic sequence RET/PTC. *Proc. Nat. Acad. Sci.* 89: 1616–1620, 1992.

[31931] 5483. Rodrigues, G. A.; Park, M.: Dimerization mediated through a leucine zipper activates the oncogenic potential of the met receptor tyrosine kinase. *Molec. Cell. Biol.* 13: 6711–6722, 1993.

[31932] 5484. Romeo, G.; Ronchetto, P.; Luo, Y.; Barone, V.; Seri, M.; Ceccherini, I.; Pasini, B.; Bocciardi, R.; Lerone, M.; Kaariainen, H.; Martucciello, G.: Point mutations affecting the tyrosine kinase domain of the RET proto-oncogene in Hirschsprung's disease. *Nature* 367: 377–378, 1994.

[31933] 5485. Salvatore, D.; Barone, M. V.; Salvatore, G.; Melillo, R. M.; Chiappetta, G.; Mineo, A.; Fenzi, G.; Vecchio, G.; Fusco, A.; Santoro, M.: Tyrosines 1015 and 1062 are in vivo autophosphorylation sites in Ret and Ret-derived oncoproteins. *J. Clin. Endocr. Metab.* 85:

3898–3907, 2000.

- [31934] 5486. Robinson, M. F.; Cote, G. J.; Nunziata, V.; Brandi, M. L.; Ferrer, J. P.; Martins Bugalho, M. J. G.; Almeida Ruas, M. M.; Chik, C.; Colantuoni, V.; Gagel, R. F.: Mutation of a specific codon of the RET proto-oncogene in the multiple endocrine neoplasia type 2A/cutaneous lichen amyloidosis syndrome. (Abstract) Fifth International Workshop on Multiple Endocrine Neoplasia, Stockholm, Archipelago, 1994.
- [31935] 5487. Boyer, S. H.: Human organ alkaline phosphatases: discrimination by several means including starch gel electrophoresis of antienzyme-enzyme supernatant fluids. *Ann. N.Y. Acad. Sci.* 103: 938–950, 1963.
- [31936] 5488. Deutman, A. F.; van Blommestein, J. D. A.; Henkes, H. E.; Waardenburg, P. J.; Solleveld-van Driest, E.: Butterfly-shaped pigment dystrophy of the fovea. *Arch. Ophthalmol.* 83: 558–569, 1970.
- [31937] 5489. Okuda, T.; Cleveland, J. L.; Downing, J. R.: PCTAIRE-1 and PCTAIRE-3, two members of a novel cdc2/CDC28-related protein kinase gene family. *Oncogene* 7:2249–2258, 1992.
- [31938] 5490. Okuda, T.; Valentine, V. A.; Shapiro, D. N.; Downing, J. R.: Cloning of genomic loci and chromosomal localization

tion of the human PCTAIRE-1 and -3 protein kinase genes. Genomics 21: 217-221, 1994.

[31939] 5491. Donald, L. J.: The genetics of placental alkaline phosphatase: a possible 'null' allele. Ann. Hum. Genet. 38: 7-18, 1974.

[31940] 5492. Donald, L. J.; Robson, E. B.: Rare variants of placental alkaline phosphatase. Ann. Hum. Genet. 37: 303-313, 1974.

[31941] 5493. Edwards, J. H.; Wingham, J.: Data on linkage between the locus determining placental alkaline phosphatase and other markers. Ann. Hum. Genet. 30: 233-237, 1967.

[31942] 5494. Garattini, E.; Margolis, J.; Heimer, E.; Felix, A.; Udenfriend, S.: Human placental alkaline phosphatase in liver and intestine. Proc. Nat. Acad. Sci. 82: 6080-6084, 1985.

[31943] 5495. Gogolin, K. J.; Slaughter, C. A.; Harris, H.: Electrophoresis of enzyme-mono-clonal antibody complexes: studies of human placental alkaline phosphatase polymorphism. Proc. Nat. Acad. Sci. 78: 5061-5065, 1981.

[31944] 5496. Henthorn, P. S.; Knoll, B. J.; Raducha, M.; Rothblum, K. N.; Slaughter, C.; Weiss, M.; Lafferty, M. A.; Fischer, T.; Harris, H.: Products of two common alleles at the locus for human placental alkaline phosphatase differ by seven amino acids. Proc. Nat. Acad. Sci. 83: 5597-5601, 1986.

- [31945] 5497.Kam, W.; Clauser, E.; Kim, Y. S.; Kan, Y. W.; Rutter, W. J.:Cloning, sequencing, and chromosomal localization of human term placentalalkaline phosphatase cDNA. *Proc. Nat. Acad. Sci.* 82: 8715–8719,1985.
- [31946] 5498.Knoll, B. J.; Rothblum, K. N.; Longley, M.: Nucleotide sequenceof the human placental alkaline phosphatase gene: evolution of the5–prime flanking region by deletion/substitution. *J. Biol. Chem.* 263:12020–12027, 1988.
- [31947] 5499.Lucarelli, P.; Scacchi, R.; Corbo, R. M.; Benincasa, A.; Palmarino,R.: Human placental alkaline phosphatase electrophoretic alleles:quantitative studies. *Am. J. Hum. Genet.* 34: 331–336, 1982.
- [31948] 5500.Martin, D.; Spurr, N. K.; Trowsdale, J.: RFLP of the human placentalalkaline phosphatase gene (PLAP). *Nucleic Acids Res.* 15: 9104 only,1987.
- [31949] 5501.Martin, D.; Tucker, D. F.; Gorman, P.; Sheer, D.; Spurr, N. K.;Trowsdale, J.: The human placental alkaline phosphatase gene andrelated sequences map to chromosome 2 band q37. *Ann. Hum. Genet.* 51:145–152, 1987.
- [31950] 5502.Millan, J. L.; Beckman, G.; Jeppsson, A.; Stigbrand, T.: Geneticvariants of placental alkaline phosphatase as detected by a monoclonalantibody. *Hum. Genet.* 60: 145–149, 1982.

- [31951] 5503.Millan, J. L.; Stigbrand, T.: Antigenic determinants of humanplacental and testicular placental-like alkaline phosphatases as mappedby monoclonal antibodies. *Europ. J. Biochem.* 136: 1-7, 1983.
- [31952] 5504.Palmarino, R.; Corbo, R. M.; Lucarelli, P.: Human placental alkalinephosphatase: analysis of genetically determined rare variants. *Hum.Biol.* 51: 341-352, 1979.
- [31953] 5505.Raimondi, E.; Talarico, D.; Moro, L.; Rutter, W. J.; Della Valle,G.; De Carli, L.: Regional mapping of the human placental alkalinephosphatase gene (ALPP) to 2q37 by in situ hybridization. *Cytogenet.Cell Genet.* 47: 98-99, 1988.
- [31954] 5506.Robinson, J. C.; Goldsmith, L. A.: Genetically determined variantsof serum alkaline phosphatase: a review. *Vox Sang.* 13: 289-307,1967.
- [31955] 5507.Robson, E. B.; Harris, H.: Genetics of the alkaline phosphatasepolymorphism of the human placenta. *Nature* 207: 1257-1259, 1965.
- [31956] 5508.Slaughter, C. A.; Gogolin, K. J.; Coseo, M. C.; Meyer, L. J.;Lesko, J.; Harris, H.: Discrimination of human placental alkalinephosphatase allelic variants by monoclonal antibodies. *Am. J. Hum.Genet.* 35: 1-20, 1983.
- [31957] 5509.Almind, K.; Delahaye, L.; Hansen, T.; Van Ob-

berghen, E.; Pedersen, O.; Kahn, C. R.: Characterization of the Met326Ile variant of phosphatidylinositol3-kinase p85-alpha. Proc. Nat. Acad. Sci. 99: 2124-2128, 2002.

[31958] 5510. Cannizzaro, L. A.; Skolnik, E. Y.; Margolis, B.; Croce, C. M.; Schlesinger, J.; Huebner, K.: The human gene encoding phosphatidylinositol3-kinase associated p85-alpha is at chromosome region 5q12-13. Cancer Res. 51: 3818-3820, 1991.

[31959] 5511. Vagnarelli, P.; Raimondi, E.; Mazzieri, R.; De Carli, L.; Mignatti, P.: Assignment of the human urokinase receptor gene (PLAUR) to 19q13. Cytogenet. Cell Genet. 60: 197-199, 1992.

[31960] 5512. Chase, P. B.; Yang, J.-M.; Thompson, F. H.; Halonen, M.; Regan, J. W.: Regional mapping of the human platelet-activating factor receptor gene (PTAFR) to 1p35-p34.3 by fluorescence in situ hybridization. Cytogenet. Cell Genet. 72: 205-207, 1996.

[31961] 5513. Prescott, S. M.; Zimmerman, G. A.; McIntyre, T. M.: Platelet-activating factor. J. Biol. Chem. 265: 17381-17384, 1990.

[31962] 5514. Seyfried, C. E.; Schweickart, V. L.; Godiska, R.; Gray, P. W.: The human platelet-activating factor receptor gene (PTAFR) contains no introns and maps to chromosome 1.

Genomics 13: 832–834, 1992.

- [31963] 5515.Abe, A.; Emi, N.; Tanimoto, M.; Terasaki, H.; Marunouchi, T.; Saito,H.: Fusion of the platelet-derived growth factor receptor beta to a novel gene CEV14 in acute myelogenous leukemia after clonal evolution. Blood 90:4271–4277, 1997.
- [31964] 5516.Buchberg, A. M.; Jenkins, N. A.; Copeland, N. G.: Localization of the murine macrophage colony-stimulating factor gene to chromosome 3 using interspecific backcross analysis. Genomics 5: 363–367, 1989.
- [31965] 5517.Claesson-Welsh, L.; Eriksson, A.; Moren, A.; Severinsson, L.; Ek,B.; Ostman, A.; Betsholtz, C.; Heldin, C.-H.: cDNA cloning and expression of a human platelet-derived growth factor (PDGF) receptor specific for B-chain-containing PDGF molecules. Molec. Cell. Biol. 8: 3476–3486,1988.
- [31966] 5518.Escobedo, J. A.; Fried, V. A.; Daniel, T. O.; Williams, L. T.:Primary structure of the platelet-derived growth factor. (Abstract) Clin.Res. 34: 544A, 1986.
- [31967] 5519.Gronwald, R. G. K.; Grant, F. J.; Haldeman, B. A.; Hart, C. E.;O'Hara, P. J.; Hagen, F. S.; Ross, R.; Bowen-Pope, D. F.; Murray,M. J.: Cloning and expression of a cDNA coding for the human platelet-derived growth factor

receptor: evidence for more than one receptor class.

Proc.Nat. Acad. Sci. 85: 3435–3439, 1988.

[31968] 5520.Klinghoffer, R. A.; Mueting–Nelsen, P. F.; Faerman, A.; Shani,M.; Soriano, P.: The two PDGF receptors maintain conserved signalingin vivo despite divergent embryological functions. Molec. Cell 7:343–354, 2001.

[31969] 5521.Kulkarni, S.; Heath, C.; Parker, S.; Chase, A.; Iqbal, S.; Pocock,C. F.; Kaeda, J.; Cwynarski, K.; Goldman, J. M.; Cross, N. C. P.:Fusion of H4/D10S170 to the platelet–derived growth factor receptorbeta in BCR–ABL–negative myeloproliferative disorders with a t(5;10)(q33;q21). CancerRes. 60: 3592–3598, 2000.

[31970] 5522.Leal, F.; Williams, L. T.; Robbins, K. C.; Aaronson, S. A.: Evidencethat the v–sis gene product transforms by interaction with the receptorfor platelet–derived growth factor. Science 230: 327–330, 1985.

[31971] 5523.Magnusson, M. K.; Meade, K. E.; Brown, K. E., Arthur, D. C.; Krueger,L. A.; Barrett, A. J.; Dunbar, C. E.: Rabaptin–5 is a novel fusionpartner to platelet–derived growth factor beta receptor in chronicmyelomonocytic leukemia. Blood 98: 2518–2525, 2001.

[31972] 5524.Matsui, T.; Heidaran, M.; Miki, T.; Popescu, N.; La Rochelle,W.; Kraus, M.; Pierce, J.; Aaronson, S.: Isolation of

a novel receptor cDNA establishes the existence of two PDGF receptor genes. *Science* 243:800–804, 1989.

- [31973] 5525. Ross, T. S.; Bernard, O. A.; Berger, R.; Gilliland, D. G.: Fusion of Huntingtin interacting protein 1 to platelet-derived growth factor-beta receptor (PDGF-beta-R) in chronic myelomonocytic leukemia with t(5;7)(q33;q11.2). *Blood* 91:4419–4426, 1998.
- [31974] 5526. Steer, E. J.; Cross, N. C. P.: Myeloproliferative disorders with translocations of chromosome 5q31–35: role of the platelet-derived growth factor receptor beta. *Acta Haemat.* 107: 113–122, 2002.
- [31975] 5527. Treacher Collins Syndrome Collaborative Group: Positional cloning of a gene involved in the pathogenesis of Treacher Collins syndrome. *Nature Genet.* 12: 130–136, 1996.
- [31976] 5528. Yarden, Y.; Escobedo, J. A.; Kuang, W.-J.; Yang-Feng, T. L.; Daniel, T. O.; Tremble, P. M.; Chen, E. Y.; Ando, M. E.; Harkins, R. N.; Francke, U.; Fried, V. A.; Ullrich, A.; Williams, L. T.: Structure of the receptor for platelet-derived growth factor helps define a family of closely related growth factor receptors. *Nature* 323: 226–232, 1986.
- [31977] 5529. Berthet, M.; Denjoy, I.; Donger, C.; Demay, L.; Hamoude, H.; Klug, D.; Schulze-Bahr, E.; Richard, P.; Funke,

H.; Schwartz, K.; Coumel, P.; Hainque, B.; Guicheney, P.: C-terminal HERG mutations: the role of hypokalemia and a KCNQ1-associated mutation in cardiac event occurrence. *Circulation* 99:1464–1470, 1999.

[31978] 5530. Allcock, R. J. N.; Williams, J. H.; Price, P.: The central MHC gene, BAT1, may encode a protein that down-regulates cytokine production. *Genes Cells* 6: 487–494, 2001.

[31979] 5531. Nunes, M.; Peelman, L.; Vaiman, M.; Bourgeaux, N.; Chardon, P.: Characterization of six new loci within the swine major histocompatibility complex class III region. *Mammalian Genome* 5: 616–622, 1994.

[31980] 5532. Peelman, L. J.; Chardon, P.; Nunes, M.; Renard, C.; Geffrotin, C.; Vaiman, M.; Van Zeveren, A.; Coppieters, W.; van de Weghe, A.; Bouquet, Y.; Choy, W. W.; Strominger, J. L.; Spies, T.: The BAT1 gene in the MHC encodes an evolutionarily conserved putative nuclear RNA helicase of the DEAD family. *Genomics* 26: 210–218, 1995.

[31981] 5533. Spies, T.; Blanck, G.; Bresnahan, M.; Sands, J.; Strominger, J. L.: A new cluster of genes within the human major histocompatibility complex. *Science* 243: 214–217, 1989.

[31982] 5534. Spies, T.; Bresnahan, M.; Strominger, J. L.: Human

major histocompatibility complex contains a minimum of 19 genes between the complement cluster and HLA-B.

Proc. Nat. Acad. Sci. 86: 8955–8958, 1989.

[31983] 5535. Colosimo, A.; Calabrese, G.; Gennarelli, M.; Ruzzo, A. M.; Sangiuolo, F.; Magnani, M.; Palka, G.; Novelli, G.; Dallapiccola, B.: Assignment of the hexokinase type 3 gene (HK3) to human chromosome band 5q35.3 by somatic cell hybrids and in situ hybridization. Cytogenet. Cell Genet. 74: 187–188, 1996.

[31984] 5536. Furuta, H.; Nishi, S.; Le Beau, M. M.; Fernald, A. A.; Yano, H.; Bell, G. I.: Sequence of human hexokinase III cDNA and assignment of the human hexokinase III gene (HK3) to chromosome band 5q35.2 by fluorescence in situ hybridization. Genomics 36: 206–209, 1996.

[31985] 5537. Harris, H.; Hopkinson, D. A.: Average heterozygosity per locus in man: an estimate based on the incidence of enzyme polymorphism. Ann. Hum. Genet. 36: 9–20, 1972.

[31986] 5538. Hopkinson, D. A.; Edwards, Y. H.; Harris, H.: The distribution of subunit numbers and subunit sizes of enzymes: a study of the products of 100 gene loci. Ann. Hum. Genet. 39: 383–411, 1976.

[31987] 5539. Povey, S.; Corney, G.; Harris, H.: Genetically determined polymorphism in a form of hexokinase, HK III,

found in human leukocytes. *Ann.Hum. Genet.* 38:
407–415, 1975.

- [31988] 5540. Buhr, A.; Bianchi, M. T.; Baur, R.; Courtet, P.; Pignay, V.; Boulenger, J. P.; Gallati, S.; Hinkle, D. J.; Macdonald, R. L.; Sigel, E.: Functional characterization of the new human GABA(A) receptor mutation beta-3(R192H). *Hum.Genet.* 111: 154–160, 2002.
- [31989] 5541. Buxbaum, J. D.; Silverman, J. M.; Smith, C. J.; Greenberg, D. A.; Kilifarski, M.; Reichert, J.; Cook, E. H., Jr.; Fang, Y.; Song, C.-Y.; Vitale, R.: Association between a GABRB3 polymorphism and autism. *Molec.Psychiat.* 7: 311–316, 2002.
- [31990] 5542. Holopainen, I. E.; Metsähonkala, E.-L.; Kokkonen, H.; Parkkola, R. K.; Manner, T. E.; Nagren, K.; Korpi, E. R.: Decreased binding of [11C]flumazenil in Angelman syndrome patients with GABA-A receptor beta-3 subunit deletions. *Ann. Neurol.* 49: 110–113, 2001.
- [31991] 5543. Homanics, G. E.; DeLorey, T. M.; Firestone, L. L.; Quinlan, J. J.; Handforth, A.; Harrison, N. L.; Krasowski, M. D.; Rick, C. E. M.; Korpi, E. R.; Makela, R.; Brilliant, M. H.; Hagiwara, N.; Ferguson, C.; Snyder, K.; Olsen, R. W.: Mice devoid of gamma-aminobutyrate type A receptor beta3 subunit have epilepsy, cleft palate, and hypersensitive be-

havior. Proc. Nat. Acad. Sci. 94: 4143–4148, 1997.

- [31992] 5544. Knoll, J. H. M.; Cheng, S.-D.; Lalande, M.: Allele specificity of DNA replication timing in the Angelman/Prader-Willi syndrome imprinted chromosomal region. Nature Genet. 6: 41–46, 1994.
- [31993] 5545. Laposky, A. D.; Homanics, G. E.; Baile, A.; Mendelson, W. B.: Deletion of the GABA(A) receptor beta 3 subunit eliminates the hypnotic actions of oleamide in mice. Neuroreport 12: 4143–4147, 2001.
- [31994] 5546. Meguro, M.; Mitsuya, K.; Sui, H.; Shigenami, K.; Kugoh, H.; Nakao, M.; Oshimura, M.: Evidence for uniparental, paternal expression of the human GABA-A receptor subunit genes, using microcell-mediated chromosome transfer. Hum. Molec. Genet. 6: 2127–2133, 1997.
- [31995] 5547. Saitoh, S.; Kubota, T.; Ohta, T.; Jinno, Y.; Niikawa, N.; Sugimoto, T.; Wagstaff, J.; Lalande, M.: Familial Angelman syndrome caused by imprinted submicroscopic deletion encompassing GABA(A) receptor beta(3)-subunit gene. (Letter) Lancet 339: 366–367, 1992.
- [31996] 5548. Scapoli, L.; Martinelli, M.; Pezzetti, F.; Carinci, F.; Bodo, M.; Tognon, M.; Carinci, P.: Linkage disequilibrium between GABRB3 gene and nonsyndromic familial cleft lip

with or without cleft palate. Hum.Genet. 110: 15–20, 2002.

[31997] 5549.Sinnott, D.; Wagstaff, J.; Glatt, K.; Woolf, E.; Kirkness, E.J.; Lalande, M.: High-resolution mapping of the gamma-aminobutyricacid receptor subunit beta-3 and alpha-5 gene cluster on chromosome15q11–q13, and localization of breakpoints in two Angelman syndrome patients. Am. J. Hum. Genet. 52: 1216–1229, 1993.

[31998] 5550.Tanabe, A.; Taketani, S.; Endo-Ichikawa, Y.; Tokunaga, R.; Ogawa, Y.; Hiramoto, M.: Analysis of the candidate genes responsible for non-syndromic cleft lip and palate in Japanese people. Clin. Sci. 99:105–111, 2000.

[31999] 5551.Wagstaff, J.; Chaillet, J. R.; Lalande, M.: The GABA(A) receptor beta-3 subunit gene: characterization of a human cDNA from chromosome15q11q13 and mapping to a region of conserved synteny on mouse chromosome7. Genomics 11: 1071–1078, 1991.

[32000] 5552.Wagstaff, J.; Knoll, J. H. M.; Fleming, J.; Kirkness, E. F.; Martin-Gallardo, A.; Greenberg, F.; Graham, J. M., Jr.; Menninger, J.; Ward, D.; Venter, J. C.; Lalande, M.: Localization of the gene encoding the GABA(A) receptor beta-3 subunit to the Angelman/Prader-Willi region of human chromosome 15. Am. J. Hum. Genet. 49: 330–337,

1991.

- [32001] 5553. Seeburg, P. H.: The human growth hormone gene family: nucleotide sequences show recent divergence and predict a new polypeptide hormone. *DNA* 1:239–249, 1982.
- [32002] 5554. Schneider, P. M.; Witzel-Schlomp, K.; Rittner, C.; Zhang, L.: The endogenous retroviral insertion in the human complement C4 gene modulates the expression of homologous genes by antisense inhibition. *Immunogenetics* 53:1–9, 2001.
- [32003] 5555. Suto, Y.; Tokunaga, K.; Watanabe, Y.; Hirai, M.: Visual demonstration of the organization of the human complement C4 and 21-hydroxylase genes by high-resolution fluorescence in situ hybridization. *Genomics* 33:321–324, 1996.
- [32004] 5556. Teisberg, P.; Akesson, I.; Olaisen, B.; Gedde-Dahl, T., Jr.; Thorsby, E.: Genetic polymorphism of C4 in man and localization of a structural C4 locus to the HLA gene complex of chromosome 6. *Nature* 264: 253–254, 1976.
- [32005] 5557. Boerkoel, C. F.; Takashima, H.; Stankiewicz, P.; Garcia, C. A.; Leber, S. M.; Rhee-Morris, L.; Lupski, J. R.: Periaxin mutations cause recessive Dejerine-Sottas neuropathy. *Am. J. Hum. Genet.* 68:325–333, 2001. Note: Erra-

tum: Am. J. Hum. Genet. 68: 557 only, 2001.

- [32006] 5558. Pischel, K. D.; Marlin, S. D.; Springer, T. A.; Woods, V. L., Jr.; Bluestein, H. G.: Polymorphism of lymphocyte function-associated antigen-1 demonstrated by a lupus patient's alloantiserum. J. Clin. Invest. 79: 1607–1614, 1987.
- [32007] 5559. Akey, J. M.; Wang, H.; Xiong, M.; Wu, H.; Liu, W.; Shriver, M. D.; Jin, L.: Interaction between the melanocortin-1 receptor and P genes contributes to inter-individual variation in skin pigmentation phenotypes in a Tibetan population. Hum. Genet. 108: 516–520, 2001.
- [32008] 5560. Chottiner, E. G.; Shewach, D. S.; Datta, N. S.; Ashcraft, E.; Gribbin, D.; Ginsburg, D.; Fox, I. H.; Mitchell, B. S.: Cloning and expression of human deoxycytidine kinase cDNA. Proc. Nat. Acad. Sci. 88: 1531–1535, 1991.
- [32009] 5561. Huang, S.-H.; Tomich, J. M.; Wu, H.; Jong, A.; Holcenberg, J.: Human deoxycytidine kinase: sequence of cDNA clones and analysis of expression in cell lines with and without enzyme activity. J. Biol. Chem. 264: 14762–14768, 1989. Note: Correction: J. Biol. Chem. 266: 5353 only, 1991.
- [32010] 5562. Johansson, M.; Brismar, S.; Karlsson, A.: Human deoxycytidine kinase is located in the cell nucleus. Proc. Nat.

Acad. Sci. 94:11941–11945, 1997.

- [32011] 5563. Song, J. J.; Walker, S.; Chen, E.; Johnson, E. E., II; Spychala, J.; Gribbin, T.; Mitchell, B. S.: Genomic structure and chromosomal localization of the human deoxycytidine kinase gene. *Proc. Nat. Acad. Sci.* 90: 431–434, 1993.
- [32012] 5564. Nurnberg, P.; Thiele, H.; Chandler, D.; Hohne, W.; Cunningham, M. L.; Ritter, H.; Leschik, G.; Uhlmann, K.; Mischung, C.; Harrop, K.; Goldblatt, J.; Borochowitz, Z. U.; Kotzot, D.; Westermann, F.; Mundlos, S.; Braun, H.-S.; Laing, N.; Tinschert, S.: Heterozygous mutations in ANKH, the human ortholog of the mouse progressive ankylosis-gene, result in craniometaphyseal dysplasia. *Nature Genet.* 28: 37–41, 2001.
- [32013] 5565. Nurnberg, P.; Tinschert, S.; Mrug, M.; Hampe, J.; Muller, C. R.; Fuhrmann, E.; Braun, H.-S.; Reis, A.: The gene for autosomal dominant craniometaphyseal dysplasia maps to chromosome 5q and is distinct from the growth hormone-receptor gene. *Am. J. Hum. Genet.* 61: 918–923, 1997.
- [32014] 5566. Soriano, P.; Montgomery, C.; Geske, R.; Bradley, A.: Targeted disruption of the c-src proto-oncogene leads to osteopetrosis in mice. *Cell* 64:693–702, 1991.
- [32015] 5567. Stegmann, A. P. A.; Honders, M. W.; Bolk, M. W. J.;

Wessels, J.; Willemze, R.; Landegent, J. E.: Assignment of the human deoxycytidine kinase (DCK) gene to chromosome 4 band q13.3–q21.1. *Genomics* 17:528–529, 1993.

[32016] 5568. Sanchez–Madrid, F.; Nagy, J.; Robbins, E.; Simon, P.; Springer, T. A.: A human leukocyte differentiation antigen family with distinct alpha subunits and a common beta subunit: the lymphocyte function–associated antigen (LFA–1), the C3b complement receptor (OKM1/Mac–1), and the p150,95 molecule. *J. Exp. Med.* 158: 1785–1803, 1983.

[32017] 5569. Saint–Ruf, C.; Panigada, M.; Azogui, O.; Debey, P.; von Boehmer, H.; Grassi, F.: Different initiation of pre–TCR and gamma–delta–TCR signalling. *Nature* 406: 524–527, 2000.

[32018] 5570. Delague, V.; Bareil, C.; Tuffery, S.; Bouvagnet, P.; Chouery, E.; Koussa, S.; Maisonobe, T.; Loiselet, J.; Megarbane, A.; Claustres, M.: Mapping of a new locus for autosomal recessive demyelinating Charcot–Marie–Tooth disease to 19q13.1–13.3 in a large consanguineous Lebanese family: exclusion of MAG as a candidate gene. *Am. J. Hum. Genet.* 67: 236–243, 2000.

[32019] 5571. Guilbot, A.; Williams, A.; Ravise, N.; Verny, C.; Brice, A.; Sherman, D. L.; Brophy, P. J.; LeGuern, E.; Delague, V.;

Bareil, C.; Megarbane, A.; Claustres, M.: A mutation in periaxin is responsible for CMT4F, an autosomal recessive form of Charcot-Marie-Tooth disease. *Hum. Molec. Genet.* 10: 415-421, 2001.

[32020] 5572. Sugino, N.; Suzuki, T.; Kashida, S.; Karube, A.; Takiguchi, S.; Kato, H.: Expression of Bcl-2 and Bax in the human corpus luteum during the menstrual cycle in early pregnancy: regulation by human chorionic gonadotropin. *J. Clin. Endocr. Metab.* 85: 4379-4386, 2000.

[32021] 5573. Vaskivuo, T. E.; Anttonen, M.; Herva, R.; Billig, H.; Dorland, M.; Te Velde, E. R.; Stenback, F.; Heikinheimo, M.; Tapanainen, J. S.: Survival of human ovarian follicles from fetal to adult life: apoptosis, apoptosis-related proteins, and transcription factor GATA-4. *J. Clin. Endocr. Metab.* 86: 3421-3429, 2001.

[32022] 5574. Cleary, M. L.; Mellentin, J. D.; Spies, J.; Smith, S. D.: Chromosomal translocation involving the beta T cell receptor gene in acute leukemia. *J. Exp. Med.* 167: 682-687, 1988.

[32023] 5575. Kuo, S. S.; Mellentin, J. D.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Cleary, M. L.: Structure, chromosome mapping, and expression of the mouse Lyl-1 gene. *Oncogene* 6: 961-968, 1991.

- [32024] 5576. Inoguchi, K.; Yoshioka, H.; Khaleduzzaman, M.; Ninomiya, Y.: ThemRNA for alpha-1(XIX) collagen chain, a new member of FACITs, contains a long unusual 3-prime untranslated region and displays many unique splicing variants. *J. Biochem.* 117: 137–146, 1995.
- [32025] 5577. Khaleduzzaman, M.; Sumiyoshi, H.; Ueki, Y.; Inoguchi, K.; Ninomiya, Y.; Yoshioka, H.: Structure of the human type XIX collagen (COL19A1) gene, which suggests it has arisen from an ancestor gene of the FACIT family. *Genomics* 45: 304–312, 1997.
- [32026] 5578. Myers, J. C.; Sun, M. J.; D'Ippolito, J. A.; Jabs, E. W.; Neilson, E. G.; Dion, A. S.: Human cDNA clones transcribed from an unusually high molecular weight RNA encode a new collagen chain. *Gene* 123: 211–217, 1993.
- [32027] 5579. Yoshioka, H.; Zhang, H.; Ramirez, F.; Mattei, M.-G.; Moradi-Ameli, M.; van der Rest, M.; Gordon, M. K.: Synteny between the loci for a novel FACIT-like collagen (D6S228E) and alpha 1(IX) collagen (COL9A1) on 6q12–q14 in humans. *Genomics* 13: 884–886, 1992.
- [32028] 5580. Gerecke, D. R.; Olson, P. F.; Koch, M.; Knoll, J. H. M.; Taylor, R.; Hudson, D. L.; Champlaud, M.-F.; Olsen, B. R.; Burgeson, R. E.: Complete primary structure of two splice variants of collagen XII, and assignment of alpha-1(XII)

collagen (COL12A1), alpha-1(IX) collagen(COL9A1), and alpha-1(XIX) collagen (COL19A1) to human chromosome6q12-q13. Genomics 41: 236-242, 1997.

- [32029] 5581.Copeland, N. G.; Jenkins, N. A.; Gilbert, D. J.; Eppig, J. T.;Maltais, L. J.; Miller, J. C.; Dietrich, W. F.; Weaver, A.; Lincoln,S. E.; Steen, R. G.; Stein, L. D.; Nadeau, J. H.; Lander, E. S.:A genetic linkage map of the mouse: current applications and futureprospects. Science 262: 57-66, 1993.
- [32030] 5582.Tong, Q.; Dalgin, G.; Xu, H.; Ting, C.-N.; Leiden, J. M.; Hotamisligil,G. S.: Function of GATA transcription factors in preadipocyte-adipocytetransition. Science 290: 134-138, 2000.
- [32031] 5583.Litt, M.; Buroker, N. E.; Kondoleon, S.; Douglass, J.; Liston,D.; Sheehy, R.; Magenis, R. E.: Chromosomal localization of the humanproenkephalin and prodynorphin genes. Am. J. Hum. Genet. 42: 327-334,1988.
- [32032] 5584.Litt, M.; Buroker, N. E.; Kondoleon, S. K.; Liston, D.; Douglass,J.; Sheehy, R.; Magenis, R. E.: Chromosomal localization of the humanproenkephalin and prodynorphin genes. (Abstract) Cytogenet. CellGenet. 46: 651 only, 1987.
- [32033] 5585.Berkovic, S. F.; McIntosh, A.; Howell, R. A.; Mitchell, A.; Sheffield,L. J.; Hopper, J. L.: Familial temporal lobe

epilepsy: a common disorder identified in twins. *Ann. Neurol.* 40: 227–235, 1996.

- [32034] 5586. Cendes, F.; Lopes-Cendes, I.; Andermann, E.; Andermann, F.: Familial temporal lobe epilepsy: a clinically heterogeneous syndrome. *Neurology* 50:554–557, 1998.
- [32035] 5587. Horikawa, S.; Takai, T.; Toyosato, M.; Takahashi, H.; Noda, M.; Kakidani, H.; Kubo, T.; Hirose, T.; Inayama, S.; Hayashida, H.; Miyata, T.; Numa, S.: Isolation and structural organization of the human preproenkephalin B gene. *Nature* 306: 611–614, 1983.
- [32036] 5588. Grigorenko, E. L.; Wood, F. B.; Meyer, M. S.; Hart, L. A.; Speed, W. C.; Shuster, A.; Pauls, D. L.: Susceptibility loci for distinct components of developmental dyslexia on chromosomes 6 and 15. *Am. J. Hum. Genet.* 60: 27–39, 1997.
- [32037] 5589. Giuli, G.; Scholl, U.; Bulle, F.; Guellaen, G.: Molecular cloning of the cDNAs coding for the two subunits of soluble guanylyl cyclase from human brain. *FEBS Lett.* 304: 83–88, 1992.
- [32038] 5590. Feder, J. N.; Li, L.; Jan, L. Y.; Jan, Y. N.: Genomic cloning and chromosomal localization of HRY, the human homolog of the *Drosophila* segmentation gene, hairy. *Genomics* 20: 56–61, 1994.

- [32039] 5591.Votruba, M.; Payne, A.; Moore, A. T.; Bhattacharya, S. S.: Dominant optic atrophy: exclusion and fine genetic mapping of the candidate gene, HRY. *Mammalian Genome* 9: 784–787, 1998.
- [32040] 5592.Yan, B.; Heus, J.; Lu, N.; Nichols, R. C.; Raben, N.; Plotz, P.H.: Transcriptional regulation of the human acid alpha-glucosidase gene: identification of a repressor element and its transcription factors Hes-1 and YY1. *J. Biol. Chem.* 276: 1789–1793, 2001.
- [32041] 5593.Yan, B.; Raben, N.; Plotz, P. H.: Hes-1, a known transcriptional repressor, acts as a transcriptional activator for the human acid alpha-glucosidase gene in human fibroblast cells. *Biochem. Biophys. Res. Commun.* 291: 582–587, 2002.
- [32042] 5594.Blanchette, F.; Day, R.; Dong, W.; Laprise, M.-H.; Dubois, C. M.: TGF-beta-1 regulates gene expression of its own converting enzyme furin. *J. Clin. Invest.* 99: 1974–1983, 1997.
- [32043] 5595.Copeland, N. G.; Gilbert, D. J.; Chretien, M.; Seidah, N. G.; Jenkins, N. A.: Regional localization of three convertases, PC1 (Nec-1), PC2 (Nec-2), and furin (Fur), on mouse chromosomes. *Genomics* 13: 1356–1358, 1992.
- [32044] 5596.Dubois, C. M.; Laprise, M.-H.; Blanchette, F.; Gentry,

L. E.; Leduc, R.: Processing of transforming growth factor beta-1 precursor by human furin convertase. *J. Biol. Chem.* 270: 10618-10624, 1995.

[32045] 5597. Hendy, G. N.; Bennett, H. P. J.; Gibbs, B. F.; Lazure, C.; Day, R.; Seidah, N. G.: Proparathyroid hormone is preferentially cleaved to parathyroid hormone by the prohormone convertase furin: a mass spectrometric study. *J. Biol. Chem.* 270: 9517-9525, 1995.

[32046] 5598. Mbikay, M.; Seidah, N. G.; Chretien, M.; Simpson, E. M.: Chromosomal assignment of the genes for proprotein convertases PC4, PC5, and PACE4 in mouse and human. *Genomics* 26: 123-129, 1995.

[32047] 5599. Roebroek, A. J. M.; Schalken, J. A.; Leunissen, J. A. M.; Onnekink, C.; Bloemers, H. P. J.; Van de Ven, W. J. M.: Evolutionary conserved close linkage of the c-fes/fps proto-oncogene and genetic sequences encoding a receptor-like protein. *EMBO J.* 5: 2197-2202, 1986.

[32048] 5600. Schalken, J. A.; Roebroek, A. J. M.; Oomen, P. P. C. A.; Wagenaar, S. S.; Debruyne, F. M. J.; Bloemers, H. P. J.; Van de Ven, W. J. M.: FUR gene expression as a discriminating marker for small cell and non-small cell lung carcinomas. *J. Clin. Invest.* 80: 1545-1549, 1987.

[32049] 5601. Seidah, N. G.; Mattei, M. G.; Gaspar, L.; Benjannet,

S.; Mbikay, M.; Chretien, M.: Chromosomal assignments of the genes for neuroendocrine convertase PC1 (NEC1) to human 5q15-21, neuroendocrine convertase PC2 (NEC2) to human 20p11.1-11.2, and furin (mouse 7[D1-E2] region). *Genomics* 11:103-107, 1991.

[32050] 5602. Curtis, E. J.; Fraser, F. C.; Warburton, D.: Congenital cleft lip and palate. *Am. J. Dis. Child.* 102: 853-857, 1961.

[32051] 5603. Awdeh, Z. L.; Alper, C. A.: Inherited structural polymorphism of the fourth component of human complement. *Proc. Nat. Acad. Sci.* 77:3576-3580, 1980.

[32052] 5604. Awdeh, Z. L.; Ochs, H. D.; Alper, C. A.: Genetic analysis of C4 deficiency. *J. Clin. Invest.* 67: 260-263, 1981.

[32053] 5605. Brade, V.; Hall, R. E.; Colten, H. R.: Biosynthesis of pro-C3, a precursor of the third component of complement. *J. Exp. Med.* 146:759-765, 1977.

[32054] 5606. Carroll, M. C.; Campbell, R. D.; Bentley, D. R.; Porter, R. R.: A molecular map of the human major histocompatibility complex class III region linking complement genes C4, C2 and factor B. *Nature* 307:237-241, 1984.

[32055] 5607. Carroll, M. C.; Porter, R. R.: Cloning of a human complement component C4 gene. *Proc. Nat. Acad. Sci.* 80: 264-267, 1983.

[32056] 5608. Chan, A. C.; Mitchell, K. R.; Munns, T. W.; Karp, D.

R.; Atkinson, J. P.: Identification and partial characterization of the secreted form of the fourth component of human complement: evidence that it is different from major plasma form. *Proc. Nat. Acad. Sci.* 80: 268–272, 1983.

[32057] 5609. Cream, J. J.; Olaisen, B.; Teisberg, P.; Soler, A. V.; Thompson, R. A.: Genetic basis of acquired C4 deficiency. *Clin. Genet.* 16:297–300, 1979.

[32058] 5610. Cunningham-Rundles, C.; Dupont, B.; Jersild, C.; Tegoli, C.; Whitsett, C.; Good, R. A.: Are HLA and Chido related antigenic groups? *Transplant. Proc.* 9: 33–38, 1977.

[32059] 5611. Cunningham-Rundles, C.; Tegoli, J.; Dupont, B.; Whitsett, C.; Good, R. A.: Chemical studies on the Chido antigen. *Transplant. Proc.* 9:647–652, 1977.

[32060] 5612. Curman, B.; Ostberg, L.; Sandberg, L.; Malmheden-Eriksson, I.; Stalenheim, G.; Rask, L.; Peterson, P. A.: H-2 linked Ss protein is C-4 component of complement. *Nature* 258: 243–245, 1975.

[32061] 5613. Ellman, L.; Green, I.; Frank, M.: Genetically controlled total deficiency of the fourth component of complement in the guinea pig. *Science* 170:74–75, 1970.

[32062] 5614. Fontaine, M.; Daveau, M.; Lebreton, J. P.: A common antigenic determinant on human C4b and C3b. *Molec. Immun.* 17: 1075–1078, 1980.

- [32063] 5615.Giles, C. M.: A new genetic variant for Chido. *Vox Sang.* 46:149–156, 1984.
- [32064] 5616.Hall, R. E.; Colten, H. R.: Genetic defect in biosynthesis of the precursor form of the fourth component of complement. *Science* 199:69–70, 1978.
- [32065] 5617.Hall, R. E.; Colten, H. R.: Cell-free synthesis of the fourth component of guinea pig complement (C4): identification of a precursor of serum C4 (pro-C4). *Proc. Nat. Acad. Sci.* 74: 1707–1710, 1977.
- [32066] 5618.Harris, J. P.; Tegoli, J.; Swanson, J.; Fisher, N.; Gavin, J.; Noades, J.: A nebulous antibody responsible for cross-matching difficulties (Chido). *Vox Sang.* 12: 140–142, 1967.
- [32067] 5619.Hobart, M. J.; Lachmann, P. J.: Allotypes of complement components in man. *Transplant. Rev.* 32: 26–42, 1976.
- [32068] 5620.Kramer, J.; Fulop, T.; Rajczy, K.; Ahn Tuan, N.; Fust, G.: A marked drop in the incidence of the null allele of the B gene of the fourth component of complement (C4B*Q0) in elderly subjects: C4B*Q0 as a probable negative selection factor for survival. *Hum. Genet.* 86:595–598, 1991.
- [32069] 5621.Kronke, M.; Geezy, A. F.; Hadding, U.; Bitter-Suermann, D.: Linkage of C4 and C4 deficiency to Bf and

GPLA. Immunogenetics 5: 461–466,1977.

[32070] 5622.Lachmann, P. J.; Grennan, D.; Martin, A.; Demant, P.: Identification of Ss protein as murine C4. Nature 258: 242–243, 1975.

[32071] 5623.Lhotta, K.; Schlogl, A.; Uring-Lambert, B.; Kronenberg, F.; Konig, P.: Complement C4 phenotypes in patients with end-stage renal disease. Nephron 72:442–446, 1996.

[32072] 5624.Mascart-Lemone, F.; Hauptmann, G.; Goetz, J.; Duchateau, J.; Delespesse, G.; Vray, B.; Dab, I.: Genetic deficiency of C4 presenting with recurrent infections and a SLE-like disease: genetic and immunologic studies. Am.J. Med. 75: 295–304, 1983.

[32073] 5625.Meo, T.; Krasteff, T.; Shreffler, D. C.: Immunochemical characterization of murine H-2 controlled Ss (serum substance) protein through identification of its human homologue as the fourth component of complement. Proc.Nat. Acad. Sci. 72: 4536–4540, 1975.

[32074] 5626.Middleton, J.; Crookston, M. C.: Chido-substance in plasma. VoxSang. 23: 256–261, 1972.

[32075] 5627.Middleton, J.; Crookston, M. C.; Falk, J. A.; Robson, E. B.; Cook, P. J. L.; Batchelor, J. R.; Bodmer, J.; Ferrara, G. B.; Festenstein, J.; Harris, H.; Kissmeyer-Nielsen, F.; Lawler,

S. D.; Sachs, J. A.; Wolf, E.: Linkage of Chido and HL-A.
Tissue Antigens 4: 366–373, 1974.

[32076] 5628.O'Neill, G. J.: The genetic control of Chido and
Rodgers bloodgroup substances. Seminars Hemat. 18:
32–38, 1981.

[32077] 5629.O'Neill, G. J.; Yang, S. Y.; Dupont, B.: Two HLA-
linked loci controlling the fourth component of human
complement. Proc. Nat.Acad. Sci. 75: 5165–5169, 1978.

[32078] 5630.O'Neill, G. J.; Yang, S. Y.; Dupont, B.: Chido and
Rodgers bloodgroups: relationships to C4 and HLA.
Transplant. Proc. 10: 749–751, 1978.

[32079] 5631.O'Neill, G. J.; Yang, S. Y.; Tegoli, J.; Berger, R.;
Dupont, B.: Chido and Rodgers blood groups are distinct
antigenic components of human C4. Nature 273: 668–670,
1978.

[32080] 5632.Ochs, H. D.; Rosenfeld, S. I.; Thomas, E. D.; Giblett,
E. R.; Alper, C. A.; Dupont, B.; Schaller, J. G.; Gilliland, B. C.;
Hansen, J. A.; Wedgwood, R. J.: Linkage between the gene
(or genes) controlling synthesis of the fourth component of
complement and the major histocompatibility complex.
New Eng. J. Med. 296: 470–475, 1977.

[32081] 5633.Olaisen, B.; Teisberg, P.; Nordhagen, R.; Michaelsen,
T.; Gedde-Dahl, T., Jr.: Human complement C4 locus is

duplicated on some chromosomes. *Nature* 279:736–737, 1979.

- [32082] 5634. Partanen, J.; Kere, J.; Wessberg, S.; Koskimies, S.: Determination of deletion sizes in the MHC-linked complement C4 and steroid 21-hydroxylase genes by pulsed-field gel electrophoresis. *Genomics* 5: 345–349, 1989.
- [32083] 5635. Partanen, J.; Koskimies, S.; Johansson, E.: C4 null phenotypes among lupus erythematosus patients are predominantly the result of deletions covering C4 and closely linked 21-hydroxylase A genes. *J. Med. Genet.* 25: 387–391, 1988.
- [32084] 5636. Petersen, G. B.; Sorensen, I. J.; Buskjaer, L.; Lamm, L. U.: Genetic studies of complement C4 in man. *Hum. Genet.* 53: 31–36, 1979.
- [32085] 5637. Kanai, Y.; Hediger, M. A.: Primary structure and functional characterization of a high-affinity glutamate transporter. *Nature* 360: 467–471, 1992.
- [32086] 5638. Lin, C. G.; Orlov, I.; Ruggiero, A. M.; Dykes-Hoberg, M.; Lee, A.; Jackson, M.; Rothstein, J. D.: Modulation of the neuronal glutamate transporter EAAC1 by the interacting protein GTRAP3-18. *Nature* 410: 84–88, 2001.
- [32087] 5639. Smith, C. P.; Weremowicz, S.; Kanai, Y.; Stelzner, M.; Morton, C. C.; Hediger, M. A.: Assignment of the gene

coding for the human high-affinity glutamate transporter EAAC1 to 9p24: potential role in dicarboxylic aminoaciduria and neurodegenerative disorders. *Genomics* 20:335–336, 1994.

- [32088] 5640. Hall, C. R.; Cole, W. G.; Haynes, R.; Hecht, J. T.: Reevaluation of a genetic model for the development of exostosis in hereditary multiple exostosis. *Am. J. Med. Genet.* 112: 1–5, 2002.
- [32089] 5641. Kirby, M. L.; Waldo, K. L.: Neural crest and cardiovascular patterning. *Circ. Res.* 77: 211–215, 1995.
- [32090] 5642. Kurihara, Y.; Kurihara, H.; Oda, H.; Maemura, K.; Nagai, R.; Ishikawa, T.; Yazaki, Y.: Aortic arch malformations and ventricular septal defect in mice deficient in endothelin-1. *J. Clin. Invest.* 96: 293–300, 1995.
- [32091] 5643. Lacey, S. W.; Sanders, J. M.; Rothberg, K. G.; Anderson, R. G. W.; Kamen, B. A.: Complementary DNA for the folate binding protein correctly predicts anchoring to the membrane by glycosyl-phosphatidylinositol. *J. Clin. Invest.* 84: 715–720, 1989.
- [32092] 5644. Istvan, E. S.; Deisenhofer, J.: Structural mechanism for statin inhibition of HMG-CoA reductase. *Science* 292: 1160–1164, 2001.
- [32093] 5645. Piedrahita, J. A.; Oetama, B.; Bennett, G. D.; van

Waes, J.; Kamen, B. A.; Richardson, J.; Lacey, S. W.; Anderson, R. G. W.; Finnell, R. H.: Mice lacking the folic acid-binding protein Folbp1 are defective in early embryonic development. *Nature Genet.* 23: 228–232, 1999.

[32094] 5646. Shaw, G. M.; Jensvold, N. G.; Wasserman, C. R.; Lammer, E. J.: Epidemiologic characteristics of phenotypically distinct neural tube defects among 0.7 million California births, 1983–1987. *Teratology* 49:143–149, 1994.

[32095] 5647. Werler, M. M.; Shapiro, S.; Mitchell, A. A.: Periconceptional folic acid exposure and risk of occurrent neural tube defects. *J.A.M.A.* 269:1257–1261, 1993.

[32096] 5648. Aittomaki, K.: The genetics of XX gonadal dysgenesis. *Am. J. Hum. Genet.* 54: 844–851, 1994.

[32097] 5649. Aittomaki, K.; Herva, R.; Stenman, U.–H.; Juntunen, K.; Ylostalo, P.; Hovatta, O.; de la Chapelle, A.: Clinical features of primary ovarian failure caused by a point mutation in the follicle-stimulating hormone receptor gene. *J. Clin. Endocr. Metab.* 81: 3722–3726, 1996.

[32098] 5650. Aittomaki, K.; Lucena, J. L. D.; Pakarinen, P.; Sistonen, P.; Tapanainen, J.; Gromoll, J.; Kaskikari, R.; Sankila, E.–M.; Lehvaslaiho, H.; Engel, A. R.; Nieschlag, E.; Huh-taniemi, I.; de la Chapelle, A.: Mutation in the follicle-stimulating hormone receptor gene causes hereditary hy-

pergonadotropic ovarian failure. Cell 82: 959–968, 1995.

[32099] 5651.Al-Hendy, A.; Moshynska, O.; Saxena, A.; Feyles, V.: Association between mutations of the follicle-stimulating-hormone receptor and repeated twinning. Lancet 356: 914 only, 2000.

[32100] 5652.Arey, B. J.; Stevis, P. E.; Deecher, D. C.; Shen, E. S.; Frail, D. E.; Negro-Vilar, A.; Lopez, F. J.: Induction of promiscuous Gprotein coupling of the follicle-stimulating hormone (FSH) receptor: a novel mechanism for transducing pleiotropic actions of FSH isoforms. Molec. Endocr. 11: 517–526, 1997.

[32101] 5653.Beau, I.; Touraine, P.; Meduri, G.; Gougeon, A.; Desroches, A.; Matuchansky, C.; Milgrom, E.; Kuttann, F.; Misrahi, M.: A novel phenotype related to partial loss of function mutations of the follicle stimulating hormone receptor. J. Clin. Invest. 102: 1352–1359, 1998.

[32102] 5654.Doherty, E.; Pakarinen, P.; Tiitinen, A.; Kiilavuori, A.; Huhtaniemi, I.; Forrest, S.; Aittomaki, K.: A novel mutation in the FSH receptor inhibiting signal transduction and causing primary ovarian failure. J. Clin. Endocr. Metab. 87: 1151–1155, 2002.

[32103] 5655.Gromoll, J.; Pekel, E.; Nieschlag, E.: The structure and organization of the human follicle-stimulating hor-

mone receptor (FSHR) gene. *Genomics* 35:308–311, 1996.

[32104] 5656.Gromoll, J.; Ried, T.; Holtgreve-Grez, H.; Nieschlag, E.; Gudermann, T.: Localization of the human FSH receptor to chromosome 2p21 using a genomic probe comprising exon 10. *J. Molec. Endocr.* 12: 265–271, 1994.

[32105] 5657.Gromoll, J.; Simoni, M.: Follicle-stimulating-hormone receptor and twinning. (Letter) *Lancet* 357: 230 only, 2001.

[32106] 5658.Heckert, L. L.; Daley, I. J.; Griswold, M. D.: Structural organization of the follicle-stimulating hormone receptor gene. *Molec. Endocr.* 6:70–80, 1992.

[32107] 5659.Kelton, C. A.; Cheng, S. V. Y.; Nugent, N. P.; Schweickhardt, R. L.; Rosenthal, J. L.; Overton, S. A.; Wands, G. D.; Kuzeja, J.B.; Luchette, C. A.; Chappel, S. C.: The cloning of the human folliclestimulating hormone receptor and its expression in COS-7, CHO, and Y-1 cells. *Molec. Cell. Endocr.* 89: 141–151, 1992.

[32108] 5660.Kotlar, T. J.; Young, R. H.; Albanese, C.; Crowley, W. F., Jr.; Scully, R. E.; Jameson, J. L.: A mutation in the follicle-stimulating hormone receptor occurs frequently in human ovarian sex cord tumors. *J.Clin. Endocr. Metab.* 82: 1020–1026, 1997.

[32109] 5661.Minegishi, T.; Nakamura, K.; Takakura, Y.; Ibuki, Y.;

Igarashi,M.: Cloning and sequencing of human FSH receptor cDNA. Biochem.Biophys. Res. Commun. 175: 1125–1130, 1991.

[32110] 5662.Jiang, M.; Aittomaki, K.; Nilsson, C.; Pakarinen, P.; Iltia, A.;Torresani, T.; Simonsen, H.; Goh, V.; Pettersson, K.; de la Chapelle,A.; Huhtaniemi, I.: The frequency of an inactivating point mutation(566C–T) of the human follicle–stimulating hormone receptor gene in four populations using allele–specific hybridization and time–resolvedfluorometry. J. Clin. Endocr. Metab. 83: 4338–4343, 1998.

[32111] 5663.Oktay, K.; Briggs, D.; Gosden, R. G.: Ontogeny of follicle–stimulatinghormone receptor gene expression in isolated human ovarian follicles. J.Clin. Endocr. Metab. 82: 3748–3751, 1997.

[32112] 5664.Rousseau–Merck, M. F.; Atger, M.; Loosfelt, H.; Milgrom, E.; Berger,R.: The chromosomal localization of the human follicle–stimulatinghormone receptor gene (FSHR) on 2p21–p16 is similar to that of the luteinizing hormone receptor gene. Genomics 15: 222–224, 1993.

[32113] 5665.Simoni, M.; Gromoll, J.; Hoppner, W.; Kamischke, A.; Krafft, T.;Stahle, D.; Nieschlag, E.: Mutational analysis of the follicle–stimulatinghormone (FSH) receptor in normal

and infertile men: identification and characterization of two discrete FSH receptor isoforms. *J. Clin. Endocr. Metab.* 84: 751–755, 1999.

[32114] 5666. Sipila, K.; Aula, P.: Database for the mutations of the Finnish disease heritage. *Hum. Mutat.* 19: 16–22, 2002.

[32115] 5667. Tapanainen, J. S.; Aittomaki, K.; Min, J.; Vaskivuo, T.; Huhtaniemi, I. T.: Men homozygous for an inactivating mutation of the follicle-stimulating hormone (FSH) receptor gene present variable suppression of spermatogenesis and fertility. *Nature Genet.* 15: 205–206, 1997.

[32116] 5668. Tortoriello, D. V.; Sidis, Y.; Holtzman, D. A.; Holmes, W. E.; Schneyer, A. L.: Human follistatin-related protein: a structural homologue of follistatin with nuclear localization. *Endocrinology* 142: 3426–3434, 2001.

[32117] 5669. Lee, H.-K.; Barbarosie, M.; Kameyama, K.; Bear, M. F.; Huganir, R. L.: Regulation of distinct AMPA receptor phosphorylation sites during bidirectional synaptic plasticity. *Nature* 405: 955–959, 2000.

[32118] 5670. Mack, V.; Burnashev, N.; Kaiser, K. M. M.; Rozov, A.; Jensen, V.; Hvalby, O.; Seeburg, P. H.; Sakmann, B.; Sprengel, R.: Conditional restoration of hippocampal synaptic potentiation in GluR-A-deficient mice. *Science* 292: 2501–2504, 2001.

- [32119] 5671. Puckett, C.; Gomez, C. M.; Korenberg, J. R.; Tung, H.; Meier, T. J.; Chen, X. N.; Hood, L.: Molecular cloning and chromosomal localization of one of the human glutamate receptor genes. *Proc. Nat. Acad. Sci.* 88:7557–7561, 1991.
- [32120] 5672. Shi, S.-H.; Hayashi, Y.; Petralla, R. S.; Zaman, S. H.; Wenthold, R. J.; Svoboda, K.; Malinow, R.: Rapid spine delivery and redistribution of AMPA receptors after synaptic NMDA receptor activation. *Science* 284:1811–1816, 1999.
- [32121] 5673. Zamanillo, D.; Sprengel, R.; Hvalby, O.; Jensen, V.; Burnashev, N.; Rozov, A.; Kaiser, K. M. M.; Koster, H. J.; Borchardt, T.; Worley, P.; Lubke, J.; Frotscher, M.; Kelly, P. H.; Sommer, B.; Andersen, P.; Seeburg, P. H.; Sakmann, B.: Importance of AMPA receptors for hippocampal synaptic plasticity but not for spatial learning. *Science* 284:1805–1811, 1999.
- [32122] 5674. Hardingham, G. E.; Fukunaga, Y.; Bading, H.: Extrasynaptic NMDARs oppose synaptic NMDARs by triggering CREB shut-off and cell death pathways. *Nature Neurosci.* 5: 405–414, 2002.
- [32123] 5675. Takano, H.; Onodera, O.; Tanaka, H.; Mori, H.; Sakimura, K.; Hori, T.; Kobayashi, H.; Mishina, M.; Tsuji, S.: Chromosomal localization of the epsilon-1, epsilon-3, and

zeta-1 subunit genes of the human NMDA receptor channel. *Biochem. Biophys. Res. Commun.* 197: 922-926, 1993.

- [32124] 5676. Meera Khan, P.; Doppert, B. A.; Hagemeijer, A.; Westerveld, A.: The human loci for phosphopyruvate hydratase and guanylate kinase are syntenic with the PGD-PGM1 linkage group in man-Chinese hamster somatic cell hybrids. *Cytogenet. Cell Genet.* 13: 130-131, 1974.
- [32125] 5677. Berkovitz, G. D.; Guerami, A.; Brown, T. R.; MacDonald, P. C.; Migeon, C. J.: Familial gynecomastia with increased extraglandular aromatization of plasma carbon(19)-steroids. *J. Clin. Invest.* 75: 1763-1769, 1985.
- [32126] 5678. Bloch, D. B.; Bloch, K. D.; Iannuzzi, M.; Collins, F. S.; Neer, E. J.; Seidman, J. G.; Morton, C. C.: The gene for the alpha-i-1 subunit of human guanine nucleotide binding protein maps near the cystic fibrosis locus. *Am. J. Hum. Genet.* 42: 884-888, 1988.
- [32127] 5679. Bray, P.; Carter, A.; Guo, V.; Puckett, C.; Kamholz, J.; Spiegel, A.; Nirenberg, M.: Human cDNA clones for an alpha subunit of G(i) signal-transduction protein. *Proc. Nat. Acad. Sci.* 84: 5115-5119, 1987.
- [32128] 5680. Itoh, H.; Toyama, R.; Kozasa, T.; Tsukamoto, T.; Matsuoka, M.; Kaziro, Y.: Presence of three distinct molec-

ular species of G(i)protein alpha subunit: structure of rat cDNAs and human genomic DNAs. J.Biol. Chem. 263: 6656–6664, 1988.

- [32129] 5681.Suki, W. N.; Abramowitz, J.; Mattera, R.; Codina, J.; Birnbaumer, L.: The human genome encodes at least three non-allelic G proteins with alpha-i-type subunits. FEBS Lett. 220: 187–192, 1987.
- [32130] 5682.Sullivan, K. A.; Liao, Y.-C.; Alborzi, A.; Beiderman, B.; Chang, F.-H.; Masters, S. B.; Levinson, A. D.; Bourne, H. R.: Inhibitory and stimulatory G proteins of adenylate cyclase: cDNA and amino acid sequences of the alpha chains. Proc. Nat. Acad. Sci. 83: 6687–6691, 1986.
- [32131] 5683.Jiang, M.; Gold, M. S.; Boulay, G.; Spicher, K.; Peyton, M.; Brabet, P.; Srinivasan, Y.; Rudolph, U.; Ellison, G.; Birnbaumer, L.: Multiple neurological abnormalities in mice deficient in the G protein G(o). Proc. Nat. Acad. Sci. 95: 3269–3274, 1998.
- [32132] 5684.Kroll, S. D.; Chen, J.; De Vivo, M.; Carty, D. J.; Buku, A.; Premont, R. T.; Iyengar, R.: The Q205L Go-alpha subunit expressed in NIH-3T3 cells induces transformation. J. Biol. Chem. 267: 23183–23188, 1992.
- [32133] 5685.Murtagh, J. J., Jr.; Eddy, R.; Shows, T. B.; Moss, J.; Vaughan, M.: Different forms of Go alpha mRNA arise by

alternative splicing of transcripts from a single gene on human chromosome 16. *Molec. Cell. Biol.* 11: 1146–1155, 1991.

[32134] 5686. Ram, P. T.; Horvath, C. M.; Iyengar, R.:

Stat3-mediated transformation of NIH-3T3 cells by the constitutively active Q205L G- α (O) protein. *Science* 287:142–144, 2000.

[32135] 5687. Strathmann, M.; Wilkie, T. M.; Simon, M. I.: Alternative splicing produces transcripts encoding two forms of the α subunit of GTP-binding protein G(o). *Proc. Nat. Acad. Sci.* 87: 6477–6481, 1990.

[32136] 5688. Tsukamoto, T.; Toyama, R.; Itoh, H.; Kozasa, T.; Matsuoka, M.; Kaziro, Y.: Structure of the human gene and two rat cDNAs encoding the α chain of GTP-binding regulatory protein G(o): two different mRNAs are generated by alternative splicing. *Proc. Nat. Acad. Sci.* 88:2974–2978, 1991.

[32137] 5689. Valenzuela, D.; Han, X.; Mende, U.; Fankhauser, C.; Mashimo, H.; Huang, P.; Pfeffer, J.; Neer, E. J.; Fishman, M. C.: G- α -o is necessary for muscarinic regulation of Ca(2+) channels in mouse heart. *Proc. Nat. Acad. Sci.* 94: 1727–2732, 1997.

[32138] 5690. Belluscio, L.; Gold, G. H.; Nemes, A.; Axel, R.: Mice

deficient in G(olf) are anosmic. *Neuron* 20: 69–81, 1998.

[32139] 5691. Buck, L. B.: Information coding in the mammalian olfactory system. *ColdSpring Harbor Symp. Quant. Biol.* 61: 147–155, 1996.

[32140] 5692. Jones, D. T.; Reed, R. R.: Golf: an olfactory neuron specific G protein involved in odorant signal transduction. *Science* 244:790–795, 1989.

[32141] 5693. Lindgren, V.; Luskey, K. L.; Russell, D. W.; Francke, U.: Human genes involved in cholesterol metabolism: chromosomal mapping of the loci for the low density lipoprotein receptor and 3-hydroxy-3-methylglutaryl-coenzyme A reductase with cDNA probes. *Proc. Nat. Acad. Sci.* 82: 8567–8571, 1985.

[32142] 5694. Luskey, K. L.: Conservation of promoter sequence but not complex intron splicing pattern in human and hamster genes for 3-hydroxy-3-methylglutarylcoenzyme A reductase. *Molec. Cell. Biol.* 7: 1881–1893, 1987.

[32143] 5695. Mohandas, T.; Heinzmann, C.; Sparkes, R. S.; Wasmuth, J.; Edwards, P.; Lusk, A. J.: Assignment of human 3-hydroxy-3-methylglutarylcoenzyme A reductase gene to q13–q23 region of chromosome 5. *Somat. Cell Molec. Genet.* 12: 89–94, 1986.

[32144] 5696. Osborne, T. F.; Goldstein, J. L.; Brown, M. S.:

5-prime end of HMG CoA reductase gene contains sequences responsible for cholesterol-mediated inhibition of transcription. *Cell* 42: 203–212, 1985.

- [32145] 5697. Van Doren, M.; Broihier, H. T.; Moore, L. A.; Lehmann, R.: HMG-CoA reductase guides migrating primordial germ cells. *Nature* 396: 466–469, 1998.
- [32146] 5698. Hou, J.; Parrish, J.; Ludecke, H.-J.; Sapru, M.; Wang, Y.; Chen, W.; Hill, A.; Siegel-Bartelt, J.; Northrup, H.; Elder, F. F. B.; Chinault, C.; Horsthemke, B.; Wagner, M. J.; Wells, D. E.: A 4-megabase YAC contig that spans the Langer-Giedion syndrome region on human chromosome 8q24.1: use in refining the location of the trichorhinophalangeal syndrome and multiple exostoses genes (TRPS1 and EXT1). *Genomics* 29:87–97, 1995.
- [32147] 5699. Ludecke, H.-J.; Wagner, M. J.; Nardmann, J.; La Pillo, B.; Parrish, J. E.; Willems, P. J.; Haan, E. A.; Frydman, M.; Hamers, G. J. H.; Wells, D. E.; Horsthemke, B.: Molecular dissection of a contiguous gene syndrome: localization of the genes involved in the Langer-Giedion syndrome. *Hum. Molec. Genet.* 4: 31–36, 1995.
- [32148] 5700. Koizumi, T.; Hendel, E.; Lalley, P. A.; Tchetgen, M.-B. N.; Nadeau, J. H.: Homologs of genes and anonymous loci on human chromosome 13 map to mouse chromosomes 8

and 14. Mammalian Genome 6: 263–268,1995.

- [32149] 5701.Devriendt, K.; Zhang, J.; van Leuven, F.; van den Berghe, H.; Cassiman,J. J.; Marynen, P.: A cluster of alpha 2–macroglobulin–related genes(alpha 2 M) on human chromosome 12p: cloning of the pregnancy–zoneprotein gene and an alpha 2M pseudogene. Gene 81: 325–334, 1989.
- [32150] 5702.Dow, D. J.; Lindsey, N.; Cairns, N. J.; Brayne, C.; Robinson, D.;Huppert, F. A.; Paykel, E. S.; Xuereb, J.; Wilcock, G.; Whittaker,J. L.; Rubinsztein, D. C.: Alpha–2 macroglobulin polymorphism andAlzheimer disease risk in the UK. (Letter) Nature Genet. 22: 16–17,1999.
- [32151] 5703.Fukushima, Y.; Bell, G. I.; Shows, T. B.: The polymorphic humanalpha–2–macroglobulin gene (A2M) is located in chromosome region 12p12.3–p13.3. Cytogenet.Cell Genet. 48: 58–59, 1988.
- [32152] 5704.Gallango, M. L.; Castillo, O.: Alpha–2–macroglobulin polymorphism:a new genetic system detected by immuno–electrophoresis. J. Immunogenet. 1:147–151, 1974.
- [32153] 5705.Hilliker, C.; Overbergh, L.; Petit, P.; Van Leuven, F.; Van denBerghe, H.: Assignment of mouse alpha–2–macroglobulin gene to chromosome6 band F1–G3.

Mammalian Genome 3: 469–471, 1992.

- [32154] 5706.Kan, C.-C.; Solomon, E.; Belt, K. T.; Chain, A. C.; Hiorns, L.R.; Fey, G.: Nucleotide sequence of cDNA encoding human alpha-2-macroglobulin and assignment of the chromosomal locus. *Proc. Nat. Acad. Sci.* 82:2282–2286, 1985.
- [32155] 5707.Leikola, J.; Fudenberg, H. H.; Kasukawa, R.; Milgrom, F.: A new genetic polymorphism of human serum: alpha(2) macroglobulin (AL-M). *Am.J. Hum. Genet.* 24: 134–144, 1972.
- [32156] 5708.Liao, A.; Nitsch, R. M.; Greenberg, S. M.; Finckh, U.; Blacker, D.; Albert, M.; Rebeck, G. W.; Gomez-Isla, T.; Clatworthy, A.; Binetti, G.; Hock, C.; Mueller-Thomsen, T.; Mann, U.; Zuchowski, K.; Beisiegel, U.; Staehelin, H.; Growdon, J. H.; Tanzi, R. E.; Hyman, B. T.: Genetic association of an alpha-2-macroglobulin (val1000ile) polymorphism and Alzheimer's disease. *Hum. Molec. Genet.* 7: 1953–1956, 1998.
- [32157] 5709.Marynen, P.; Bell, G. I.; Cavalli-Sforza, L. L.: Three RFLPs associated with the human alpha-2-macroglobulin gene (A2M). *Nucleic Acids Res.* 13: 8287 only, 1985.
- [32158] 5710.Marynen, P.; Zhang, J.; Devriendt, K.; Cassiman, J.-J.: Alpha-2-macroglobulin, pregnancy zone protein and an

alpha-2-macroglobulin pseudogene map to chromosome 12p12.2-p13. (Abstract) Cytogenet. Cell Genet. 51:1040 only, 1989.

[32159] 5711. Mattheijs, G.; Devriendt, K.; Cassiman, J.-J.; van den Berghe, H.; Marynen, P.: Structure of the human alpha-2 macroglobulin gene and its promoter (sic). Biochem. Biophys. Res. Commun. 184: 596-603, 1992.

[32160] 5712. Mattheijs, G.; Marynen, P.: A deletion polymorphism in the human alpha-2-macroglobulin (A2M) gene. Nucleic Acids Res. 19: 5102 only, 1991.

[32161] 5713. Mattheijs, G.; Marynen, P. A.: A deletion polymorphism in the human alpha-2-macroglobulin (A2M) gene. Nucleic Acids Res. 19: 5102 only, 1991.

[32162] 5714. Poller, W.; Barth, J.; Voss, B.: Detection of an alteration of the alpha-2-macroglobulin gene in a patient with chronic lung disease and serum alpha-2-macroglobulin deficiency. Hum. Genet. 83: 93-96, 1989.

[32163] 5715. Poller, W.; Faber, J.-P.; Klobeck, G.; Olek, K.: Cloning of the human alpha-2-macroglobulin gene and detection of mutations in two functional domains: the bait region and the thiolester site. Hum. Genet. 88: 313-319, 1992.

[32164] 5716. Rogaeva, E. A.; Premkumar, S.; Grubbs, J.; Serneels, L.; Scott, W. K.; Kawarai, T.; Song, Y.; Hill, D. M.; Abou-

Donia, S. M.; Martin, E. R.; Vance, J. J.; Yu, G.; and 18 others: An alpha-2-macroglobulin insertion-deletion polymorphism in Alzheimer disease. (Letter) *Nature Genet.* 22: 19–21, 1999.

[32165] 5717. Rudrasingham, V.; Wavrant-De Vrieze, F.; Lambert, J.-C.; Chakraverty, S.; Kehoe, P.; Crook, R.; Amouyel, P.; Wu, W.; Rice, F.; Perez-Tur, J.; Frigard, B.; Morris, J. C.; and 11 others: Alpha-2 macroglobulin gene and Alzheimer disease. (Letter) *Nature Genet.* 22: 17–19, 1999.

[32166] 5718. Sottrup-Jensen, L.; Stepanik, T. M.; Kristensen, T.; Lonblad, P. B.; Jones, C. M.; Wierzbicki, D. M.; Magnusson, S.; Domdey, H.; Wetsel, R. A.; Lundwall, A.; Tack, B. F.; Fey, G. H.: Common evolutionary origin of alpha-2-macroglobulin and complement components C3 and C4. *Proc. Nat. Acad. Sci.* 82: 9–13, 1985.

[32167] 5719. Umans, L.; Serneels, L.; Hilliker, C.; Stas, L.; Overbergh, L.; De Strooper, B.; Van Leuven, F.; Van den Berghe, H.: Molecular cloning of the mouse gene coding for alpha-2-macroglobulin and targeting of the gene in embryonic stem cells. *Genomics* 22: 519–529, 1994.

[32168] 5720. Buckler, A. J.; Chang, D. D.; Graw, S. L.; Brook, J. D.; Haber, D. A.; Sharp, P. A.; Housman, D. E.: Exon amplification: a strategy to isolate mammalian genes based on RNA

splicing. Proc. Nat. Acad.Sci. 88: 4005–4009, 1991.

- [32169] 5721.Brennan, R. W.; Dehejia, H.; Kutt, H.; Verebely, K.; McDowell,F.: Diphenylhydantoin intoxication attendant to slow inactivationof isoniazid. Neurology 20: 687–693, 1970.
- [32170] 5722.Brown, S.; Chalmers, D. E.: Microsomal epoxide hydrolase activityin human x mouse hybrid cells. Biochem. Biophys. Res. Commun. 137:775–780, 1986.
- [32171] 5723.Buehler, B. A.; Delimont, D.; van Waes, M.; Finnell, R. H.: Prenatalprediction of risk of the fetal hydantoin syndrome. New Eng. J. Med. 322:1567–1571, 1990.
- [32172] 5724.Chodirker, B. N.; Chudley, A. E.; Reed, M. H.; Persaud, T. V. N.: Possible prenatal hydantoin effect in a child born to a nonepilepticmother. Am. J. Med. Genet. 27: 373–378, 1987.
- [32173] 5725.De Smet, L.; Debeer, P.: Fetal hydantoin syndrome with unilateralatypical cleft hand: additional evidence for vascular disruption. Genet.Counsel. 13: 157–161, 2002.
- [32174] 5726.de Wolff, F. A.; Vermeij, P.; Ferrari, M. D.; Buruma, O. J. S.;Breimer, D. D.: Impairment of phenytoin parahydroxylation as a causeof severe intoxication. Ther. Drug Monit. 5: 213–215, 1983.
- [32175] 5727.Gennis, M. A.; Vemuri, R.; Burns, E. A.; Hill, J. V.;

Miller, M.A.; Spielberg, S. P.: Familial occurrence of hypersensitivity to phenytoin. *Am. J. Med.* 91: 631–634, 1991.

[32176] 5728. Goldman, A. S.; Van Dyke, D. C.; Gupta, C.; Katsumata, M.: Elevated glucocorticoid receptor levels in lymphocytes of children with the fetal hydantoin syndrome (FHS). *Am. J. Med. Genet.* 28: 607–618, 1987.

[32177] 5729. Hanson, J. W.; Myrianthopoulos, N. C.; Sedgwick, M. H. A.; Smith, D. W.: Risks to the offspring of women treated with hydantoin anticonvulsants, with emphasis on the fetal hydantoin syndrome. *J. Pediatr.* 89: 662–668, 1976.

[32178] 5730. Hartsfield, J. K., Jr.; Sutcliffe, M. J.; Everett, E. T.; Hassett, C.; Omiecinski, C. J.; Saari, J. A.: Assignment of microsomal epoxide hydrolase (EPHX1) to human chromosome 1q42.1 by in situ hybridization. *Cytogenet. Cell Genet.* 83: 44–45, 1998.

[32179] 5731. Dorner, M. H.; Salfeld, J.; Will, H.; Leibold, E. A.; Vass, J. K.; Munro, H. N.: Structure of human ferritin light subunit messenger RNA: comparison with heavy subunit message and functional implications. *Proc. Nat. Acad. Sci.* 82: 3139–3143, 1985.

[32180] 5732. Filie, J. D.; Buckler, C. E.; Kozak, C. A.: Genetic mapping of the mouse ferritin light chain gene and 11 pseudo-

genes on 11 mouse chromosomes. *Mammalian Genome* 9: 111–113, 1998.

- [32181] 5733. Girelli, D.; Bozzini, C.; Zecchina, G.; Tinazzi, E.; Bosio, S.; Piperno, A.; Ramenghi, U.; Peters, J.; Levi, S.; Camaschella, C.; Corrocher, R.: Clinical, biochemical and molecular findings in a series of families with hereditary hyperferritinaemia–cataract syndrome. *Brit. J. Haemat.* 115: 334–340, 2001.
- [32182] 5734. Girelli, D.; Corrocher, R.; Bisceglia, L.; Olivieri, O.; De Franceschi, L.; Zelante, L.; Gasparini, P.: Molecular basis for the recently described hereditary hyperferritinemia–cataract syndrome: A mutation in the iron–responsive element of ferritin L–subunit gene (the 'Verona mutation'). *Blood* 86: 4050–453, 1995.
- [32183] 5735. Girelli, D.; Corrocher, R.; Bisceglia, L.; Olivieri, O.; Zelante, L.; Panozzo, G.; Gasparini, P.: Hereditary hyperferritinemia–cataract syndrome caused by a 29–base pair deletion in the iron responsive element of ferritin L–subunit gene. *Blood* 90: 2084–2088, 1997.
- [32184] 5736. Girelli, D.; Olivieri, O.; De Franceschi, L.; Corrocher, R.; Bergamaschi, G.; Cazzola, M.: A linkage between hereditary hyperferritinaemia not related to iron overload and autosomal dominant congenital cataract. *Brit. J.*

Haemat. 90: 931–934, 1995.

- [32185] 5737. Lebo, R. V.; Kan, Y. W.; Cheung, M.-C.; Jain, S. K.; Drysdale, J.: Human ferritin light chain gene sequences mapped to several sorted chromosomes. Hum. Genet. 71: 325–328, 1985.
- [32186] 5738. Martin, M. E.; Fargion, S.; Brissot, P.; Pellat, B.; Beaumont, C.: A point mutation in the bulge of the iron-responsive element of the L ferritin gene in two families with the hereditary hyperferritinemia-cataracts syndrome. Blood 91: 319–323, 1998.
- [32187] 5739. McGill, J. R.; Boyd, D.; Barrett, K. J.; Drysdale, J. W.; Moore, C. M.: Localization of human ferritin H (heavy) and L (light) subunits by in situ hybridization. (Abstract) Am. J. Hum. Genet. 36: 146S, 1984.
- [32188] 5740. McLeod, J. L.; Craig, J.; Gumley, S.; Roberts, S.; Kirkland, M. A.: Mutation spectrum in Australian pedigrees with hereditary hyperferritinaemia-cataracts syndrome reveals novel and de novo mutations. Brit. J. Haemat. 118: 1179–1182, 2002.
- [32189] 5741. Mumford, A. D.; Vulliamy, T.; Lindsay, J.; Watson, A.: Hereditary hyperferritinemia-cataract syndrome: two novel mutations in the L-ferritin iron-responsive element. (Letter) Blood 91: 367–368, 1998.

- [32190] 5742. Munro, H. N.; Aziz, N.; Leibold, E. A.; Murray, M.; Rogers, J.; Vass, J. K.; White, K.: The ferritin genes: structure, expression, and regulation. *Ann. N.Y. Acad. Sci.* 526: 113–123, 1988.
- [32191] 5743. Santoro, C.; Marone, M.; Ferrone, M.; Costanzo, F.; Colombo, M.; Minganti, C.; Cortese, R.; Silengo, L.: Cloning of the gene coding for human L apoferritin. *Nucleic Acids Res.* 14: 2863–2876, 1986.
- [32192] 5744. Watanabe, N.; Drysdale, J. W.: Evidence for distinct mRNAs for ferritin subunits. *Biochem. Biophys. Res. Commun.* 98: 507–511, 1981.
- [32193] 5745. Usala, A.-L.; Madigan, T.; Burguera, B.; Sinha, M. K.; Caro, J. F.; Cunningham, P.; Powell, J. G.; Butler, P. C.: Treatment of insulin-resistant diabetic ketoacidosis with insulin-like growth factor I in an adolescent with insulin-dependent diabetes. *New Eng. J. Med.* 327: 853–857, 1992.
- [32194] 5746. Vaessen, N.; Janssen, J. A.; Heutink, P.; Hofman, A.; Lamberts, S. W. J.; Oostra, B. A.; Pols, H. A. P.; van Duijn, C. M.: Association between genetic variation in the gene for insulin-like growth factor-1 and low birthweight. *Lancet* 359: 1036–1037, 2002.
- [32195] 5747. Van Wyk, J. J.; Svoboda, M. E.; Underwood, L. E.: Evi-

dence from radioligand assays that somatomedin-C and insulin-like growth factor-I are similar to each other and different from other somatomedins. J.Clin. Endocr. Metab. 50: 206-208, 1980.

- [32196] 5748. Hochwald, G. M.; Thorbecke, G. J.: Abnormal metabolism or reduced transport of CSF gamma-trace microprotein in hereditary cerebral hemorrhage with amyloidosis. (Letter) New Eng. J. Med. 312: 1127-1128, 1985.
- [32197] 5749. Jensson, O.; Arnason, A.; Thorsteinsson, L.; Petursdottir, I.; Gudmundsson, G.; Blondal, H.; Grubb, A.; Lofberg, H.; Luyendijk, W.; Bots, G. T. A. M.; Frangione, B.: Cystatin C (gamma-trace) amyloidosis. In: Turk, V.: Cysteine Proteinases and their Inhibitors. New York: Walter de Gruyter and Co. (pub.) 1986.
- [32198] 5750. Jensson, O.; Gudmundsson, G.; Arnason, A.; Blondal, H.; Petursdottir, I.; Thorsteinsson, L.; Grubb, A.; Lofberg, H.; Cohen, D.; Frangione, B.: Hereditary cystatin C (gamma-trace) amyloid angiopathy of the CNS causing cerebral hemorrhage. Acta Neurol. Scand. 76: 102-114, 1987.
- [32199] 5751. Jensson, O.; Palsdottir, A.; Thorsteinsson, L.; Arnason, A.: The saga of cystatin C gene mutation causing amyloid angiopathy and brain hemorrhage--clinical genet-

ics in Iceland. Clin. Genet. 36:368–377, 1989.

- [32200] 5752.Lofberg, H.; Grubb, A. O.; Nilsson, E. K.; Jensson, O.; Gudmundsson,G.; Blondal, H.; Arnason, A.; Thorsteinsson, L.: Immunohistochemical characterization of the amyloid deposits and quantitation of pertinent cerebrospinal fluid proteins in hereditary cerebral hemorrhage with amyloidosis. Stroke 18: 431–440, 1987.
- [32201] 5753.Palsdottir, A.; Abrahamson, M.; Thorsteinsson, L.; Arnason, A.; Olafsson, I.; Grubb, A.; Jensson, O.: Mutation in cystatin C gene causes hereditary brain haemorrhage. Lancet II: 603–604, 1988.
- [32202] 5754.Zalin, A. M.; Jones, S.; Fitch, N. J. S.; Ramsden, D. B.: Familial nephropathic non-neuropathic amyloidosis: clinical features, immunohistochemistry and chemistry. Quart. J. Med. 81: 945–956, 1991.
- [32203] 5755.Ceccherini, I.; Romei, C.; Barone, V.; Pacini, F.; Martino, E.; Loviselli, A.; Pinchera, A.; Romeo, G.: Identification of the cys634-to-tyr mutation of the RET proto-oncogene in a pedigree with multiple endocrine neoplasia type 2A and localized cutaneous lichen amyloidosis. J.Endocr. Invest. 17: 201–204, 1994.
- [32204] 5756.Winkelmann, B. R.; Nauck, M.; Klein, B.; Russ, A. P.; Bohm, B.O.; Siekmeier, R.; Ihnken, K.; Verho, M.; Gross,

W.; Marz, W.: Deletion polymorphism of the angiotensin I-converting enzyme gene is associated with increased plasma angiotensin-converting enzyme activity but not with increased risk for myocardial infarction and coronary artery disease. *Ann. Intern. Med.* 125: 19–25, 1996.

[32205] 5757. Woods, D.; Onambele, G.; Woledge, R.; Skelton, D.; Bruce, S.; Humphries, S. E.; Montgomery, H.: Angiotensin-I converting enzyme genotype-dependent benefit from hormone replacement therapy in isometric muscle strength and bone mineral density. *J. Clin. Endocr. Metab.* 86: 2200–2204, 2001.

[32206] 5758. Yoshioka, T.; Xu, Y.; Yoshida, H.; Shiraga, H.; Muraki, T.; Ito, K.: Deletion polymorphism of the angiotensin converting enzyme gene predicts persistent proteinuria in Henoch-Schönlein purpura nephritis. *Arch. Dis. Child.* 79: 394–399, 1998.

[32207] 5759. Zhu, X.; Bouzekri, N.; Southam, L.; Cooper, R. S.; Adeyemo, A.; McKenzie, C. A.; Luke, A.; Chen, G.; Elston, R. C.; Ward, R.: Linkage and association analysis of angiotensin I-converting enzyme (ACE)-gene polymorphisms with ACE concentration and blood pressure. *Am. J. Hum. Genet.* 68: 1139–1148, 2001.

[32208] 5760. Zhu, X.; McKenzie, C. A.; Forrester, T.; Nickerson, D.

A.; Broeckel,U.; Schunkert, H.; Doering, A.; Jacob, H. J.; Cooper, R. S.; Rieder,M. J.: Localization of a small genomic region associated with elevatedACE. Am. J. Hum. Genet. 67: 1144–1153, 2000. Note: Erratum: Am.J. Hum. Genet. 67: 1365 only, 2000.

- [32209] 5761.Axton, R.; Hanson, I.; Danes, S.; Sellar, G.; van Heyningen, V.;Prosser, J.: The incidence of PAX6 mutation in patients with simpleaniridia: an evaluation of mutation detection in 12 cases. J. Med.Genet. 34: 279–286, 1997.
- [32210] 5762.Beauchamp, G. R.: Anterior segment dysgenesis keratolenticularadhesion and aniridia. J. Pediat. Ophthal. Strabismus 17: 55–58,1978.
- [32211] 5763.Crolla, J. A.; Cross, I.; Atkey, N.; Wright, M.; Oley, C. A.:FISH studies in a patient with sporadic aniridia and t(7;11)(q31.2;p13). J.Med. Genet. 33: 66–68, 1996.
- [32212] 5764.Fantes, J.; Redeker, B.; Breen, M.; Boyle, S.; Brown, J.; Fletcher,J.; Jones, S.; Bickmore, W.; Fukushima, Y.; Man-nens, M.; Danes, S.;van Heyningen, V.; Hanson, I.: Aniridia-associated cytogenetic rearrangementssuggest that a position effect may cause the mutant phenotype. Hum.Molec. Genet. 4: 415–422, 1995.
- [32213] 5765.Sarkar, F. H.; Gupta, S. L.: Receptors for human gamma interferon:binding and crosslinking of

125-I-labeled recombinant human gamma interferon to receptors on WISH cells. Proc. Nat. Acad. Sci. 81:5160–5164, 1984.

[32214] 5766.Slate, D. L.; Ruddle, F. H.: Antibodies to chromosome 21 coded cell surface components can block response to human interferon. Cytogenet. Cell Genet. 22: 265–269, 1978.

[32215] 5767.Slate, D. L.; Shulman, L.; Lawrence, J. B.; Revel, M.; Ruddle, F. H.: Presence of human chromosome 21 alone is sufficient for hybrid cell sensitivity to human interferon. J. Virol. 25: 319–325, 1978.

[32216] 5768.Takaoka, A.; Mitani, Y.; Suemori, H.; Sato, M.; Yokochi, T.; Noguchi, S.; Tanaka, N.; Taniguchi, T.: Cross talk between interferon-gamma and -alpha/beta signaling components in caveolar membrane domains. Science 288:2357–2360, 2000.

[32217] 5769.Tan, Y. H.: Chromosome 21 and the cell growth inhibitory effect of human interferon preparations. Nature 260: 141–143, 1976.

[32218] 5770.Tan, Y. H.; Schneider, E. L.; Tischfield, J.; Epstein, C. J.; Ruddle, F. H.: Human chromosome 21 dosage: effect on the expression of the interferon induced antiviral state. Science 186: 61–63, 1974.

- [32219] 5771.Tan, Y. H.; Tischfield, J.; Ruddle, F. H.: The linkage of genes for the human interferon-induced antiviral protein and indophenoloxidase-B traits to chromosome G-21. *J. Exp. Med.* 37: 317-330, 1973.
- [32220] 5772.Weil, J.; Tucker, G.; Epstein, L. B.; Epstein, C. J.: Interferon induction of (2-prime-5-prime) oligoisoadenylate synthetase in diploid and trisomy 21 human fibroblasts: relation to dosage of the interferon receptor gene (IFRC). *Hum. Genet.* 65: 108-111, 1983.
- [32221] 5773.Wiranowska-Stewart, M.; Stewart, W. E., II: The role of human chromosome 21 in sensitivity to interferons. *J. Gen. Virol.* 37:629-633, 1977.
- [32222] 5774.Aguet, M.; Dembic, Z.; Merlin, G.: Molecular cloning and expression of the human interferon-gamma receptor. *Cell* 55: 273-280, 1988.
- [32223] 5775.Alcaide-Loridan, C.; Le Coniat, M.; Bono, R.; Benech, P.; Couillin, P.; Van Cong, N.; Fisher, D. N.; Berger, R.; Fellous, M.: Mapping of the human interferon gamma response. (Abstract) *Cytogenet. Cell Genet.* 51: 949 only, 1989.
- [32224] 5776.Branca, A. A.; Baglioni, C.: Evidence that types I and II interferons have different receptors. *Nature* 294: 768-770, 1981.

- [32225] 5777.Casanova, J.-L.; Jouanguy, E.; Lamhamedi, S.; Blanche, S.; Fischer, A.: Immunological conditions of children with BCG disseminated infection.(Letter) Lancet 346: 581 only, 1995.
- [32226] 5778.Celada, A.; Allen, R.; Esparza, I.; Gray, P. W.; Schreiber, R.D.: Demonstration and partial characterization of the interferon-gammareceptor on human mononuclear phagocytes. J. Clin. Invest. 76: 2196-2205, 1985.
- [32227] 5779.Lee, W. M.; Galbraith, R. M.: The extracellular actin-scavengersystem and actin toxicity. New Eng. J. Med. 326: 1335-1341, 1992.
- [32228] 5780.Pilz, A.; Moseley, H.; Peters, J.; Abbott, C.: Comparative mapping of mouse chromosome 2 and human chromosome 9q: the genes for gelsolin and dopamine beta-hydroxylase map to mouse chromosome 2. Genomics 12:715-719, 1992.
- [32229] 5781.Brahe, C.; Servidei, S.; Zappata, S.; Ricci, E.; Tonali, P.; Neri, G.: Genetic homogeneity between childhood-onset and adult-onset autosomal recessive spinal muscular atrophy. Lancet 346: 741-742, 1995.
- [32230] 5782.Eglen, R. M.; Whiting, R. L.: Muscarinic receptor subtypes: a critique of the current classification and a pro-

posal for a working nomenclature. J. Auton. Pharm. 6: 323–346, 1986.

[32231] 5783. Goyal, R. K.: Muscarinic receptor subtypes: physiology and clinical implications. New Eng. J. Med. 321: 1022–1029, 1989.

[32232] 5784. Liao, C. F.; Themmen, A. P.; Joho, R.; Barberis, C.; Birnbaumer, M.; Birnbaumer, L.: Molecular cloning and expression of a fifth muscarinic acetylcholine receptor. J. Biol. Chem. 264: 7328–7337, 1989.

[32233] 5785. Peralta, E. G.; Ashkenazi, A.; Winslow, J. W.; Smith, D. H.; Ramachandran, J.; Capon, D. J.: Distinct primary structures, ligand-binding properties and tissue-specific expression of four human muscarinic acetylcholine receptors. EMBO J. 6: 3923–3929, 1987.

[32234] 5786. Burchill, S. A.; Wheeldon, J.; Cullinane, C.; Lewis, I. J.: EWS-FLI1 fusion transcripts identified in patients with typical neuroblastoma. Europ. J. Cancer 33: 239–243, 1997.

[32235] 5787. Pan, T.-C.; Sasaki, T.; Zhang, R.-Z.; Fassler, R.; Timpl, R.; Chu, M.-L.: Structure and expression of fibulin-2, a novel extracellular matrix protein with multiple EGF-like repeats and consensus motifs for calcium binding. J. Cell Biol. 123: 1269–1277, 1993.

[32236] 5788. Fling, S. P.; Arp, B.; Pious, D.: HLA-DMA and -DMB

genes are both required for MHC class II/peptide complex formation in antigen-presenting cells. *Nature* 368: 554–558, 1994.

[32237] 5789. Morris, P.; Shaman, J.; Attaya, M.; Amaya, M.; Goodman, S.; Bergman, C.; Monaco, J. J.; Mellins, E.: An essential role for HLA-DM in antigen presentation by class II major histocompatibility molecules. *Nature* 368:551–554, 1994.

[32238] 5790. Hiroi, S.; Harada, H.; Nishi, H.; Satoh, M.; Nagai, R.; Kimura, A.: Polymorphisms in the SOD2 and HLA-DRB1 genes are associated with nonfamilial idiopathic dilated cardiomyopathy in Japanese. *Biochem. Biophys. Res. Commun.* 261: 332–339, 1999.

[32239] 5791. Nishi, H.; Koga, Y.; Koyanagi, T.; Harada, H.; Imaizumi, T.; Toshima, H.; Sasazuki, T.; Kimura, A.: DNA typing of HLA class II genes in Japanese patients with dilated cardiomyopathy. *J. Molec. Cell. Cardiol.* 27:2385–2392, 1995.

[32240] 5792. Delattre, O.; Zucman, J.; Plougastel, B.; Desmaze, C.; Melot, T.; Peter, M.; Kovar, H.; Joubert, I.; de Jong, P.; Rouleau, G.; Aurias, A.; Thomas, G.: Gene fusion with an ETS DNA-binding domain caused by chromosome translocation in human tumours. *Nature* 359: 162–165, 1992.

[32241] 5793. Douglass, E. C.; Valentine, M.; Green, A. A.; Hayes, F.

A.; Thompson, E. I.: t(11;22) and other chromosomal rearrangements in Ewing's sarcoma. *J. Nat. Cancer Inst.* 77: 1211–1213, 1986.

[32242] 5794. Ewing, J.: Diffuse endothelioma of bone. *Proc. N. Y. Path. Soc.* 21:17–24, 1921.

[32243] 5795. Gerald, W. L.; Rosai, J.; Ladanyi, M.: Characterization of the genomic breakpoint and chimeric transcripts in the EWS–WT1 gene fusion of desmoplastic small round cell tumor. *Proc. Nat. Acad. Sci.* 92:1028–1032, 1995.

[32244] 5796. Geurts van Kessel, A.; Turc–Carel, C.; de Klein, A.; Grosveld, G.; Lenoir, G.; Bootsma, D.: Translocation of oncogene c–sis from chromosome 22 to chromosome 11 in a Ewing sarcoma–derived cell line. *Molec. Cell. Biol.* 5: 427–429, 1985.

[32245] 5797. Gill, S.; McManus, A. P.; Crew, A. J.; Benjamin, H.; Sheer, D.; Gusterson, B. A.; Pinkerton, C. R.; Patel, K.; Cooper, C. S.; Shipley, J. M.: Fusion of the EWS gene to a DNA segment from 9q22–31 in a human myxoid chondrosarcoma. *Genes Chromosomes Cancer* 12: 307–310, 1995.

[32246] 5798. Ginsberg, J. P.; de Alava, E.; Ladanyi, M.; Wexler, L. H.; Kovar, H.; Paulussen, M.; Zoubek, A.; Dockhorn–Dworniczak, B.; Juergens, H.; Wunder, J. S.; Andrulis, I. L.;

Malik, R.; Sorensen, P. H. B.; Womer, R. B.; Barr, F. G.: EWS-FLI1 and EWS-ERG gene fusions are associated with similar clinical phenotypes in Ewing's sarcoma. *J. Clin. Oncol.* 17: 1809-1814, 1999.

[32247] 5799. Griffin, C. A.; McKeon, C.; Israel, M. A.; Geggion, A.; Ghysdael, J.; Stehelin, D.; Douglass, E. C.; Green, A. A.; Emanuel, B. S.: Comparison of constitutional and tumor-associated 11;22 translocations: nonidentical breakpoints on chromosomes 11 and 22. *Proc. Nat. Acad. Sci.* 83: 6122-6126, 1986.

[32248] 5800. Jeon, I.-S.; Davis, J. N.; Braun, B. S.; Sublett, J. E.; Roussel, M. F.; Denny, C. T.; Shapiro, D. N.: A variant Ewing's sarcoma translocation(7;22) fuses the EWS gene to the ETS gene ETV1. *Oncogene* 10: 1229-1234, 1995.

[32249] 5801. Maletz, N.; McMorro, L. E.; Greco, M. A.; Wolman, S. R.: Ewing's sarcoma: pathology, tissue culture, and cytogenetics. *Cancer* 58:252-257, 1986.

[32250] 5802. Mastrangelo, T.; Modena, P.; Tornielli, S.; Bullrich, F.; Testi, M. A.; Mezzelani, A.; Radice, P.; Azzarelli, A.; Pilotti, S.; Croce, C. M.; Pierotti, M. A.; Sozzi, G.: A novel zinc finger gene is fused to EWS in small round cell tumor. *Oncogene* 19: 3799-3804, 2000.

[32251] 5803. May, W. A.; Gishizky, M. L.; Lessnick, S. L.; Lunsford,

L. B.; Lewis, B. C.; Delattre, O.; Zucman, J.; Thomas, G.; Denny, C. T.: Ewing sarcoma 11;22 translocation produces a chimeric transcription factor that requires the DNA-binding domain encoded by FLI1 for transformation. Proc. Nat. Acad. Sci. 90: 5752–5756, 1993.

[32252] 5804. Morohoshi, F.; Ootsuka, Y.; Arai, K.; Ichikawa, H.; Mitani, S.; Munakata, N.; Ohki, M.: Genomic structure of the human RBP56/hTAF(II)68 and FUS/TLS genes. Gene 221: 191–198, 1998.

[32253] 5805. Mugneret, F.; Aurias, A.; Lizard, S.; Turc-Carel, C.: Der(16)t(1;16)(q11;q11.1) is a consistent secondary chromosome change in Ewing's sarcoma. (Abstract) Cytogenet. Cell Genet. 46: 665 only, 1987.

[32254] 5806. Mugneret, F.; Lizard, S.; Aurias, A.; Turc-Carel, C.: Chromosomes in Ewing's sarcoma. II. Nonrandom additional changes, trisomy 8 and der(16)t(1;16). Cancer Genet. Cytogenet. 32: 239–245, 1988.

[32255] 5807. Ohno, T.; Ouchida, M.; Lee, L.; Gatalica, Z.; Rao, V. N.; Reddy, E. S. P.: The EWS gene, involved in Ewing family of tumors, malignant melanoma of soft parts and desmoplastic small round cell tumors, codes for an RNA binding protein with novel regulatory domains. Oncogene 9: 3087–3097, 1994.

- [32256] 5808. Peter, M.; Couturier, J.; Pacquement, H.; Michon, J.; Thomas, G.; Magdelenat, H.; Delattre, O.: A new member of the ETS family fused to EWS in Ewing tumors. *Oncogene* 14: 1159–1164, 1997.
- [32257] 5809. Plougastel, B.; Mattei, M.-G.; Thomas, G.; Delattre, O.: Cloning and chromosome localization of the mouse Ews gene. *Genomics* 23:278–281, 1994.
- [32258] 5810. Plougastel, B.; Zucman, J.; Peter, M.; Thomas, G.; Delattre, O.: Genomic structure of the EWS gene and its relationship to EWSR1, a site of tumor-associated chromosome translocation. *Genomics* 18:609–615, 1993.
- [32259] 5811. Selleri, L.; Hermanson, G. G.; Eubanks, J. H.; Lewis, K. A.; Evans, G. A.: Molecular localization of the t(11;22)(q24;q12) translocation of Ewing sarcoma by chromosomal in situ suppression hybridization. *Proc. Nat. Acad. Sci.* 88: 887–891, 1991.
- [32260] 5812. Tanaka, K.; Iwakuma, T.; Harimaya, K.; Sato, H.; Iwamoto, Y.: EWS–Fli1 antisense oligodeoxynucleotide inhibits proliferation of human Ewing's sarcoma and primitive neuroectodermal tumor cells. *J. Clin. Invest.* 99: 239–247, 1997.
- [32261] 5813. Tilly, H.; Bastard, C.; Chevallier, B.; Halkin, E.; Monconduit, M.: Chromosomal abnormalities in secondary Ew-

ing's sarcoma. (Letter) Lancet II:812 only, 1984.

- [32262] 5814. Triche, T. J.; Askin, F. B.: Neuroblastoma and the differential diagnosis of small-, round-, blue-cell tumors. Hum. Path. 14: 569–595, 1983.
- [32263] 5815. Turc-Carel, C.; Aurias, A.; Mugneret, F.; Lizard, S.; Sidaner, I.; Philip, I.; Philip, T.; Lenoir, G. M.; Mazabraud, A.: Outstanding consistency of the chromosomal break-points 11q24 and 22q12 in Ewing's sarcoma. (Abstract) Cytogenet. Cell Genet. 46: 706 only, 1987.
- [32264] 5816. Turc-Carel, C.; Aurias, A.; Mugneret, F.; Lizard, S.; Sidaner, I.; Volk, C.; Thiery, J. P.; Olschwang, S.; Philip, I.; Berger, M. P.; Philip, T.; Lenoir, G. M.; Mazabraud, A.: Chromosomes in Ewing's sarcoma. I. An evaluation of 85 cases and remarkable consistency of t(11;22)(q24;q12). Cancer Genet. Cytogenet. 32: 229–238, 1988.
- [32265] 5817. Turc-Carel, C.; Philip, I.; Berger, M. P.; Philip, T.; Lenoir, G. M.: Chromosomal translocations in Ewing's sarcoma. New Eng. J. Med. 309: 497–498, 1983.
- [32266] 5818. Turc-Carel, C.; Philip, I.; Berger, M. P.; Philip, T.; Lenoir, G. M.: Chromosome study of Ewing's sarcoma (ES) cell lines: consistency of a reciprocal translocation t(11;22)(q24;q12). Cancer Genet. Cytogenet. 12:1–19, 1984.

- [32267] 5819.Whang–Peng, J.; Freter, C. E.; Knutsen, T.; Nanfro, J. J.; Gazdar,A.: Translocation t(11;22) in esthesioneuroblas–toma. *Cancer Genet.Cytogenet.* 29: 155–157, 1987.
- [32268] 5820.Whang–Peng, J.; Triche, T. J.; Knutsen, T.; Miser, J.; Douglass,E. C.; Israel, M. A.: Chromosome translocation in peripheral neuroepithelioma. *NewEng. J. Med.* 311: 584–585, 1984.
- [32269] 5821.Zhang, R.–Z.; Pan, T.–C.; Zhang, Z.–Y.; Mattei, M.–G.; Timpl, R.;Chu, M.–L.: Fibulin–2 (FBLN2): human cDNA sequence, mRNA expression,and mapping of the gene on human and mouse chromosomes. *Genomics* 22:425–430, 1994.
- [32270] 5822.Baden, H. P.; Roth, S. I.; Goldsmith, L. A.; Lee, L. D.: Keratohyalinprotein in disorders of keratinization. *J. In–vest. Derm.* 62: 411–414,1974.
- [32271] 5823.Gan, S.–Q.; McBride, O. W.; Idler, W. W.; Markova, N.; Steinert,P. M.: Organization, structure, and polymor–phisms of the human profilaggringene. *Biochemistry* 29: 9432–9440, 1990.
- [32272] 5824.Holbrook, K. A.; Dale, B. A.; Brown, K. S.: Abnormal epidermalkeratinization in the repeated epilation mutant mouse. *J. Cell Biol.* 92:387–397, 1982.
- [32273] 5825.McKinley–Grant, L. J.; Idler, W. W.; Bernstein, I. A.;

Parry, D.A. D.; Cannizzaro, L.; Croce, C. M.; Huebner, K.; Lessin, S. R.; Steinert, P. M.: Characterization of a cDNA clone encoding human filaggrin and localization of the gene to chromosome region 1q21. *Proc. Nat. Acad. Sci.* 86: 4848–4852, 1989.

[32274] 5826. Rothnagel, J. A.; Longley, M. A.; Bundman, D. S.; Naylor, S. L.; Lalley, P. A.; Jenkins, N. A.; Gilbert, D. J.; Copeland, N. G.; Roop, D. R.: Characterization of the mouse loricrin gene: linkage with profilaggrin and the flaky tail and soft coat mutant loci on chromosome 3. *Genomics* 23: 450–456, 1994.

[32275] 5827. Sybert, V. P.; Dale, B. A.; Holbrook, K. A.: Ichthyosis vulgaris: identification of a defect in synthesis of filaggrin correlated with an absence of keratohyaline granules. *J. Invest. Derm.* 84: 191–194, 1985.

[32276] 5828. Volz, A.; Korge, B. P.; Compton, J. G.; Ziegler, A.; Steinert, P. M.; Mischke, D.: Physical mapping of a functional cluster of epidermal differentiation genes on chromosome 1q21. *Genomics* 18: 92–99, 1993.

[32277] 5829. Barbosa, C. A. A.; Koury, W. H.; Krieger, H.: Linkage data on MN and the Hb beta locus. *Am. J. Hum. Genet.* 27: 797–801, 1975.

[32278] 5830. Riddell, D. C.; Wang, H.; Umbenhauer, D. R.;

Beaume, P. H.; Guengerich, F. P.; Hamerton, J. L.: Regional assignment for the genes encoding human P450III A3 (CYP3) and P450IIC9 (CYP2C). (Abstract) *Cytogenet. Cell Genet.* 46: 682, 1987.

[32279] 5831. Yoder, J. A.; Yen, R.-W. C.; Vertino, P. M.; Bestor, T. H.; Baylin, S. B.: New 5-prime regions of the murine and human genes for DNA (cytosine-5)-methyltransferase. *J. Biol. Chem.* 271: 31092-31097, 1996.

[32280] 5832. Hofmann, S.; Lichtner, P.; Schuffenhauer, S.; Gerbitz, K.-D.; Meitinger, T.: Assignment of the human genes coding for cytochrome c oxidase subunits Va (COX5A), VIc (COX6C) and VIIc (COX7C) to chromosome bands 15q25, 8q22-q23 and 5q14 and of three pseudogenes (COX5AP1, COX6CP1, COX7CP1) to 14q22, 16p12 and 13q14-q21 by FISH and radiation hybrid mapping. *Cytogenet. Cell Genet.* 83: 226-227, 1998.

[32281] 5833. Martin-Gallardo, A.; McCombie, W. R.; Gocayne, J. D.; FitzGerald, M. G.; Wallace, S.; Lee, B. M. B.; Lamerdin, J.; Trapp, S.; Kelley, J. M.; Liu, L.-I.; Dubnick, M.; Johnston-Dow, L. A.; Kerlavage, A. R.; de Jong, P.; Carrano, A.; Fields, C.; Venter, J. C.: Automated DNA sequencing and analysis of 106 kilobases from human chromosome 19q13.3. *Nature Genet.* 1: 34-39, 1992.

- [32282] 5834.Schellens, J. H. M.; Soons, P. A.; Breimer, D. D.: Lack of bimodality in nifedipine plasma kinetics in a large population of healthy subjects. *Biochem.Pharm.* 37: 2507–2510, 1988.
- [32283] 5835.Shet, M. S.; Fisher, C. W.; Holmans, P. L.; Estabrook, R. W.: Human cytochrome P450 3A4: enzymatic properties of a purified recombinant fusion protein containing NADPH–P450 reductase. *Proc. Nat. Acad.Sci.* 90: 11748–11752, 1993.
- [32284] 5836.Shimada, T.; Guengerich, F. P.: Evidence for cytochrome P–450(NF), the nifedipine oxidase, being the principal enzyme involved in the bioactivation of aflatoxins in human liver. *Proc. Nat. Acad. Sci.* 86:462–465, 1989.
- [32285] 5837.Spurr, N. K.; Gough, A. C.; Stevenson, K.; Wolf, C. R.: The human cytochrome P450 CYP3 locus: assignment to chromosome 7q22–qter. *Hum.Genet.* 81: 171–174, 1989.
- [32286] 5838.Walker, A. H.; Jaffe, J. M.; Gunasegaram, S.; Cummings, S. A.; Huang, C.–S.; Chern, H.–D.; Olopade, O. I.; Weber, B. L.; Rebbeck, T. R.: Characterization of an allelic variant in the nifedipine–specific element of CYP3A4: ethnic distribution and implications for prostate cancer risk. *Hum. Mutat.* 12: 289–293, 1998.
- [32287] 5839.Watkins, P. B.; Wrighton, S. A.; Maurel, P.; Schuetz, E.

G.; Mendez-Picon, G.; Parker, G. A.; Guzelian, P. S.: Identification of an inducible form of cytochrome P-450 in human liver. *Proc. Nat. Acad. Sci.* 82:6310-6314, 1985.

[32288] 5840. Wolf, C. R.; Smith, C. A. D.; Gough, A. C.; Moss, J. E.; Vallis, K. A.; Howard, G.; Carey, F. J.; Mills, K.; McNee, W.; Carmichael, J.; Spurr, N. K.: Relationship between the debrisoquine hydroxylase polymorphism and cancer susceptibility. *Carcinogenesis* 13: 1035-1038, 1992.

[32289] 5841. Wrighton, S. A.; Stevens, J. C.: The human hepatic cytochromes P450 involved in drug metabolism. *Crit. Rev. Toxicol.* 22: 1-21, 1992.

[32290] 5842. Wrighton, S. A.; Vandenbranden, M.: Isolation and characterization of human fetal liver cytochrome P450HLp2: a third member of the P450III gene family. *Arch. Biochem. Biophys.* 268: 144-151, 1989.

[32291] 5843. Xie, W.; Barwick, J. L.; Downes, M.; Blumberg, B.; Simon, C. M.; Nelson, M. C.; Neuschwander-Tetri, B. A.; Brunt, E. M.; Guzelian, P. S.; Evans, R. M.: Humanized xenobiotic response in mice expressing nuclear receptor SXR. *Nature* 406: 435-439, 2000.

[32292] 5844. Gray, I. C.; Nobile, C.; Muresu, R.; Ford, S.; Spurr, N. K.: A 2.4-megabase physical map spanning the CYP2C gene cluster on chromosome 10q24. *Genomics* 28:

328–332, 1995.

- [32293] 5845. Guengerich, F. P.; Distlerath, L. M.; Reilly, P. E. B.; Wolff, T.; Shimada, T.; Umbenhauer, D. R.; Martin, M. V.: Human–liver cytochromes P–450 involved in polymorphisms of drug oxidation. *Xenobiotica* 16:367–378, 1986.
- [32294] 5846. Lenka, N.; Vijayasathy, C.; Mullick, J.; Avadhani, N. G.: Structural organization and transcription regulation of nuclear genes encoding the mammalian cytochrome c oxidase complex. *Prog. Nucleic Acid Res. Molec. Biol.* 61: 309–344, 1998.
- [32295] 5847. Otsuka, M.; Mizuno, Y.; Yoshida, M.; Kagawa, Y.; Ohta, S.: Nucleotide sequence of cDNA encoding human cytochrome c oxidase subunit VIc. *Nucleic Acids Res.* 16: 10916 only, 1988.
- [32296] 5848. Akbari, O.; Freeman, G. J.; Meyer, E. H.; Greenfield, E. A.; Chang, T. T.; Sharpe, A. H.; Berry, G.; DeKruyff, R. H.; Umetsu, D. T.: Antigen–specific regulatory T cells develop via the ICOS–ICOS–ligand pathway and inhibit allergen–induced airway hyperreactivity. *Nature Med.* 8: 1024–1032, 2002.
- [32297] 5849. Cox, D. R.; Burmeister, M.; Price, E. R.; Kim, S.; Myers, R. M.: Radiation hybrid mapping: a somatic cell genetic method for constructing high–resolution maps of

mammalian chromosomes. *Science* 250: 245–250,1990.

[32298] 5850.Crawley, J. B.; Williams, L. M.; Mander, T.; Brennan, F. M.; Foxwell,B. M.: Interleukin–10 stimulation of phosphatidylinositol 3–kinaseand p70 S6 kinase is required for the proliferative but not the antiinflammatoryeffects of the cytokine. *J. Biol. Chem.* 271: 16357–16362, 1996.

[32299] 5851.Eskdale, J.; Gallagher, G.; Verweij, C. L.; Keijsers, V.; Westendorp,R. G. J.; Huizinga, T. W. J.: Interleukin 10 secretion in relationto human IL–10 locus haplotypes. *Proc. Nat. Acad. Sci.* 95: 9465–9470,1998.

[32300] 5852.Eskdale, J.; Kube, D.; Tesch, H.; Gallagher, G.: Mapping of thehuman IL10 gene and further characterization of the 5–prime flankingsequence. *Immunogenetics* 46: 120–128, 1997.

[32301] 5853.Farmer, M. A.; Sundberg, J. P.; Bristol, I. J.; Churchill, G. A.;Li, R.; Elson, C. O.; Leiter, E. H.: A major quantitative trait locus on chromosome 3 controls colitis severity in IL–10–deficient mice. *Proc.Nat. Acad. Sci.* 98: 13820–13825, 2001.

[32302] 5854.Franchimont, D.; Martens, H.; Hagelstein, M.–T.; Louis, E.; Dewe,W.; Chrousos, G. P.; Belaiche, J.; Geenen, V.: Tumor necrosis factoralpha decreases, and interleukin–10 increases, the sensitivity ofhuman monocytes to

dexamethasone: potential regulation of the glucocorticoid receptor. *J. Clin. Endocr. Metab.* 84: 2834–2839, 1999.

- [32303] 5855. Gesser, B.; Leffers, H.; Jinqian, T.; Vestergaard, C.; Kirstein, N.; Sindet-Pedersen, S.; Jensen, S. L.; Thestrup-Pedersen, K.; Larsen, C. G.: Identification of functional domains on human interleukin 10. *Proc. Nat. Acad. Sci.* 94: 14620–14625, 1997.
- [32304] 5856. Gibson, A. W.; Edberg, J. C.; Wu, J.; Westendorp, R. G. J.; Huizinga, T. W. J.; Kimberly, R. P.: Novel single nucleotide polymorphisms in the distal IL-10 promoter affect IL-10 production and enhance the risk of systemic lupus erythematosus. *J. Immun.* 166: 3915–3922, 2001.
- [32305] 5857. Goudy, K.; Song, S.; Wasserfall, C.; Zhang, Y. C.; Kapturczak, M.; Muir, A.; Powers, M.; Scott-Jorgensen, M.; Campbell-Thompson, M.; Crawford, J. M.; Ellis, T. M.; Flotte, T. R.; Atkinson, M. A.: Adeno-associated virus vector-mediated IL-10 gene delivery prevents type 1 diabetes in NOD mice. *Proc. Nat. Acad. Sci.* 98: 13913–13918, 2001.
- [32306] 5858. Grove, J.; Daly, A. K.; Bassendine, M. F.; Gilvarry, E.; Day, C. P.: Interleukin 10 promoter region polymorphisms and susceptibility to advanced alcoholic liver disease. *Gut*

46: 540–545, 2000.

- [32307] 5859.Kim, J. M.; Brannan, C. I.; Copeland, N. G.; Jenkins, N. A.; Khan,T. A.; Moore, K. W.: Structure of the mouse Il-10 gene and chromosomallocalization of the mouse and human genes. J. Immun. 148: 3618–3623,1992.
- [32308] 5860.Kitagawa, N.; Goto, M.; Kurozumi, K.; Maruo, S.; Fukayama, M.;Naoe, T.; Yasukawa, M.; Hino, K.; Suzuki, T.; Todo, S.; Takada, K.: Epstein–Barr virus–encoded poly(A)–RNA supports Burkitt's lymphomagrowth through inter-leukin–10 induction. EMBO J. 19: 6742–6750,2000.
- [32309] 5861.Kuhn, R.; Lohler, J.; Rennick, D.; Rajewsky, K.; Muller, K.:Interleukin–10–deficient mice develop chronic enterocolitis. Cell 75:263–274, 1993.
- [32310] 5862.Lee, T.–S.; Chau, L.–Y.: Heme oxygenase–1 mediates the anti–inflammatoryeffect of interleukin–10 in mice. Nature Med. 8: 240–246, 2002.
- [32311] 5863.Meng, X.; Sawamura, D.; Tamai, K.; Hanada, K.; Ishida, H.; Hashimoto,I.: Keratinocyte gene therapy for systemic diseases: circulatinginterleukin 10 released from gene–transferred keratinocytes inhibitscontact hypersensitivity at distant areas of the skin. J. Clin. Invest. 101:1462–1467, 1998.
- [32312] 5864.Pinderski Oslund, L. J.; Hedrick, C. C.; Olvera, T.;

Hagenbaugh,A.; Territo, M.; Berliner, J. A.; Fyfe, A. I.: Interleukin-10 blocks atherosclerotic events in vitro and in vivo. *Arterioscler. Thromb.Vasc. Biol.* 19: 2847-2853, 1999.

[32313] 5865.Rosenwasser, L. J.; Borish, L.: Genetics of atopy and asthma:the rationale behind promoter-based candidate gene studies (IL-4 and IL-10). *Am. J. Resp. Crit. Care Med.* 156: S152-S155, 1997.

[32314] 5866.Shin, H. D.; Winkler, C.; Stephens, J. C.; Bream, J.; Young, H.;Goedert, J. J.; O'Brien, T. R.; Vlahov, D.; Buchbinder, S.; Giorgi,J.; Rinaldo, C.; Donfield, S.; Willoughby, A.; O'Brien, S. J.; Smith,M. W.: Genetic restriction of HIV-1 pathogenesis to AIDS by promoter alleles of IL10. *Proc. Nat. Acad. Sci.* 97: 14467-14472, 2000.

[32315] 5867.Terkeltaub, R. A.: IL-10: an 'immunologic scalpel' for atherosclerosis?(Editorial) *Atheroscler. Thromb. Vasc. Biol.* 19: 2823-2825, 1999.

[32316] 5868.Turner, D. M.; Williams, D. M.; Sankaran, D.; Lazarus, M.; Sinnott,P. J.; Hutchinson, I. V.: An investigation of polymorphism in the interleukin-10 gene promoter. *Europ. J. Immunogenet.* 24: 1-8, 1997.

[32317] 5869.Vieira, P.; de Waal-Malefyt, R.; Dang, M.-N.; Johnson, K. E.;Kastelein, R.; Fiorentino, D. F.; deVries, J. E.;

Roncarolo, M.-G.; Mosmann, T. R.; Moore, K. W.: Isolation and expression of human cytokinesynthesis inhibitory factor cDNA clones: homology to Epstein-Barrvirus open reading frame BCRF1. *Proc. Nat. Acad. Sci.* 88: 1172-1176, 1991.

[32318] 5870. Westendorp, R. G. J.; Langermans, J. A. M.; Huizinga, T. W. G.; Elouali, A. H.; Boomsma, D. I.; Verweij, C. L.; Van-denbroucke, J. P.: Genetic influence on cytokine production and fatal meningococcal disease. *Lancet* 349: 170-173, 1997.

[32319] 5871. Armstrong, E.; Cannizzaro, L.; Bergman, M.; Huebner, K.; Alitalo, K.: The c-src tyrosine kinase (CSK) gene, a potential antioncogene, localizes to human chromosome region 15q23-q25. *Cytogenet. Cell Genet.* 60:119-120, 1992.

[32320] 5872. Cloutier, J.-F.; Veillette, A.: Association of inhibitory tyrosineprotein kinase p50(csk) with protein tyrosine phosphatase PEP in Tcells and other hemopoietic cells. *EMBO J.* 15: 4909-4918, 1996.

[32321] 5873. Partanen, J.; Armstrong, E.; Bergman, M.; Makela, T. P.; Hirvonen, H.; Huebner, K.; Alitalo, K.: Cyl encodes a putative cytoplasmic tyrosine kinase lacking the conserved tyrosine autophosphorylation site (Y416-src). *Oncogene* 6:

2013–2018, 1991.

- [32322] 5874. Choubey, D.; Snoddy, J.; Chaturvedi, V.; Toniato, E.; Opdenakker, G.; Thakur, A.; Samanta, H.; Engel, D. A.; Lengyel, P.: Interferon- γ gene activators: indications for repeated gene duplication during the evolution of a cluster of interferon-activatable genes on murine chromosome 1. *J. Biol. Chem.* 264: 17182–17189, 1989.
- [32323] 5875. Ahuja, H. G.; Felix, C. A.; Aplan, P. D.: The t(11;20)(p15;q11) chromosomal translocation associated with therapy-related myelodysplastic syndrome results in an NUP98–TOP1 fusion. *Blood* 94: 3258–3261, 1999.
- [32324] 5876. Zhang, F. R.; Delattre, O.; Rouleau, G.; Couturier, J.; Lefrançois, D.; Thomas, G.; Aurias, A.: The neuroepithelioma breakpoint on chromosome 22 is proximal to the meningioma locus. *Genomics* 6: 174–177, 1990.
- [32325] 5877. Siciliano, M. J.; Bachinski, L.; Dolf, G.; Carrano, A. V.; Thompson, L. H.: Chromosomal assignments of human DNA repair genes that complement Chinese hamster ovary (CHO) cell mutants. (Abstract) *Cytogenet. Cell Genet.* 46: 691–692, 1987.
- [32326] 5878. Thompson, L. H.; Carrano, A. V.; Sato, K.; Salazar, E. P.; White, B. F.; Stewart, S. A.; Minkler, J. L.; Siciliano, M. J.: Identification of nucleotide-excision-repair genes on hu-

man chromosomes 2 and 13 by functional complementation in hamster-human hybrids. *Somat. Cell Molec. Genet.* 13: 539–551, 1987.

- [32327] 5879. Sul, H. S.; Wise, L. S.; Brown, M. L.; Rubin, C. S.: Cloning of cDNA sequences for murine ATP-citrate lyase: construction of recombinant plasmids using an immunopurified mRNA template and evidence for the nutritional regulation of ATP-citrate lyase mRNA content in mouse-liver. *J. Biol. Chem.* 259: 1201–1205, 1984.
- [32328] 5880. Brandt, P.; Ibrahim, E.; Bruns, G. A. P.; Neve, R. L.: Determination of the nucleotide sequence and chromosomal localization of the ATP2B2 gene encoding human Ca(2+)-pumping ATPase isoform PMCA2. *Genomics* 14: 484–487, 1992.
- [32329] 5881. Kozel, P. J.; Friedman, R. A.; Erway, L. C.; Yamoah, E. N.; Liu, L. H.; Riddle, T.; Duffy, J. J.; Doetschman, T.; Miller, M. L.; Cardell, E. L.; Shull, G. E.: Balance and hearing deficits in mice with a null mutation in the gene encoding plasma membrane Ca(2+)-ATPase isoform 2. *J. Biol. Chem.* 273: 18693–18696, 1998.
- [32330] 5882. Richards, F. M.; Phipps, M. E.; Latif, F.; Yao, M.; Crossey, P. A.; Foster, K.; Linehan, W. M.; Affara, N. A.; Lerman, M. I.; Zbar, B.; Ferguson-Smith, M. A.; Maher, E.

R.: Mapping the von Hippel–Lindau disease tumour suppressor gene: identification of germline deletions by pulsed field gel electrophoresis. *Hum. Molec. Genet.* 2: 879–882, 1993.

[32331] 5883. Street, V. A.; McKee–Johnson, J. W.; Fonseca, R. C.; Tempel, B. L.; Noben–Trauth, K.: Mutations in a plasma membrane $\text{Ca}(2+)$ –ATPase gene cause deafness in deaf-waddler mice. *Nature Genet.* 19: 390–394, 1998.

[32332] 5884. Wang, M. G.; Yi, H.; Hilfiker, H.; Carafoli, E.; Strehler, E. E.; McBride, O. W.: Localization of two genes encoding plasma membrane $\text{Ca}(2+)$ –ATPases isoforms 2 (ATP2B2) and 3 (ATP2B3) to human chromosomes 3p26–p25 and Xq28, respectively. *Cytogenet. Cell Genet.* 67: 41–45, 1994.

[32333] 5885. Shi, G.–P.; Munger, J. S.; Meara, J. P.; Rich, D. H.; Chapman, H. A.: Molecular cloning and expression of human alveolar macrophage cathepsin S, an elastinolytic cysteine protease. *J. Biol. Chem.* 267: 7258–7262, 1992.

[32334] 5886. Shi, G.–P.; Webb, A. C.; Foster, K. E.; Knoll, J. H. M.; Lemere, C. A.; Munger, J. S.; Chapman, H. A.: Human cathepsin S: chromosomal localization, gene structure, and tissue distribution. *J. Biol. Chem.* 269: 11530–11536, 1994.

- [32335] 5887.Grundmann, U.; Nerlich, C.; Rein, T.; Zettlmeissl, G.: CompletecDNA sequence encoding human beta-galactoside alpha-2,6-sialyltransferase. *NucleicAcids Res.* 18: 667 only, 1990.
- [32336] 5888.Wang, X.; Vertino, A.; Eddy, R. L.; Byers, M. G.; Jani-Sait, S.N.; Shows, T. B.; Lau, J. T. Y.: Chromosome mapping and organizationof the human beta-galactoside alpha-2,6-sialyltransferase gene: differentialand cell-type specific usage of upstream exon sequences in B-lymphoblastoidcells. *J. Biol. Chem.* 268: 4355-4361, 1993.
- [32337] 5889.Xiang, M.; Zhou, L.; Macke, J. P.; Yoshioka, T.; Hendry, S. H.C.; Eddy, R. L.; Shows, T. B.; Nathans, J.: The Brn-3 family of POU-domainfactors: primary structure, binding specificity, and expression insubsets of retinal ganglion cells and somatosensory neurons. *J. Neurosci.* 15:4762-4785, 1995.
- [32338] 5890.Bouillaud, F.; Villarroya, F.; Hentz, E.; Raimbault, S.; Cassard,A.-M.; Ricquier, D.: Detection of brown adipose tissue uncouplingprotein mRNA in adult patients by a human genomic probe. *Clin. Sci.* 75:21-27, 1988.
- [32339] 5891.Bouillaud, F.; Weissenbach, J.; Ricquier, D.: Complete cDNA-derivedamino acid sequence of rat brown fat un-

coupling protein. J. Biol.Chem. 261: 1487–1491, 1986.

- [32340] 5892.Cassard, A. M.; Bouillaud, F.; Mattei, M. G.; Hentz, E.; Raimbault,S.; Thomas, M.; Ricquier, D.: Human uncoupling protein gene: structure,comparison with rat gene, and assignment to the long arm of chromosome
- [32341] 5893.Echtay, K. S.; Roussel, D.; St-Pierre, J.; Jekabsons, M. B.; Cadenas,S.; Stuart, J. A.; Harper, J. A.; Roebuck, S. J.; Morrison, A.; Pickering,S.; Clapham, J. C.; Brand, M. D.: Superoxide activates mitochondrialuncoupling proteins. Nature 415: 96–99, 2002.
- [32342] 5894.Echtay, K. S.; Winkler, E.; Klingenberg, M.: Coenzyme Q is anobligatory cofactor for uncoupling protein function. Nature 408:609–613, 2000.
- [32343] 5895.J. Cell. Biochem. 43: 255–264, 1990.4. Clement, K.; Ruiz, J.; Cassard-Doulcier, A. M.; Bouillaud, F.;Ricquier, D.; Basdevant, A.; Guy-Grand, B.; Froguel, P.: Additiveeffect of A-G (–3826) variant of the uncoupling protein gene and thetrp64arg mutation of the beta 3-adrenergic receptor gene on weightgain in morbid obesity. Int. J. Obes. Relat. Metab. Disord. 20:1062–1066, 1996.
- [32344] 5896.Enerback, S.; Jacobsson, A.; Simpson, E. M.; Guerra, C.; Yamashita,H.; Harper, M.–E.; Kozak, L. P.: Mice lacking mitochondrial uncouplingprotein are cold-sensitive but

not obese. *Nature* 387: 90–93, 1997.

- [32345] 5897. Fletcher, C.; Norman, D. J.; Germond, E.; Heintz, N.: A multilocus linkage map of mouse chromosome 8. *Genomics* 9: 737–741, 1991.
- [32346] 5898. Jacobson, A.; Stadler, U.; Glotzer, M. A.; Kozak, L. P.: Mitochondrial uncoupling protein from mouse brown fat: molecular cloning, genetic mapping and mRNA expression. *J. Biol. Chem.* 260: 16250–16254, 1985.
- [32347] 5899. Lowell, B. B.; S-Susulic, V.; Hamann, A.; Lawitts, J. A.; Himms-Hagen, J.; Boyer, B. B.; Kozak, L. P.; Flier, J. S.: Development of obesity in transgenic mice after genetic ablation of brown adipose tissue. *Nature* 366: 740–742, 1993.
- [32348] 5900. Oppert, J. M.; Vohl, M. C.; Chagnon, M.; Dionne, F. T.; Cassard-Doucier, A. M.; Ricquier, D.; Perusse, L.; Bouchard, C.: DNA polymorphism in the uncoupling protein (UCP) gene and human body fat. *Int. J. Obes. Relat. Metab. Disord.* 18: 526–531, 1994.
- [32349] 5901. Ridley, R. G.; Patel, H. V.; Gerber, G. E.; Morton, R. C.; Freeman, K. B.: Complete nucleotide and derived amino acid sequence of cDNA encoding the mitochondrial uncoupling protein of rat brown adipose tissue: lack of mitochondrial targeting presequence. *Nucleic Acids Res.* 14:

4025–4035, 1986.

- [32350] 5902.Urhammer, S. A.; Fridberg, M.; Sorensen, T. I.; Echwald, S. M.; Andersen, T.; Tybjaerg-Hansen, A.; Clausen, J. O.; Pedersen, O.:Studies of genetic variability of the uncoupling protein 1 gene inCaucasian subjects with juvenile-onset obesity. J. Clin. Endocr.Metab. 82: 4069–4074, 1997.
- [32351] 5903.Rodriguez de Cordoba, S.; Rubinstein, P.; Ferreira, A.: Highresolution isoelectric focusing of immunoprecipitated proteins underdenaturing conditions: a simple analytical method applied to the studyof complement component polymorphisms. J. Immun. Methods 69: 165–172,1984.
- [32352] 5904.Muller, U.; Kupke, K. G.: The genetics of primary torsion dystonia. Hum.Genet. 84: 107–115, 1990.
- [32353] 5905.Di Marzo, V.; Goparaju, S. K.; Wang, L.; Liu, J.; Batkai, S.; Jarai,Z.; Fezza, F.; Miura, G. I.; Palmiter, R. D.; Sugiura, T.; Kunos,G.: Leptin-regulated endocannabinoids are involved in maintainingfood intake. Nature 410: 822–825, 2001.
- [32354] 5906.Gerard, C. M.; Mollereau, C.; Vassart, G.; Parmentier, M.: Molecularcloning of a human cannabinoid receptor which is also expressed intestis. Biochem. J. 279:

129–134, 1991.

- [32355] 5907.Hoehe, M. R.; Caenazzo, L.; Martinez, M. M.; Hsieh, W.-T.; Modi,W. S.; Gershon, E. S.; Bonner, T. I.: Genetic and physical mappingof the human cannabinoid receptor gene to chromosome 6q14–q15. *NewBiologist* 3: 880–885, 1991.
- [32356] 5908.Ledent, C.; Valverde, O.; Cossu, G.; Petitet, F.; Aubert, J.-F.;Beslot, F.; Bohme, G. A.; Imperato, A.; Pedrazzini, T.; Roques, B.P.; Vassart, G.; Fratta, W.; Parmentier, M.: Unresponsiveness tocannabinoids and reduced addictive effects of opiates in CB(1) receptorknockout mice. *Science* 283 401–404, 1999.
- [32357] 5909.Marsicano, G.; Wotjak, C. T.; Azad, S. C.; Bisogno, T.; Rammes,G.; Cascio, M. G.; Hermann, H.; Tang, J.; Hofmann, C.; Zieglgansberger,W.; Di Marzo, V.; Lutz, B.: The endogenous cannabinoid system controlsextinction of aversive memories. *Nature* 418: 530–534, 2002.
- [32358] 5910.Matsuda, L. A.; Lolait, S. J.; Brownstein, M. J.; Young, A. C.;Bonner, T. I.: Structure of a cannabinoid receptor and functionalexpression of the cloned cDNA. *Nature* 346: 561–564, 1990.
- [32359] 5911.Modi, W. S.; Bonner, T. I.: Localization of the cannabinoid (sic)receptor locus using non–isotopic in situ hy–

bridization. (Abstract) Cytogenet.Cell Genet. 58: 1915 only, 1991.

[32360] 5912.Panikashvili, D.; Simeonidou, C.; Ben-Shabat, S.; Hanus, L.; Breuer,A.; Mechoulam, R.; Shohami, E.: An endogenous cannabinoid (2-AG)is neuroprotective after brain injury. Nature 413: 527-531, 2001.

[32361] 5913.Beechey, C.; Tweedie, S.; Spurr, N.; Ball, S.; Peters, J.; Edwards,Y.: Mapping of mouse carbonic anhydrase-3, Car-3: another locus inthe homologous region of mouse chromosome 3 and human chromosome 8. Genomics 6:692-696, 1990.

[32362] 5914.Carter, N.; Jeffery, S.; Shiels, A.; Edwards, Y.; Tipler, T.; Hopkinson,D. A.: Characterization of human carbonic anhydrase III from skeletal muscle. Biochem. Genet. 17: 837-854, 1979.

[32363] 5915.Edwards, Y. H.; Lloyd, J.; Parkar, M.; Povey, S.: Human musclespecific carbonic anhydrase, CA3, is on chromosome 8. (Abstract) Cytogenet.Cell Genet. 40: 621 only, 1985.

[32364] 5916.Edwards, Y. H.; Lloyd, J. C.; Parkar, M.; Povey, S.: The gene for human muscle specific carbonic anhydrase (CAIII) is assigned to chromosome 8. Ann. Hum. Genet. 50: 41-47, 1986.

- [32365] 5917.Heath, R.; Carter, N. D.; Jeffery, S.; Edwards, R. J.; Watts, D.C.; Watts, R. L.: Evaluation of carrier detection of Duchenne muscular dystrophy using carbonic anhydrase III and creatine kinase. *Am. J. Med. Genet.* 21: 291–296, 1985.
- [32366] 5918.Lloyd, J.; Brownson, C.; Tweedie, S.; Charlton, J.; Edwards, Y.H.: Human muscle carbonic anhydrase: gene structure and DNA methylation patterns in fetal and adult tissues. *Genes Dev.* 1: 594–602, 1987.
- [32367] 5919.Lloyd, J.; McMillan, S.; Hopkinson, D.; Edwards, Y. H.: Nucleotide sequence and derived amino acid sequence of a cDNA encoding human muscle carbonic anhydrase. *Gene* 41: 233–239, 1986.
- [32368] 5920.Lloyd, J. C.; Isenberg, H.; Hopkinson, D. A.; Edwards, Y. H.: Isolation of a cDNA clone for the human muscle specific carbonic anhydrase, CA III. *Ann. Hum. Genet.* 49: 241–251, 1985.
- [32369] 5921.Wade, R.; Gunning, P.; Eddy, R.; Shows, T.; Kedes, L.: Nucleotide sequence, tissue-specific expression, and chromosome location of human carbonic anhydrase III: the human CAIII gene is located on the same chromosome as the closely linked CAI and CAII genes. *Proc. Nat. Acad. Sci.* 83: 9571–9575, 1986.

- [32370] 5922.Hall, C.; Monfries, C.; Smith, P.; Lim, H. H.; Kozma, R.; Ahmed,S.; Vanniasingham, V.; Leung, T.; Lim, L.: Novel human brain cDNAencoding a 34,000 M(r) protein n-chimaerin, related to both the regulatorydomain of protein kinase C and BCR, the product of the breakpointcluster region gene. *J. Molec. Biol.* 211: 11–16, 1990.
- [32371] 5923.Hall, C.; Sin, W. C.; Teo, M.; Michael, G. J.; Smith, P.; Dong,J. M.; Lim, H. H.; Manser, E.; Spurr, N. K.; Jones, T. A.; Lim, L.: Alpha-2-chimerin, an SH2-containing GTPase-activating protein forthe ras-related protein p21-rac derived by alternate splicing of thehuman n-chimerin gene, is selectively expressed in brain regions andtestes. *Molec. Cell. Biol.* 13: 4986–4998, 1993.
- [32372] 5924.Brewer, C.; Holloway, S.; Zawalnyski, P.; Schinzel, A.; FitzPatrick,D.: A chromosomal deletion map of human malformations. *Am. J. Hum.Genet.* 63: 1153–1159, 1998.
- [32373] 5925.Brewer, C. M.; Leek, J. P.; Green, A. J.; Holloway, S.; Bonthron,D. T.; Markham, A. F.; FitzPatrick, D. R.: A locus for isolated cleftpalate, located on human chromosome 2q32. *Am. J. Hum. Genet.* 65:387–396, 1999.
- [32374] 5926.Carter, C. O.; Evans, K.; Coffey, R.; Roberts, J. A. F.; Buck,A.; Roberts, M. F.: A family study of isolated cleft palate. *J.Med. Genet.* 19: 329–331, 1982.

- [32375] 5927.Christensen, K.; Holm, N. V.; Olsen, J.; Kock, K.; Fogh-Andersen, P.: Selection bias in genetic-epi-demiological studies of cleft lip and palate. *Am. J. Hum. Genet.* 51: 654-659, 1992.
- [32376] 5928.Christensen, K.; Mitchell, L. E.: Familial recurrence-pattern analysis of nonsyndromic isolated cleft palate: a Danish registry study. *Am. J. Hum. Genet.* 58: 182-190, 1996.
- [32377] 5929.Jenkins, M.; Stady, C.: Dominant inheritance of cleft of the soft palate. *Hum. Genet.* 53: 341-342, 1980.
- [32378] 5930.Shields, E. D.; Bixler, D.; Fogh-Andersen, P.: Cleft palate: a genetic and epidemiologic investigation. *Clin. Genet.* 20: 13-24, 1981.
- [32379] 5931.Hwang, S. J.; Beaty, T. H.; Panny, S. R.; Street, N. A.; Joseph, J. M.; Gordon, S.; McIntosh, I.; Francomano, C. A.: Association study of transforming growth factor alpha (TGF-alpha) TaqI polymorphism and oral clefts: indication of gene-environment interaction in a population-based sample of infants with birth defects. *Am. J. Epidemiol.* 141: 629-636, 1995.
- [32380] 5932.Van Dyke, D. C.; Goldman, A. S.; Spielman, R. S.; Zmijewski, C. M.: Segregation of HLA in families with oral clefts: evidence against linkage between isolated cleft

palate and HLA. *Am. J. Med. Genet.* 15:85–88, 1983.

[32381] 5933.Donald, L. J.; Wang, H. S.; Hamerton, J. L.: Are there additionalCKBB loci?(Abstract) *Cytogenet. Cell Genet.* 32: 267–268, 1982.

[32382] 5934.McLellan, R. A.; Oscarson, M.; Alexandrie, A.–K.; Seidegard, J.;Evans, D. A. P.; Rannug, A.; Ingelman–Sundberg, M.: Characterizationof a human glutathione S–transferase mu cluster containing a duplicatedGSTM1 gene that causes ultrarapid enzyme activity. *Molec. Pharm.* 52:958–965, 1997.

[32383] 5935.Pearson, W. R.; Vorachek, W. R.; Xu, S.; Berger, R.; Hart, I.;Vannais, D.; Patterson, D.: Identification of class–mu glutathionetransferase genes GSTM1–GSTM5 on human chromosome 1p13. *Am. J. Hum.Genet.* 53: 220–233, 1993.

[32384] 5936.Sanchez–Corral, P.; Pardo–Manuel de Villena, F.; Rey–Campos, J.;Rodriguez de Cordoba, S.: C4BPAL1, a member of the human regulatorof complement activation (RCA) gene cluster that resulted from theduplication of the gene coding for the alpha–chain of C4b–bindingprotein. *Genomics* 17: 185–193, 1993.

[32385] 5937.Hillarp, A.; Dahlback, B.: Novel subunit in C4b–binding proteinrequired for protein S binding. *J. Biol.*

Chem. 263: 12759–12764,1988.

- [32386] 5938.Hillarp, A.; Dahlback, B.: Cloning of cDNA coding for the beta-chain of human complement component C4b-binding protein: sequence homology with the alpha chain. Proc. Nat. Acad. Sci. 87: 1183–1187, 1990.
- [32387] 5939.Hillarp, A.; Pardo-Manuel, F.; Ramos Ruiz, R.; Rodriguez de Cordoba, R.; Dahlback, B.: The human C4b-binding protein beta-chain gene. J.Biol. Chem. 268: 15017–15023, 1993.
- [32388] 5940.Pardo-Manuel, F.; Rey-Campos, J.; Hillarp, A.; Dahlback, B.; Rodriguez de Cordoba, S.: Human genes for the alpha and beta chains of complement C4b-binding protein are closely linked in a head-to-tail arrangement. Proc.Nat. Acad. Sci. 87: 4529–4532, 1990.
- [32389] 5941.Rodriguez de Cordoba, S.; Perez-Blas, M.; Ramos-Ruiz, R.; Sanchez-Corral, P.; Pardo-Manuel de Villena, F.; Rey-Campos, J.: The gene coding for the beta-chain of C4b-binding protein (C4BPB) has become a pseudogene in the mouse. Genomics 21: 501–509, 1994.
- [32390] 5942.Collaborative Study on the Genetics of Asthma: A genome-wide search for asthma susceptibility loci in ethnically diverse populations. Nature Genet. 15: 389–392, 1997.

- [32391] 5943.Gavett, S. H.; O'Hearn, D. J.; Li, X.; Huang, S. K.; Finkelman, F. D.; Wills-Karp, M.: Interleukin 12 inhibits antigen-induced airway hyperresponsiveness, inflammation, and Th2 cytokine expression in mice. *J. Exp. Med.* 182: 1527–1536, 1995.
- [32392] 5944.Karp, C. L.; Grupe, A.; Schadt, E.; Ewart, S. L.; Keane-Moore, M.; Cuomo, P. J.; Kohl, J.; Wahl, L.; Kuperman, D.; Germer, S.; Aud, D.; Peltz, G.; Wills-Karp, M.: Identification of complement factor 5 as a susceptibility locus for experimental allergic asthma. *Nature Immun.* 1: 221–226, 2000.
- [32393] 5945.Ober, C.; Cox, N. J.; Abney, M.; Di Rienzo, A.; Lander, E. S.; Changyaleket, B.; Gidley, H.; Kurtz, B.; Lee, J.; Nance, M.; Pettersson, A.; Prescott, J.; Richardson, A.; Schlenker, E.; Summerhill, E.; Willadsen, S.; Parry, R.; Collaborative Study on the Genetics of Asthma: Genome-wide search for asthma susceptibility loci in a founder population. *Hum. Molec. Genet.* 7: 1393–1398, 1998.
- [32394] 5946.Gennarelli, M.; Novelli, G.; Cobo, A.; Baiget, M.; Dalapiccola, B.: 3-Prime creatine kinase (M-type) polymorphisms linked to myotonic dystrophy in Italian and Spanish populations. *Hum. Genet.* 87: 654–656, 1991.
- [32395] 5947.Nigro, J. M.; Schweinfest, C. W.; Rajkovic, A.; Pavlovic, J.; Jamal, S.; Dottin, R. P.; Hart, J. T.; Kamarck, M.

E.; Rae, P. M. M.; Carty, M. D.; Martin-DeLeon, P.: cDNA cloning and mapping of the human creatine kinase M gene to 19q13. *Am. J. Hum. Genet.* 40: 115–125, 1987.

- [32396] 5948. Perryman, M. B.; Kerner, S. A.; Bohlmeier, T. J.; Roberts, R.: Isolation and sequence analysis of a full-length cDNA for human M creatine kinase. *Biochem. Biophys. Res. Commun.* 140: 981–989, 1986.
- [32397] 5949. Povey, S.; Inwood, M.; Tanyar, A.; Bobrow, M.: The expression of the BB isozyme of creatine kinase. (Abstract) *Cytogenet. Cell Genet.* 25: 198 only, 1979.
- [32398] 5950. Povey, S.; Inwood, M.; Tanyar, A.; Bobrow, M.: The expression of creatine kinase isozymes in human cultured cells. *Ann. Hum. Genet.* 43: 15–26, 1979.
- [32399] 5951. Roman, D.; Billadello, J.; Gordon, J.; Grace, A.; Sobel, B.; Strauss, A.: Complete nucleotide sequence of dog heart creatine kinase mRNA: conservation of amino acid sequence within and among species. *Proc. Nat. Acad. Sci.* 82: 8394–8398, 1985.
- [32400] 5952. Rosenberg, U. B.; Kunz, G.; Frischauf, A.; Lehrach, H.; Mahr, R.; Eppenberger, H. M.; Perriard, J.-C.: Molecular cloning and expression during myogenesis of sequences coding for M-creatine kinase. *Proc. Nat. Acad. Sci.* 79: 6589–6592, 1982.

- [32401] 5953.Schweinfest, C. W.; Nigro, J. M.; Rajkovic, A.; Dottin, R. P.;Hart, J. M.; Karmack, M. E.; Rae, P. M. M.: Localization of the humancreatine kinase–M gene to chromosome 19. (Abstract) Cytogenet. CellGenet. 40: 740–741, 1985.
- [32402] 5954.Smeets, H.; Bachinski, L.; Coerwinkel, M.; Schepens, J.; Hoeijmakers,J.; van Duin, M.; Grzeschik, K.–H.; Weber, C. A.; de Jong, P.; Siciliano,M. J.; Wieringa, B.: A long–range restriction map of the human chromosome19q13 region: close physical linkage between CKMM and the ERCC1 andERCC2 genes. Am. J. Hum. Genet. 46: 492–501, 1990.
- [32403] 5955.Stallings, R. L.; Olson, E.; Strauss, A. W.; Thompson, L. H.;Bachinski, L. L.; Siciliano, M. J.: Human creatine ki–nase genes onchromosomes 15 and 19, and proximity of the gene for the muscle formto the genes for apolipoprotein C2 and excision repair. Am. J. Hum.Genet. 43: 144–151, 1988.
- [32404] 5956.Steeghs, K.; Benders, A.; Oerlemans, F.; de Haan, A.; Heerschap,A.; Ruitenbeek, W.; Jost, C.; van Deursen, J.; Perryman, B.; Pette,D.; Bruckwilder, M.; Koudijs, J.; Jap, P.; Veerkamp, J.; Wieringa,B.: Altered Ca(2+) responses in muscles with combined mitochondrialand cytosolic creatine kinase deficiencies. Cell 89: 93–103, 1997.

- [32405] 5957.Watts, D. C.: Creatine kinase (adenosine 5–prime–triphosphate–creatinephosphotransferase).In: Boyer, P. D.: The Enzymes. New York: AcademicPress (pub.) (3rd ed.) 8: 1973. Pp. 384–455.
- [32406] 5958.Weil, D.; Van Cong, N.; Gross, M.–S.; Foubert, C.; Frezal, J.: Localisation du gene de la creatine kinase BB sur le chromosome14 par l'analyse des hybrides homme–rongeur. Ann. Genet. 23: 150–154,1980.
- [32407] 5959.Kishi, K.; Yasuda, T.; Awazu, S.; Mizuta, K.: Genetic polymorphismof human urine deoxyribonuclease I. Hum. Genet. 81: 295–297, 1989.
- [32408] 5960.Kishi, K.; Yasuda, T.; Ikehara, Y.; Sawazaki, K.; Sato, W.; Iida,R.: Human serum deoxyribonuclease I (DNase I) polymorphism: patternsimilarities among isozymes from serum, urine, kidney, liver, andpancreas. Am. J. Hum. Genet. 47: 121–126, 1990.
- [32409] 5961.Napirei, M.; Karsunky, H.; Zevnik, B.; Stephan, H.; Mannherz, H.G.; Moroy, T.: Features of systemic lupus erythematosus in Dnase1–deficientmice. Nature Genet. 25: 177–181, 2000.
- [32410] 5962.Yasuda, T.; Awazu, S.; Sato, W.; Iida, R.; Tanaka, Y.; Kishi, K.: Human genetically polymorphic deoxyribonucle–ase: purification, characterization,and multiplicity of urine

deoxyribonuclease I. J. Biochem. 108:393–398, 1990.

- [32411] 5963.Yasuda, T.; Kishi, K.; Yanagawa, Y.; Yoshida, A.: Structure of the human deoxyribonuclease I (DNase I) gene: identification of the nucleotide substitution that generates its classical genetic polymorphism. Ann.Hum. Genet. 59: 1–15, 1995.
- [32412] 5964.Yasuda, T.; Nadano, D.; Iida, R.; Takeshita, H.; Lane, S. A.; Callen, D. F.; Kishi, K.: Chromosomal assignment of the human deoxyribonuclease I gene, DNASE1 (DNL1), to band 16p13.3 using the polymerase chain reaction. Cytogenet. Cell Genet. 70: 221–223, 1995.
- [32413] 5965.Yasutomo, K.; Horiuchi, T.; Kagami, S.; Tsukamoto, H.; Hashimura, C.; Urushihara, M.; Kuroda, Y.: Mutation of DNASE1 in people with systemic lupus erythematosus. Nature Genet. 28: 313–314, 2001.
- [32414] 5966.Muller, U.; Steinberger, D.; Nemeth, A. H.: Clinical and molecular genetics of primary dystonias. Neurogenetics 1: 165–177, 1998.
- [32415] 5967.Chambers, I.; Frampton, J.; Goldfarb, P.; Affara, N.; McBain, W.; Harrison, P. R.: The structure of the mouse glutathione peroxidase gene: the selenocysteine in the active site is encoded by the 'termination' codon, TGA. EMBO J. 5: 1221–1227, 1986.

- [32416] 5968.Chu, F.-F.; Esworthy, R. S.; Doroshov, J. H.; Doan, K.; Liu, X.-F.: Expression of glutathione peroxidase in human liver in addition to kidney, heart, lung, and breast in humans and rodents. *Blood* 79:3233–3238, 1992.
- [32417] 5969.Takahashi, K.; Akasaka, M.; Yamamoto, Y.; Kobayashi, C.; Mizoguchi, J.; Koyama, J.: Primary structure of human plasma glutathione peroxidase deduced from cDNA sequences. *J. Biochem.* 108: 145–148, 1990.
- [32418] 5970.Kelner, M. J.; Montoya, M. A.: Structural organization of the human selenium-dependent phospholipid hydroperoxide glutathione peroxidase gene (GPX4): chromosomal localization to 19p13.3. *Biochem. Biophys. Res. Commun.* 249: 53–55, 1998.
- [32419] 5971.Roveri, A.; Casasco, A.; Maiorino, M.; Dalan, P.; Caligaro, A.; Ursini, F.: Phospholipid hydroperoxide glutathione peroxidase of rat testis. *J. Biol. Chem.* 267: 6142–6146, 1992.
- [32420] 5972.Comstock, K. E.; Johnson, K. J.; Rifken, D.; Henner, W. D.: Isolation and analysis of the gene and cDNA for a human mu class glutathione S-transferase, GSTM4. *J. Biol. Chem.* 268: 16958–16965, 1993.
- [32421] 5973.Ross, V. L.; Board, P. G.: Molecular cloning and heterologous expression of an alternatively spliced human mu

class glutathione S-transferase transcript. *Biochem. J.* 294: 373–380, 1993.

[32422] 5974. Ross, V. L.; Board, P. G.; Webb, G. C.: Chromosomal mapping of the human mu class glutathione S-transferases to 1p13. *Genomics* 18:87–91, 1993.

[32423] 5975. Taylor, J. B.; Oliver, J.; Sherrington, R.; Pemble, S. E.: Structure of human glutathione S-transferase class mu genes. *Biochem. J.* 274:587–593, 1991.

[32424] 5976. Board, P.; Coggan, M.; Johnston, P.; Ross, V.; Suzuki, T.; Webb, G.: Genetic heterogeneity of the human glutathione transferases: a complex of gene families. *Pharm. Therap.* 48: 357–369, 1990.

[32425] 5977. Chen, C.-L.; Liu, Q.; Relling, M. V.: Simultaneous characterization of glutathione S-transferase M1 and T1 polymorphisms by polymerase chain reaction in American whites and blacks. *Pharmacogenetics* 6:187–191, 1996.

[32426] 5978. DeJong, J. L.; Chang, C.-M.; Whang-Peng, J.; Knutsen, T.; Tu, C.-P. D.: The human liver glutathione S-transferase gene superfamily: expression and chromosome mapping of an H(b) subunit cDNA. *Nucleic Acids Res.* 16:8541–8554, 1988.

[32427] 5979. DeJong, J. L.; Mohandas, T.; Tu, C.-P. D.: The human H(b) (mu) class glutathione S-transferases are encoded by

a dispersed gene family. *Biochem.Biophys. Res. Commun.* 180: 15–22, 1991.

- [32428] 5980.Godschalk, R. W. L.; Dallinga, J. W.; Wikman, H.; Risch, A.; Kleinjans,J. C. S.; Bartsch, H.; Van Schooten, F.–J.: Modulation of DNA andprotein adducts in smokers by genetic polymorphisms in GSTM1, GSTT1,NAT1 and NAT2. *Pharmacogenetics* 11: 389–398, 2001.
- [32429] 5981.Harada, S.; Abei, M.; Tanaka, N.; Agarwal, D. P.; Goedde, H. W.: Liver glutathione S–transferase polymorphism in Japanese and itspharmacogenetic importance. *Hum. Genet.* 75: 322–325, 1987.
- [32430] 5982.Lee, K. A.; Kim, S. H.; Woo, H. Y.; Hong, Y. J.; Cho, H. C.: Increasedfrequencies of glutathione S–transferase (GSTM1 and GSTT1) gene deletionsin Korean patients with acquired aplastic anemia. *Blood* 98: 3483–3485,2001.
- [32431] 5983.Mannervik, B.: The isozymes of glutathione transferase. *Adv.Enzym. Relat. Areas Molec. Biol.* 57: 357–417, 1985.
- [32432] 5984.Mannervik, B.; Awasthi, Y. C.; Board, P. G.; Hayes, J. D.; Dillio, C.; Ketterer, B.; Listowsky, I.; Morgenstern, R.; Muramatsu,M.; Pearson, W. R.; Pickett, C. B.; Sato, K.; Widersten, M.; Wolf,R. C.: Nomenclature for human glutathione transferases. (Letter) *Biochem.J.* 282: 305–306,

1992.

- [32433] 5985.Arce, M. A.; Thompson, E. S.; Wagner, S.; Coyne, K. E.; Ferdman,B. A.; Lublin, D. M.: Molecular cloning of RhD cDNA derived from a gene present in RhD-positive, but not RhD-negative individuals. *Blood* 82:651–655, 1993.
- [32434] 5986.Bennett, P. R.; Le Van Kim, C.; Colin, Y.; Warwick, R. M.; Cherif-Zahar,B.; Fisk, N. M.; Cartron, J.-P.: Prenatal determination of fetalRhD type by DNA amplification. *New Eng. J. Med.* 329: 607–610, 1993.
- [32435] 5987.Bowman, J. M.: RhD hemolytic disease of the newborn. (Editorial) *NewEng. J. Med.* 339: 1775–1777, 1998.
- [32436] 5988.Cartron, J.-P.: Defining the Rh blood group antigens: biochemistryand molecular genetics. *Blood Rev.* 8: 199–212, 1994.
- [32437] 5989.Colin, Y.; Cherif-Zahar, B.; Le Van Kim, C.; Raynal, V.; Van Huffel,V.; Cartron, J.-P.: Genetic basis of the RhD-positive and RhD-negativeblood group polymorphism as determined by Southern analysis. *Blood* 78:2747–2752, 1991.
- [32438] 5990.Diamond, L. K.; Blackfan, K. D.; Baty, J. M.: Erythroblastosisfetalis and its association with universal edema of the fetus, icterusgravis neonatorum and anemia of the newborn. *J. Pediat.* 1: 269–309,1932.

- [32439] 5991.du Toit, E. D.; Martell, R. W.; Botha, I.; Kriel, C. J.: Anti-Dantibodies in the Rh-positive mothers. S. Afr. Med. J. 75: 452,1989.
- [32440] 5992.Garratty, G.: Severe reactions associated with trans- fusion ofpatients with sickle cell disease. Transfusion 37: 357-361, 1997.
- [32441] 5993.Huang, C.-H.: Personal Communication. New York City, N. Y. 10/11/1996.
- [32442] 5994.Huang, C.-H.; Chen, Y.; Reid, M.; Ghosh, S.: Genetic recombinationat the human RH locus: a family study of the red-cell Evans phenotypereveals a transfer of exons 2-6 from the RHD to the RHCE gene. Am.J. Hum. Genet. 59: 825-833, 1996.
- [32443] 5995.Huang, C.-H.; Reid, M. E.; Chen, Y.; Coghlan, G.; Okubo, Y.:Molecular definition of red cell Rh haplotypes by tightly linked SphIRFLPs. Am. J. Hum. Genet. 58: 133-142, 1996.
- [32444] 5996.Hyland, C. A.; Wolter, L. C.; Liew, Y. W.; Saul, A.: A Southernanalysis of Rh blood group genes: association between restrictionfragment length polymorphism pat- terns and Rh serotypes. Blood 83:566-572, 1994.
- [32445] 5997.Issitt, P. D.: The Rh blood group system, 1988: eight new antigensin nine years and some observations on the

biochemistry and genetics of the system. *Transfusion Med. Rev.* 3: 1–12, 1989.

- [32446] 5998. Kemp, T. J.; Poulter, M.; Carritt, B.: A recombination hot spot in the Rh genes revealed by analysis of unrelated donors with the rare D[–] phenotype. *Am. J. Hum. Genet.* 59: 1066–1073, 1996.
- [32447] 5999. Legler, T. J.; Eber, S. W.; Lakomek, M.; Lynen, R.; Maas, J. H.; Pekrun, A.; Repas-Humpe, M.; Schroter, W.; Kohler, M.: Application of RHD and RHCE genotyping for correct blood group determination in chronically transfused patients. *Transfusion* 39: 852–855, 1999.
- [32448] 6000. Le Van Kim, C.; Cherif-Zahar, B.; Raynal, V.; Mouro, I.; Lopez, M.; Cartron, J. P.; Colin, Y.: Multiple Rh messenger RNA isoforms are produced by alternative splicing. *Blood* 80: 1074–1078, 1992.
- [32449] 6001. Le Van Kim, C.; Mouro, I.; Cherif-Zahar, B.; Raynal, V.; Cherrier, C.; Cartron, J.-P.; Colin, Y.: Molecular cloning and primary structure of the human blood group RhD polypeptide. *Proc. Nat. Acad. Sci.* 89: 10925–10929, 1992.
- [32450] 6002. Levine, P.; Katzin, E. M.; Burnham, L.: Isoimmunization in pregnancy: its possible bearing on the etiology of erythroblastosis foetalis. *J.A.M.A.* 116: 825–827, 1941.
- [32451] 6003. Lo, Y. M. D.; Hjelm, N. M.; Fidler, C.; Sargent, I. L.;

Murphy, M. F.; Chamberlain, P. F.; Poon, P. M. K.; Redman, C. W. G.; Wainscoat, J. S.: Prenatal diagnosis of fetal RhD status by molecular analysis of maternal plasma. *New Eng. J. Med.* 339: 1734–1738, 1998.

[32452] 6004. Miyoshi, O.; Yabe, R.; Wakui, K.; Fukushima, Y.; Koizumi, S.; Uchikawa, M.; Kajii, T.; Numakura, C.; Takahashi, S.; Hayasaka, K.; Niikawa, N.: Two cases of mosaic RhD blood-group phenotypes and paternal isodisomy for chromosome 1. *Am. J. Med. Genet.* 104: 250–256, 2001.

[32453] 6005. Okuda, H.; Kawano, M.; Iwamoto, S.; Tanaka, M.; Seno, T.; Okubo, Y.; Kajii, E.: The RHD gene is highly detectable in RhD-negative Japanese donors. *J. Clin. Invest.* 100: 373–379, 1997.

[32454] 6006. Race, R. R.: An 'incomplete' antibody in human serum. (Letter) *Nature* 153: 771–772, 1944.

[32455] 6007. Race, R. R.; Sanger, R.: *Blood Groups in Man*. Oxford: Blackwell (pub.) (6th ed.): 1975.

[32456] 6008. Rouillac, C.; Le Van Kim, C.; Beolet, M.; Cartron, J.-P.; Colin, Y.: Leu110-to-pro substitution in the RhD polypeptide is responsible for the D(VII) category blood group phenotype. *Am. J. Hemat.* 49: 87–88, 1995.

[32457] 6009. Smythe, J. S.; Avent, N. D.; Judson, P. A.; Parsons, S. F.; Martin, P. G.; Anstee, D. J.: Expression of RHD and RHCE

gene products using retroviral transduction of K562 cells establishes the molecular basis of Rh blood group antigens. *Blood* 87: 2968–2973, 1996.

- [32458] 6010.Spanos, T.; Karageorga, M.; Ladis, V.; Peristeri, J.; Hatziliami, A.; Kattamis, C.: Red cell alloantibodies in patients with thalassemia. *VoxSang.* 58: 50–55, 1990.
- [32459] 6011.Wagner, F. F.; Flegel, W. A.: RHD gene deletion occurred in the Rhesus box. *Blood* 95: 3662–3668, 2000.
- [32460] 6012.Wagner, F. F.; Gassner, C.; Muller, T. H.; Schonitzer, D.; Schunter, F.; Flegel, W. A.: Molecular basis of weak D phenotypes. *Blood* 93:385–393, 1999.
- [32461] 6013.Wagner, F. F.; Ladewig, B.; Angert, K. S.; Heymann, G. A.; Eicher, N. I.; Flegel, W. A.: The DAU allele cluster of the RHD gene. *Blood* 100:306–311, 2002.
- [32462] 6014.Wiener, A. S.: The Rh series of allelic genes. *Science* 100:595–597, 1944.
- [32463] 6015.Wang, X.; Chan, S. J.; Eddy, R. L.; Byers, M. G.; Fukushima, Y.; Henry, W. M.; Haley, L. L.; Steiner, D. F.; Shows, T. B.: Chromosome assignment of cathepsin B (CTSB) to 8p22 and cathepsin H (CTSH) to 15q24–q25. (Abstract) *Cytogenet. Cell Genet.* 46: 710–711, 1987.
- [32464] 6016.Tyynela, J.; Sohar, I.; Sleat, D. E.; Gin, R. M.; Donnelly, R. J.; Baumann, M.; Haltia, M.; Lobel, P.: A mutation

in the ovine cathepsinD gene causes a congenital lysosomal storage disease with profound neurodegeneration. EMBO J. 19: 2786–2792, 2000.

- [32465] 6017. Gelb, B. D.; Shi, G.-P.; Heller, M.; Weremowicz, S.; Morton, C.; Desnick, R. J.; Chapman, H. A.: Structure and chromosomal assignment of the human cathepsin K gene. Genomics 41: 258–262, 1997.
- [32466] 6018. Shiang, R.; Lidral, A. C.; Ardinger, H. H.; Buetow, K. H.; Romitti, P. A.; Munger, R. G.; Murray, J. C.: Association of transforming growth-factor alpha gene polymorphisms with nonsyndromic cleft palate only (CPO). Am. J. Hum. Genet. 53: 836–843, 1993.
- [32467] 6019. Shields, E. D.; Bixler, D.; Fogh-Andersen, P.: Facial clefts in Danish twins. Cleft Palate J. 16: 1–6, 1979.
- [32468] 6020. Pollack, M. S.; Ochs, H. D.; Dupont, B.: HLA typing of cultured amniotic cells for the prenatal diagnosis of complement C4 deficiency. Clin. Genet. 18: 197–200, 1980.
- [32469] 6021. Porter, R. R.; Reid, K. B. M.: The biochemistry of complement. Nature 275: 699–704, 1978.
- [32470] 6022. Rittner, C.; Bertrams, J.: On the significance of C2, C4, and factor B polymorphisms in disease. Hum. Genet. 56: 235–247, 1981.
- [32471] 6023. Rittner, C.; Hauptmann, G.; Grosse-Wilde, H.;

Grosshans, E.; Tongio, M. M.; Mayer, S.: Linkage between HL-A (major histocompatibility complex) and genes controlling the fourth component of complement. In: Histocompatibility Testing 1975. Copenhagen: Munksgaard (pub.) 1976. Pp. 945–953.

[32472] 6024. Roos, M. H.; Mollenhauer, E.; Demant, P.; Rittner, C.: A molecular basis for the two locus model of human complement component C4. *Nature* 298:854–856, 1982.

[32473] 6025. Rosenfeld, S. I.; Ruddy, S.; Austen, K. F.: Structural polymorphism of the fourth component of human complement. *J. Clin. Invest.* 48:2283–2292, 1969.

[32474] 6026. Roychoudhury, A. K.; Nei, M.: Human Polymorphic Genes: World Distribution. New York: Oxford Univ. Press (pub.) 1988.

[32475] 6027. Schaller, J. G.; Gilliland, B. G.; Ochs, H. D.; Leddy, J. P.; Agodoa, L. C. Y.; Rosenfeld, S. I.: Severe systemic lupus erythematosus with nephritis in a boy with deficiency of the fourth component of complement. *Arthritis Rheum.* 20: 1519–1525, 1977.

[32476] 6028. Shevach, E. M.; Frank, M. M.; Green, I.: Linkage of gene controlling the synthesis of the fourth component of complement to the major histocompatibility complex of the guinea pig. *Immunogenetics* 3: 595–602, 1976.

- [32477] 6029.Shreffler, D. C.: The S region of the mouse major histocompatibilitycomplex (H-2): genetic variation and functional role in complementsystem. Transplant. Rev. 32: 140-167, 1976.
- [32478] 6030.Torisu, M.; Sonozaki, H.; Inai, S.; Arata, M.: Deficiency ofthe fourth component of complement in man. J. Immunogenet. 104:728-737, 1970.
- [32479] 6031.Wank, R.; Schendel, D. J.; O'Neill, G. J.; Riethmuller, G.; Held,E.; Feucht, H. E.: Rare variant of complement C4 is seen in highfrequency in patients with primary glomerulonephritis. Lancet I:872-874, 1984.
- [32480] 6032.Welch, T. R.; Beischel, L.: C4 uremic variant: an acquired C4allotype. Immunogenetics 22: 553-562, 1985.
- [32481] 6033.Welch, T. R.; Beischel, L. S.; Choi, E.; Balakrishnan, K.; Bishof,N. A.: Uniparental isodisomy 6 associated with deficiency of thefourth component of complement. J. Clin. Invest. 86: 675-678, 1990.
- [32482] 6034.Whitehead, A. S.; Colten, H. R.; Chang, C. C.; Demars, R.: Localizationof the human MHC-linked complement genes between HLA-B and HLA-DRby using HLA mutant cell lines. J. Immun. 134: 641-643, 1985.
- [32483] 6035.Wilson, W. A.; Perez, M. C.: Complete C4B deficiency in blackAmericans with systemic lupus erythematosus. J.

Rheum. 15: 1855–1858,1988.

- [32484] 6036.Andersson, A.; Dahlback, B.; Hanson, C.; Hillarp, A.; Levan, G.;Szpirer, J.; Szpirer, C.: Genes for C4b-binding protein alpha- andbeta-chains (C4BPA and C4BPB) are located on chromosome 1, band 1q32,in humans and on chromosome 13 in rats. Somat. Cell Molec. Genet. 16:493–500, 1990.
- [32485] 6037.Aso, T.; Okamura, S.; Matsuguchi, T.; Sakamoto, N.; Sata, T.; Niho,Y.: Genomic organization of the alpha chain of the human C4b-bindingprotein gene. Biochem. Biophys. Res. Commun. 174: 222–227, 1991.
- [32486] 6038.Barnum, S. R.; Kristensen, T.; Chaplin, D. D.; Seldin, M. F.; Tack,B. F.: Molecular analysis of the murine C4b-binding protein gene:chromosome assignment and partial gene organization. Biochemistry 28:8312–8317, 1989.
- [32487] 6039.Gigli, I.; Fujita, T.; Nussenzweig, V.: Modulation of the classicalpathway C3 convertase by plasma proteins C4 binding protein and C3binactivator. Proc. Nat. Acad. Sci. 76: 6596–6600, 1979.
- [32488] 6040.Hatch, J. A.; Atkinson, J. P.; Suarez, B. K.; Dykman, T. R.: Evaluationof linkage of the human C3b/C4b receptor to HLA. J. Immun. 132:2168–2169, 1984.

- [32489] 6041.Hing, S.; Day, A. J.; Linton, S. J.; Ripoche, J.; Sim, R. B.; Reid, K. B. M.; Solomon, E.: Assignment of complement components C4 binding protein (C4BP) and factor H (FH) to human chromosome 1q, using cDNA probes. *Ann. Hum. Genet.* 52: 117–122, 1988.
- [32490] 6042.Kaidoh, T.; Natsuume–Sakai, S.; Takahashi, M.: Murine binding protein of the fourth component of complement: structural polymorphism and its linkage to the major histocompatibility complex. *Proc. Nat. Acad. Sci.* 78: 3794–3798, 1981.
- [32491] 6043.Matsuguchi, T.; Okamura, S.; Aso, T.; Sata, T.; Niho, Y.: Molecular cloning of the cDNA coding for proline-rich protein (PRP): identity of PRP as C4b-binding protein. *Biochem. Biophys. Res. Commun.* 165:138–144, 1989.
- [32492] 6044.Nagasawa, S.; Stroud, R. M.: Purification and characterization of a macromolecular weight cofactor for C3b-inactivator, C4bC3bINA-cofactor, of human plasma. *Molec. Immun.* 17: 1365–1372, 1980.
- [32493] 6045.Rey–Campos, J.; Rubinstein, P.; Rodriguez de Cordoba, S.: A physical map of the human regulator of complement activation gene cluster linking the complement genes CR1, CR2, DAF, and C4BP. *J. Exp. Med.* 167:664–669, 1988.

- [32494] 6046.Rodriguez de Cordoba, S.; Dykman, T. R.; Ginsberg-Fellner, F.;Ercilla, G.; Aqua, M.; Atkinson, J. P.; Rubinstein, P.: Evidencefor linkage between the loci coding for the binding protein for thefourth component of human complement (C4BP) and for the C3b/C4b receptor. Proc.Nat. Acad. Sci. 81: 7890–7892, 1984.
- [32495] 6047.Rodriguez de Cordoba, S.; Ferreira, A.; Nussen-zweig, V.; Rubinstein,P.: Genetic polymorphism of human C4-binding protein. J. Immun. 131:1565–1569, 1983.
- [32496] 6048.Rodriguez de Cordoba, S.; Lublin, D. M.; Rubinstein, P.; Atkinson,J. P.: Human genes for three complement components that regulatethe activation of C3 are tightly linked. J. Exp. Med. 161: 1189–1195,1985.
- [32497] 6049.Sullivan, F. X.; Kumar, R.; Kriz, R.; Stahl, M.; Xu, G.–Y.; Rouse,J.; Chang, X.; Boodhoo, A.; Potvin, B.; Cum-ming, D. A.: Molecularcloning of human GDP-mannose 4,6-dehydratase and reconstitution ofGDP-fucose biosyn-thesis in vitro. J. Biol. Chem. 273: 8193–8202,1998.
- [32498] 6050.Justice, M. J.; Siracusa, L. D.; Gilbert, D. J.; Heis-terkamp, N.;Groffen, J.; Chada, K.; Silan, C. M.; Copeland, N. G.; Jenkins, N.A.: A genetic linkage map of mouse chromosome 10: localization ofeighteen molecular mark-ers using a single interspecific backcross. Genetics

125:855–866, 1990.

- [32499] 6051. Mouillet–Richard, S.; Ermonval, M.; Chebassier, C.; Laplanche, J. L.; Lehmann, S.; Launay, J. M.; Kellermann, O.: Signal transduction through prion protein. *Science* 289: 1925–1928, 2000.
- [32500] 6052. Parravicini, V.; Gadina, M.; Kovarova, M.; Odom, S.; Gonzalez–Espinosa, C.; Furumoto, Y.; Saitoh, S.; Samelson, L. E.; O'Shea, J. J.; Rivera, J.: Fyn kinase initiates complementary signals required for IgE–dependent mast cell degranulation. *Nature Immun.* 3: 741–748, 2002.
- [32501] 6053. Casari, G.; Barlassina, C.; Cusi, D.; Zagato, L.; Muirhead, R.; Righetti, M.; Nembri, P.; Amar, K.; Gatti, M.; Macciardi, F.; Binelli, G.; Bianchi, G.: Association of the alpha-adducin locus with essential hypertension. *Hypertension* 25: 320–326, 1995.
- [32502] 6054. Crosby, A. H.; Edwards, S. J.; Murray, J. C.; Dixon, M. J.: Genomic organization of the human osteopontin gene: exclusion of the locus from a causative role in the pathogenesis of dentinogenesis imperfecta type II. *Genomics* 27: 155–160, 1995.
- [32503] 6055. Abernethy, T. J.; Avery, O. T.: The occurrence during acute infection of a protein not normally present in the blood. I. Distribution of the reactive protein in patients'

sera and the effect of calcium on the flocculation reaction with C polysaccharide of pneumococcus. J. Exp. Med. 73: 173–182, 1941.

[32504] 6056. Floyd-Smith, G. A.; Whitehead, A. S.; Colten, H. R.; Francke, U.: The human C-reactive protein gene (CRP) and serum amyloid P component gene (APCS) are located on the proximal long arm of chromosome 1. Immunogenetics 24:171–176, 1986.

[32505] 6057. Tillett, W. S.; Francis, T., Jr.: Serological reactions in pneumonia with a nonprotein somatic fraction of pneumococcus. J. Exp. Med. 52:561–585, 1930.

[32506] 6058. Benger, J. C.; Teshima, I.; Walter, M. A.; Brubacher, M. G.; Daouk, G. H.; Cox, D. W.: Localization and genetic linkage of the human immunoglobulin heavy chain genes and the creatine kinase brain (CKB) gene: identification of a hot spot for recombination. Genomics 9:614–622, 1991.

[32507] 6059. Brubacher, M. G.; Benger, J. C.; Billingsley, G. D.; Hofker, M. H.; Nakamura, Y.; White, R.; Cox, D. W.: A genetic linkage map of the distal region of human chromosome 14. (Abstract) Am. J. Hum. Genet. 45 (suppl.): A133 only, 1989.

[32508] 6060. Hoo, J. J.; Goedde, H. W.: Determination of brain type creatine kinase for diagnosis of perinatal asphyxia–

–choice of method. (Letter) *Pediat.Res.* 16: 806 only, 1982.

[32509] 6061.Kaye, F. J.; McBride, O. W.; Battey, J. F.; Gazdar, A. F.; Sausville,E. A.: Human creatine kinase–B complementary DNA: nucleotide sequence, gene expression in lung cancer, and chromosomal assignment to two distinct loci. *J. Clin. Invest.* 79: 1412–1420, 1987.

[32510] 6062.Ma, T. S.; Ifegwu, J.; Watts, L.; Siciliano, M. J.; Roberts, R.;Perryman, M. B.: Serial Alu sequence transposition interrupting a human B creatine kinase pseudogene. *Genomics* 10: 390–399, 1991.

[32511] 6063.Mariman, E. C. M.; Schepens, J. T. G.; Wieringa, B.: Complete nucleotide sequence of the human creatine kinase B gene. *Nucleic Acids Res.* 17: 6385 only, 1989.

[32512] 6064.Pfeiffer, F. E.; Homburger, H. A.; Yanagihara, T.: Creatine kinase BB isoenzyme in CSF in neurologic diseases: measurement by radioimmunoassay. *Arch.Neurol.* 40: 169–172, 1983.

[32513] 6065.Stallings, R. L.; Olson, E.; Strauss, A. W.; Thompson, L. H.; Bachinski, L. L.; Siciliano, M. J.: Human creatine kinase genes on chromosomes 15 and 19, and proximity of the gene for the muscle form to the genes for apolipoprotein C2 and excision repair. *Am. J. Hum. Genet.*

43:144–151, 1988.

- [32514] 6066.Villarreal–Levy, G.; Ma, T. S.; Kerner, S. A.; Roberts, R.; Perryman,M. B.: Human creatine kinase: isolation and sequence analysis of cDNA clones for the B subunit, development of subunit specific probes, and determination of gene copy number. *Biochem. Biophys. Res. Commun.* 144:1116–1127, 1987.
- [32515] 6067.Bark, C. J.: Mitochondrial creatine kinase: a poor prognostic sign. *J.A.M.A.* 243: 2058–2060, 1980.
- [32516] 6068.Steeghs, K.; Merks, G.; Wieringa, B.: The ubiquitous mitochondrial creatine kinase gene maps to a conserved region on human chromosome 15q15 and mouse chromosome 2 bands F1–F3. *Genomics* 24: 193–195, 1994.
- [32517] 6069.Klein, S. C.; Haas, R. C.; Perryman, M. B.; Billadello, J. J.; Strauss, A. W.: Regulatory element analysis and structural characterization of the human sarcomeric mitochondrial creatine kinase gene. *J. Biol.Chem.* 266: 18058–18065, 1991.
- [32518] 6070.Richard, I.; Devaud, C.; Cherif, D.; Cohen, D.; Beckmann, J. S.: The gene for creatine kinase, mitochondrial 2 (sarcomeric; CKMT2), maps to chromosome 5q13.3. *Genomics* 18: 134–136, 1993.
- [32519] 6071.Bailly, J.; MacKenzie, A. E.; Leblond, S.; Korneluk, R.

G.: Assessment of a creatine kinase isoform M defect as a cause of myotonic dystrophy and the characterization of two novel CKMM polymorphisms. *Hum. Genet.* 86:457–462, 1991.

[32520] 6072. Böhner, J.; Stein, W.; Kuhlmann, E.; Eggstein, M.: Serum creatine kinase BB linked to immunoglobulin G. *Clin. Chim. Acta* 97: 83–88, 1979.

[32521] 6073. Dawson, D. M.; Eppenberger, H. M.; Eppenberger, M. E.: Multiple molecular forms of creatine kinases. *Ann. N.Y. Acad. Sci.* 151: 616–626, 1968.

[32522] 6074. Schonk, D.; van Dijk, P.; Riegmann, P.; Trapman, J.; Holm, C.; Willcocks, T. C.; Sillekens, P.; van Venrooij, W.; Wimmer, E.; Geurts van Kessel, A.; Ropers, H.-H.; Wieringa, B.: Assignment of seven genes to distinct intervals on the midportion of human chromosome 19q surrounding the myotonic dystrophy gene region. *Cytogenet. Cell Genet.* 54: 15–19, 1990.

[32523] 6075. Coles, L. S.; Diamond, P.; Occhiodoro, F.; Vadas, M. A.; Shannon, M. F.: Cold shock domain proteins repress transcription from the GM-CSF promoter. *Nucleic Acids Res.* 24: 2311–2317, 1996.

[32524] 6076. Didier, D. K.; Schiffenbauer, J.; Woulfe, S. L.; Zacheis, M.; Schwartz, B. D.: Characterization of the cDNA encoding

a protein binding to the major histocompatibility complex class II Y box. *Proc. Nat. Acad. Sci.* 85: 7322–7326, 1988.

[32525] 6077. Kudo, S.; Mattei, M.-G.; Fukuda, M.: Characterization of the gene for dbpA, a family member of the nucleic-acid-binding proteins containing a cold-shock domain. *Europ. J. Biochem.* 231: 72–82, 1995.

[32526] 6078. Makino, Y.; Ohga, T.; Toh, S.; Koike, K.; Okumura, K.; Wada, M.; Kuwano, M.; Kohno, K.: Structural and functional analysis of the human Y-box binding protein (YB-1) gene promoter. *Nucleic Acid Res.* 24: 1873–1878, 1996.

[32527] 6079. Sakura, H.; Maekawa, T.; Imamoto, F.; Yasuda, K.; Ishii, S.: Two human genes isolated by a novel method encode DNA-binding proteins containing a common region of homology. *Gene* 73: 499–507, 1988.

[32528] 6080. Spitkovsky, D. D.; Royer-Pokora, B.; Delius, H.; Kisseljov, F.; Jenkins, N. A.; Gilbert, D. J.; Copeland, N. G.; Royer, H.-D.: Tissue restricted expression and chromosomal localization of the YB-1 gene encoding a 42kD nuclear CCAAT binding protein. *Nucleic Acids Res.* 20: 797–803, 1992.

[32529] 6081. Levi-Strauss, M.; Carroll, M. C.; Steinmetz, M.; Meo, T.: A previously undetected MHC gene with an unusual periodic structure. *Science* 240: 201–204, 1988.

- [32530] 6082.Speiser, P. W.; White, P. C.: Structure of the human RD gene:a highly conserved gene in the class III region of the major histocompatibilitycomplex. DNA 8: 745–751, 1989.
- [32531] 6083.Surowy, C. S.; Gosink, J. J.; Spritz, R. A.: cDNA cloning of thehuman 'RD' protein, a possible novel RNA-binding protein. (Abstract) Am.J. Hum. Genet. 43: A203 only, 1988.
- [32532] 6084.Yamaguchi, Y.; Takagi, T.; Wada, T.; Yano, K.; Furuya, A.; Sugimoto,S.; Hasegawa, J.; Handa, H.: NELF, a multisubunit complex containingRD, cooperates with DSIF to repress RNA polymerase II elongation. Cell 97:41–51, 1999.
- [32533] 6085.Gross, M.–S.; Guyonnet–Duperat, V.; Porchet, N.; Bernheim, A.;Aubert, J. P.; Nguyen, V. C.: Mucin 4 (MUC4) gene: regional assignment(3q29) and RFLP analysis. Ann. Genet. 35: 21–26, 1992.
- [32534] 6086.Porchet, N.; Van Cong, N.; Dufosse, J.; Audie, J. P.; Guyonnet–Duperat,V.; Gross, M. S.; Denis, C.; Degand, P.; Bernheim, A.; Aubert, J.P.: Molecular cloning and chromosomal localization of a novel humantracheo–bronchial mucin cDNA containing tandemly repeated sequencesof 48 base pairs. Biochem. Biophys. Res. Commun. 175:

414–422,1991.

- [32535] 6087. Van Cong, N.; Aubert, J.-P.; Gross, M.-S.; Porchet, N.; Degand, P.; Bernheim, A.: Tracheobronchial mucin 4 (MUC4) gene: assignment to 3q29 and polymorphism of VNTR type. (Abstract) Cytogenet. Cell Genet. 58: 1879–1880, 1991.
- [32536] 6088. Pigny, P.; Pratt, W. S.; Laine, A.; Leclercq, A.; Swallow, D. M.; Nguyen, V. C.; Aubert, J. P.; Porchet, N.: The MUC5AC gene: RFLP analysis with the Jer58 probe. Hum. Genet. 96: 367–368, 1995.
- [32537] 6089. Garcia, J. V.; Jones, C.; Miller, A. D.: Localization of the amphotropic murine leukemia virus receptor gene to the pericentromeric region of human chromosome 8. J. Virol. 65: 6316–6319, 1991.
- [32538] 6090. van Zeijl, M.; Johann, S. V.; Closs, E.; Cunningham, J.; Eddy, R.; Shows, T. B.; O'Hara, B.: A human amphotropic retrovirus receptor is a second member of the gibbon ape leukemia virus receptor family. Proc. Nat. Acad. Sci. 91: 1168–1172, 1994.
- [32539] 6091. van Zeijl, M.; Johann, S. V.; Eddy, R. L.; Shows, T. B.; O'Hara, B.: Assignment of GLVR2, a receptor for murine amphotropic virus to human chromosome 8. (Abstract) Human Genome Mapping Workshop 93 18 only, 1993.

- [32540] 6092.Cawthon, R. M.; O'Connell, P.; Buchberg, A. M.; Viskochil, D.;Weiss, R. B.; Culver, M.; Stevens, J.; Jenkins, N. A.; Copeland, N.G.; White, R.: Identification and characterization of transcriptsfrom the neurofibromatosis 1 region: the sequence and genomic structureof EVI2 and mapping of other transcripts. *Genomics* 7: 555–565, 1990.
- [32541] 6093.Collins, F. S.: Personal Communication. Bethesda, Md. 11/17/1993.
- [32542] 6094.O'Connell, P.; Buchberg, A.; Cawthon, R. M.; Culver, M.; Stevens,J.; Viskochil, D.; Carey, J. C.; Fountain, J.; Wallace, M.; Jenkins,N.; Copeland, N.; Collins, F. S.; White, R.: The human homologueof the murine Evi–2 oncogene lies between von Recklinghausen NF translocationbreakpoints. (Abstract) *Am. J. Hum. Genet.* 45 (suppl.): A210 only,1989.
- [32543] 6095.O'Connell, P.; Viskochil, D.; Buchberg, A. M.; Fountain, J.; Cawthon,R. M.; Culver, M.; Stevens, J.; Rich, D. C.; Ledbetter, D. H.; Wallace,M.; Carey, J. C.; Jenkins, N. A.; Copeland, N. G.; Collins, F. S.;White, R.: The human homolog of murine Evi–2 lies between two vonRecklinghausen neurofibromatosis translocations. *Genomics* 7: 547–554,1990.

- [32544] 6096. Blanck, O.; Perrin, C.; Mziaut, H.; Darbon, H.; Mattei, M. G.; Miquelis, R.: Molecular cloning, cDNA analysis, and localization of a monomer of the N-acetylglucosamine-specific receptor of the thyroid, NAGR1, to chromosome 19p13.3-13.2. *Genomics* 21: 18-26, 1994. Note: Erratum: *Genomics* 27: 561 only, 1995.
- [32545] 6097. Datar, K. V.; Dreyfuss, G.; Swanson, M. S.: The human hnRNP M proteins: identification of a methionine/arginine-rich repeat motif in ribonucleoproteins. *Nucleic Acids Res.* 21: 439-446, 1993.
- [32546] 6098. Hull, E.; Sarkar, M.; Spruijt, M. P. N.; Hoppener, J. W. M.; Dunn, R.; Schachter, H.: Organization and localization to chromosome 5 of the human UDP-N-acetylglucosamine:alpha-3-D-mannoside beta-1,2-N-acetylglucosaminyltransferase 1 gene. *Biochem. Biophys. Res. Commun.* 176: 608-615, 1991.
- [32547] 6099. Ioffe, E.; Stanley, P.: Mice lacking N-acetylglucosaminyltransferase I activity die at mid-gestation, revealing an essential role for complex or hybrid N-linked carbohydrates. *Proc. Nat. Acad. Sci.* 91: 728-732, 1994.
- [32548] 6100. Kumar, R.; Stanley, P.: Transfection of a human gene

that corrects the Lec1 glycosylation defect: evidence for transfer of the structural gene for N-acetylglucosaminyltransferase I. *Molec. Cell. Biol.* 9:5713–5717, 1989. Note: Erratum: *Molec. Cell. Biol.* 10: 3857 only, 1990.

[32549] 6101. Kumar, R.; Yang, J.; Eddy, R. L.; Byers, M. G.; Shows, T. B.; Stanley, P.: Cloning and expression of the murine gene and chromosomal location of the human gene encoding N-acetylglucosaminyltransferase I. *Glycobiology* 2:383–393, 1992.

[32550] 6102. Kumar, R.; Yang, J.; Larsen, R. D.; Stanley, P.: Cloning and expression of N-acetylglucosaminyltransferase I, the medial Golgi transferase that initiates complex N-linked carbohydrate formation. *Proc. Nat. Acad. Sci.* 87: 9948–9952, 1990.

[32551] 6103. Metzler, M.; Gertz, A.; Sarkar, M.; Schachter, H.; Schrader, J.W.; Marth, J. D.: Complex asparagine-linked oligosaccharides are required for morphogenic events during post-implantation development. *EMBOJ.* 13: 2056–2065, 1994.

[32552] 6104. Pownall, S.; Kozak, C. A.; Schappert, K.; Sarkar, M.; Hull, E.; Schachter, H.; Marth, J. D.: Molecular cloning and characterization of the mouse UDP–

N-acetylglucosamine:alpha-3-D-mannoside beta-1,2-N-acetylglucosaminyltransferase I gene. Genomics 12: 699-704, 1992.

[32553] 6105. Puthalakath, H.; Burke, J.; Gleeson, P. A.: Glycosylation defect in lec1 Chinese hamster ovary mutant is due to a point mutation in N-acetylglucosaminyltransferase 1 gene. J. Biol. Chem. 271: 27818-27822, 1996.

[32554] 6106. Shows, T. B.: Personal Communication. Buffalo, N.Y. 4/13/1999.

[32555] 6107. Yip, B.; Chen, S.-H.; Mulder, H.; Hoppener, J. W. M.; Schachter, H.: Organization of the human beta-1,2-N-acetylglucosaminyltransferase I gene (MGAT1), which controls complex and hybrid N-glycan synthesis. Biochem. J. 321: 465-474, 1997.

[32556] 6108. Jaiswal, A. K.; Bell, D. W.; Radjendirane, V.; Testa, J. R.: Localization of human NQO1 gene to chromosome 16q22 and NQO2-6p25 and associated polymorphisms. Pharmacogenetics 9: 413-418, 1999.

[32557] 6109. Jaiswal, A. K.; Burnett, P.; Adesnik, M.; McBride, O. W.: Nucleotide and deduced amino acid sequence of a human cDNA (NQO2) corresponding to a second member of the NAD(P)H:quinone oxidoreductase gene family: extensive polymorphism at the NQO2 gene locus on

chromosome 6. (Abstract) *Biochemistry* 29:1899–1906, 1990.

[32558] 6110. Tan, J.; D'Agostaro, G. A. F.; Bendiak, B.; Reck, F.; Sarkar, M.; Squire, J. A.; Leong, P.; Schachter, H.: The human UDP-N-acetylglucosamine:alpha-6-D-mannoside-beta-1,2-N-acetylglucosaminyltransferase II gene (MGAT2): cloning of genomic DNA, localization to chromosome 14q21, expression in insect cells and purification of the recombinant protein. *Eur. J. Biochem.* 231: 317–328, 1995.

[32559] 6111. Kondo, M.; Scherer, D. C.; Miyamoto, T.; King, A. G.; Akashi, K.; Sugamura, K.; Weissman, I. L.: Cell-fate conversion of lymphoid-committed progenitors by instructive actions of cytokines. *Nature* 407: 383–386, 2000.

[32560] 6112. Le Beau, M. M.; Westbrook, C. A.; Diaz, M. O.; Larson, R. A.; Rowley, J. D.; Gasson, J. C.; Golde, D. W.; Sherr, C. J.: Evidence for the involvement of GM-CSF and FMS in the deletion (5q) in myeloid disorders. *Science* 231: 984–987, 1986.

[32561] 6113. Morgan, R.; Hecht, B. K.; Sandberg, A. A.; Hecht, F.; Smith, S. D.: Chromosome 5q35 breakpoint in malignant histiocytosis. (Letter) *New Eng. J. Med.* 314: 1322 only,

1986.

- [32562] 6114.Ridge, S. A.; Worwood, M.; Oscier, D.; Jacobs, A.; Padua, R. A.: FMS mutations in myelodysplastic, leukemic, and normal subjects. *Proc.Nat. Acad. Sci.* 87: 1377–1380, 1990.
- [32563] 6115.Roberts, W. M.; Look, A. T.; Roussel, M. F.; Sherr, C. J.: Tandemlinkage of human CSF–1 receptor (c–fms) and PDGF receptor genes. *Cell* 55:655–661, 1988.
- [32564] 6116.Sapi, E.; Flick, M. B.; Kacinski, B. M.: The first intron of human c–fms proto–oncogene contains a processed pseudogene (RPL7P) for ribosomal protein L7. *Genomics* 22: 641–645, 1994.
- [32565] 6117.Verbeek, J. S.; Roebroek, A. J. M.; van den Ouweland, A. M. W.; Bloemers, H. P. J.; Van de Ven, W. J. M.: Human c–fms proto–oncogene: comparative analysis with an abnormal allele. *Molec. Cell. Biol.* 5:422–426, 1985.
- [32566] 6118.Verbeek, J. S.; van Heerikhuizen, H.; de Pauw, B. E.; Haanen, C.; Bloemers, H. P. J.; Van de Ven, W. J. M.: A hereditary abnormal c–fms proto–oncogene in a patient with acute lymphocytic leukaemia and congenital hypothyroidism. *Brit. J. Haemat.* 61: 135–138, 1985.
- [32567] 6119.Yarden, Y.; Ullrich, A.: Growth factor receptor tyrosine kinases. *Ann.Rev. Biochem.* 57: 443–478, 1988.

- [32568] 6120. Brown, J. R.; Ye, H.; Bronson, R. T.; Dikkes, P.; Greenberg, M.E.: A defect in nurturing in mice lacking the immediate early gene *fosB*. *Cell* 86: 297–309, 1996.
- [32569] 6121. Forsdyke, D.: Personal Communication. Kingston, Ontario, Canada 6/8/1992.
- [32570] 6122. Lazo, P. S.; Dorfman, K.; Noguchi, T.; Mattei, M. G.; Bravo, R.: Structure and mapping of the *fosB* gene: *FosB* downregulates the activity of the *fosB* promoter. *Nucleic Acids Res.* 20: 343–350, 1992.
- [32571] 6123. Nakabeppu, Y.; Nathans, D.: A naturally occurring truncated form of *FosB* that inhibits *Fos/Jun* transcriptional activity. *Cell* 64: 751–759, 1991.
- [32572] 6124. Siderovski, D. P.; Blum, S.; Forsdyke, R. E.; Forsdyke, D. R.: A set of human putative lymphocyte G0/G1 switch genes includes genes homologous to rodent cytokine and zinc finger protein–encoding genes. *Dev Cell Biol.* 9: 579–587, 1990.
- [32573] 6125. Kelz, M. B.; Chen, J.; Carlezon, W. A., Jr.; Whisler, K.; Gilden, L.; Beckmann, A. M.; Steffen, C.; Zhang, Y.-J.; Marotti, L.; Self, D. W.; Tkatch, T.; Baranauskas, G.; Surmeier, D. J.; Neve, R. L.; Duman, R. S.; Picciotto, M. R.; Nestler, E. J.: Expression of the transcription factor *delta-FosB* in the brain controls sensitivity to cocaine. *Nature*

401: 272–276, 1999.

[32574] 6126.Boyd, M. T.; Vlatkovic, N.; Haines, D. S.: A novel cellular protein(MTBP) binds to MDM2 and induces a G1 arrest that is suppressed byMDM2. J. Biol. Chem. 275: 31883–31890, 2000.

[32575] 6127.Bueso–Ramos, C. E.; Yang, Y.; deLeon, E.; McCown, P.; Stass, S.A.; Albitar, M.: The human MDM–2 oncogene is overexpressed in leukemias. Blood 82:2617–2623, 1993.

[32576] 6128.Buschmann, T.; Fuchs, S. Y.; Lee, C.–G.; Pan, Z.–Q.; Ronai, Z.: SUMO–1 modification of Mdm2 prevents its self–ubiquitination andincreases Mdm2 ability to ubiquitinate p53. Cell 101: 753–762, 2000.Note: Erratum: Cell 107: 549 only, 2001.

[32577] 6129.Hoffmeyer, S.; Burk, O.; von Richter, O.; Arnold, H. P.; Brockmoller,J.; Johne, A.; Cascorbi, I.; Gerloff, T.; Roots, I.; Eichelbaum, M.;Brinkmann, U.: Functional polymorphisms of the human multidrug–resistancegene: multiple sequence variations and correlation of one allele withP–glycoprotein expression and activity in vivo. Proc. Nat. Acad.Sci. 97: 3473–3478, 2000.

[32578] 6130.Fruman, D. A.; Snapper, S. B.; Yballe, C. M.; Davidson, L.; Yu,J. Y.; Alt, F. W.; Cantley, L. C.: Impaired B cell development andproliferation in absence of phospho–

inositide 3-kinase p85- α . *Science* 283:393–397, 1999.

- [32579] 6131. Fukao, T.; Tanabe, M.; Terauchi, Y.; Ota, T.; Matsuda, S.; Asano, T.; Kadowaki, T.; Takeuchi, T.; Koyasu, S.: PI3K-mediated negative feedback regulation of IL-12 production in DCs. *Nature Immun.* 3:875–881, 2002.
- [32580] 6132. Fukao, T.; Yamada, T.; Tanabe, M.; Terauchi, Y.; Ota, T.; Takayama, T.; Asano, T.; Takeuchi, T.; Kadowaki, T.; Hata, J.; Koyasu, S.: Selective loss of gastrointestinal mast cells and impaired immunity in PI3K-deficient mice. *Nature Immun.* 3: 295–304, 2002.
- [32581] 6133. Hoyle, J.; Yulug, I. G.; Egan, S. E.; Fisher, E. M. C.: The gene that encodes the phosphatidylinositol-3 kinase regulatory subunit (p85- α) maps to chromosome 13 in the mouse. *Genomics* 24: 400–402, 1994.
- [32582] 6134. Otsu, M.; Hiles, I.; Gout, I.; Fry, M. J.; Ruiz-Larrea, F.; Panayotou, G.; Thompson, A.; Dhand, R.; Hsuan, J.; Totty, N.; Smith, A. D.; Morgan, S. J.; Courtneidge, S. A.; Parker, P. J.; Waterfield, M. D.: Characterization of two 85 kd proteins that associate with receptor tyrosine kinases, middle-T/pp60(c-src) complexes, and PI3-kinase. *Cell* 65: 91–104, 1991.
- [32583] 6135. Skolnik, E. Y.; Margolis, B.; Mohammadi, M.; Lowen-

stein, E.; Fischer, R.; Drepps, A.; Ullrich, A.; Schlessinger, J.: Cloning of PI3-kinase-associated p85 utilizing a novel method for expression/cloning of target proteins for receptor tyrosine kinases. *Cell* 65: 83–90, 1991.

[32584] 6136. Suzuki, H.; Terauchi, Y.; Fujiwara, M.; Aizawa, S.; Yazaki, Y.; Kadowaki, T.; Koyasu, S.: Xid-like immunodeficiency in mice with disruption of the p85- α subunit of phosphoinositide 3-kinase. *Science* 283:390–392, 1999.

[32585] 6137. Terauchi, Y.; Tsuji, Y.; Satoh, S.; Minoura, H.; Murakami, K.; Okuno, A.; Inukai, K.; Asano, T.; Kaburagi, Y.; Ueki, K.; Nakajima, H.; Hanafusa, T.; and 18 others: Increased insulin sensitivity and hypoglycaemia in mice lacking the p85- α subunit of phosphoinositide 3-kinase. *Nature Genet.* 21: 230–235, 1999.

[32586] 6138. Volinia, S.; Patracchini, P.; Otsu, M.; Hiles, I.; Gout, I.; Calzolari, E.; Bernardi, F.; Rooke, L.; Waterfield, M. D.: Chromosomal localization of human p85- α , a subunit of phosphatidylinositol 3-kinase, and its homologue p85- β . *Oncogene* 7: 789–793, 1992.

[32587] 6139. Bonne, G.; Di Barletta, M. R.; Varnous, S.; Becane, H.-M.; Hammouda, E.-H.; Merlini, L.; Muntoni, F.; Greenberg, C. R.; Gary, F.; Urtizberea, J.-A.; Duboc, D.; Fardeau, M.; Toniolo, D.; Schwartz, K.: Mutations in the gene encod-

ing lamin A/C cause autosomal dominant Emery–Dreifussmuscular dystrophy. *Nature Genet.* 21: 285–288, 1999.

- [32588] 6140. Budarf, M. L.; Konkle, B. A.; Ludlow, L. B.; Michaud, D.; Li, M.; Yamashiro, D. J.; McDonald–McGinn, D.; Zackai, E. H.; Driscoll, D. A.: Identification of a patient with Bernard–Soulier syndrome and a deletion in the DiGeorge/velo–cardio–facial chromosomal region in 22q11.2. *Hum. Molec. Genet.* 4: 763–766, 1995.
- [32589] 6141. Kelly, M. D.; Essex, D. W.; Shapiro, S. S.; Meloni, F. J.; Druck, T.; Huebner, K.; Konkle, B. A.: Complementary DNA cloning of the alternatively expressed endothelial cell glycoprotein Ib–beta (GPIb–beta) and localization of the GPIb–beta gene to chromosome 22. *J. Clin. Invest.* 93: 2417–2424, 1994.
- [32590] 6142. Kunishima, S.; Lopez, J. A.; Kobayashi, S.; Imai, N.; Kamiya, T.; Saito, H.; Naoe, T.: Missense mutations of the glycoprotein (GP)Ib–beta gene impairing the GPIb alpha/beta disulfide linkage in a family with giant platelet disorder. *Blood* 89: 2404–2412, 1997.
- [32591] 6143. Lopez, J. A.; Chung, D. W.; Fujikawa, K.; Hagen, F. S.; Davie, E. W.; Roth, G. J.: The alpha and beta chains of human platelet glycoprotein Ib are both transmembrane pro–

teins containing a leucine-rich aminoacid sequence. Proc. Nat. Acad. Sci. 85: 2135–2139, 1988.

[32592] 6144.Lopez, J. A.; Chung, D. W.; Fujikawa, K.; Hagen, F. S.; Papayannopoulou,T.; Roth, G. J.: Cloning of the alpha chain of human platelet glycoproteinIb: a transmembrane protein with homology to leucine-rich alpha 2-glycoprotein. Proc.Nat. Acad. Sci. 84: 5615–5619, 1987.

[32593] 6145.Roth, G. J.: The wanderings of a platelet gene: what is 'neo'Ib-beta telling us? (Editorial) J. Clin. Invest. 93: 2301–2302,1994.

[32594] 6146.Yagi, M.; Edelhoff, S.; Disteché, C. M.; Roth, G. J.: Structuralcharacterization and chromosomal location of the gene encoding humanplatelet glycoprotein Ib-beta. J. Biol. Chem. 269: 17424–17427,1994.

[32595] 6147.Zieger, B.; Hashimoto, Y.; Ware, J.: Alternative expression ofplatelet glycoprotein Ib-beta mRNA from an adjacent 5-prime gene withan imperfect polyadenylation signal sequence. J. Clin. Invest. 99:520–525, 1997.

[32596] 6148.Bakker, E.; Pearson, P. L.; Meera Khan, P.; Schreuder, G. M. T.;Madan, K.: Orientation of major histocompatibility (MHC) genes relativeto the centromere of human chromosome 6. Clin. Genet. 15: 198–202,1979.

- [32597] 6149.Bender, K.; Grzeschik, K. H.: Assignment of the genes for humanglyoxalase I to chromosome 6 and for human esterase D to chromosome13. *Cytogenet. Cell Genet.* 16: 93–96, 1976.
- [32598] 6150.Beretta, M.; Schiliro, G.; Russo, A.; Barbujani, G.; Mazzetti,P.; Russo, G.; Barrai, I.: A new rare variant of the glyoxalase I system of the red cell: GLO–Sicily. *Am. J. Hum. Genet.* 35: 1042–1047,1983.
- [32599] 6151.Blanche, H.; Zoghbi, H. Y.; Jabs, E. W.; de Gouyon, B.; Zunec,R.; Dausset, J.; Cann, H. M.: A centromere–based genetic map of the short arm of human chromosome 6. *Genomics* 9: 420–428, 1991.
- [32600] 6152.Carter, N. D.; West, C. M.; Bernard, J. M.; Farid, N. R.; Larsen,B.; Marshall, W. H.: Linkage of glyoxalase I and HLA in two Newfoundland communities. *Hum. Hered.* 28: 397–400, 1978.
- [32601] 6153.Giblett, E. R.; Lewis, M.: Gene linkage studies on glyoxalaseI. *Cytogenet. Cell Genet.* 16: 313 only, 1976.
- [32602] 6154.Goldman, D.; O'Brien, S. J.; Lucas–Derse, S.; Dean, M.: Linkagemapping of human polymorphic proteins identified by two–dimensionalelectrophoresis. *Genomics* 11: 875–884, 1991.
- [32603] 6155.Hansen, H. E.; Eriksen, B.: HLA–GLO linkage analysis

in 57 informative families. Hum. Hered. 29: 355–360, 1979.

[32604] 6156. Karlsson, S.; Arnason, A.; Jensson, O.: GLO polymorphism in Iceland. Hum. Hered. 30: 383–385, 1980.

[32605] 6157. Kavathas, P.; DeMars, R.: A new variant glyoxalase I allele that is readily detectable in stimulated lymphocytes and lymphoblastoid cell lines but not in circulating lymphocytes or erythrocytes. Am. J. Hum. Genet. 33: 935–945, 1981.

[32606] 6158. Kompf, J.; Bissbort, S.; Gussmann, S.; Ritter, H.: Polymorphism of red cell glyoxalase I (E.C.4.4.1.5), a new genetic marker in man: investigation of 169 mother–child combinations. Humangenetik 27: 141–143, 1975.

[32607] 6159. Kompf, J.; Bissbort, S.; Ritter, H.: Red cell glyoxalase I (E.C.4.4.1.5): formal genetics and linkage relations. Humangenetik 28: 249–251, 1975.

[32608] 6160. Kompf, J.; Siebert, G.; Ritter, H.; Heilbronner, H.; Schunter, F.; Wernet, P.; Gupta, D.; Moeller, H.: Data on linkage relations between GLO and 21-hydroxylase. Hum. Genet. 54: 419–420, 1980.

[32609] 6161. Meo, T.; Douglas, T.; Rijnbeek, A.–M.: Glyoxalase I polymorphism in the mouse: a new genetic marker linked to H-2. Science 198: 311–313, 1977.

- [32610] 6162.Olaisen, B.; Gedde-Dahl, T., Jr.; Thorsby, E.: Localization of the human GLO gene locus. *Hum. Genet.* 32: 301–304, 1976.
- [32611] 6163.Parr, C. W.; Bagster, I. A.; Welch, S. G.: Human red cell glyoxalase polymorphism. *Biochem. Genet.* 15: 109–114, 1977.
- [32612] 6164.Reinsmoen, N. L.; Friend, P. S.; Miller, W. V.; Burgdorf, A.; Giblett, E. R.; Yunis, E. J.: Inheritance of recombinant HLA-GLO haplotype suggesting the gene sequence. *Nature* 267: 276–278, 1977.
- [32613] 6165.Rubinstein, P.; Suciú-Foca, N.: Glyoxalase 1: a possible 'null' allele. *Hum. Hered.* 29: 217–220, 1979.
- [32614] 6166.Schimandle, C. M.; Vander Jagt, D. L.: Isolation and kinetic analysis of the multiple forms of glyoxalase-1 from human erythrocytes. *Arch. Biochem. Biophys.* 195: 261–268, 1979.
- [32615] 6167.Sparkes, R. S.; Sparkes, M. C.; Crist, M.; Anderson, C. E.: Glyoxalase 'null' allele in a new family: identification by abnormal segregation pattern and quantitative assay. *Hum. Genet.* 64: 146–147, 1983.
- [32616] 6168.Teng, Y. S.; Tan, S. G.; Lopez, C. G.: Red cell glyoxalase and placental soluble aconitase polymorphisms in the three major ethnic groups of Malaysia. *Jpn. J. Hum.*

Genet. 23: 211–215, 1978.

- [32617] 6169. Kartner, N.; Evernden-Porelle, D.; Bradley, G.; Ling, V.: Detection of P-glycoprotein in multidrug-resistant cell lines by monoclonal antibodies. *Nature* 316: 820–823, 1985.
- [32618] 6170. Kim, R. B.; Fromm, M. F.; Wandel, C.; Leake, B.; Wood, A. J. J.; Roden, D. M.; Wilkinson, G. R.: The drug transporter P-glycoprotein limits oral absorption and brain entry of HIV-1 protease inhibitors. *J. Clin. Invest.* 101: 289–294, 1998.
- [32619] 6171. Lankas, G. R.; Wise, L. D.; Cartwright, M. E.; Pippert, T.; Umbenhauer, D. R.: Placental P-glycoprotein deficiency enhances susceptibility to chemically induced birth defects in mice. *Reprod. Toxicol.* 12: 457–463, 1998.
- [32620] 6172. Martinsson, T.; Levan, G.: Localization of the multidrug resistance-associated 170 kDa P-glycoprotein gene to mouse chromosome 5 and to homogeneously staining regions in multidrug-resistant mouse cells by in situ hybridization. *Cytogenet. Cell Genet.* 45: 99–101, 1987.
- [32621] 6173. Mealey, K. L.; Bentjen, S. A.; Gay, J. M.; Cantor, G. H.: Ivermectin sensitivity in collies is associated with a deletion mutation of the *mdr1* gene. *Pharmacogenetics* 11: 727–733, 2001.

- [32622] 6174.Mickley, L. A.; Spengler, B. A.; Knutsen, T. A.; Biedler, J. L.;Fojo, T.: Gene rearrangement: a novel mechanism for MDR-1 gene activation. J.Clin. Invest. 99: 1947-1957, 1997.
- [32623] 6175.Pastan, I.; Gottesman, M.: Multiple-drug resistance in humancancer. New Eng. J. Med. 316: 1388-1393, 1987.
- [32624] 6176.Pulliam, J. D.; Seward, R. L.; Henry, R. T.; Steinberg, S. A.: Investigating ivermectin toxicity in collies. Vet. Med. 80: 33-40,1985.
- [32625] 6177.Randolph, G. J.; Beaulieu, S.; Pope, M.; Sugawara, I.; Hoffman,L.; Steinman, R. M.; Muller, W. A.: A physiologic function for p-glycoprotein(MDR-1) during the migration of dendritic cells from skin via afferentlymphatic vessels. Proc. Nat. Acad. Sci. 95: 6924-6929, 1998.
- [32626] 6178.Riordan, J. R.; Deuchars, K.; Kartner, N.; Alon, N.; Trent, J.;Ling, V.: Amplification of P-glycoprotein genes in multidrug-resistantmammalian cell lines. Nature 316: 817-819, 1985.
- [32627] 6179.Roninson, I. B.; Chin, J. E.; Choi, K.; Gros, P.; Housman, D.E.; Fojo, A.; Shen, D.; Gottesman, M. M.; Pastan, I.: Isolation ofhuman mdrl DNA sequences amplified in multidrug-resistant KB carcinomacells. Proc. Nat. Acad. Sci. 83: 4538-4542, 1986.

- [32628] 6180. Ruiz, J. C.; Choi, K.; Von Hoff, D. D.; Roninson, I. B.; Wahl, G. M.: Autonomously replicating episomes contain MDR1 genes in a multidrug-resistant human cell line. *Molec. Cell. Biol.* 9: 109–115, 1989.
- [32629] 6181. Safa, A. R.; Stern, R. K.; Choi, K.; Agresti, M.; Tamai, I.; Mehta, N. D.; Roninson, I. B.: Molecular basis of preferential resistance to colchicine in multidrug-resistant human cells conferred by Gly185-to-Val185 substitution in P-glycoprotein. *Proc. Nat. Acad. Sci.* 87: 7225–7229, 1990.
- [32630] 6182. Schinkel, A. H.; Smit, J. J. M.; van Tellingen, O.; Beijnen, J. H.; Wagenaar, E.; van Deemter, L.; Mol, C. A. A. M.; van der Valk, M. A.; Robanus-Maandag, E. C.; te Riele, H. P. J.; Berns, A. J. M.; Borst, P.: Disruption of the mouse *mdr1a* P-glycoprotein gene leads to a deficiency in the blood-brain barrier and to increased sensitivity to drugs. *Cell* 77: 491–502, 1994.
- [32631] 6183. Shen, D.-W.; Fojo, A.; Chin, J. E.; Roninson, I. B.; Richert, N.; Pastan, I.; Gottesman, M. M.: Human multidrug-resistant cell lines: increased *mdr1* expression can precede gene amplification. *Science* 232: 643–645, 1986.
- [32632] 6184. Slovak, M. L.; Hoeltge, G. A.; Trent, J. M.: Cytogenetic alterations associated with the acquisition of doxorubicin resistance: possible significance of chromosome 7

alterations. *Cancer Res.* 47: 6646–6652, 1987.

- [32633] 6185. Smit, J. W.; Huisman, M. T.; van Tellingen, O.; Wiltshire, H. R.; Schinkel, A. H.: Absence or pharmacological blocking of placental P-glycoprotein profoundly increases fetal drug exposure. *J. Clin. Invest.* 104: 1441–1447, 1999.
- [32634] 6186. Synold, T. W.; Dussault, I.; Forman, B. M.: The orphan nuclear receptor SXR coordinately regulates drug metabolism and efflux. *Nature Med.* 7: 584–590, 2001.

- [32635] 6187. Taylor, H. R.; Pacque, M.; Munoz, B.; Greene, B. M.: Impact of mass treatment of onchocerciasis with ivermectin on the transmission of infection. *Science* 250: 116–118, 1990.
- [32636] 6188. Trent, J.; Bell, D.; Willard, H.; Ling, V.: Chromosomal localization in normal human cells and CHO cells of a sequence derived from P-glycoprotein (PGY1). (Abstract) *Cytogenet. Cell Genet.* 40: 761–762, 1985.
- [32637] 6189. Trent, J. M.; Witkowski, C. M.: Clarification of the chromosomal assignment of the human P-glycoprotein/mdr1 gene: possible coincidence with the cystic fibrosis and c-met oncogene. *Cancer Genet. Cytogenet.* 26: 187–190, 1987.
- [32638] 6190. Trezise, A. E. O.; Romano, P. R.; Gill, D. R.; Hyde, S. C.; Sepulveda, F. V.; Buchwald, M.; Higgins, C. F.: The multidrug resistance and cystic fibrosis genes have complementary patterns of epithelial expression. *EMBO J.* 11: 4291–4303, 1992.
- [32639] 6191. Ueda, K.; Cardarelli, C.; Gottesman, M. M.; Pastan, I.: Expression of a full-length cDNA for the human 'MDR1' gene confers resistance to colchicine, doxorubicin, and vinblastine. *Proc. Nat. Acad. Sci.* 84: 3004–3008, 1987.
- [32640] 6192. Ueda, K.; Clark, D. P.; Chen, C.; Roninson, I. B.;

Gottesman, M. M.; Pastan, I.: The human multidrug resistance (mdr1) gene: cDNA cloning and transcription initiation. *J. Biol. Chem.* 262: 505–508, 1987.

[32641] 6193. Ueda, K.; Cornwell, M. M.; Gottesman, M. M.; Pastan, I.; Roninson, I. B.; Ling, V.; Riordan, J. R.: The mdr1 gene, responsible for multidrug-resistance, codes for P-glycoprotein. *Biochem. Biophys. Res. Commun.* 141: 956–962, 1986.

[32642] 6194. Ueda, K.; Pastan, I.; Gottesman, M. M.: Isolation and sequence of the promoter region of the human multidrug-resistance (P-glycoprotein) gene. *J. Biol. Chem.* 262: 17432–17436, 1987.

[32643] 6195. Umbenhauer, D. R.; Lankas, G. R.; Pippert, T. R.; Wise, L. D.; Cartwright, M. E.; Hall, S. J.; Beare, C. M.: Identification of a P-glycoprotein-deficient subpopulation in the CF-1 mouse strain using a restriction fragment length polymorphism. *Toxicol. Appl. Pharm.* 146: 88–94, 1997.

[32644] 6196. van Helvoort, A.; Smith, A. J.; Sprong, H.; Fritzsche, I.; Schinkel, A. H.; Borst, P.; van Meer, G.: MDR1 P-glycoprotein is a lipid translocase of broad specificity, while MDR3 P-glycoprotein specifically translocates phosphatidylcholine. *Cell* 87: 507–517, 1996.

[32645] 6197. Woods, K. A.; Camacho-Hubner, C.; Bergman, R. N.;

Barter, D.;Clark, A. J. L.; Savage, M. O.: Effects of insulin-like growth factor I (IGF-I) therapy on body composition and insulin resistance in IGF-I gene deletion. *J. Clin. Endocr. Metab.* 85: 1407–1411, 2000.

[32646] 6198.Woods, K. A.; Camacho-Hubner, C.; Savage, M. O.; Clark, A. J.L.: Intrauterine growth retardation and postnatal growth failure associated with deletion of the insulin-like growth factor I gene. *NewEng. J. Med.* 335: 1363–1367, 1996.

[32647] 6199.Yang-Feng, T. L.; Brissenden, J. E.; Ullrich, A.; Francke, U.: Sub-regional localization of human genes for insulin-like growth factors I (IGF1) and II (IGF2) by in situ hybridization. (Abstract) *Cytogenet.Cell Genet.* 40: 782 only, 1985.

[32648] 6200.Yanovski, J. A.; Sovik, K. N.; Nguyen, T. T.; Sebring, N. G.:Insulin-like growth factors and bone mineral density in African American and white girls. *J. Pediat.* 137: 826–832, 2000.

[32649] 6201.Zhu, J.; Kahn, C. R.: Analysis of a peptide hormone-receptor interaction in the yeast two-hybrid system. *Proc. Nat. Acad. Sci.* 94:13063–13068, 1997.

[32650] 6202.Kobayashi, M.; Takamatsu, K.; Saitoh, S.; Miura, M.; Noguchi, T.: Molecular cloning of hippocalcin, a novel cal-

cium-binding protein of the recoverin family exclusively expressed in hippocampus. *Biochem. Biophys. Res. Commun.* 189: 511–517, 1992.

[32651] 6203. Takamatsu, K.; Kobayashi, M.; Saitoh, S.; Fujishiro, M.; Noguchi, T.: Molecular cloning of human hippocalcin cDNA and chromosomal mapping of its gene. *Biochem. Biophys. Res. Commun.* 200: 606–611, 1994.

[32652] 6204. Borrego, S.; Ruiz, A.; Saez, M. E.; Gimm, O.; Gao, X.; Lopez-Alonso, M.; Hernandez, A.; Wright, F. A.; Antinolo, G.; Eng, C.: RET genotypes comprising specific haplotypes of polymorphic variants predispose to isolated Hirschsprung disease. *J. Med. Genet.* 37: 572–578, 2000.

[32653] 6205. Borrego, S.; Saez, M. E.; Ruiz, A.; Gimm, O.; Lopez-Alonso, M.; Antinolo, G.; Eng, C.: Specific polymorphisms in the RET proto-oncogene are over-represented in patients with Hirschsprung disease and may represent loci modifying phenotypic expression. *J. Med. Genet.* 36: 771–774, 1999.

[32654] 6206. Goldstein, J. L.; Brown, M. S.; Stone, N. J.: Genetics of the LDL receptor: evidence that the mutations affecting binding and internalization are allelic. *Cell* 12: 629–641, 1977.

[32655] 6207. Goldstein, J. L.; Dana, S. E.; Brunschede, G. Y.;

Brown, M. S.: Genetic heterogeneity in familial hypercholesterolemia: evidence for two different mutations affecting functions of low-density lipoprotein receptor. *Proc. Nat. Acad. Sci.* 72: 1092–1096, 1975.

[32656] 6208. Hobbs, H. H.; Brown, M. S.; Goldstein, J. L.: Molecular genetics of the LDL receptor gene in familial hypercholesterolemia. *Hum. Mutat.* 1:445–466, 1992.

[32657] 6209. Hobbs, H. H.; Brown, M. S.; Goldstein, J. L.; Russell, D. W.: Deletion of exon encoding cysteine-rich repeat of low density lipoprotein receptor alters its binding specificity in a subject with familial hypercholesterolemia. *J. Biol. Chem.* 261: 13114–13120, 1986.

[32658] 6210. Hobbs, H. H.; Russell, D. W.; Brown, M. S.; Goldstein, J. L.: The LDL receptor locus in familial hypercholesterolemia: mutational analysis of a membrane protein. *Annu. Rev. Genet.* 24: 133–170, 1990.

[32659] 6211. Horsthemke, B.; Beisiegel, U.; Dunning, A.; Havinga, J. R.; Williamson, R.; Humphries, S.: Unequal crossing-over between two Alu-repetitive DNA sequences in the low-density-lipoprotein-receptor gene: a possible mechanism for the defect in a patient with familial hypercholesterolaemia. *Europ. J. Biochem.* 164: 77–81, 1987.

[32660] 6212. Khachadurian, A. K.: The inheritance of essential fa-

milial hypercholesterolemia. *Am.J. Med.* 37: 402–407, 1964.

[32661] 6213.Kingsley, D. M.; Krieger, M.: Receptor-mediated endocytosis of low density lipoprotein: somatic cell mutants define multiple genes required for expression of surface-receptor activity. *Proc. Nat.Acad. Sci.* 81: 5454–5458, 1984.

[32662] 6214.Knight, B. L.; Gavigan, S. J. P.; Soutar, A. K.; Patel, D. D.: Defective processing and binding of low-density lipoprotein receptors in fibroblasts from a familial hypercholesterolaemic subject. *Europ.J. Biochem.* 179: 693–698, 1989.

[32663] 6215.Knoblauch, H.; Muller-Myhsok, B.; Busjahn, A.; Ben Avi, L.; Bähring, S.; Baron, H.; Heath, S. C.; Uhlmann, R.; Faulhaber, H.–D.; Shpitzen, S.; Aydin, A.; Reshef, A.; and 11 others: A cholesterol-lowering gene maps to chromosome 13q. *Am. J. Hum. Genet.* 66: 157–166, 2000.

[32664] 6216.Komuro, I.; Kato, H.; Nakagawa, T.; Takahashi, K.; Mimori, A.; Takeuchi, F.; Nishida, Y.; Miyamoto, T.: Case report: the longest-lived patient with homozygous familial hypercholesterolemia secondary to a defect in internalization of the LDL receptor. *Am. J. Med. Sci.* 294:341–345, 1987.

- [32665] 6217.Kotze, M. J.; Langenhoven, E.; Retief, A. E.; Steyn, K.; Marais,M. P.; Grobbelaar, J. J.; Oosthuizen, C. J. J.; Weich, H. F. H.; Benade,A. J. S.: Haplotype associations of three DNA polymorphisms at the human low density lipoprotein receptor gene locus in familial hypercholesterolemia. J.Med. Genet. 24: 750–755, 1987.
- [32666] 6218.Grundy, H. O.; Peltz, G.; Moore, K. W.; Golbus, M. S.; Jackson,L. G.; Lebo, R. V.: The polymorphic Fc-gamma receptor II gene maps to human chromosome 1q. Immunogenetics 29: 331–339, 1989.
- [32667] 6219.Kudo, A.; Melchers, F.: A second gene, Vpre-B in the lambda 5 locus of the mouse, which appears to be selectively expressed in pre-B lymphocytes. EMBO J. 6: 2267–2272, 1987.
- [32668] 6220.Mattei, M.-G.; Fumoux, F.; Roeckel, N.; Fougereau, M.; Schiff,C.: The human pre-B-specific lambda-like cluster is located in the 22q11.2–22q12.3 region, distal to the IgC-lambda locus. Genomics 9:544–546, 1991.
- [32669] 6221.Minegishi, Y.; Coustan-Smith, E.; Wang, Y.-H.; Cooper, M. D.; Campana,D.; Conley, M. E.: Mutations in the human lambda-5/14.1 gene result in B cell deficiency and agammaglobulinemia. J. Exp. Med. 187: 71–77,1998.
- [32670] 6222.Pillai, S.; Baltimore, D.: The omega and iota surro-

gate immunoglobulin light chains. *Curr. Top. Microbiol. Immun.* 137: 136–139, 1988.

[32671] 6223. Sakaguchi, N.; Melchers, F.: Lambda 5, a new light-chain-related locus selectively expressed in pre-B lymphocytes. *Nature* 324: 579–582, 1986.

[32672] 6224. Schiff, C.; Milili, M.; Fougereau, M.: Isolation of early immunoglobulin lambda-like gene transcripts in human fetal liver. *Europ. J. Immun.* 19:1873–1878, 1989.

[32673] 6225. Lin, P.-F.; Slate, D. L.; Lawyer, F. C.; Ruddle, F. H.: Assignment of the murine interferon sensitivity and cytoplasmic superoxide dismutase genes to chromosome 16. *Science* 209: 285–287, 1980.

[32674] 6226. Holmes, W. E.; Lee, J.; Kuang, W.-J.; Rice, G. C.; Wood, W. I.: Structure and functional expression of a human interleukin-8 receptor. *Science* 253:1278–1280, 1991.

[32675] 6227. Sligh, J. E., Jr.; Ballantyne, C. M.; Rich, S. S.; Hawkins, H.K.; Smith, C. W.; Bradley, A.; Beaudet, A. L.: Inflammatory and immune responses are impaired in mice deficient in intercellular adhesion molecule 1. *Proc. Nat. Acad. Sci.* 90: 8529–8533, 1993.

[32676] 6228. Nishimura, D.; Buetow, K. H.; Yamada, Y.; Murray, J. C.: RFLPs and linkage relationships of the human laminin

B2 gene. Genomics 3:393–395, 1988.

- [32677] 6229. Karaplis, A. C.; Luz, A.; Glowacki, J.; Bronson, R. T.; Tybulewicz, V. L. J.; Kronenberg, H. M.; Mulligan, R. C.: Lethal skeletal dysplasia from targeted disruption of the parathyroid hormone–related peptide gene. Genes Dev. 8: 277–289, 1994.
- [32678] 6230. Bevilacqua, G.; Sobel, M. E.; Liotta, L. A.; Steeg, P. S.: Association of low nm23 RNA levels in human primary infiltrating ductal breast carcinomas with lymph node involvement and other histopathological indicators of high metastatic potential. Cancer Res. 49: 5185–5190, 1989.
- [32679] 6231. Chang, C. L.; Zhu, X.; Thoraval, D. H.; Ungar, D.; Rawwas, J.; Hora, N.; Strahler, J. R.; Hanash, S. M.; Radany, E.: nm23–H1 mutation in neuroblastoma. (Letter) Nature 370: 335–336, 1994.
- [32680] 6232. Dooley, S.; Seib, T.; Engel, M.; Theisinger, B.; Janz, H.; Piontek, K.; Zang, K.–D.; Welter, C.: Isolation and characterization of the human genomic locus coding for the putative metastasis control gene nm23–H1. Hum. Genet. 93: 63–66, 1994.
- [32681] 6233. Eddy, R. L.; Mendola, C. E.; Fairhurst, J. L.; Shows, T. B.; O'Hara, B.; Kovesdi, I.; Backer, J. M.: Metastasis suppressor gene is mapped to human chromosome

17p11-qter. (Abstract) Cytogenet. Cell Genet. 58:2005 only, 1991.

- [32682] 6234. Gilles, A.-M.; Presecan, E.; Vonica, A.; Lascu, I.: Nucleosidediphosphate kinase from human erythrocytes: structural characterization of the two polypeptide chains responsible for heterogeneity of the hexameric enzyme. J. Biol. Chem. 266: 8784–8789, 1991.
- [32683] 6235. Keim, D.; Hailat, N.; Melhem, R.; Zhu, X. X.; Lascu, I.; Veron, M.; Strahler, J.; Hanash, S. M.: Proliferation-related expression of p19/nm23 nucleoside diphosphate kinase. J. Clin. Invest. 89:919–924, 1992.
- [32684] 6236. Drayna, D.; Fielding, C.; McLean, J.; Baer, B.; Castro, G.; Chen, E.; Comstock, L.; Henzel, W.; Kohr, W.; Rhee, L.; Wion, K.; Lawn, R.: Cloning and expression of human apolipoprotein D cDNA. J. Biol. Chem. 261: 16535–16539, 1986.
- [32685] 6237. Drayna, D.; Scott, J. D.; Lawn, R.: Multiple RFLPs at the human apolipoprotein D (APOD) locus. Nucleic Acids Res. 15: 9617 only, 1987.
- [32686] 6238. Whittington, J. E.; Keats, B. J. B.; Jackson, J. F.; Currier, R. D.; Terasaki, P. I.: Linkage studies on glyoxalase I (GLO), pepsinogen (PG), spinocerebellar ataxia (SCA1), and HLA. Cytogenet. Cell Genet. 28:145–150, 1980.

- [32687] 6239.Ziegler, A.; Fonatsch, C.; Kompf, J.: Mapping of the locus for glyoxalase 1 (GLO1) on human chromosome 6 using mutant cell lines.(Abstract) *Cytogenet. Cell Genet.* 40: 787 only, 1985.
- [32688] 6240.Agar, N. S.; Board, P. G.; Bell, K.: Studies of erythrocyte glyoxalase II in various domestic species: discovery of glyoxalase II deficiency in the horse. *Animal Blood Groups Biochem. Genet.* 15: 67–70, 1984.
- [32689] 6241.Ball, J. C.; Vander Jagt, D. L.: Purification of S-2-hydroxyacylglutathione hydrolase (glyoxalase II) from rat erythrocytes. *Anal. Biochem.* 98:472–477, 1979.
- [32690] 6242.Board, P. G.: Genetic polymorphism of human erythrocyte glyoxalase II. *Am. J. Hum. Genet.* 32: 690–694, 1980.
- [32691] 6243.Board, P. G.; Gibbs, C. J., Jr.; Gajdusek, D. C.: Polymorphism of erythrocyte glyoxalase II in anthropoid primates. *Folia Primatol.* 36:138–143, 1981.
- [32692] 6244.Honey, N. K.; Shows, T. B.: Assignment of the glyoxalase II gene (HAGH) to human chromosome 16. *Hum. Genet.* 58: 358–361, 1981.
- [32693] 6245.Mulley, J. C.; Barton, N.; Callen, D. F.: Localisation of human PGP and HAGH genes to 16p13.3. *Cytogenet. Cell Genet.* 53: 175–176, 1990.

- [32694] 6246. Mulley, J. C.; Callen, D. F.: New regional localisations for HAGHand PGP on human chromosome 16. *Hum. Genet.* 74: 423–424, 1986.
- [32695] 6247. Ridderstrom, M.; Saccucci, F.; Hellman, U.; Bergman, T.; Principato, G.; Mannervik, B.: Molecular cloning, heterologous expression, and characterization of human glyoxalase II. *J. Biol. Chem.* 271: 319–323, 1996.
- [32696] 6248. Valentine, W. N.; Paglia, D. E.; Neerhout, R. C.; Konrad, P. N.: Erythrocyte glyoxalase II deficiency with coincidental hereditary elliptocytosis. *Blood* 36: 797–808, 1970.
- [32697] 6249. Beranova, M.; Oliveira, L. M. B.; Bedecarrats, G. Y.; Schipani, E.; Vallejo, M.; Ammini, A. C.; Quintos, J. B.; Hall, J. E.; Martin, K. A.; Hayes, F. J.; Pitteloud, N.; Kaiser, U. B.; Crowley, W. F., Jr.; Seminara, S. B.: Prevalence, phenotypic spectrum, and modes of inheritance of gonadotropin-releasing hormone receptor mutations in idiopathic hypogonadotropic hypogonadism. *J. Clin. Endocr. Metab.* 86: 1580–1588, 2001.
- [32698] 6250. Caron, P.; Chauvin, S.; Christin-Maitre, S.; Bennet, A.; Lahlou, N.; Counis, R.; Bouchard, P.; Kottler, M.-L.: Resistance of hypogonadic patients with mutated GnRH receptor genes to pulsatile GnRH administration. *J. Clin. Endocr. Metab.* 84: 990–996, 1999.

- [32699] 6251.Costa, E. M. F.; Bedecarrats, G. Y.; Mendonca, B. B.; Arnhold, I. J. P.; Kaiser, U. B.; Latronico, A. C.: Two novel mutations in the gonadotropin-releasing hormone receptor gene in Brazilian patients with hypogonadotropic hypogonadism and normal olfaction. *J. Clin. Endocr. Metab.* 86: 2680–2686, 2001.
- [32700] 6252.de Roux, N.; Young, J.; Brailly-Tabard, S.; Misrahi, M.; Milgrom, E.; Schaison, G.: The same molecular defects of the gonadotropin-releasing hormone receptor determine a variable degree of hypogonadism in affected kindred. *J. Clin. Endocr. Metab.* 84: 567–572, 1999.
- [32701] 6253.de Roux, N.; Young, J.; Misrahi, M.; Genet, R.; Chanson, P.; Schaison, G.; Milgrom, E.: A family with hypogonadotropic hypogonadism and mutations in the gonadotropin-releasing hormone receptor. *New Eng. J. Med.* 337: 1597–1602, 1997.
- [32702] 6254.Fan, N. C.; Jeung, E.-B.; Peng, C.; Olofsson, J. I.; Krisinger, J.; Leung, P. C. K.: The human gonadotropin-releasing hormone (GnRH) receptor gene: cloning, genomic organization and chromosomal assignment. *Molec. Cell. Endocr.* 103: R1–R6, 1994.
- [32703] 6255.Iwashita, T.; Murakami, H.; Asai, N.; Takahashi, M.: Mechanism of Ret dysfunction by Hirschsprung mutations

affecting its extracellular domain. *Hum. Molec. Genet.* 5: 1577–1580, 1996.

- [32704] 6256. Lehrman, M. A.; Goldstein, J. L.; Brown, M. S.; Russell, D. W.; Schneider, W. J.: Internalization-defective LDL receptors produced by genes with nonsense and frameshift mutations that truncate the cytoplasmic domain. *Cell* 41: 735–743, 1985.
- [32705] 6257. Bird, A. P.: CpG islands as gene markers in the vertebrate nucleus. *Trends Genet.* 3: 342–347, 1987.
- [32706] 6258. Maartmann-Moe, K.; Wang, H. S.; Donald, L. J.; Hamerton, J. L.; Berg, K.: Data from hybrid cell lines raise the possibility that factors controlling the low density lipoprotein receptor activity may reside on human chromosome 21, 5 or both. (Abstract) *Cytogenet. Cell Genet.* 32: 295–296, 1982.
- [32707] 6259. Miyake, Y.; Tajima, S.; Funahashi, T.; Yamamoto, A.: Analysis of a recycling-impaired mutant of low density lipoprotein receptor in familial hypercholesterolemia. *J. Biol. Chem.* 264: 16584–16590, 1989.
- [32708] 6260. Seftel, H. C.; Baker, S. G.; Sandler, M. P.; Forman, M. B.; Joffe, B. I.; Mendelsohn, D.; Jenkins, T.; Mieny, C. J.: A host of hypercholesterolaemic homozygotes in South Africa. *Brit. Med. J.* 281: 633–636, 1980.

- [32709] 6261.Grundy, H. O.; Peltz, G.; Barsh, G.; Moore, K.; Golbus, M. S.;Lebo, R. V.: Immunoglobulin G Fc receptor II and Fc receptor IIIgenes map to chromosome 1 by spot-blot chromosome analysis. (Abstract) Am.J. Hum. Genet. 43: A145, 1988.
- [32710] 6262.Hibbs, M. L.; Bonadonna, L.; Scott, B. M.; McKenzie, I. F. C.;Hogarth, P. M.: Molecular cloning of a human immunoglobulin G Fcreceptor. Proc. Nat. Acad. Sci. 85: 2240-2244, 1988.
- [32711] 6263.Hibbs, M. L.; Hogarth, P. M.; McKenzie, I. F. C.: The mouse Ly-17locus identifies a polymorphism of the Fc receptor. Immunogenetics 22:335-348, 1985.
- [32712] 6264.Lebo, R. V.; Lynch, E. D.; Wiegant, J.; Moore, K.; Trounstein,M.; van der Ploeg, M.: Multicolor fluorescence in situ hybridizationand pulsed field electrophoresis dissect CMT1B gene region. Hum.Genet. 88: 13-20, 1991.
- [32713] 6265.Moser, K. L.; Neas, B. R.; Salmon, J. E.; Yu, H.; Gray-McGuire,C.; Asundi, N.; Bruner, G. R.; Fox, J.; Kelly, J.; Henshall, S.; Bacino,D.; Dietz, M.; Hogue, R.; Koelsch, G.; Nightingale, L.; Shaver, T.;Abdou, N. I.; Albert, D. A.; Carson, C.; Petri, M.; Treadwell, E.L.; James, J. A.; Harley, J. B.: Genome scan of human systemic lupuserythematosus: evidence for linkage on chromosome 1q in African-

American pedigrees. Proc. Nat. Acad. Sci. 95:
14869–14874, 1998.

- [32714] 6266. Oakey, R. J.; Watson, M. L.; Seldin, M. F.: Construction of a physical map on mouse and human chromosome 1: comparison of 13 Mb of mouse and 11 Mb of human DNA. Hum. Molec. Genet. 1: 613–620, 1992.
- [32715] 6267. Peltz, G. A.; Grundy, H. O.; Lebo, R. V.; Yssel, H.; Barsh, G. S.; Moore, K. W.: Human Fc-gamma-RIII: cloning, expression, and identification of the chromosomal locus of two Fc receptors for IgG. Proc. Nat. Acad. Sci. 86: 1013–1017, 1989.
- [32716] 6268. Qiu, W. Q.; de Bruin, D.; Brownstein, B. H.; Pearce, R.; Ravetch, J. V.: Organization of the human and mouse low-affinity Fc-gamma-R genes: duplication and recombination. Science 248: 732–735, 1990.
- [32717] 6269. Salmon, J. E.; Millard, S.; Schachter, L. A.; Arnett, F. C.; Ginzler, E. M.; Gourley, M. F.; Ramsey-Goldman, R.; Peterson, M. G. E.; Kimberly, R. P.: Fc-gamma-RIIA alleles are heritable risk factors for lupus nephritis in African Americans. J. Clin. Invest. 97: 1348–1354, 1996.
- [32718] 6270. Sammartino, L.; Webber, L. M.; Hogarth, P. M.; McKenzie, I. F. C.; Garson, O. M.: Assignment of the gene coding for human FcRII (CD32) to bands q23q24 on chro-

mosome 1. Immunogenetics 28: 380–381,1988.

- [32719] 6271.Stein, M.–P.; Edberg, J. C.; Kimberly, R. P.; Mangan, E. K.; Bharadwaj,D.; Mold, C.; Du Clos, T. W.: C–reactive protein binding to Fc–gamma–RIIaon human monocytes and neutrophils is allele–specific. J. Clin. Invest. 105:369–376, 2000.
- [32720] 6272.Unkeless, J. C.: Function and heterogeneity of human Fc receptorsfor immunoglobulin G. J. Clin. Invest. 83: 355–361, 1989.
- [32721] 6273.Chang–Yeh, A.; Jabs, E. W.; Li, X.; Dracopoli, N. C.; Huang, R.C. C.: The IPP gene is assigned to human chromosome 1p32–1p22. Genomics 15:239–241, 1993.
- [32722] 6274.Chang–Yeh, A.; Mold, D. E.; Brilliant, M. H.; Huang, R. C. C.:The mouse intracisternal A particle–promoted placental gene retrotranspositionis mouse–strain–specific. Proc. Nat. Acad. Sci. 90: 292–296, 1993.
- [32723] 6275.Chang–Yeh, A.; Mold, D. E.; Huang, R. C. C.: Identification ofa novel murine IAP–promoted placenta–expressed gene. Nucleic AcidsRes. 19: 3667–3672, 1991.
- [32724] 6276.Mold, D. E.; Chang–Yeh, A.; Huang, R. C. C.: Cell lineage–specificexpression of the MIPP gene. Biochem. Biophys. Res. Commun. 177:1062–1067, 1991.
- [32725] 6277.Cama, A.; de la Luz Sierra, M.; Quon, M. J.; Ottini, L.;

Gorden,P.; Taylor, S. I.: Substitution of glutamic acid for alanine 1135 in the putative 'catalytic loop' of the tyrosine kinase domain of the human insulin receptor: a mutation that impairs proteolytic processing into subunits and inhibits receptor tyrosine kinase activity. *J.Biol. Chem.* 268: 8060–8069, 1993.

[32726] 6278.Cama, A.; Quon, M. J.; de la Luz Sierra, M.; Taylor, S. I.: Substitution of isoleucine for methionine at position 1153 in the beta-subunit of the human insulin receptor. *J. Biol. Chem.* 267: 8383–8389, 1992.

[32727] 6279.Caro, J. F.; Raju, S. M.; Sinha, M. K.; Goldfine, I. D.; Dohm, G. L.: Heterogeneity of human liver, muscle, and adipose tissue insulin receptor. *Biochem. Biophys. Res. Commun.* 151: 123–129, 1988.

[32728] 6280.Christiansen, K.; Tranum-Jensen, J.; Carlsen, J.; Vinten, J.: A model for the quaternary structure of human placental insulin receptor deduced from electron microscopy. *Proc. Nat. Acad. Sci.* 88: 249–252, 1991.

[32729] 6281.Cocozza, S.; Porcellini, A.; Riccardi, G.; Monticelli, A.; Condorelli, G.; Ferrara, A.; Pianese, L.; Miele, C.; Capaldo, B.; Beguinot, F.; Varrone, S.: NIDDM associated with mutation in tyrosine kinase domain of insulin receptor gene. *Diabetes* 41: 521–526, 1992.

- [32730] 6282.Due, C.; Simonsen, M.; Olsson, L.: The major histocompatibility complex class I heavy chain as a structural subunit of the human cell membrane insulin receptor: implications for the range of biological functions of histocompatibility antigens. *Proc. Nat. Acad. Sci.* 83:6007–6011, 1986.
- [32731] 6283.Ebina, Y.; Ellis, L.; Jarnagin, K.; Edery, M.; Graf, L.; Clauser, E.; Ou, J.-H.; Masiarz, F.; Kan, Y. W.; Goldfine, I. D.; Roth, R.A.; Rutter, W. J.: The human insulin receptor cDNA: the structural basis for hormone activated trans-membrane signalling. *Cell* 40: 747–758, 1985.
- [32732] 6284.Elbein, S. C.; Corsetti, L.; Ullrich, A.; Permutt, M. A.: Multiple restriction fragment length polymorphisms at the insulin receptor locus: a highly informative marker for linkage analysis. *Proc. Nat. Acad. Sci.* 83: 5223–5227, 1986.
- [32733] 6285.Elbein, S. C.; Sorensen, L. K.; Schumacher, M. C.: Methionine for valine substitution in exon 17 of the insulin receptor gene in a pedigree with familial NIDDM. *Diabetes* 42: 429–434, 1993.
- [32734] 6286.Ferrannini, E.; Muggeo, M.; Navalesi, R.; Pilo, A.: Impaired insulin degradation in a patient with insulin resistance and acanthosis nigricans. *Am. J. Med.* 73: 148–154,

1982.

- [32735] 6287. Grigorescu, F.; Flier, J. S.; Kahn, C. R.: Characterization of binding and phosphorylation defects of erythrocyte insulin receptors in the type A syndrome of insulin resistance. *Diabetes* 35: 127–138, 1986.
- [32736] 6288. Grigorescu, F.; Flier, J. S.; Kahn, C. R.: Defect in insulin receptor phosphorylation in erythrocytes and fibroblasts associated with severe insulin resistance. *J. Biol. Chem.* 259: 15003–15006, 1984.
- [32737] 6289. Grunberger, G.; Zick, Y.; Gordon, G.: Defect in phosphorylation of insulin receptors in cells from an insulin-resistant patient with normal insulin binding. *Science* 223: 932–934, 1984.
- [32738] 6290. Hone, J.; Accili, D.; Al-Gazali, L. I.; Lestringant, G.; Orban, T.; Taylor, S. I.: Homozygosity for a new mutation (ile119-to-met) in the insulin receptor gene in five sibs with familial insulin resistance. *J. Med. Genet.* 31: 715–716, 1994.
- [32739] 6291. Kadowaki, T.; Bevins, C. L.; Cama, A.; Ojamaa, K.; Marcus-Samuels, B.; Kadowaki, H.; Beitz, L.; McKeon, C.; Taylor, S. I.: Two mutant alleles of the insulin receptor gene in a patient with extreme insulin resistance. *Science* 240: 787–790, 1988.

- [32740] 6292.Kadowaki, T.; Kadowaki, H.; Accili, D.; Taylor, S. I.: Substitution of lysine for asparagine at position 15 in the alpha-subunit of the human insulin receptor: a mutation that impairs transport of receptors to the cell surface and decreases the affinity of insulin binding. *J. Biol. Chem.* 265: 19143–19150, 1990.
- [32741] 6293.Kadowaki, T.; Kadowaki, H.; Accili, D.; Yazaki, Y.; Taylor, S.I.: Substitution of arginine for histidine at position 209 in the alpha-subunit of the human insulin receptor: a mutation that impairs receptor dimerization and transport of receptors to the cell surface. *J. Biol. Chem.* 266: 21224–21231, 1991.
- [32742] 6294.Kadowaki, T.; Kadowaki, H.; Rechler, M. M.; Serrano-Rios, M.; Roth, J.; Gorden, P.; Taylor, S. I.: Five mutant alleles of the insulin receptor gene in patients with genetic forms of insulin resistance. *J. Clin. Invest.* 86: 254–264, 1990.
- [32743] 6295.Kadowaki, T.; Kadowaki, H.; Taylor, S. I.: A nonsense mutation causing decreased levels of insulin receptor mRNA: detection by a simplified technique for direct sequencing of genomic DNA amplified by the polymerase chain reaction. *Proc. Nat. Acad. Sci.* 87: 658–662, 1990.
- [32744] 6296.Kahn, C. R.; Flier, J. S.; Bar, R. S.; Archer, J. A.; Gor-

den,P.; Martin, M. M.; Roth, J.: The syndromes of insulin resistanceand acanthosis nigricans: insulin receptor disorders in man. NewEng. J. Med. 294: 739–745, 1976.

[32745] 6297.Kahn, C. R.; Goldstein, B. J.: Molecular defects in insulin action. Science 245:13 only, 1989.

[32746] 6298.Kahn, C. R.; White, M. F.: The insulin receptor and the molecularmechanism of insulin action. J. Clin. Invest. 82: 1151–1156, 1988.

[32747] 6299.Takehi, T.; Hisatomi, A.; Kuzuya, H.; Yoshimasa, Y.; Okamoto,M.; Yamada, K.; Nishimura, H.; Kosaki, A.; Nawata, H.; Umeda, F.;Ibayashi, H.; Imura, H.: Defective processing of insulin–receptorprecursor in cultured lymphocytes from a patient with extreme insulinresistance. J. Clin. Invest. 81: 2020–2022, 1988.

[32748] 6300.Kittur, D.; Shimizu, Y.; DeMars, R.; Edidin, M.: Insulin bindingto human B lymphoblasts is a function of HLA haplotype. Proc. Nat.Acad. Sci. 84: 1351–1355, 1987.

[32749] 6301.Klinkhamer, M. P.; Groen, N. A.; van der Zon, G. C. M.; Lindhout,D.; Sandkuyl, L. A.; Krans, H. M. J.; Moller, W.; Maassen, J. A.:A leucine–to–proline mutation in the insulin receptor in a familywith insulin resistance. EMBO J. 8: 2503–2507, 1989.

[32750] 6302.Krook, A.; Brueton, L.; O'Rahilly, S.: Homozygous

nonsense mutation in the insulin receptor gene in infant with leprechaunism. *Lancet* 342:277–278, 1993.

[32751] 6303. Kulkarni, R. N.; Bruning, J. C.; Winnay, J. N.; Postic, C.; Magnuson, M. A.; Kahn, C. R.: Tissue-specific knockout of the insulin receptor in pancreatic beta cells creates an insulin secretory defect similar to that in type 2 diabetes. *Cell* 96: 329–339, 1999.

[32752] 6304. Chan, A. M.-L.; Hilkens, J.; Kroezen, V.; Mitchell, P. J.; Scambler, P.; Wainwright, B. J.; Williamson, R.; Cooper, C. S.: Molecular cloning and localization to chromosome 6 of mouse INT1L1 gene. *Somat. Cell Molec. Genet.* 15: 555–562, 1989.

[32753] 6305. Huguet, E. L.; McMahon, J. A.; McMahon, A. P.; Bicknell, R.; Harris, A. L.: Differential expression of human Wnt genes 2, 3, 4, and 7B in human breast cell lines and normal and disease states of human breast tissue. *Cancer Res.* 54: 2615–2621, 1994.

[32754] 6306. McCoy, P. A.; Shao, Y.; Wolpert, C. M.; Donnelly, S. L.; Ashley-Koch, A.; Abel, H. L.; Ravan, S. A.; Abramson, R. K.; Wright, H. H.; DeLong, G. R.; Cuccaro, M. L.; Gilbert, J. R.; Pericak-Vance, M. A.: No association between the WNT2 gene and autistic disorder. *Am. J. Med. Genet. (Neuropsychiat. Genet.)* 114: 106–109, 2002.

- [32755] 6307.Nusse, R.; Brown, A.; Papkoff, J.; Scambler, P.; Shackleford, G.; McMahon, A.; Moon, R.; Varmus, H.: A new nomenclature for int-1 and related genes: the Wnt gene family. *Cell* 64: 231–232, 1991.
- [32756] 6308.Wainwright, B. J.; Scambler, P. J.; Stanier, P.; Watson, E. K.; Bell, G.; Wicking, C.; Estivill, X.; Courtney, M.; Bour, A.; Pedersen, P. S.; Williamson, R.; Farrall, M.: Isolation of a human gene with protein sequence similarity to human and murine int-1 and the Drosophila segment polarity mutant wingless. *EMBO J.* 7: 1743–1748, 1988.
- [32757] 6309.Wassink, T. H.; Piven, J.; Vieland, V. J.; Huang, J.; Swiderski, R. E.; Pietila, J.; Braun, T.; Beck, G.; Folstein, S. E.; Haines, J. L.; Sheffield, V. C.: Evidence supporting WNT2 as an autism susceptibility gene. *Am. J. Med. Genet.* 105: 406–413, 2001.
- [32758] 6310.Kluck, P. M. C.; Wiegant, J.; Jansen, R. P. M.; Bolk, M. W. J.; Raap, A. K.; Willemze, R.; Landegent, J. E.: The human interleukin-6 receptor alpha-chain gene is localized on chromosome 1 band q21. *Hum. Genet.* 90: 542–544, 1993.
- [32759] 6311.Szpirer, J.; Szpirer, C.; Riviere, M.; Houart, C.; Baumann, M.; Fey, G. H.; Poli, V.; Cortese, R.; Islam, M. Q.; Levan, G.: The interleukin-6-dependent DNA-binding pro-

tein gene (transcription factor 5: TCF5) maps to human chromosome 20 and rat chromosome 3, the IL6 receptor locus (IL6R) to human chromosome 1 and rat chromosome 2, and the rat IL6 gene to rat chromosome 4. Genomics 10: 539–546, 1991.

- [32760] 6312. Yamasaki, K.; Taga, T.; Hirata, Y.; Yawata, H.; Kawanishi, Y.; Seed, B.; Taniguchi, T.; Hirano, T.; Kishimoto, T.: Cloning and expression of the human interleukin-6 (BSF-2/IFN-beta-2) receptor. Science 241:825–828, 1988.
- [32761] 6313. Alli, C.; Consalez, G. G.: Linkage mapping of Csrp to proximal mouse chromosome 3. Mammalian Genome 9: 172 only, 1998.
- [32762] 6314. Erdel, M.; Weiskirchen, R.: Assignment of CSRP1 encoding the LIM domain protein CRP1, to human chromosome 1q32 by fluorescence in situ hybridization. Cytogenet. Cell Genet. 83: 10–11, 1998.
- [32763] 6315. Liebhaber, S. A.; Emery, J. G.; Urbanek, M.; Wang, X.; Cooke, N.E.: Characterization of a human cDNA encoding a widely expressed and highly conserved cysteine-rich protein with an unusual zinc-finger motif. Nucleic Acids Res. 18: 3871–3879, 1990.
- [32764] 6316. Weiskirchen, R.; Pino, J. D.; Macalima, T.; Bister, K.;

Beckerle, M. C.: The cysteine-rich protein family of highly related LIM domain proteins. *J. Biol. Chem.* 270: 28946–28954, 1995.

- [32765] 6317. Yamauchi, M.; Yamauchi, N.; Phear, G.; Spurr, N. K.; Martinsson, T.; Weith, A.; Meuth, M.: Genomic organization and chromosomal localization of the human CTP synthetase gene (CTPS). *Genomics* 11: 1088–1096, 1991.
- [32766] 6318. Wang, X.; Lee, G.; Liebhaber, S. A.; Cooke, N. E.: Human cysteine-rich protein: a member of the LIM/double-finger family displaying coordinate serum induction with c-myc. *J. Biol. Chem.* 267: 9176–9184, 1992.
- [32767] 6319. Wang, X.; Ray, K.; Szpirer, J.; Levan, G.; Liebhaber, S. A.; Cooke, N. E.: Analysis of the human cysteine-rich protein gene (CSRP), assignment to chromosome 1q24–1q32, and identification of an associated MspI polymorphism. *Genomics* 14: 391–397, 1992.
- [32768] 6320. Courseaux, A.; Grosgeorge, J.; Gaudray, P.; Pannett, A. A. J.; Forbes, S. A.; Williamson, C.; Bassett, D.; Thakker, R. V.; Teh, B. T.; Farnebo, F.; Shepherd, J.; Skogseid, B.; Larsson, C.; Giraud, S.; Zhang, C. X.; Salandre, J.; Calender, A.: Definition of the minimal MEN1 candidate area based on a 5-Mb integrated map of proximal 11q13. *Genomics* 37: 354–365, 1996.

- [32769] 6321.Bullock, S.; Hayward, C.; Manson, J.; Brock, D. J. H.; Raeburn, J. A.: Quantitative immunoassays for diagnosis and carrier detection in cystic fibrosis. Clin. Genet. 21: 336–341, 1982.
- [32770] 6322.Dorin, J. R.; Emslie, E.; van Heyningen, V.: Related calcium-binding proteins map to the same subregion of chromosome 1q and to an extended region of synteny on mouse chromosome 3. Genomics 8: 420–426, 1990.
- [32771] 6323.Dorin, J. R.; Novak, M.; Hill, R. E.; Brock, D. J. H.; Secher, D. S.; van Heyningen, V.: A clue to the basic defect in cystic fibrosis from cloning the CF antigen gene. Nature 326: 614–617, 1987.
- [32772] 6324.Frizzell, R. A.; Rechkemmer, G.; Shoemaker, R. L.: Altered regulation of airway epithelial cell chloride channels in cystic fibrosis. Science 233:558–560, 1986.
- [32773] 6325.Manson, J. C.; Brock, D. J. H.: Development of a quantitative immunoassay for the cystic fibrosis gene. Lancet I: 330–331, 1980.
- [32774] 6326.Herzig, K. H.; Schon, I.; Tatemoto, K.; Ohe, Y.; Li, Y.; Folsch, U. R.; Owyang, C.: Diazepam binding inhibitor is a potent cholecystokinin-releasing peptide in the intestine. Proc. Nat. Acad. Sci. 93: 7927–7932, 1996.
- [32775] 6327.Li, Y.; Hao, Y.; Owyang, C.: Diazepam-binding in-

hibitor mediates feedback regulation of pancreatic secretion and postprandial release of cholecystokinin. *J. Clin. Invest.* 105: 351–359, 2000.

- [32776] 6328. Rose, T. M.; Schultz, E. R.; Todaro, G. J.: Molecular cloning of the gene for the yeast homolog (ACB) of diazepam binding inhibitor/enkephalinase. *Proc. Nat. Acad. Sci.* 89: 11287–11291, 1992.
- [32777] 6329. Spannagel, A. W.; Green, G. M.; Guan, D.; Liddle, R. A.; Faull, K.; Reeve, J. R.: Purification and characterization of a luminal cholecystokinin-releasing factor from rat intestinal secretion. *Proc. Nat. Acad. Sci.* 93: 4415–4420, 1996.
- [32778] 6330. Todd, S.; Naylor, S. L.: New chromosomal mapping assignments for arginine succinate synthetase pseudogene 1, interferon- β -3 gene, and the diazepam binding inhibitor gene. *Somat. Cell Molec. Genet.* 18: 381–385, 1992.
- [32779] 6331. Webb, N. R.; Rose, T. M.; Malik, N.; Marquardt, H.; Shoyab, M.; Todaro, G. J.; Lee, D. C.: Bovine and human cDNA sequences encoding a putative benzodiazepine receptor ligand. *DNA* 6: 71–79, 1987.
- [32780] 6332. Anagnou, N. P.; Antonarakis, S. E.; O'Brien, S. J.;

Modi, W. S.; Nienhuis, A. W.: Chromosomal localization and racial distribution of the polymorphic human dihydrofolate reductase pseudogene (DHFRPI). *Am.J. Hum. Genet.* 42: 345–352, 1988.

[32781] 6333. Anagnou, N. P.; Antonarakis, S. E.; O'Brien, S. J.; Nienhuis, A.W.: Chromosomal localization and racial distribution of the polymorphic DHFR-psi-1 pseudogene. (Abstract) *Clin. Res.* 33: 328A only, 1985.

[32782] 6334. Anagnou, N. P.; Antonarakis, S. E.; O'Brien, S. J.; Nienhuis, A.W.: A novel form of human polymorphism involving the hDHFR-psi-1 pseudogene identifies three RFLPs. *Nucleic Acids Res.* 15: 5501 only, 1987.

[32783] 6335. Anagnou, N. P.; O'Brien, S. J.; Shimada, T.; Nash, W. G.; Chen, M.-J.; Nienhuis, A. W.: Chromosomal organization of the human dihydrofolate reductase genes: dispersion, selective amplification and a novel form of polymorphism. *Proc. Nat. Acad. Sci.* 81: 5170–5174, 1984.

[32784] 6336. Blakley, R. L.; Sorrentino, B. P.: In vitro mutations in dihydrofolate reductase that confer resistance to methotrexate: potential for clinical application. *Hum. Mutat.* 11: 259–263, 1998.

[32785] 6337. Chen, M.-J.; Shimada, T.; Moulton, A. D.; Harrison, M.; Nienhuis, A. W.: Intronless human dihydrofolate reduc-

tase genes are derived from processed RNA molecules.

Proc. Nat. Acad. Sci. 79: 7435–7439, 1982.

[32786] 6338. Craik, C. S.; Rutter, W. J.; Fletterick, R.: Splice junctions: association with variation in protein structure. Science 220: 1125–1129, 1983.

[32787] 6339. Erbe, R. W.: Inborn errors of folate metabolism. New Eng. J. Med. 293: 753–757 and 807–812, 1975.

[32788] 6340. Funanage, V. L.; Myoda, T. T.; Moses, P. A.; Cowell, H. R.: Assignment of the human dihydrofolate reductase gene to the q11–q22 region of chromosome 5. Molec. Cell. Biol. 4: 2010–2016, 1984.

[32789] 6341. Hoffbrand, A. V.; Tripp, E.; Jackson, B. F. A.; Luck, W. E.; Frater-Schroder, M.: Hereditary abnormal transcobalamin II previously diagnosed as congenital dihydrofolate reductase deficiency. (Letter) New Eng. J. Med. 310: 789–790, 1984.

[32790] 6342. Killary, A. M.; Leach, R. J.; Moran, R. G.; Fournier, R. E. K.: Assignment of genes encoding dihydrofolate reductase and hexosaminidase B to Mus musculus chromosome 13. Somat. Cell Molec. Genet. 12: 641–648, 1986.

[32791] 6343. Maurer, B.; Barker, P. E.; Masters, J. N.; D'Eustachio, P.; Ruddle, F. H.; Attardi, G.: Chromosomal location of the normal human DHFR gene and of its amplified copies in

methotrexate resistant cell variants.(Abstract) Cytogenet. Cell Genet. 37: 534 only, 1984.

- [32792] 6344.Maurer, B. J.; Barker, P. E.; Masters, J. N.; Ruddle, F. H.; Attardi,G.: Human dihydrofolate reductase gene is located in chromosome 5and is unlinked to the related pseudogenes. Proc. Nat. Acad. Sci. 81:1484–1488, 1984.
- [32793] 6345.Maurer, B. J.; Carlock, L.; Wasmuth, J.; Attardi, G.: Assignmentof human dihydrofolate reductase gene to band q23 of chromosome 5and of related pseudogene psiHD1 to chromosome 3. Somat. Cell Molec.Genet. 11: 79–85, 1985.
- [32794] 6346.Myoda, T. T.; Funanage, V. L.: Personal Communication. Wilmington,Del. 10/7/1983.
- [32795] 6347.Singer, M. J.; Mesner, L. D.; Friedman, C. L.; Trask, B. J.; Hamlin,J. L.: Amplification of the human dihydrofolate reductase gene viadouble minutes is initiated by chromosome breaks. Proc. Nat. Acad.Sci. 97: 7921–7926, 2000.
- [32796] 6348.Tauro, G. P.; Danks, D. M.; Rowe, P. B.; Van der Weyden, M. B.;Schwarz, M. A.; Collins, V. L.; Neal, B. W.: Dihydrofolate reductasedeficiency causing megaloblastic anemia in two families. New Eng.J. Med. 294: 466–470, 1976.
- [32797] 6349.Walters, T. R.: Congenital megaloblastic anemia responsive toN(5)–formyltetrahydrofolic acid administration.

J. Pediat. 70: 686–687,1967.

[32798] 6350.Ali, G.; Wasco, W.; Cai, X.; Szabo, P.; Sheu, K.–F. R.; Cooper,A. J. L.; Gaston, S. M.; Gusella, J. F.; Tanzi, R. E.; Blass, J. P.: Isolation, characterization, and mapping of gene encoding dihydrolipoylsuccinyltransferase (E2k) of human alpha–ketoglutarate dehydrogenasecomplex. Somat. Cell Molec. Genet. 20: 99–105, 1994.

[32799] 6351.Nakano, K.; Matuda, S.; Sakamoto, T.; Takase, C.; Nakagawa, S.;Ohta, S.; Ariyama, T.; Inazawa, J.; Abe, T.; Miyata, T.: Human dihydrolipoamidesuccinyltransferase: cDNA cloning and localization on chromosome 14q24.2–q24.3.Biochim. Biophys. Acta 1216: 360–368, 1993.

[32800] 6352.Nakano, K.; Takase, C.; Sakamoto, T.; Ohta, S.; Nakagawa, S.; Ariyama,T.; Inazawa, J.; Abe, T.; Matuda, S.: An unspliced cDNA for humandihydrolipoamide succinyltransferase: characterization and mappingof the gene to chromosome 14q24.2–q24.3. Biochem. Biophys. Res.Commun. 196: 527–533, 1993.

[32801] 6353.Patel, M. S.; Harris, R. A.: Mammalian alpha–keto acid dehydrogenasecomplexes: gene regulation and genetic defects. FASEB J. 9: 1164–1172,1995.

[32802] 6354.McKusick, V. A.: Heritable Disorders of Connective

Tissue. St. Louis: C. V. Mosby (pub.) (4th ed.): 1972.

[32803] 6355. Palmer, G.; Manen, D.; Bonjour, J.-P.; Caverzasio, J.: Characterization of the human Glvr-1 phosphate transporter/retrovirus receptor gene and promoter region. *Gene* 226: 25-33, 1999.

[32804] 6356. Fan, N. C.; Peng, C.; Krisinger, J.; Leung, P. C. K.: The human gonadotropin-releasing hormone receptor gene: complete structure including multiple promoters, transcription initiation sites, and polyadenylation signals. *Molec. Cell. Endocr.* 107: R1-R8, 1995.

[32805] 6357. Boghosian-Sell, L.; Comings, D. E.; Overhauser, J.: Tourette syndrome in a pedigree with a 7;18 translocation: identification of a YAC spanning the translocation breakpoint at 18q22.3. *Am. J. Hum. Genet.* 59:999-1005, 1996.

[32806] 6358. Grosse, R.; Schoneberg, T.; Schultz, G.; Gudermann, T.: Inhibition of gonadotropin-releasing hormone receptor signaling by expression of a splice variant of the human receptor. *Molec. Endocr.* 11: 1305-1318, 1997.

[32807] 6359. Kaiser, U. B.; Dushkin, H.; Altherr, M. R.; Beier, D. R.; Chin, W. W.: Chromosomal localization of the gonadotropin-releasing hormone receptor gene to human chromosome 4q13.1-q21.1 and mouse chromosome 5. *Genomics* 20: 506-508, 1994.

- [32808] 6360.Kakar, S. S.; Musgrove, L. C.; Devor, D. C.; Sellers, J. C.; Neill, J. D.: Cloning, sequencing, and expression of human gonadotropin-releasing hormone (GnRH) receptor. *Biochem. Biophys. Res. Commun.* 189:289–295, 1992.
- [32809] 6361.Kakar, S. S.; Neill, J. D.: The human gonadotropin-releasing hormone receptor gene (GNRHR) maps to chromosome band 4q13. *Cytogenet. Cell Genet.* 70: 211–214, 1995.
- [32810] 6362.Kottler, M.-L.; Chauvin, S.; Lahlou, N.; Harris, C. E.; Johnston, C. J.; Lagarde, J.-P.; Bouchard, P.; Farid, N. R.; Counis, R.: A new compound heterozygous mutation of the gonadotropin-releasing hormone receptor (L314X, Q106R) in a woman with complete hypogonadotropic hypogonadism: chronic estrogen administration amplifies the gonadotropin defect. *J. Clin. Endocr. Metab.* 85: 3002–3008, 2000.
- [32811] 6363.Kottler, M.-L.; Counis, R.; Bouchard, P.: Mutations of the GnRH receptor gene: a new cause of autosomal-recessive hypogonadotropic hypogonadism. *Arch. Med. Res.* 30: 481–485, 1999.
- [32812] 6364.Kottler, M. L.; Lorenzo, F.; Bergametti, F.; Comeron, P.; Souchier, C.; Counis, R.: Subregional mapping of the human gonadotropin-releasing hormone receptor

(GnRH-R) gene to 4q between the markers D4S392 and D4S409. Hum. Genet. 96: 477–480, 1995.

- [32813] 6365. Layman, L. C.; Cohen, D. P.; Jin, M.; Xie, J.; Li, Z.; Reindollar, R. H.; Bolbolan, S.; Bick, D. P.; Sherins, R. R.; Duck, L. W.; Musgrove, L. C.; Sellers, J. C.; Neill, J. D.: Mutations in gonadotropin-releasing hormone receptor gene cause hypogonadotropic hypogonadism. (Letter) Nature-Genet. 18: 14–15, 1998.
- [32814] 6366. Leung, P. C. K.; Squire, J.; Peng, C.; Fan, N.; Hayden, M. R.; Olofsson, J. I.: Mapping of the gonadotropin-releasing hormone (GnRH) receptor gene to human chromosome 4q21.2 by fluorescence in situ hybridization. Mammalian Genome 6: 309–310, 1995.
- [32815] 6367. Mason, A. J.; Hayflick, J. S.; Zoeller, R. T.; Young, W. S., III; Phillips, H. S.; Nikolics, K.; Seeburg, P. H.: A deletion truncating the gonadotropin-releasing hormone gene is responsible for hypogonadism in the 'hpg' mouse. Science 234: 1366–1371, 1986.
- [32816] 6368. Morrison, N.; Sellar, R. E.; Boyd, E.; Eidne, K. A.; Connor, J. M.: Assignment of the gene encoding the human gonadotropin-releasing hormone receptor to 4q13.2–13.3 by fluorescence in situ hybridization. Hum. Genet. 93: 714–715, 1994.

- [32817] 6369. Pitteloud, N.; Boepple, P. A.; DeCruz, S.; Valkenburg, S. B.; Crowley, W. F., Jr.; Hayes, F. J.: The fertile eunuch variant of idiopathic hypogonadotropic hypogonadism: spontaneous reversal associated with a homozygous mutation in the gonadotropin-releasing hormone receptor. *J. Clin. Endocr. Metab.* 86: 2470–2475, 2001.
- [32818] 6370. Pralong, F. P.; Gomez, F.; Castillo, E.; Cotechia, S.; Abuin, L.; Aubert, M. L.; Portmann, L.; Gaillard, R. C.: Complete hypogonadotropic hypogonadism associated with a novel inactivating mutation of the gonadotropin-releasing hormone receptor. *J. Clin. Endocr. Metab.* 84: 3811–3816, 1999.
- [32819] 6371. Szende, B.; Srkalovic, G.; Timar, J.; Mulchahey, J. J.; Neill, J. D.; Lapis, K.; Csikos, A.; Szepeshazi, K.; Schally, A. V.: Localization of receptors for luteinizing hormone-releasing hormone in pancreatic and mammary cancer cells. *Proc. Nat. Acad. Sci.* 88: 4153–4156, 1991.
- [32820] 6372. Smith, M.; Herrell, S.; Lusher, M.; Lako, L.; Simpson, C.; Wiestner, A.; Skoda, R.; Ireland, M.; Strachan, T.: Genomic organisation of the human chordin gene and mutation screening of candidate Corneli de Lange syndrome genes. *Hum. Genet.* 105: 104–111, 1999.
- [32821] 6373. Spemann, H.; Mangold, H.: Ueber induktion von em-

bryonalanlagen durch implantation artfremder Organisatoren. Arch. Mikroskopische Anat. Entwicklungsmechanik 100: 599–638, 1924.

- [32822] 6374. Zech, L.; Haglund, U.; Nilsson, K.; Klein, G.: Characteristic chromosomal abnormalities in biopsies and lymphoid-cell lines from patients with Burkitt and non-Burkitt lymphomas. Int. J. Cancer 17:47–56, 1976.
- [32823] 6375. Bahler, M.; Kehrer, I.; Gordon, L.; Stoffler, H.-E.; Olsen, A.S.: Physical mapping of human myosin-IXB (MYO9B), the human orthologue of the rat myosin myr 5, to chromosome 19p13.1. Genomics 43: 107–109, 1997.
- [32824] 6376. Miyajima, N.; Kadowaki, Y.; Fukushige, S.; Shimizu, S.; Semba, K.; Yamanashi, Y.; Matsubara, K.; Toyoshima, K.; Yamamoto, T.: Identification of two novel members of erbA superfamily by molecular cloning: the gene products of the two are highly related to each other. Nucleic Acids Res. 16: 11057–11073, 1988.
- [32825] 6377. Qiu, Y.; Krishnan, V.; Zeng, Z.; Gilbert, D. J.; Copeland, N. G.; Gibson, L.; Yang-Feng, T.; Jenkins, N. A.; Tsai, M.-J.; Tsai, S. Y.: Isolation, characterization, and chromosomal localization of mouse and human COUP-TF I and II genes. Genomics 29: 240–246, 1995.
- [32826] 6378. MacDonald, N. J.; Freije, J. M. P.; Stracke, M. L.; Man-

row, R.E.; Steeg, P. S.: Site-directed mutagenesis of nm23-H1: mutation of proline 96 or serine 120 abrogates its motility inhibitory activity upon transfection into human breast carcinoma cells. *J. Biol. Chem.* 271:25107–25116, 1996.

[32827] 6379. Okabe-Kado, J.; Kasukabe, T.; Honma, Y.; Hayashi, M.; Henzel, W.J.; Hozumi, M.: Identity of a differentiation inhibiting factor from mouse myeloid leukemia cells with NM23/nucleoside diphosphate kinase. *Biochem. Biophys. Res. Commun.* 182: 987–994, 1992.

[32828] 6380. Postel, E. H.: Modulation of c-myc transcription by triple helix formation. *Ann. N.Y. Acad. Sci.* 660: 57–63, 1992.

[32829] 6381. Postel, E. H.; Berberich, S. J.; Flint, S. J.; Ferrone, C. A.: Human c-myc transcription factor PuF identified as nm23-H2 nucleoside diphosphate kinase, a candidate suppressor of tumor metastasis. *Science* 261:478–480, 1993.

[32830] 6382. Petek, E.; Windpassinger, C.; Vincent, J. B.; Cheung, J.; Boright, A. P.; Scherer, S. W.; Kroisel, P. M.; Wagner, K.: Disruption of a novel gene (IMMP2L) by a breakpoint in 7q31 associated with Tourette syndrome. *Am. J. Hum. Genet.* 848–858, 2001.

- [32831] 6383.Kugler, S.; Plenz, G.; Muller, P. K.: Two additional 5-prime exons in the human vigilin gene distinguish it from the chicken gene and provide the structural basis for differential routes of gene expression. *Europ.J. Biochem.* 238: 410–417, 1996.
- [32832] 6384.LeBoeuf, R. C.; Xia, Y.-R.; Oram, J. F.; Lusi, A. J.: Mapping of the gene for high-density lipoprotein binding protein (Hdlbp) to proximal mouse chromosome 1. *Genomics* 23: 296–298, 1994.
- [32833] 6385.Plenez, G.; Kugler, S.; Schnittger, S.; Rieder, H.; Fonatsch, C.; Muller, P. K.: The human vigilin gene: identification, chromosomal localization and expression pattern. *Hum. Genet.* 93: 575–582, 1994.
- [32834] 6386.Xia, Y.-R.; Klisak, I.; Sparkes, R. S.; Oram, J.; Lusi, A. J.: Localization of the gene for high-density lipoprotein binding protein (HDLBP) to human chromosome 2q37. *Genomics* 16: 524–525, 1993.
- [32835] 6387.Kaelbling, M.; Eddy, R.; Shows, T. B.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Klinger, H. P.; O'Hara, B.: Localization of the human gene allowing infection by gibbon ape leukemia virus to human chromosome region 2q11–q14 and to the homologous region on mouse chromosome 2. *J. Virol.* 65: 1743–1747, 1991.

- [32836] 6388.Kavanaugh, M. P.; Miller, D. G.; Zhang, W.; Law, W.; Kozak, S.L.; Kabat, D.; Miller, A. D.: Cell-surface receptors for gibbon apeleukemia virus and amphotropic murine retrovirus are inducible sodium-dependentphosphate symporters. *Proc. Nat. Acad. Sci.* 91: 7071–7075, 1994.
- [32837] 6389.O'Hara, B.; Johann, S. V.; Klinger, H. P.; Blair, D. G.; Robinson,H.; Dunn, K. J.; Sass, P.; Vitek, S. M.; Robins, T.: Characterizationof a human gene conferring sensitivity to infection by gibbon apeleukemia virus. *Cell Growth Differ.* 1: 119–127, 1990.
- [32838] 6390.Palmer, G.; Manen, D.; Bonjour, J.–P.; Caverzasio, J.: Species-specificmechanisms control the activity of the Pit1/PIT1 phosphate transporter gene promoter in mouse and human. *Gene* 279: 49–62, 2001.
- [32839] 6391.Hayashi, K.; Yano, H.; Hashida, T.; Takeuchi, R.; Takeda, O.; Asada,K.; Takahashi, E.; Kato, I.; Sobue, K.: Genomic structure of thehuman caldesmon gene. *Proc. Nat. Acad. Sci.* 89: 12122–12126, 1992.
- [32840] 6392.Humphrey, M. B.; Herrera–Sosa, H.; Gonzalez, G.; Lee, R.; Bryan,J.: Cloning of cDNAs encoding human caldesmons. *Gene* 112: 197–204,1992.
- [32841] 6393.Dickson, K. M.; Bergeron, J. J. M.; Shames, I.; Colby, J.; Nguyen,D. T.; Chevet, E.; Thomas, D. Y.; Snipes, G. J.:

Association of calnexin with mutant peripheral myelin protein-22 ex vivo: a basis for 'gain-of-function' ER diseases. Proc. Nat. Acad. Sci. 99: 9852–9857, 2002.

- [32842] 6394. Gray, P. W.; Byers, M. G.; Eddy, R. L.; Shows, T. B.: The assignment of the calnexin gene to the q35 region of chromosome 5. (Abstract) Human Genome Mapping Workshop 93 9 only, 1993.
- [32843] 6395. Schrag, J. D.; Bergeron, J. J. M.; Li, Y.; Borisova, S.; Hahn, M.; Thomas, D. Y.; Cygler, M.: The structure of calnexin, an ER chaperone involved in quality control of protein folding. Molec. Cell 8: 633–644, 2001.
- [32844] 6396. Tjoelker, L. W.; Seyfried, C. E.; Eddy, R. L., Jr.; Byers, M. G.; Shows, T. B.; Calderon, J.; Schreiber, R. B.; Gray, P. W.: Human, mouse, and rat calnexin cDNA cloning: identification of potential calcium binding motifs and gene localization to human chromosome 5. Biochemistry 33: 3229–3236, 1994.
- [32845] 6397. Richard, I.; Beckmann, J. S.: Molecular cloning of mouse canp3, the gene associated with limb-girdle muscular dystrophy 2A in human. Mammalian Genome 7: 377–379, 1996.
- [32846] 6398. Bataille, N.; Schmitt, N.; Aumercier-Maes, P.; Ollivier, B.; Lucas-Heron, B.; Lestienne, P.: Molecular cloning of hu-

man calmitine, a mitochondrialcalcium binding protein, reveals identity with calsequestrine. *Biochem.Biophys. Res. Commun.* 203: 1477–1482, 1994.

[32847] 6399.Fujii, J.; Willard, H. F.; MacLennan, D. H.: Characterizationand localization to human chromosome 1 of human fast-twitch skeletal muscle calsequestrin gene. *Somat. Cell Molec. Genet.* 16: 185–189,1990.

[32848] 6400.Chen, C.-S.; Bejeck, B. E.; Kersey, J. H.: A mapping study of13 genes on human chromosome bands 4q11–q25. *Cytogenet. Cell Genet.* 69:260–265, 1995.

[32849] 6401.Fujiwara, Y.; Miwa, M.; Nogami, M.; Okumura, K.; Nobori, T.; Suzuki,T.; Ueda, M.: Genomic organization and chromosomal localization ofthe human casein gene family. *Hum. Genet.* 99: 368–373, 1997.

[32850] 6402.Rijnkels, M.; Meershoek, E.; de Boer, H. A.; Pieper, F. R.: Physicalmap and localization of the human casein gene locus. *Mammalian Genome* 8:285–286, 1997.

[32851] 6403.Voglino, G. F.; Ponzzone, A.: Polymorphism in human casein. *NatureN.B.* 238: 149 only, 1972.

[32852] 6404.Ferretti, L.; Leone, P.; Sgaramella, V.: Long range restrictionanalysis of the bovine casein genes. *Nucleic Acids Res.* 18: 6829–6833,1990.

[32853] 6405.Threadgill, D. W.; Womack, J. E.: Genomic analysis of

the major bovine milk protein genes. *Nucleic Acids Res.* 18: 6935–6942, 1990.

- [32854] 6406. Kurotaki, N.; Imaizumi, K.; Harada, N.; Masuno, M.; Kondoh, T.; Nagai, T.; Ohashi, H.; Naritomi, K.; Tsukahara, M.; Makita, Y.; Sugimoto, T.; Sonoda, T.; and 11 others: Haploinsufficiency of NSD1 causes Sotos syndrome. *Nature Genet.* 30: 365–366, 2002.
- [32855] 6407. Whang-Peng, J.; Triche, T. J.; Knutsen, T.; Miser, J.; Kao-Shan, S.; Tsai, S.; Israel, M. A.: Cytogenetic characterization of selected small round cell tumors of childhood. *Cancer Genet. Cytogenet.* 21:185–208, 1986.
- [32856] 6408. Mount, L. A.; Reback, S.: Familial paroxysmal choreoathetosis: preliminary report on a hitherto undescribed clinical syndrome. *Arch. Neurol. Psychiat.* 44: 841–847, 1940.
- [32857] 6409. Lee, S.; Lin, M.; Mele, A.; Cao, Y.; Farmer, J.; Russo, D.; Redman, C.: Proteolytic processing of big endothelin-3 by the Kell blood group protein. *Blood* 94: 1440–1450, 1999.
- [32858] 6410. Cyr, C.; Huebner, K.; Druck, T.; Kris, R.: Cloning and chromosomal localization of a human endothelin ETA receptor. *Biochem. Biophys. Res. Commun.* 181: 184–190, 1991.

- [32859] 6411.Hosoda, K.; Nakao, K.; Tamura, N.; Arai, H.; Ogawa, Y.; Suga, S.;Nakanishi, S.; Imura, H.: Organization, structure, chromosomal assignment,and expression of the gene encoding the human endothelin–A receptor. *J.Biol. Chem.* 267: 18797–18804, 1992.
- [32860] 6412.Tzourio, C.; El Amrani, M.; Poirier, O.; Nicaud, V.; Bousser, M.–G.;Alperovitch, A.: Association between migraine and endothelin typeA receptor (ETA –231 A/G) gene polymorphism. *Neurology* 56: 1273–1277,2001.
- [32861] 6413.Amiel, J.; Attie, T.; Jan, D.; Pelet, A.; Edery, P.; Bidaud, C.;Lacombe, D.; Tam, P.; Simeoni, J.; Flori, E.; Nihoul–Fekete, C.; Munnich,A.; Lyonnet, S.: Heterozygous endothelin receptor B (EDNRB) mutationsin isolated Hirschsprung disease. *Hum. Molec. Genet.* 5: 355–357,1996.
- [32862] 6414.Arai, H.; Nakao, K.; Takaya, K.; Hosoda, K.; Ogawa, Y.; Nakanishi,S.; Imura, H.: The human endothelin–B receptor gene: structural organizationand chromosomal assignment. *J. Biol. Chem.* 268: 3463–3470, 1993.
- [32863] 6415.Attie, T.; Till, M.; Pelet, A.; Amiel, J.; Edery, P.; Boutrand,L.; Munnich, A.; Lyonnet, S.: Mutation of the endothelin–receptorB gene in Waardenburg–Hirschsprung disease. *Hum. Molec. Genet.* 4:2407–2409, 1995.

- [32864] 6416. Auricchio, A.; Griseri, P.; Carpentieri, M. L.; Betsos, N.; Staiano, A.; Tozzi, A.; Priolo, M.; Thompson, H.; Boccia-
rdi, R.; Romeo, G.; Ballabio, A.; Ceccherini, I.: Double het-
erozygosity for a RET substitution inferring with splicing
and an EDNRB missense mutation in Hirschsprung disease.
(Letter) Am. J. Hum. Genet. 64: 1216–1221, 1999.
- [32865] 6417. Carrasquillo, M. M.; McCallion, A. S.; Puffenberger,
E. G.; Kashuk, C. S.; Nouri, N.; Chakravarti, A.: Genome-
wide association study and mouse model identify interac-
tion between RET and EDNRB pathways in Hirschsprung
disease. Nature Genet. 32: 237–244, 2002.
- [32866] 6418. Ceccherini, I.; Zhang, A. L.; Matera, I.; Yang, G.; De-
voto, M.; Romeo, G.; Cass, D. T.: Interstitial deletion of the
endothelin-B receptor gene in the spotting lethal (sl) rat.
Hum. Molec. Genet. 4: 2089–2096, 1995.
- [32867] 6419. Chakravarti, A.: Endothelin receptor-mediated sig-
naling in Hirschsprung disease. Hum. Molec. Genet. 5:
303–307, 1996.
- [32868] 6420. Elshourbagy, N. A.; Adamou, J. E.; Gagnon, A. W.;
Wu, H.-L.; Pullen, M.; Nambi, P.: Molecular characterization
of a novel human endothelin receptor splice variant. J. Biol.
Chem. 271: 25300–25307, 1996.
- [32869] 6421. Gariepy, C. E.; Cass, D. T.; Yanagisawa, M.: Null mu-

tation of endothelin receptor type B gene in spotting lethal rats causes aganglionic megacolon and white coat color. Proc. Nat. Acad. Sci. 93: 867–872, 1996.

[32870] 6422. Gariepy, C. E.; Ohuchi, T.; Williams, S. C.; Richardson, J. A.; Yanagisawa, M.: Salt-sensitive hypertension in endothelin-B receptor-deficient rats. J. Clin. Invest. 105: 925–933, 2000.

[32871] 6423. Gariepy, C. E.; Williams, S. C.; Richardson, J. A.; Hammer, R. E.; Yanagisawa, M.: Transgenic expression of the endothelin-B receptor prevents congenital intestinal aganglionosis in a rat model of Hirschsprung disease. J. Clin. Invest. 102: 1092–1101, 1998.

[32872] 6424. Gross, A.; Kunze, J.; Maier, R. F.; Stoltenburg-Didinger, G.; Grimmer, I.; Obladen, M.: Autosomal-recessive neural crest syndrome with albinism, black lock, cell migration disorder of the neurocytes of the gut, and deafness: ABCD syndrome. Am. J. Med. Genet. 56: 322–326, 1995.

[32873] 6425. Hofstra, R. M. W.; Osinga, J.; Buys, C. H. C. M.: Mutations in Hirschsprung disease: when does a mutation contribute to the phenotype? Europ. J. Hum. Genet. 5: 180–185, 1997.

[32874] 6426. Hosoda, K.; Hammer, R. E.; Richardson, J. A.; Bay-

nash, A. G.;Cheung, J. C.; Giaid, A.; Yanagisawa, M.: Targeted and natural (piebald-lethal)mutations of endothelin-B receptor gene produce megacolon associatedwith spotted coat color in mice. *Cell* 79: 1267-1276, 1994.

[32875] 6427.Charlton, P.; Guida, L.; Copeland, N.; Jenkins, N.; Munroe, D.;Greenberg, F.; Fiedorek, F. T.; Nicholls, R. D.: Genetic approachto function of the neuropeptide galanin. (Abstract) *Am. J. Hum. Genet.* 53(suppl.): A1137, 1993.

[32876] 6428.Evans, H.; Baumgartner, M.; Shine, J.; Herzog, H.: Genomic organizationand localization of the gene encoding human preprogalanin. *Genomics* 18:473-477, 1993.

[32877] 6429.Evans, H. F.; Shine, J.: Human galanin: molecular cloning reveals a unique structure. *Endocrinology* 129: 1682-1684, 1991.

[32878] 6430.Harris, G. W.: Neural control of the pituitary gland. *Physiol.Rev.* 28: 139-179, 1948.

[32879] 6431.Holmes, F. E.; Mahoney, S.; King, V. R.; Bacon, A.; Kerr, N. C.H.; Pachnis, V.; Curtis, R.; Priestley, J. V.; Wynick, D.: Targetedisruption of the galanin gene reduces the number of sensory neuronsand their regenerative capacity. *Proc. Nat. Acad. Sci.* 97: 11563-11568,2000.

[32880] 6432.Lopez, F. J.; Merchenthaler, I.; Ching, M.; Wisniewski, M. G.;Negro-Vilar, A.: Galanin: a hypothalamic-hy-

pophysiotropic hormonemodulating reproductive functions. Proc. Nat. Acad. Sci. 88: 4508–4512,1991.

- [32881] 6433.Lundkvist, J.; Land, T.; Kahl, U.; Bedecs, K.; Bartfai, T.: cDNAsequence, ligand binding, and regulation of galanin/GMAP in mousebrain. Neurosci. Lett. 200: 121–124, 1995.
- [32882] 6434.O'Meara, G.; Coumis, U.; Ma, S. Y.; Kehr, J.; Mahoney, S.; Bacon,A.; Allen, S. J.; Holmes, F.; Kahl, U.; Wang, F. H.; Kearns, I. R.;Ove–Ogren, S.; Dawbarn, D.; Mufson, E. J.; Davies, C.; Dawson, G.;Wynick, D.: Galanin regulates the postnatal survival of a subsetof basal forebrain cholinergic neurons. Proc. Nat. Acad. Sci. 97:11569–11574, 2000.
- [32883] 6435.Rattan, S.: Role of galanin in the gut. Gastroenterology 100:1762–1768, 1991.
- [32884] 6436.Schmidt, W. E.; Kratzin, H.; Eckart, K.; Drevs, D.; Mundkowski,G.; Clemens, A.; Katsoulis, S.; Schafer, H.; Gallwitz, B.; Creutzfeldt,W.: Isolation and primary structure of pituitary human galanin, a30–residue nonamidated neuropeptide. Proc. Nat. Acad. Sci. 88: 11435–11439,1991.
- [32885] 6437.Steiner, R. A.; Hohmann, J. G.; Holmes, A.; Wrenn, C. C.; Cadd,G.; Jureus, A.; Clifton, D. K.; Luo, M.; Gutshall,

M.; Ma, S. Y.; Mufson, E. J.; Crawley, J. N.: Galanin transgenic mice display cognitive and neurochemical deficits characteristic of Alzheimer's disease. *Proc. Nat. Acad. Sci.* 98: 4184–4189, 2001.

[32886] 6438. Wynick, D.; Small, C. J.; Bacon, A.; Holmes, F. E.; Norman, M.; Ormandy, C. J.; Kilic, E.; Kerr, N. C. H.; Ghatel, M.; Talamantes, F.; Bloom, S. R.; Pachnis, V.: Galanin regulates prolactin release and lactotroph proliferation. *Proc. Nat. Acad. Sci.* 95: 12671–12676, 1998.

[32887] 6439. Puech, A.; Saint-Jore, B.; Funke, B.; Gilbert, D. J.; Sirotkin, H.; Copeland, N. G.; Jenkins, N. A.; Kucherlapati, R.; Morrow, B.; Skoultschi, A. I.: Comparative mapping of the human 22q11 chromosomal region and the orthologous region in mice reveals complex changes in gene organization. *Proc. Nat. Acad. Sci.* 94: 14608–14613, 1997.

[32888] 6440. van Hille, B.; Vanek, M.; Richener, H.; Green, J. R.; Bilbe, G.: Cloning and tissue distribution of subunits C, D, and E of the human vacuolar H(+)-ATPase. *Biochem. Biophys. Res. Commun.* 97: 15–21, 1993.

[32889] 6441. Lewis, J.; Dickson, D. W.; Lin, W.-L.; Chisholm, L.; Corral, A.; Jones, G.; Yen, S.-H.; Sahara, N.; Skipper, L.; Yager, D.; Eckman, C.; Hardy, J.; Hutton, M.; McGowan, E.: Enhanced neurofibrillary degeneration in transgenic mice

expressing mutant tau and APP. *Science* 293:1487–1491, 2001.

[32890] 6442.Denker, S. P.; Huang, D. C.; Orlowski, J.; Furthmayr, H.; Barber, D. L.: Direct binding of the Na–H exchanger NHE1 to ERM proteins regulates the cortical cytoskeleton and cell shape independently of H(+) translocation. *Molec. Cell* 6: 1425–1436, 2000.

[32891] 6443.Dudley, C. R. K.; Giuffra, L. A.; Tippet, P.; Kidd, K. K.; Reeder, S. T.: The Na⁺/H⁺ antiporter: a 'melt' polymorphism allows regional mapping to the short arm of chromosome 1. *Hum. Genet.* 86: 79–83, 1990.

[32892] 6444.Franchi, A.; Perucca–Lostanlen, D.; Pouyssegur, J.: Functional expression of a human Na⁺/H⁺ antiporter gene transfected into antiporter–deficient mouse L cells. *Proc. Nat. Acad. Sci.* 83: 9388–9392, 1986.

[32893] 6445.Lifton, R. P.; Sardet, C.; Pouyssegur, J.; Lalouel, J.–M.: Cloning of the human genomic amiloride–sensitive Na⁺/H⁺ antiporter gene, identification of genetic polymorphisms, and localization on the genetic map of chromosome 1p. *Genomics* 7: 131–135, 1990.

[32894] 6446.Mattei, M.–G.; Galloni, M.; Sardet, C.; Franchi, A.; Counillon, L.; Passage, E.; Pouyssegur, J.: Localization of the antiporter gene (APNH) and chromosomal homology

between human 1p, mouse 4 and Chinese hamster 2q.

(Abstract) Cytogenet. Cell Genet. 51: 1041, 1989.

[32895] 6447. Mattei, M.-G.; Sardet, C.; Franchi, A.; Pouyssegur, J.: Chromosomal mapping of the amiloride-sensitive Na^+/H^+ antiporter gene. (Abstract) Cytogenet. Cell Genet. 46: 658–659, 1987.

[32896] 6448. Mattei, M.-G.; Sardet, C.; Franchi, A.; Pouyssegur, J.: The human amiloride-sensitive Na^+/H^+ antiporter: localization to chromosome 1 by in situ hybridization. Cytogenet. Cell Genet. 48: 6–8, 1988.

[32897] 6449. Mendoza, S. A.: The Na^+/H^+ antiport is a mediator of cell proliferation. Acta Paediat. Scand. 76: 545–547, 1987.

[32898] 6450. Morahan, G.; Rakar, S.: Localization of the mouse Na^+/H^+ exchanger gene on distal chromosome 4. Genomics 15: 231–232, 1993.

[32899] 6451. Sardet, C.; Franchi, A.; Pouyssegur, J.: Molecular cloning, primary structure, and expression of the human growth factor-activatable Na^+/H^+ antiporter. Cell 56: 271–280, 1989.

[32900] 6452. Davare, M. A.; Avdonin, V.; Hall, D. D.; Peden, E. M.; Burette, A.; Weinberg, R. J.; Horne, M. C.; Hoshi, T.; Hell, J. W.: A beta-2 adrenergic receptor signaling complex as–

sembled with the Ca(2+) channelCa(V)1.2. Science 293: 98–101, 2001.

[32901] 6453.Jacobs, P. A.; Brunton, M.; Frackiewicz, A.; Newton, M.; Cook,P. J. L.; Robson, E. B.: Studies on a family with three cytogeneticmarkers. Ann. Hum. Genet. 33: 325–336, 1970.

[32902] 6454.Brown, A.; Bernier, G.; Mathieu, M.; Rossant, J.; Kothary, R.:The mouse dystonia musculorum gene is a neural isoform of bullouspemphigoid antigen 1. Nature Genet. 10: 301–306, 1995.

[32903] 6455.Brown, A.; Dalpe, G.; Mathieu, M.; Kothary, R.: Cloning and characterizationof the neural isoforms of human dystonin. Genomics 29: 777–780,1995.

[32904] 6456.Copeland, N. G.; Gilbert, D. J.; Li, K.; Sawamura, D.; Giudice,G. J.; Chu, M.–L.; Jenkins, N. A.; Uitto, J.: Chromosomal localizationof mouse bullous pemphigoid antigens, BPAG1 and BPAG2: identificationof a new region of homology between mouse and human chromosomes. Genomics 15:180–181, 1993.

[32905] 6457.Diaz, L. A.; Ratrie, H., III; Saunders, W. S.; Futamura, S.; Squiquera,H. L.; Anhalt, G. J.; Giudice, G. J.: Isolation of a human epidermalcDNA corresponding to the 180–kD autoantigen recognized by bullouspemphigoid and herpes

gestationis sera: immunolocalization of this protein to the hemidesmosome. *J. Clin. Invest.* 86: 1088–1094, 1990.

[32906] 6458. Guo, L.; Degenstein, L.; Dowling, J.; Yu, Q.-C.; Wollmann, R.; Perman, B.; Fuchs, E.: Gene targeting of BPAG1: abnormalities in mechanical strength and cell migration in stratified epithelia and neurologic degeneration. *Cell* 81: 233–243, 1995.

[32907] 6459. Minoshima, S.; Amagai, M.; Kudoh, J.; Fukuyama, R.; Hashimoto, T.; Nishikawa, T.; Shimizu, N.: Localization of the human gene for 230-kDa bullous pemphigoid autoantigen to the pter-q15 region of chromosome 6. (Abstract) *Cytogenet. Cell Genet.* 58: 1914–1915, 1991.

[32908] 6460. Minoshima, S.; Amagai, M.; Kudoh, J.; Fukuyama, R.; Hashimoto, T.; Nishikawa, T.; Shimizu, N.: Localization of the human gene for 230-kDa bullous pemphigoid autoantigen (BPAG1) to chromosome 6pter-q15. *Cytogenet. Cell Genet.* 57: 30–32, 1991.

[32909] 6461. Ryynanen, M.; Knowlton, R. G.; Kero, M.; Sawamura, D.; Li, K.; Giudice, G. J.; Diaz, L. A.; Uitto, J.: Bullous pemphigoid antigens (BPAGs): identification of RFLPs in human BPAG1 and BPAG2, and exclusion as candidate genes in a large kindred with dominant epidermolysis bullosa simplex. *Genomics* 11: 1025–1029, 1991.

- [32910] 6462.Sawamura, D.; Nomura, K.; Sugita, Y.; Mattei, M.–G.; Chu, M.–L.; Knowlton, R.; Uitto, J.: Bullous pemphigoid antigen (BPAG1): cDNA cloning and mapping of the gene to the short arm of human chromosome 6. *Genomics* 8: 722–726, 1990.
- [32911] 6463.Stanley, J. R.; Tanaka, T.; Mueller, S.; Klaus–Kovtun, V.; Roop, D.: Isolation of complementary DNA for bullous pemphigoid antigen by use of patients' autoantibodies. *J. Clin. Invest.* 82: 1864–1870, 1988.
- [32912] 6464.Tamai, K.; Sawamura, D.; Do, H. C.; Tamai, Y.; Li, K.; Uitto, J.: The human 230–kD bullous pemphigoid antigen gene (BPAG1): exon–intron organization and identification of regulatory tissue specific elements in the promoter region. *J. Clin. Invest.* 92: 814–822, 1993.
- [32913] 6465.Yang, Y.; Bauer, C.; Strasser, G.; Wollman, R.; Julien, J.–P.; Fuchs, E.: Integrators of the cytoskeleton that stabilize microtubules. *Cell* 98:229–238, 1999.
- [32914] 6466.Tashian, R. E.: Genetics of the mammalian carbonic anhydrases. *Adv.Genet.* 30: 321–356, 1992.
- [32915] 6467.Montgomery, J. C.; Shows, T. B.; Venta, P. J.; Tashian, R. E.: Gene for novel human carbonic anhydrase (CA) isozyme on chromosome 16 is unlinked to the CA1/CA2/CA3 gene cluster. (Abstract) *Am. J. Hum. Genet.*

41: A229 only, 1987.

- [32916] 6468. Montgomery, J. C.; Venta, P. J.; Eddy, R. L.; Fukushima, Y.-S.; Shows, T. B.; Tashian, R. E.: Characterization of the human gene for a newly discovered carbonic anhydrase, CA VII, and its localization to chromosome 16. *Genomics* 11: 835–848, 1991.
- [32917] 6469. Venta, P. J.; Montgomery, J. C.; Tashian, R. E.: Molecular genetics of carbonic anhydrase isozymes. *Isozymes: Curr. Top. Biol. Med. Res.* 14: 59–72, 1987.
- [32918] 6470. Blake, N. M.: Genetic variants of carbonic anhydrase in the Asian-Pacific area. *Ann. Hum. Biol.* 5: 557–568, 1978.
- [32919] 6471. Blake, N. M.; Kirk, R. L.: Widespread distribution of variant forms of carbonic anhydrase in Australian aborigines. *Med. J. Aust.* 1: 183–185, 1978.
- [32920] 6472. Butterworth, P.; Barlow, J.; Konialis, C.; Povey, S.; Edwards, Y. H.: The assignment of human erythrocyte carbonic anhydrase CA I to chromosome 8. (Abstract) *Cytogenet. Cell Genet.* 40: 597 only, 1985.
- [32921] 6473. Carter, N. D.: Carbonic anhydrase isozymes in *Cavia porcellus*, *Cavia aperea* and their hybrids. *Comp. Biochem. Physiol. B* 43: 743–747, 1972.
- [32922] 6474. Carter, N. D.: Carbonic anhydrase II polymorphism

in Africa. Hum.Hered. 22: 539–541, 1972.

- [32923] 6475.Carter, N. D.; Tashian, R. E.; Huntsman, R. G.; Sacker, L.: Characterization of two new variants of red cell carbonic anhydrase in the British population: Ca le Portsmouth and Ca le Hull. Am. J. Hum. Genet. 24:330–338, 1972.
- [32924] 6476.Davis, M. B.; West, L. F.; Barlow, J. H.; Butterworth, P. H. W.; Lloyd, J. C.; Edwards, Y. H.: Regional localization of carbonic anhydrase genes CA1 and CA3 on human chromosome 8. Somat. Cell Molec. Genet. 13:173–178, 1987.
- [32925] 6477.Davis, M. B.; West, L. F.; Butterworth, P.; Edwards, Y. H.: The assignment of human carbonic anhydrases CA1 and CA3 to chromosome 8q13–22. (Abstract) 7th Int. Cong. Hum. Genet., Berlin 616 only, 1986.
- [32926] 6478.DeSimone, J.; Linde, M.; Tashian, R. E.: Evidence for linkage of carbonic anhydrase isozyme genes in the pig-tailed macaque, *Macaca nemestrina*. Nature N.B. 242: 55–56, 1973.
- [32927] 6479.Dodgson, S. J.; Tashian, R. E.; Gross, G.; Carter, N. D.: The Carbonic Anhydrases: Cellular Physiology and Molecular Genetics. New York: Plenum, 1991.
- [32928] 6480.Edwards, Y. H.; Barlow, J. H.; Konialis, C. P.; Povey, S.; Butterworth, P. H. W.: Assignment of the gene deter–

mining human carbonic anhydrase,CAI, to chromosome 8.
Ann. Hum. Genet. 50: 123–129, 1986.

[32929] 6481.Eicher, E. M.; Stern, R. H.; Womack, J. E.; Davisson, M. T.; Roderick,T. H.; Reynolds, S. C.: Evolution of mammalian carbonic anhydraseloci by tandem duplication: close linkage of Car-1 and Car-2 to thecentromere region of chromosome 3 of the mouse. Biochem. Genet. 14:651–660, 1976.

[32930] 6482.Goriki, K.; Tashian, R. E.; Stroup, S. K.; Yu, Y.-S. L.; Henriksson,D. M.: Chemical characterization of a new Japanese variant of carbonicanhydrase I, Ca 2 (Nagasaki 1) (76 arg-to-gln). Biochem. Genet. 17:449–460, 1979.

[32931] 6483.Hewett-Emmett, D.; Tashian, R. E.: Functional diversity, conservationand convergence in the evolution of the alpha-, beta-, and gamma-carbonicanhydrase gene families. Molec. Phylogenet. Evol. 5: 50–77, 1996.

[32932] 6484.Kere, J.; Ruutu, T.; Davies, K. A.; Roninson, I. B.; Watkins, P.C.; Winqvist, R.; de la Chapelle, A.: Chromosome 7 long arm deletionin myeloid disorders: a narrow breakpoint region in 7q22 defined bymolecular mapping. Blood 73: 230–234, 1989.

[32933] 6485.Neufeld, E. J.: Personal Communication. Boston, Mass. 2/21/1995.

- [32934] 6486. Neufeld, E. J.; Skalnik, D. G.; Lievens, P. M.-J.; Orkin, S. H.: Human CCAAT displacement protein is homologous to the Drosophila homeoprotein, cut. *Nature Genet.* 1: 50–55, 1992.
- [32935] 6487. Scherer, S. W.; Neufeld, E. J.; Lievens, P. M.-J.; Orkin, S. H.; Kim, J.; Tsui, L.-C.: Regional localization of the CCAAT displacement protein gene (CUTL1) to 7q22 by analysis of somatic cell hybrids. *Genomics* 15:695–696, 1993.
- [32936] 6488. Snyder, S. R.; Wang, J.; Waring, J. F.; Ginder, G. D.: Identification of CCAAT displacement protein (CDP/cut) as a locus-specific repressor of major histocompatibility complex gene expression in human tumor cells. *J. Biol. Chem.* 276: 5323–5330, 2001.
- [32937] 6489. Zeng, W. R.; Scherer, S. W.; Koutsilieris, M.; Huizenga, J. J.; Filteau, F.; Tsui, L.-C.; Nepveu, A.: Loss of heterozygosity and reduced expression of the CUTL1 gene in uterine leiomyomas. *Oncogene* 14:2355–2365, 1997.
- [32938] 6490. Blackburn, M. R.; Datta, S. K.; Kellems, R. E.: Adenosine deaminase-deficient mice generated using a two-stage genetic engineering strategy exhibit a combined immunodeficiency. *J. Biol. Chem.* 273: 5093–5100, 1998.
- [32939] 6491. Blaese, R. M.; Culver, K. W.; Miller, A. D.; Carter, C.

S.; Fleisher, T.; Clerici, M.; Shearer, G.; Chang, L., Chiang, Y.; Tolstoshev, P.; Greenblatt, J. J.; Rosenberg, S. A.; Klein, H.; Berger, M.; Mullen, C. A.; Ramsey, W. J.; Muul, L.; Morgan, R. A.; Anderson, W. F.: T lymphocyte-directed gene therapy for ADA-SCID: initial trial results after 4 years. *Science* 270: 475-480, 1995.

[32940] 6492. Bollinger, M. E.; Arredondo-Vega, F. X.; Santisteban, I.; Schwarz, K.; Hershey, M. S.; Lederman, H. M.: Hepatic dysfunction as a complication of adenosine deaminase deficiency. *New Eng. J. Med.* 334:1367-1371, 1996.

[32941] 6493. Bonthron, D. T.; Markham, A. F.; Ginsburg, D.; Orkin, S. H.: Identification of a point mutation in the adenosine deaminase gene responsible for immunodeficiency. *J. Clin. Invest.* 76: 894-897, 1985.

[32942] 6494. Bordignon, C.; Notarangelo, L. D.; Nobili, N.; Ferrari, G.; Casorati, G.; Panina, P.; Mazzolari, E.; Maggioni, D.; Rossi, C.; Servida, P.; Ugazio, A. G.; Mavilio, F.: Gene therapy in peripheral blood lymphocytes and bone marrow for ADA: immunodeficient patients. *Science* 270:470-475, 1995.

[32943] 6495. Bortin, M. M.; Rimm, A. A. (eds.): Severe combined immunodeficiency disease: characterization of the disease and results of transplantation. *J.A.M.A.* 238:591-600,

1977.

- [32944] 6496.Boss, G. R.; Thompson, L. F.; O'Connor, R. D.; Ziering, R. W.;Seegmiller, J. E.: Ecto-5-prime-nucleotidase deficiency: associationwith adenosine deaminase deficiency and nonassociation with deoxyadenosinetoxicity. Clin. Immun. Immunopath. 19: 1-7, 1981.
- [32945] 6497.Bottini, N.; De Luca, D.; Saccucci, P.; Fiumara, A.; Elia, M.;Porfirio, M. C.; Lucarelli, P.; Curatolo, P.: Autism: evidence ofassociation with adenosine deaminase genetic polymorphism. Neurogenetics 3:111-113, 2001.
- [32946] 6498.Chen, S.-H.; Ochs, H. D.; Scott, C. R.: Adenosine deaminase deficiency:disappearance of adenine deoxynucleotides from a patient's erythrocytesafter successful marrow transplantation. J. Clin. Invest. 62: 1386-1389,1978.
- [32947] 6499.Chen, S.-H.; Ochs, H. D.; Scott, C. R.; Giblett, E. R.: Adenosinedeaminase and nucleoside phosphorylase activity in patients with immunodeficiencysyndromes. Clin. Immun. Immunopath. 13: 156-160, 1979.
- [32948] 6500.Chen, S.-H.; Scott, C. R.; Giblett, E. R.: Adenosine deaminase:demonstration of a 'silent' gene associated with combined immunodeficiencydisease. Am. J. Hum. Genet. 26: 103-107, 1974.

- [32949] 6501.Cohen, A.; Hirschhorn, R.; Horowitz, S. D.; Rubinstein, A.; Polmar, S. H.; Hong, R.; Martin, D. W., Jr.: Deoxyadenosine triphosphate as a potentially toxic metabolite in adenosine deaminase deficiency. *Proc.Nat. Acad. Sci.* 75: 472–476, 1978.
- [32950] 6502.Cook, P. J. L.; Hopkinson, D. A.; Robson, E. B.: The linkage relationships of adenosine deaminase. *Ann. Hum. Genet.* 34: 187–188, 1970.
- [32951] 6503.Creagan, R. P.; Tischfield, J. A.; Nichols, E. A.; Ruddle, F.H.: Autosomal assignment of the gene for the form of adenosine deaminase which is deficient in patients with combined immunodeficiency syndrome. (Letter) *Lancet* II: 1449 only, 1973.
- [32952] 6504.Culver, K. W.: *Gene Therapy: A Primer for Physicians.* Larchmont, New York: Mary Ann Liebert, Inc. , 1996.
- [32953] 6505.Daddona, P. E.; Kelley, W. N.: Human adenosine deaminase: stoichiometry of the adenosine deaminase-binding protein complex. *Biochim. Biophys. Acta* 580: 302–311, 1979.
- [32954] 6506.Daddona, P. E.; Mitchell, B. S.; Meuwissen, H. J.; Davidson, B.L.; Wilson, J. M.; Koller, C. A.: Adenosine deaminase deficiency with normal immune function: an acidic enzyme mutation. *J. Clin. Invest.* 72: 483–492,

1983.

- [32955] 6507.Detter, J. C.; Stamatoyannopoulos, G.; Giblett, E. R.; Motulsky, A. G.: Adenosine deaminase: racial distribution and report of a new phenotype. *J. Med. Genet.* 7: 356–357, 1970.
- [32956] 6508.Dissing, J.; Knudsen, B.: Adenosine–deaminase deficiency and combined immunodeficiency syndrome. (Letter) *Lancet* II: 1316, 1972.
- [32957] 6509.Dissing, J.; Knudsen, J. B.: A new red cell adenosine deaminase phenotype in man. *Hum. Hered.* 19: 375–377, 1969.
- [32958] 6510.Dyminski, J. W.; Daoud, A.; Lampkin, B. C.; Limouze, S.; Donofrio, J.; Coleman, M. S.; Hutton, J. J.: Immunological and biochemical profiles in response to transfusion therapy in adenosine deaminase–deficient patient with severe combined immunodeficiency disease. *Clin. Immun. Immunopath.* 14: 307–326, 1979.
- [32959] 6511.Egashira, M.; Ariga, T.; Kawamura, N.; Miyoshi, O.; Niikawa, N.; Sakiyama, Y.: Visible integration of the adenosine deaminase (ADA) gene into the recipient genome after gene therapy. *Am. J. Med. Genet.* 75:314–317, 1998.
- [32960] 6512.Ellis, N. A.; Lennon, D. J.; Proytcheva, M.; Alhadeff, B.; Henderson, E. E.; German, J.: Somatic intragenic recom-

ination within the mutated locus BLM can correct the high SCE phenotype of Bloom syndrome cells. *Am.J. Hum. Genet.* 57: 1019–1027, 1995.

- [32961] 6513. Ferrari, G.; Rossini, S.; Giavazzi, R.; Maggioni, D.; Nobili, N.; Soldati, M.; Ungers, G.; Mavilio, F.; Gilboa, E.; Bordignon, C.: An in vivo model of somatic cell gene therapy for human severe combined immunodeficiency. *Science* 251: 1363–1366, 1991.
- [32962] 6514. Giblett, E. R.; Anderson, J. E.; Cohen, F.; Pollara, B.; Meuwissen, H. J.: Adenosine–deaminase deficiency in two patients with severely impaired cellular immunity. *Lancet* I: 1067–1069, 1972.
- [32963] 6515. Hart, S. L.; Lane, A. B.; Jenkins, T.: Partial adenosine deaminase deficiency: another family from southern Africa. *Hum. Genet.* 74:307–312, 1986.
- [32964] 6516. Herbschleb–Voogt, E.; Scholten, J.–W.; Meera Khan, P.: Basic defect in the expression of adenosine deaminase in ADA SCID disease. II. Deficiency of ADA–CRM detected in heterozygote human–Chinese hamster cell hybrids. *Hum. Genet.* 63: 121–125, 1983.
- [32965] 6517. Hershfield, M. S.: PEG–ADA: an alternative to haploidentical bone marrow transplantation and an adjunct to gene therapy for adenosine deaminase deficiency. *Hum.*

Mutat. 5: 107–112, 1995.

- [32966] 6518.Hershfield, M. S.; Buckley, R. H.; Greenberg, M. L.; Melton, A.L.; Schiff, R.; Hatem, C.; Kurtzberg, J.; Markert, M. L.; Kobayashi,R. H.; Kobayashi, A. L.; Abuchowski, A.: Treatment of adenosine deaminasedeficiency with polyethylene glycol–modified adenosine deaminase. NewEng. J. Med. 316: 589–596, 1987.
- [32967] 6519.Hershfield, M. S.; Kredich, N. M.: S–adenosylhomocysteine hydrolaseis an adenosine–binding protein: a target for adenosine toxicity. Science 202:757–760, 1978.
- [32968] 6520.Presley, J. F.; Ward, T. H.; Pfeifer, A. C.; Siggia, E. D.; Phair,R. D.; Lippincott–Schwartz, J.: Dissection of COPI and Arf1 dynamicsin vivo and role in Golgi membrane trans–port. Nature 417: 187–193,2002.
- [32969] 6521.McGuire, R. E.; Daiger, S. P.; Green, E. D.: Localiza–tion andcharacterization of the human ADP–ribosylation factor 5 (ARF5) gene. Genomics 41:481–484, 1997.
- [32970] 6522.Tsuchiya, M.; Price, S. R.; Tsai, S.–C.; Moss, J.; Vaughan, M.: Molecular identification of ADP–ribosylation factor mRNAs and their expression in mammalian cells. J. Biol. Chem. 266: 2772–2777, 1991.
- [32971] 6523.Chang, C.–Y.; Wu, D.–A.; Lai, C.–C.; Miller, W. L.;

Chung, B.-C.: Cloning and structure of the human adrenodoxin gene. DNA 7: 609–615, 1988.

[32972] 6524. Chang, C.-Y.; Wu, D.-A.; Mohandas, T. K.; Chung, B.-C.: Structure, sequence, chromosomal location, and evolution of the human ferredoxin gene family. DNA Cell Biol. 9: 205–212, 1990.

[32973] 6525. Lachman, H. M.; Papolos, D. F.; Saito, T.; Yu, Y. M.; Szumlanski, C. L.; Weinshilboum, R. M.: Human catechol-O-methyltransferase pharmacogenetics: description of a functional polymorphism and its potential application to neuropsychiatric disorders. Pharmacogenetics 6: 243–250, 1996.

[32974] 6526. Tiihonen, J.; Hallikainen, T.; Lachman, H.; Saito, T.; Volavka, J.; Kauhanen, J.; Salonen, J. T.; Ryyanen, O.-P.; Koulou, M.; Karvonen, M. K.; Pohjalainen, T.; Syvalahti, E.; Hietala, J.: Association between the functional variant of the catechol-O-methyltransferase (COMT) gene and type 1 alcoholism. Molec. Psychiat. 4: 286–289, 1999.

[32975] 6527. Bohren, K. M.; Bullock, B.; Wermuth, B.; Gabbay, K. H.: The aldo-ketoreductase superfamily: cDNAs and deduced amino acid sequences of human aldehyde and aldose reductases. J. Biol. Chem. 264: 9547–9551, 1989.

[32976] 6528. Charlesworth, D.: Starch-gel electrophoresis of four

enzymes from human red blood cells: glyceraldehyde-3-phosphate dehydrogenase, fructoaldolase, glyoxalase II and sorbitol dehydrogenase. *Ann. Hum. Genet.* 35: 477–484, 1972.

[32977] 6529. Frade, R.; Balbo, M.; Barel, M.: RB18A, whose gene is localized on chromosome 17q12–q21.1, regulates in vivo p53 transactivating activity. *Cancer Res.* 60: 6585–6589, 2000.

[32978] 6530. Wolffe, A. P.: Transcriptional control: sinful repression. *Nature* 387:16–17, 1997.

[32979] 6531. Wu, L.; Aster, J. C.; Blacklow, S. C.; Lake, R.; Artavanis-Tsakonas, S.; Griffin, J. D.: MAML1, a human homologue of *Drosophila* mastermind, is a transcriptional co-activator for NOTCH receptors. *Nature Genet.* 26:484–489, 2000.

[32980] 6532. Inohara, N.; Koseki, T.; del Peso, L.; Hu, Y.; Yee, C.; Chen, S.; Carrio, R.; Merino, J.; Liu, D.; Ni, J.; Nunez, G.: Nod1, an Apaf-1-like activator of caspase-9 and nuclear factor-kappa-B. *J. Biol. Chem.* 274:14560–14567, 1999.

[32981] 6533. Charroux, B.; Pellizzoni, L.; Parkinson, R. A.; Shevchenko, A.; Mann, M.; Dreyfuss, G.: Gemin3: a novel DEAD box protein that interacts with SMN, the spinal muscular atrophy gene product, and is a component of gems.

J. Cell Biol. 147: 1181–1193, 1999.

- [32982] 6534.Grundhoff, A. T.; Kremmer, E.; Tureci, O.; Gleden, A.; Gindorf,C.; Atz, J.; Mueller–Lantzsch, N.; Schubach, W. H.; Grasser, F. A.: Characterization of DP103, a novel DEAD box protein that binds tothe Epstein–Barr virus nuclear proteins EBNA2 and EBNA3C. J. Biol.Chem. 274: 19136–19144, 1999.
- [32983] 6535.Ruteshouser, E. C.; Ashworth, L. K.; Huff, V.: Absence of PPP2R1Amutations in Wilms tumor. Oncogene 20: 2050–2054, 2001.
- [32984] 6536.Everett, A. D.; Xue, C.; Stoops, T.: Developmental expressionof protein phosphatase 2A in the kidney. J. Am. Soc. Nephrol. 10:1737–1745, 1999.
- [32985] 6537.Hemmings, B. A.; Adams–Pearson, C.; Maurer, F.; Muller, P.; Goris,J.; Merlevede, W.; Hofsteenge, J.; Stone, S. R.: Alpha– and beta–formsof the 65–kDa subunit of protein phosphatase 2A have a similar 39amino acid repeating structure. Biochemistry 29: 3166–3173, 1990.
- [32986] 6538.Hendrix, P.; Turowski, P.; Mayer–Jaekel, R. E.; Goris, J.; Hofsteenge,J.; Merlevede, W.; Hemmings, B. A.: Analysis of subunit isoformsin protein phosphatase 2A holoenzymes from rabbit and Xenopus. J.Biol. Chem. 268: 7330–7337, 1993.

- [32987] 6539.Hunt, S. L.; Hsuan, J. J.; Totty, N.; Jackson, R. J.: unr, a cellularcytoplasmic RNA-binding protein with five cold-shock domains, is requiredfor internal initiation of translation of human rhinovirus RNA. *GenesDev.* 13: 437-448, 1999.
- [32988] 6540.Matsuda, S.; Katsumata, R.; Okuda, T.; Yamamoto, T.; Miyazaki,K.; Senga, T.; Machida, K.; Thant, A. A.; Nakatsugawa, S.; Hamaguchi,M.: Molecular cloning and characterization of human MAWD, a novelprotein containing WD-40 repeats frequently overexpressed in breast-cancer. *Cancer Res.* 60: 13-17, 2000.
- [32989] 6541.Levy, C.; Nechushtan, H.; Razin, E.: A new role for the STAT3inhibitor, PIAS3: a repressor of microphthalmia transcription factor. *J.Biol. Chem.* 277: 1962-1966, 2002.
- [32990] 6542.Ueki, N.; Seki, N.; Yano, K.; Saito, T.; Masuho, Y.; Muramatsu,M.: Isolation and chromosomal assignment of a human gene encodingprotein inhibitor of activated STAT3 (PIAS3). *J. Hum. Genet.* 44:193-196, 1999.
- [32991] 6543.Pasteris, N. G.; Trask, B. J.; Sheldon, S.; Gorski, J. L.: Discordantphenotype of two overlapping deletions involving the PAX3 gene inchromosome 2q35. *Hum. Molec. Genet.* 2: 953-959, 1993.
- [32992] 6544.Beeson, D.; Brydson, M.; Betty, M.; Jeremiah, S.;

Povey, S.; Vincent, A.; Newsom-Davis, J.: Primary structure of the human muscle acetylcholinereceptor cDNA cloning of the gamma and epsilon subunits. *Europ. J. Biochem.* 215: 229–238, 1993.

[32993] 6545. Ohno, K.; Hutchinson, D. O.; Milone, M.; Brengman, J. M.; Bouzat, C.; Sine, S. M.; Engel, A. G.: Congenital myasthenic syndrome caused by prolonged acetylcholine receptor channel openings due to a mutation in the M2 domain of the epsilon subunit. *Proc. Nat. Acad. Sci.* 92: 758–762, 1995.

[32994] 6546. Ohno, K.; Quiram, P. A.; Milone, M.; Wang, H.-L.; Harper, M. C.; Pruitt, J. N., II; Brengman, J. M.; Pao, L.; Fischbeck, K. H.; Crawford, T. O.; Sine, S. M.; Engel, A. G.: Congenital myasthenic syndromes due to heteroallelic nonsense/missense mutations in the acetylcholinereceptor epsilon subunit gene: identification and functional characterization of six new mutations. *Hum. Molec. Genet.* 6: 753–766, 1997.

[32995] 6547. Ohno, K.; Wang, H.-L.; Milone, M.; Bren, N.; Brengman, J. M.; Nakano, S.; Quiram, P.; Pruitt, J. N.; Sine, S. M.; Engel, A. G.: Congenital myasthenic syndrome caused by decreased agonist binding affinity due to a mutation in the acetylcholine receptor epsilon subunit. *Neuron*

17:157–170, 1996.

- [32996] 6548.Sieb, J. P.; Dorfler, P.; Tzartos, S.; Wewer, U. M.; Ruegg, M.A.; Meyer, D.; Baumann, I.; Lindemuth, R.; Jakschik, J.; Ries, F.: Congenital myasthenic syndromes in two kinships with end-plate acetylcholinereceptor and utrophin deficiency. *Neurology* 50: 54–61, 1998.
- [32997] 6549.Sieb, J. P.; Kraner, S.; Rauch, M.; Steinlein, O. K.: Immature end-plates and utrophin deficiency in congenital myasthenic syndrome caused by epsilon-AChR subunit truncating mutations. *Hum. Genet.* 107:160–164, 2000.
- [32998] 6550.Witzemann, V.; Schwarz, H.; Koenen, M.; Berberich, C.; Villarroel, A.; Wernig, A.; Brenner, H. R.; Sakmann, B.: Acetylcholine receptor epsilon-subunit deletion causes muscle weakness and atrophy in juvenile and adult mice. *Proc. Nat. Acad. Sci.* 93: 13286–13291, 1996.
- [32999] 6551.Bartels, C. F.; Zelinski, T.; Lockridge, O.: Mutation at codon 322 in the human acetylcholinesterase (ACHE) gene accounts for Y blood group polymorphism. *Am. J. Hum. Genet.* 52: 928–936, 1993.
- [33000] 6552.Coates, P. M.; Simpson, N. E.: Genetic variation in human erythrocyte acetylcholinesterase. *Science* 175: 1466–1467, 1972.
- [33001] 6553.Ehrlich, G.; Viegas-Pequignot, E.; Ginzberg, D.; Sin-

del, L.; Soreq, H.; Zakut, H.: Mapping the human acetylcholinesterase gene to chromosome 7q22 by fluorescent in situ hybridization coupled with selective PCR amplification from a somatic hybrid cell panel and chromosome-sorted DNA libraries. *Genomics* 13: 1192–1197, 1992.

[33002] 6554. Feng, G.; Krejci, E.; Molgo, J.; Cunningham, J. M.; Massoulie, J.; Sanes, J. R.: Genetic analysis of collagen Q: roles in acetylcholinesterase and butyrylcholinesterase assembly and in synaptic structure and function. *J. Cell Biol.* 144: 1349–1360, 1999.

[33003] 6555. Getman, D. K.; Eubanks, J. H.; Camp, S.; Evans, G. A.; Taylor, P.: The human gene encoding acetylcholinesterase is located on the long arm of chromosome 7. *Am. J. Hum. Genet.* 51: 170–177, 1992.

[33004] 6556. Lapidot-Lifson, Y.; Prody, C. A.; Ginzberg, D.; Meytes, D.; Zakut, H.; Soreq, H.: Coamplification of human acetylcholinesterase and butyrylcholinesterase genes in blood cells: correlation with various leukemias and abnormal megakaryocytopoiesis. *Proc. Nat. Acad. Sci.* 86: 4715–4719, 1989.

[33005] 6557. Meshorer, E.; Erb, C.; Gazit, R.; Pavlovsky, L.; Kaufer, D.; Friedman, A.; Glick, D.; Ben-Arie, N.; Soreq, H.: Alternative splicing and neuritic mRNA translocation under

long-term neuronal hypersensitivity. *Science* 295:508–512, 2002.

- [33006] 6558. Rachinsky, T. L.; Crenshaw, E. B., III; Taylor, P.: Assignment of the gene for acetylcholinesterase to distal mouse chromosome 5. *Genomics* 14:511–514, 1992.
- [33007] 6559. Rotundo, R. L.; Gomez, A. M.; Fernandez-Valle, C.; Randall, W. R.: Allelic variants of acetylcholinesterase: genetic evidence that all acetylcholinesterase forms in avian nerves and muscles are encoded by a single gene. *Proc. Nat. Acad. Sci.* 85: 7805–7809, 1988.
- [33008] 6560. Shapira, M.; Tur-Kaspa, I.; Bosgraaf, L.; Livni, N.; Grant, A. D.; Grisaru, D.; Korner, M.; Ebstein, R. P.; Soreq, H.: A transcription-activating polymorphism in the ACHE promoter associated with acute sensitivity to anti-acetylcholinesterases. *Hum. Molec. Genet.* 9: 1273–1281, 2000.
- [33009] 6561. Drayna, D. T.; McLean, J. W.; Wion, K. L.; Trent, J. M.; Drabkin, H. A.; Lawn, R. M.: Human apolipoprotein D gene: gene sequence, chromosomal localization, and homology to the alpha-2mu-globulin superfamily. *DNA* 6:199–204, 1987.
- [33010] 6562. Fielding, P. E.; Fielding, C. J.: A cholesteryl ester transfer complex in human plasma. *Proc. Nat. Acad. Sci.*

77: 3327–3330, 1980.

- [33011] 6563.Graham, C. A.; McGrew, W. C.: Menstrual synchrony in female undergraduates living on a coeducational campus. *Psychoneuroendocrinology* 5: 245–252, 1980.
- [33012] 6564.Kamboh, M. I.; Albers, J. J.; Majumder, P. P.; Ferrell, R. E.: Genetic studies of human apolipoproteins. IX. Apolipoprotein D polymorphism and its relation to serum lipoprotein lipid levels. *Am. J. Hum. Genet.* 45:147–154, 1989.
- [33013] 6565.McClintock, M. K.: Menstrual synchrony and suppression. *Nature* 229:244–245, 1971.
- [33014] 6566.McClintock, M. K.: Estrous synchrony and its mediation by airborne chemical communication (*Rattus norvegicus*). *Horm. Behav.* 10: 264–276, 1978.
- [33015] 6567.Quadagno, D. M.; Shubeita, H. E.; Deck, J.; Francoeur, D.: Influence of male social contacts, exercise and all-female living conditions on the menstrual cycle. *Psychoneuroendocrinology* 6: 239–244, 1981.
- [33016] 6568.Warden, C. H.; Diep, A.; Taylor, B. A.; Lusi, A. J.: Localization of the gene for apolipoprotein D on mouse chromosome 16. *Genomics* 12:851–852, 1992.
- [33017] 6569.Zeng, C.; Spielman, A. I.; Vowles, B. R.; Leyden, J. J.; Biemann, K.; Preti, G.: A human axillary odorant is carried

by apolipoproteinD. Proc. Nat. Acad. Sci. 93: 6626–6630, 1996.

- [33018] 6570.Hopkinson, D. A.; Coppock, J. S.; Muhlemann, M. F.; Edwards, Y.H.: The detection and differentiation of the products of the humancarbonic anhydrase loci, Ca I and Ca II, using fluorogenic substrates. Ann.Hum. Genet. 38: 155–162, 1974.
- [33019] 6571.Cleutjens, C. B. J. M.; van Eekelen, C. C. E. M.; van Dekken, H.;Smit, E. M. E.; Hagemeijer, A.; Wagner, M. J.; Wells, D. E.; Trapman,J.: The human C/EBP–delta (CRP3/CELF) gene: structure and chromosomallocalization. Genomics 16: 520–523, 1993.
- [33020] 6572.Williams, S. C.; Cantwell, C. A.; Johnson, P. F.: A family ofC/EBP–related proteins capable of forming covalently linked leucinezipper dimers in vitro. Genes Dev. 5: 1553–1567, 1991.
- [33021] 6573.Al–Awqati, Q.; Preisig, P. A.: Size does matter: will knockoutof p21(WAF1/CIP1) save the kidney by limiting compensatory renal growth?(Commentary) Proc. Nat. Acad. Sci. 96: 10551–10553, 1999.
- [33022] 6574.Chedid, M.; Michieli, P.; Lengel, C.; Huppi, K.; Givol, D.: Asingle nucleotide substitution at codon 31 (ser/arg) defines a polymorphismin a highly conserved region of

the p53-inducible gene WAF1/CIP1. *Oncogene*

9:3021–3024, 1994.

[33023] 6575.Cheng, T.; Rodrigues, N.; Shen, H.; Yang, Y.; Dom-bkowski, D.; Sykes,M.; Scadden, D. T.: Hematopoietic stem cell quiescence maintainedby p21(cip1/waf1). *Science* 287: 1804–1808, 2000.

[33024] 6576.Harper, J. W.; Adami, G. R.; Wei, N.; Keyomarsi, K.; Elledge, S.J.: The p21 Cdk-interacting protein Cip1 is a potent inhibitor ofG1 cyclin-dependent kinases. *Cell* 75: 805–816, 1993.

[33025] 6577.Huppi, K.; Siwarski, D.; Dosik, J.; Michieli, P.; Chedid, M.; Reed,S.; Mock, B.; Givol, D.; Mushinski, J. F.: Molecular cloning, sequencing,chromosomal localization and ex-pression of mouse p21 (Waf1). *Oncogene* 9:3017–3020, 1994.

[33026] 6578.Megyesi, J.; Price, P. M.; Tamayo, E.; Safirstein, R. L.: Thelack of a functional p21(WAF1/CIP1) gene ameliorates progression tochronic renal failure. *Proc. Nat. Acad. Sci.* 96: 10830–10835, 1999.

[33027] 6579.Mousses, S.; Ozcelik, H.; Lee, P. D.; Malkin, D.; Bull, S. B.;Andrulis, I. L.: Two variants of the CIP1/WAF1 gene occur togetherand are associated with human cancer. *Hum. Molec. Genet.* 4: 1089–1092,1995.

- [33028] 6580.Iwata, N.; Tsubuki, S.; Takaki, Y.; Shirotani, K.; Lu, B.; Gerard, N. P.; Gerard, C.; Hama, E.; Lee, H.-J.; Saido, T. C.: Metabolic regulation of brain A-beta by neprilysin. *Science* 292: 1550–1552, 2001.
- [33029] 6581.Lu, P.-J.; Wulf, G.; Zhou, X. Z.; Davies, P.; Lu, K. P.: The prolyl isomerase Pin1 restores the function of Alzheimer-associated phosphorylated tau protein. *Nature* 399: 784–788, 1999.
- [33030] 6582.Kageoka, T.; Hewett-Emmett, D.; Stroup, S. K.; Yu, Y.-S. L.; Tashian, R. E.: Amino acid substitution and chemical characterization of a Japanese variant of carbonic anhydrase I: CA I Hiroshima-1 (86 asp-to-gly). *Biochem. Genet.* 19: 535–549, 1981.
- [33031] 6583.Kendall, A. G.; Tashian, R. E.: Erythrocyte carbonic anhydrase I: inherited deficiency in humans. *Science* 197: 471–472, 1977.
- [33032] 6584.Lindskog, S.; Henderson, L. E.; Kannan, K. K.; Liljas, A.; Nyman, P. O.; Strandberg, B.: Carbonic anhydrase. In: Boyer, P. D.: *The Enzymes*. New York: Academic Press (pub.) 5: 1971. Pp. 587–665.
- [33033] 6585.McGeer, P. L.; McGeer, E. G.: Polymorphisms in inflammatory genes and the risk of Alzheimer disease. *Arch. Neurol.* 58: 1790–1792, 2001.

- [33034] 6586.Modi, W. S.; Masuda, A.; Yamada, M.; Oppenheim, J. J.; Matsushima,K.; O'Brien, S. J.: Chromosomal localization of the human interleukin1 alpha (IL-1-alpha) gene. *Genomics* 2: 310-314, 1988.
- [33035] 6587.Mosley, B.; Urdal, D. L.; Prickett, K. S.; Larsen, A.; Cosman,D.; Conlon, P. J.; Gillis, S.; Dower, S. K.: The interleukin-1 receptorbinds the human interleukin-1-alpha precursor but not the interleukin-1-betaprecursor. *J. Biol. Chem.* 262: 2941-2944, 1987.
- [33036] 6588.Bailly, S.; di Giovine, F. S.; Blakemore, A. I. F.; Duff, G. W.: Genetic polymorphism of human interleukin-1-alpha. *Europ. J. Immun.* 23:1240-1245, 1993.
- [33037] 6589.Boultwood, J.; Breckon, G.; Birch, D.; Cox, R.: Chromosomal localizationof murine interleukin-1 alpha and beta genes. *Genomics* 5: 481-485,1989.
- [33038] 6590.Cox, A.; Camp, N. J.; Cannings, C.; di Giovine, F. S.; Dale, M.;Worthington, J.; John, S.; Ollier, W. E. R.; Silman, A. J.; Duff,G. W.: Combined sib-TDT and TDT provide evidence for linkage of theinterleukin-1 gene cluster to erosive rheumatoid arthritis. *Hum.Molec. Genet.* 8: 1707-1713, 1999.
- [33039] 6591.Cox, A.; Camp, N. J.; Nicklin, M. J. H.; di Giovine, F. S.; Duff,G. W.: An analysis of linkage disequilibrium in the

interleukin-1 gene cluster, using a novel grouping method for multiallelic markers. *Am.J. Hum. Genet.* 62: 1180-1188, 1998.

- [33040] 6592. Diehl, S. R.; Wang, Y.; Brooks, C. N.; Burmeister, J. A.; Califano, J. V.; Wang, S.; Schenkein, H. A.: Linkage disequilibrium of interleukin-1 genetic polymorphisms with early-onset periodontitis. *J. Periodont.* 70:418-430, 1999.
- [33041] 6593. Du, Y.; Dodel, R. C.; Eastwood, B. J.; Bales, K. R.; Gao, F.; Lohmuller, F.; Muller, U.; Kurz, A.; Zimmer, R.; Evans, R. M.; Hake, A.; Gasser, T.; Oertel, W. H.; Griffin, W. S. T.; Paul, S. M.; Farlow, M. R.: Association of an interleukin 1-alpha polymorphism with Alzheimer's disease. *Neurology* 55: 480-484, 2000.
- [33042] 6594. Furutani, Y.; Notake, M.; Fukui, T.; Ohue, M.; Nomura, H.; Yamada, M.; Nakamura, S.: Complete nucleotide sequence of the gene for human interleukin 1 alpha. *Nucleic Acids Res.* 14: 3167-3179, 1986.
- [33043] 6595. Gray, P. W.; Glaister, D.; Chen, E.; Goeddel, D. V.; Pennica, D.: Two interleukin 1 genes in the mouse: cloning and expression of the cDNA for murine interleukin 1-beta. *J. Immun.* 137: 3644-3648, 1986.
- [33044] 6596. Grimaldi, L. M. E.; Casadei, V. M.; Ferri, C.; Veglia, F.; Licastro, F.; Annoni, G.; Biunno, I.; De Bellis, G.; Sorbi, S.;

Mariani, C.;Canal, N.; Griffin, W. S. T.: Association of early-onset Alzheimer'sdisease with an interleukin-1-alpha gene polymorphism. *Ann. Neurol.* 47:361-365, 2000.

[33045] 6597.Hogquist, K. A.; Nett, M. A.; Unanue, E. R.; Chaplin, D. D.:Interleukin 1 is processed and released during apoptosis. *Proc. Nat.Acad. Sci.* 88: 8485-8489, 1991.

[33046] 6598.Hurwitz, A.; Loukides, J.; Ricciarelli, E.; Botero, L.; Katz,E.; McAllister, J. M.; Garcia, J. E.; Rohan, R.; Adashi, E. Y.; Hernandez,E. R.: Human intraovarian interleukin-1 (IL-1) system: highly compartmentalizedand hormonally dependent regulation of the genes encoding IL-1, itsreceptor, and its receptor antagonist. *J. Clin. Invest.* 89: 1746-1754,1992.

[33047] 6599.Ki, C.-S.; Na, D. L.; Kim, D. K.; Kim, H. J.; Kim, J.-W.: Lackof association of the interleukin-1-alpha gene polymorphism with Alzheimer'sdisease in a Korean population. (Letter) *Ann. Neurol.* 49: 817-818,2001.

[33048] 6600.Kolsch, H.; Ptok, U.; Bagli, M.; Papassotiropoulos, A.; Schmitz,S.; Barkow, K.; Kockler, M.; Rao, M. L.; Maier, W.; Heun, R.: Genepolymorphisms of interleukin-1-alpha influence the course of Alzheimer'sdisease. (Letter) *Ann. Neurol.* 49: 818-819, 2001.

- [33049] 6601.Kornman, K. S.; Crane, A.; Wang, H.-Y.; di Giovine, F. S.; Newman,M. G.; Pirk, F. W.; Wilson, T. G., Jr.; Higginbottom, F. L.; Duff,G. W.: The interleukin-1 genotype as a severity factor in adult periodontaldisease. J. Clin. Periodont. 24: 72-77, 1997.
- [33050] 6602.Lafage, M.; Maroc, N.; Dubreuil, P.; de Waal Malefijt, R.; Pebusque,M.-J.; Carcassonne, Y.; Mannoni, P.: The human interleukin-1-alphagene is located on the long arm of chromosome 2 at band q13. Blood 73:104-107, 1989.
- [33051] 6603.Lord, P. C. W.; Wilmoth, L. M. G.; Mizel, S. B.; McCall, C. E.: Expression of interleukin-1 alpha and beta genes by human bloodpolymorphonuclear leukocytes. J. Clin. Invest. 87: 1312-1321, 1991.
- [33052] 6604.Murphy, G. M., Jr.; Claassen, J. D.; DeVoss, J. J.; Pascoe, N.;Taylor, J.; Tinklenberg, J. R.; Yesavage, J. A.: Rate of cognitivedecline in AD is accelerated by the interleukin-1-alpha -889 *1 allele. Neurology 56:1595-1597, 2001.
- [33053] 6605.Stamey, T. A.; Yang, N.; Hay, A. R.; McNeal, J. E.; Freiha, F.S.; Redwine, E.: Prostate-specific antigen as a serum marker foradenocarcinoma of the prostate. New Eng. J. Med. 317: 909-916, 1987.
- [33054] 6606.Mallinson, G.; Soo, K. S.; Schall, T. J.; Pisacka, M.; Anstee,D. J.: Mutations in the erythrocyte chemokine re-

ceptor (Duffy) gene:the molecular basis of the Fy(a)/Fy(b) antigens and identification of a deletion in the Duffy gene of an apparently healthy individual with the Fy(a-b-) phenotype. Brit. J. Haemat. 90: 823-829, 1995.

[33055] 6607. Miller, L. H.; Mason, S. J.; Clyde, D. F.; McGinnis, M. H.: The resistance factor to *Plasmodium vivax* in blacks: the Duffy blood group genotype, FyFy. New Eng. J. Med. 295: 302-304, 1976.

[33056] 6608. Miller, L. H.; Mason, S. J.; Dvorak, J. A.: Erythrocyte receptors of *Plasmodium knowlesi* malaria: Duffy blood group determinants. Science 189:561-562, 1975.

[33057] 6609. Nance, W. E.; Conneally, M.; Kang, K. W.; Reed, T. E.; Schroder, J.; Rose, S.: Genetic linkage analysis of human hemoglobin variants. Am. J. Hum. Genet. 22: 453-459, 1970.

[33058] 6610. Nichols, M. E.; Rubinstein, P.; Barnwell, J.; Rodriguez de Cordoba, S.; Rosenfield, R. E.: A new human Duffy blood group specificity defined by a murine monoclonal antibody: immunogenetics and association with susceptibility to *Plasmodium vivax*. J. Exp. Med. 166: 776-785, 1987.

[33059] 6611. Olsson, M. L.; Smythe, J. S.; Hansson, C.; Poole, J.; Mallinson, G.; Jones, J.; Avent, N. D.; Daniels, G.: The Fy(x)

phenotype is associated with a missense mutation in the Fy(b) allele predicting Arg89Cys in the Duffy glycoprotein. Brit. J. Haemat. 103: 1184–1191, 1998.

[33060] 6612. Palmer, C. G.; Christian, J. C.; Merritt, A. D.: Partial trisomy 1 due to a 'shift' and probable location of the Duffy (Fy) locus. Am. J. Hum. Genet. 29: 371–377, 1977.

[33061] 6613. Parasol, N.; Reid, M.; Rios, M.; Castilho, L.; Harari, I.; Kosower, N. S.: A novel mutation in the coding sequence of the FY*B allele of the Duffy chemokine receptor gene is associated with an altered erythrocyte phenotype. Blood 92: 2237–2243, 1998.

[33062] 6614. Pasvol, G.; Wilson, R. J. M.: The interaction of malaria parasites with red blood cells. Brit. Med. Bull. 38: 133–140, 1982.

[33063] 6615. Peiper, S. C.; Wang, Z.; Neote, K.; Martin, A. W.; Showell, H. J.; Conklyn, M. J.; Ogborne, K.; Hadley, T. J.; Lu, Z.; Hesselgesser, J.; Horuk, R.: The Duffy antigen/receptor for chemokines (DARC) is expressed in endothelial cells of Duffy negative individuals who lack the erythrocyte receptor. J. Exp. Med. 181: 1311–1317, 1995.

[33064] 6616. Ritter, H.: Zur formalen Genetik des Duffy-systems. Untersuchung von 247 Familien. Humangenetik 4: 59–61, 1967.

- [33065] 6617.Robson, E. B.; Cook, P. J. L.; Corney, G.; Hopkinson, D. A.; Noades,J.; Cleghorn, T. E.: Linkage data on Rh, PGM, PGD, peptidase C and Fy from family studies. *Ann. Hum. Genet.* 36: 393–399, 1973.
- [33066] 6618.Szabo, M. C.; Soo, K. S.; Zlotnik, A.; Schall, T. J.: Chemokine class differences in binding to the Duffy antigen–erythrocyte chemokine receptor. *J. Biol. Chem.* 270: 25348–25351, 1995.
- [33067] 6619.Tamasauskas, D.; Powell, V.; Saksela, K.; Yazdanbakhsh, K.: A homologous naturally occurring mutation in Duffy and CCR5 leading to reduced receptor expression. *Blood* 97: 3651–3654, 2001.
- [33068] 6620.Tang, T.; Owen, J. D.; Du, J.; Walker, C. L.; Richmond, A.: Molecular cloning and characterization of a mouse gene with homology to the Duffy–antigen receptor for chemokines. *DNA Seq.* 9: 129–143, 1999.
- [33069] 6621.Tournamille, C.; Kim Le Van, C.; Gane, P.; Le Pennec, P. Y.; Roubinet, F.; Babinet, J.; Cartron, J. P.; Colin, Y.: Arg89Cys substitution results in very low membrane expression of the Duffy antigen/receptor for chemokines in Fy(x) individuals. *Blood* 92: 2147–2156, 1998. Note: Erratum. *Blood* 95: 2753 only, 2000.
- [33070] 6622.Tournamille, C.; Le Van Kim, C.; Gane, P.; Cartron,

J.-P.; Colin, Y.: Molecular basis and PCR-DNA typing of the Fya/fyb blood group polymorphism. *Hum. Genet.* 95: 407–410, 1995.

[33071] 6623. Weitkamp, L. R.: Personal Communication. Rochester, N. Y. 1972.

[33072] 6624. Zimmerman, P. A.; Woolley, I.; Masinde, G. L.; Miller, S. M.; McNamara, D. T.; Hazlett, F.; Mgone, C. S.; Alpers, M. P.; Genton, B.; Boatin, B. A.; Kazura, J. W.: Emergence of FY*A(null) in a *Plasmodium vivax*-endemic region of Papua New Guinea. *Proc. Nat. Acad. Sci.* 96:13973–13977, 1999.

[33073] 6625. Anderson, S. E.; McKenzie, J. L.; McLoughlin, K.; Beard, M. E. J.; Hart, D. N. J.: The inheritance of abnormal sialoglycoproteins found in a Gerbich negative individual. *Pathology* 18: 407–412, 1986.

[33074] 6626. Anstee, D. J.; Parsons, S. F.; Ridgwell, K.; Tanner, M. J. A.; Merry, A. H.; Thomson, E. E.; Judson, P. A.; Johnson, P.; Bates, S.; Fraser, I. D.: Two individuals with elliptocytic red cells apparently lack three minor erythrocyte membrane sialoglycoproteins. *Biochem. J.* 218: 615–619, 1984.

[33075] 6627. Barnes, R.; Lewis, T. L. T.: A rare antibody (anti-Ge) causing hemolytic disease of the newborn. *Lancet* II: 1285–1286, 1961.

[33076] 6628. Bennett, V.: The membrane skeleton of human ery-

throcytes and its implications for more complex cells.

Annu. Rev. Biochem. 54: 273–304, 1985.

[33077] 6629. Bloomfield, L.; Rowe, G. P.; Green, C.: The Webb (Wb) antigen in South Wales donors. Hum. Hered. 36: 352–356, 1986.

[33078] 6630. Booth, P. B.; McLoughlin, K.: The Gerbich blood group system, especially in Melanesians. Vox Sang. 22: 73–84, 1972.

[33079] 6631. Cartron, J.-P.; Colin, Y.; Kudo, S.; Fukuda, M.: Molecular genetics of human erythrocyte sialoglycoproteins A, B, C, and D. In: Harris, J. R.: Erythroid Cells. Blood Cell Biochemistry. New York: Plenum Press (pub.) 1: 1990. Pp. 299–335.

[33080] 6632. Chang, S.; Reid, M. E.; Conboy, J.; Kan, Y. W.; Mohandas, N.: Molecular characterization of erythrocyte glycophorin C variants. Blood 77: 644–648, 1991.

[33081] 6633. Ming, G.; Wong, S. T.; Henley, J.; Yuan, X.; Song, H.; Spitzer, N. C.; Poo, M.: Adaptation in the chemotactic guidance of nerve growth cones. Nature 417: 411–418, 2002.

[33082] 6634. Ozcelik, T.; Rosenthal, A.; Francke, U.: Chromosomal mapping of brain-derived neurotrophic factor and neurotrophin-3 genes in man and mouse. Genomics 10:

569–575, 1991.

- [33083] 6635. Zuccato, C.; Ciammola, A.; Rigamonti, D.; Leavitt, B. R.; Goffredo, D.; Conti, L.; MacDonald, M. E.; Friedlander, R. M.; Silani, V.; Hayden, M. R.; Timmusk, T.; Sipione, S.; Cattaneo, E.: Loss of huntingtin-mediated BDNF gene transcription in Huntington's disease. *Science* 293: 493–498, 2001.
- [33084] 6636. Ichimura–Ohshima, Y.; Morii, K.; Ichimura, T.; Araki, K.; Takahashi, Y.; Isobe, T.; Minoshima, S.; Fukuyama, R.; Shimizu, N.; Kuwano, R.: cDNA cloning and chromosome assignment of the gene for human brain 14–3–3 protein eta chain. *J. Neurosci. Res.* 31: 600–605, 1992.
- [33085] 6637. Muratake, T.; Hayashi, S.; Ichikawa, T.; Kumanishi, T.; Ichimura, Y.; Kuwano, R.; Isobe, T.; Wang, Y.; Minoshima, S.; Shimizu, N.; Takahashi, Y.: Structural organization and chromosomal assignment of the human 14–3–3-eta chain gene (YWHAH). *Genomics* 36: 63–69, 1996.
- [33086] 6638. Tommerup, N.; Leffers, H.: Assignment of the human genes encoding 14–3–3 eta (YWHAH) to 22q12, 14–3–3 zeta (YWHAZ) to 2p25.1–p25.2, and 14–3–3 beta (YWHA B) to 20q13.1 by in situ hybridization. *Genomics* 33: 149–150, 1996.

- [33087] 6639.Watanabe, M.; Isobe, T.; Ichimura, T.; Kuwano, R.; Takahashi, Y.;Kondo, H.; Inoue, Y.: Molecular cloning of rat cDNAs for the zetaand theta subtypes of 14-3-3 protein and differential distributionsof their mRNAs in the brain. *Molec. Brain Res.* 25: 113-121, 1994.
- [33088] 6640.Yaffe, M. B.; Rittinger, K.; Volinia, S.; Caron, P. R.; Aitken,A.; Leffers, H.; Gamblin, S. J.; Smerdon, S. J.; Cantley, L. C.:The structural basis for 14-3-3:phosphopeptide binding specificity. *Cell* 91:961-971, 1997.
- [33089] 6641.Zupan, L. A.; Steffens, D. L.; Berry, C. A.; Landt, M.; Gross,R. W.: Cloning and expression of a human 14-3-3 protein mediatingphospholipolysis. *J. Biol. Chem.* 267: 8707-8710, 1992.
- [33090] 6642.Ben-Yosef, T.; Eden, A.; Benvenisty, N.: Characterization of murineBCAT genes: Bcat1, a c-Myc target, and its homolog, Bcat2. *MammalianGenome* 9: 595-597, 1998.
- [33091] 6643.Benvenisty, N.; Leder, A.; Kuo, A.; Leder, P.: An embryonicallyexpressed gene is a target for c-Myc regulation via the c-Myc-bindingsequence. *Genes Dev.* 6: 2513-2523, 1992.
- [33092] 6644.Eden, A.; Simchen, G.; Benvenisty, N.: Two yeast homologs of ECA39,a target for c-Myc regulation, code for

cytosolic and mitochondrial branched-chain amino acid aminotransferases. *J. Biol. Chem.* 271:20242–20245, 1996.

[33093] 6645. Jones, C.; Moore, E. E.: Isolation of mutants lacking branched-chain amino acid transaminase. *Somat. Cell Genet.* 2: 235–243, 1976.

[33094] 6646. Jones, C.; Moore, E. E.: Assignment of the human gene complementing the auxotrophic marker TRANS-minus (BCT1) to chromosome 12. (Abstract) *Cytogenet. Cell Genet.* 25: 168 only, 1979.

[33095] 6647. Jones, C.; Moore, E. E.: Localization of a gene which complements branched-chain amino acid transaminase deficiency to the short arm of human chromosome 12. *Hum. Genet.* 66: 206–211, 1984.

[33096] 6648. Naylor, S. L.; Shows, T. B.: Branched-chain amino-transferase genes (BCT-1 and BCT-2) assigned to human chromosomes 12 and 19 using alpha-ketoacid selection media. (Abstract) *Cytogenet. Cell Genet.* 25: 191–192, 1979.

[33097] 6649. Naylor, S. L.; Shows, T. B.: Branched-chain amino-transferase deficiency in Chinese hamster cells complemented by two independent genes on human chromosomes 12 and 19. *Somat. Cell Genet.* 6: 641–652, 1980.

- [33098] 6650.Schuldiner, O.; Eden, A.; Ben-Yosef, T.; Yanuka, O.; Simchen, G.;Benvenisty, N.: ECA39, a conserved gene regulated by c-Myc in mice,is involved in G1/S cell cycle regulation in yeast. *Proc. Nat. Acad.Sci.* 93: 7143–7148, 1996.
- [33099] 6651.Tanaka, K.; Rosenberg, L. E.: Disorders of branched chain aminoacid and organic acid metabolism.In: Stanbury, J. B.; Wyngaarden,J. B.; Fredrickson, D. S.; Goldstein, J. L.; Brown, M. S.: *The MetabolicBasis of Inherited Disease*. New York: McGraw–Hill (pub.) (5thed.): 1983. Pp. 450–451.
- [33100] 6652.Bassett, D. E., Jr.; Boguski, M. S.; Spencer, F.; Reeves, R.; Kim,S.; Weaver, T.; Hieter, P.: Genome cross-referencing and XREFdb:implications for the identification and analysis of genes mutatedin human disease. *Nature Genet.* 15: 339–344, 1997.
- [33101] 6653.Goebel, H. H.; Anderson, J. R.; Hubner, C.; Oexle, K.; Warlo, I.: Congenital myopathy with excess of thin myofilaments. *NeuromuscularDisord.* 7: 160–168, 1997.
- [33102] 6654.Cattanach, B. M.; Barr, J. A.; Evans, E. P.; Burtenshaw, M.; Beechey,C. V.; Leff, S. E.; Brannan, C. I.; Copeland, N. G.; Jenkins, N. A.;Jones, J.: A candidate mouse model for Prader–Willi syndrome whichshows an absence of *Snrpn*

expression. *Nature Genet.* 2: 270–274,1992.

- [33103] 6655.Colin, Y.; Rahuel, C.; London, J.; Romeo, P. H.; d'Auriol, L.;Galibert, F.; Cartron, J.–P.: Isolation of cDNA clones and completeamino acid sequence of human erythrocyte glycophorin C. *J. Biol.Chem.* 261: 229–233, 1986.
- [33104] 6656.Accolla, R. S.; Gross, N.; Carrel, S.; Corte, G.: Distinct formsof both alpha and beta subunits are present in the human Ia molecularpool. *Proc. Nat. Acad. Sci.* 78: 4549–4551, 1981.
- [33105] 6657.Kingsmore, S. F.; Snoddy, J.; Choubey, D.; Lengyel, P.; Seldin,M. F.: Physical mapping of a family of interferon–activated genes,serum amyloid P–component, and alpha–spectrin on mouse chromosome1. *Immunogenetics* 30: 169–174, 1989.
- [33106] 6658.Thomas, K. R.; Folger, K. R.; Capecchi, M. R. :*Cell* 44: 419–428,1986.
- [33107] 6659.Watanabe, K.; Kessler, C. A.; Bachurski, C. J.; Kanda, Y.; Richardson,B. D.; Stanek, J.; Handwerger, S.; Brar, A. K.: Identification ofa decidua–specific enhancer on the human prolactin gene with two criticalactivator protein 1 (AP–1) binding sites. *Molec. Endocr.* 15: 638–653,2001.
- [33108] 6660.Guillon, H.; de Massy, B.: An initiation site for meiotic crossing–overand gene conversion in the mouse. *Na–*

ture Genet. 32: 296–299, 2002.

- [33109] 6661.Irsch, J.; Nitsch, S.; Hansmann, M.–L.; Rajewsky, K.; Tesch, H.; Diehl, V.; Jox, A.; Kuppers, R.; Radbruch, A.: Isolation of viable Hodgkin and Reed–Sternberg cells from Hodgkin disease tissues. Proc.Nat. Acad. Sci. 95: 10117–10122, 1998.
- [33110] 6662.Hanby, A. M.; Poulsom, R.; Singh, S.; Elia, G.; Jeffery, R. E.; Wright, N. A.: Spasmolytic polypeptide is a major antral peptide: distribution of the trefoil peptides human spasmolytic polypeptide and pS2 in the stomach. Gastroenterology 105: 1110–1116, 1993.
- [33111] 6663.Blankstein, J.; Faiman, C.; Reyes, F. I.; Schroeder, M. L.; Winter, J. S. D.: Adult–onset familial adrenal 21–hydroxylase deficiency. Am.J. Med. 68: 441–448, 1980.
- [33112] 6664.Hoeg, J. M.; Osborne, J. C., Jr.; Gregg, R. E.; Brewer, H. B., Jr.: Initial diagnosis of lipoprotein lipase deficiency in a 75–year–old man. Am. J. Med. 75: 889–892, 1983.
- [33113] 6665.Batischev, A. I.; Chernyak, N. B.; Torakev, Y. N.: Detection of a new abnormal variant of glucose–6–phosphate dehydrogenase in human red cells. Bulletin Eksperimentalnoi Biologii i Meditsiny 84: 728–731, 1977.
- [33114] 6666.Parolini, S.; Bottino, C.; Falco, M.; Augugliaro, R.;

Giliani,S.; Franceschini, R.; Ochs, H. D.; Wolf, H.; Bonnefoy, J.-Y.; Biassoni,R.; Moretta, L.; Notarangelo, L. D.; Moretta, A.: X-linked lymphoproliferative disease: 2B4 molecules displaying inhibitory rather than activating function are responsible for the inability of natural killer cells to kill Epstein-Barr virus-infected cells. *J. Exp. Med.* 192: 337-346, 2000.

[33115] 6667. Hawkins, J. R.; Taylor, A.; Goodfellow, P. N.; Migeon, C. J.; Smith, K. D.; Berkovitz, G. D.: Evidence for increased prevalence of SRY mutations in XY females with complete rather than partial gonadal dysgenesis. *Am. J. Hum. Genet.* 51: 979-984, 1992.

[33116] 6668. Burchell, B.; Nebert, D. W.; Nelson, D. R.; Bock, K. W.; Iyanagi, T.; Jansen, P. L. M.; Lancet, T.; Mulder, G. J.; Chowdhury, J. R.; Siest, G.; Tephly, T. R.; Mackenzie, P. I.: The UDP-glucuronosyltransferase gene superfamily: suggested nomenclature based on evolutionary divergence. *DNACell Biol.* 10: 487-494, 1991.

[33117] 6669. Harrington, J. J.; Lieber, M. R.: The characterization of a mammalian DNA structure-specific endonuclease. *EMBO J.* 13: 1235-1246, 1994.

[33118] 6670. Yi, T.; Cleveland, J. L.; Ihle, J. N.: Identification of novel protein tyrosine phosphatases of hematopoietic cells

by polymerase chain reaction amplification. *Blood* 78: 2222–2228, 1991.

- [33119] 6671. Amano, M.; Mukai, H.; Ono, Y.; Chihara, K.; Matsui, T.; Hamajima, Y.; Okawa, K.; Iwamatsu, A.; Kaibuchi, K.: Identification of a putative target for rho as the serine–threonine kinase protein kinase N. *Science* 271:648–651, 1996.
- [33120] 6672. Gorlatov, S. N.; Stadtman, T. C.: Human thioredoxin reductase from HeLa cells: selective alkylation of seleno–cysteine in the protein inhibits enzyme activity and reduction with NADPH influences affinity to heparin. *Proc. Nat. Acad. Sci.* 95: 8520–8525, 1998.
- [33121] 6673. ten Dijke, P.; Franzen, P.; Yamashita, H.; Ichijo, H.; Heldin, C. H.; Miyazono, K.: Serine/threonine kinase receptors. *Prog. Growth Factor Res.* 5: 55–72, 1994.
- [33122] 6674. MacGrogan, D.; Levy, A.; Bova, G. S.; Isaacs, W. B.; Bookstein, R.: Structure and methylation–associated silencing of a gene within a homozygously deleted region of human chromosome band 8p22. *Genomics* 35:55–65, 1996.
- [33123] 6675. McPherron, A. C.; Lee, S.–J.: Suppression of body fat accumulation in myostatin–deficient mice. *J. Clin. Invest.* 109: 595–601, 2002.

- [33124] 6676.Klein, R. D.; Sherman, D.; Ho, W.-H.; Stone, D.; Bennett, G. L.;Moffat, B.; Vandlen, R.; Simmons, L.; Gu, Q.; Hongo, J.-A.; Devaux,B.; Poulsen, K.; Armanini, M.; Nozaki, C.; Asai, N.; Goddard, A.;Phillips, H.; Henderson, C. E.; Takahashi, M.; Rosenthal, A.: A GPI-linkedprotein that interacts with Ret to form a candidate neurturin receptor. *Nature* 387:717–721, 1997.
- [33125] 6677.Masuda, H.; Tanaka, K.; Takagi, M.; Ohgami, K.; Sakamaki, T.; Shibata,N.; Takahashi, K.: Molecular cloning and characterization of humannon-smooth muscle calponin. *J. Biochem.* 120: 415–424, 1996.
- [33126] 6678.Vlangos, C. N.; Das, P.; Patel, P. I.; Elsea, S. H.: Assignmentof developmentally regulated GTP-binding protein (DRG2) to human chromosomeband 17p11.2 with somatic cell hybrids and localization to the Smith-Magenissyndrome critical interval. *Cytogenet. Cell Genet.* 88: 283–285,2000.
- [33127] 6679.Peyrard, M.: Personal Communication. Stockholm, Sweden 1/7/1999.
- [33128] 6680.Ware, F. E.; Lehrman, M. A.: Expression cloning of a novel suppressorof the Lec15 and Lec35 glycosylation mutations of Chinese hamsterovary cells. *J. Biol. Chem.* 271: 13935–13938, 1996.

- [33129] 6681.Lowe, D. G.; Camerato, T. R.; Goeddel, D. V.: cDNA sequence of the human atrial natriuretic peptide clearance receptor. *Nucleic Acids Res.* 18: 3412 only, 1990.
- [33130] 6682.Porter, J. G.; Arfsten, A.; Fuller, F.; Miller, J. A.; Gregory, L. C.; Lewicki, J. A.: Isolation and functional expression of the human atrial natriuretic peptide clearance receptor cDNA. *Biochem. Biophys. Res. Commun.* 171: 796–803, 1990.
- [33131] 6683.Suda, M.; Ogawa, Y.; Tanaka, K.; Tamura, N.; Yasoda, A.; Takigawa, T.; Uehira, M.; Nishimoto, H.; Itoh, H.; Saito, Y.; Shiota, K.; Nakao, K.: Skeletal overgrowth in transgenic mice that overexpress brain natriuretic peptide. *Proc. Nat. Acad. Sci.* 95: 2337–2342, 1998.
- [33132] 6684.Boehm, J.; Orth, T.; Van Nguyen, P.; Soling, H. D.: Systemic lupus erythematosus is associated with increased auto-antibody titers against calreticulin and grp94, but calreticulin is not the Ro/SS-A antigen. *Europ. J. Clin. Invest.* 24: 248–257, 1994.
- [33133] 6685.Burns, K.; Duggan, B.; Atkinson, E. A.; Famulski, K. S.; Nemer, M.; Bleackley, R. C.; Michalak, M.: Modulation of gene expression by calreticulin binding to the glucocorticoid receptor. *Nature* 367:476–480, 1994.
- [33134] 6686.Telen, M. J.; Whitsett, C. F.: Erythrocyte acetyl-

cholinesterase bears the Cartwright blood group antigens.
(Abstract) Clin. Res. 40:170A only, 1992.

- [33135] 6687. Wjst, M.; Fischer, G.; Immervoll, T.; Jung, M.; Saar, K.; Rueschendorf, F.; Reis, A.; Ulbrecht, M.; Gomolka, M.; Weiss, E. H.; Jaeger, L.; Nickel, R.; and 14 others: A genome-wide search for linkage to asthma. *Genomics* 58: 1–8, 1999.
- [33136] 6688. Kalachikov, S.; Evgrafov, O.; Ross, B.; Winawer, M.; Barker-Cummings, C.; Boneschi, F. M.; Choi, C.; Morozov, P.; Das, K.; Teplitzskaya, E.; Yu, A.; Cayanis, E.; Penchaszadeh, G.; Kottmann, A. H.; Pedley, T. A.; Hauser, W. A.; Ottman, R.; Gilliam, T. C.: Mutations in *LGI1* cause autosomal-dominant partial epilepsy with auditory features. *Nature Genet.* 30: 335–341, 2002.
- [33137] 6689. Privalsky, M. L.; Ralston, R.; Bishop, J. M.: The membrane glycoprotein encoded by the retroviral oncogene *v-erb-B* is structurally related to tyrosine-specific protein kinases. *Proc. Nat. Acad. Sci.* 81:704–707, 1984.
- [33138] 6690. Gordon, M. S.; Kato, R. M.; Lansigan, F.; Thompson, A. A.; Wall, R.; Rawlings, D. J.: Aberrant B cell receptor signaling from B29(Ig-beta, CD79b) gene mutations of chronic lymphocytic leukemia B cells. *Proc. Nat. Acad. Sci.* 97: 5504–5509, 2000.

- [33139] 6691. Kuida, K.; Lippke, J. A.; Ku, G.; Harding, M. W.; Livingston, D. J.; Su, M. S.-S.; Flavell, R. A.: Altered cytokine export and apoptosis in mice deficient in interleukin-1-beta converting enzyme. *Science* 267:2000-2003, 1995.
- [33140] 6692. Morissette, J.; Rheaume, E.; Leblanc, J.-F.; Luu-The, V.; Labrie, F.; Simard, J.: Genetic linkage mapping of HSD3B1 and HSD3B2 encoding human types I and II 3-beta-hydroxysteroid dehydrogenase/delta-5-delta-4-isomerase close to D1S514 and the centromeric D1Z5 locus. *Cytogenet. Cell Genet.* 69: 59-62, 1995.
- [33141] 6693. Morrison, N.; Nickson, D. A.; McBride, M. W.; Mueller, U. W.; Boyd, E.; Sutcliffe, R. G.: Regional chromosomal assignment of human 3-beta-hydroxy-5-enesteroid dehydrogenase to 1p13.1 by non-isotopic in situ hybridisation. *Hum. Genet.* 87: 223-225, 1991.
- [33142] 6694. Race, R. R.; Sanger, R.: *Blood Groups in Man*. Oxford: Blackwell (pub.) (6th ed.): 1975. Pp. 92-138.
- [33143] 6695. Siebert, P. D.; Fukuda, M.: Molecular cloning of a human glycophorin B cDNA: nucleotide sequence and genomic relationship to glycophorin A. *Proc. Nat. Acad. Sci.*

84: 6735–6739, 1987.

- [33144] 6696.Kumar, B. V.; Aleman–Gomez, J. A.; Colwell, N.; Lopez–Candales,A.; Bosner, M. S.; Spilburg, C. A.; Lowe, M.; Lange, L. G.: Structureof the human pancreatic cholesterol esterase gene. *Biochemistry* 31:6077–6081, 1992.
- [33145] 6697.Lidberg, U.; Nilsson, J.; Stromberg, K.; Stenman, G.; Sahlin, P.;Enerback, S.; Bjursell, G.: Genomic organization, sequence analysis,and chromosomal localization of the human carboxyl ester lipase (CEL)gene and a CEL–like (CELL) gene. *Genomics* 13: 630–640, 1992.
- [33146] 6698.Galland, F.; Stefanova, M.; Pirisi, V.; Birnbaum, D.: Characterizationof a murine glyceraldehyde–3–phosphate dehydrogenase pseudogene. *Biochimie* 72:759–762, 1990.
- [33147] 6699.Lidmer, A.–S.; Kannius, M.; Lundberg, L.; Bjursell, G.; Nilsson,J.: Molecular cloning and characterization of the mouse carboxylester lipase gene and evidence for expression in the lactating mammarygland. *Genomics* 29: 115–122, 1995.
- [33148] 6700.Nilsson, J.; Hellquist, M.; Bjursell, G.: The human carboxyl esterlipase–like (CELL) gene is ubiquitously expressed and contains a hypervariableregion. *Genomics* 17: 416–422, 1993.

- [33149] 6701.Taylor, A. K.; Zambaux, J. L.; Klisak, I.; Mohandas, T.; Sparkes, R. S.; Schotz, M. C.; Lusi, A. J.: Carboxyl-ester lipase: a highly polymorphic locus on chromosome 9qter. *Genomics* 10: 425-431, 1991.
- [33150] 6702.Vanin, E. F.: Processed pseudogenes: characteristics and evolution. *Annu. Rev. Genet.* 19: 253-272, 1985.
- [33151] 6703.Gardell, S. J.; Craik, C. S.; Clauser, E.; Goldsmith, E. J.; Stewart, C. B.; Graf, M.; Rutter, W. J.: A novel rat carboxypeptidase, CPA2: characterization, molecular cloning, and evolutionary implications on substrate specificity in the carboxypeptidase gene family. *J. Biol. Chem.* 263: 17828-17836, 1988.
- [33152] 6704.Lowe, N.; Edwards, Y. H.; Edwards, M.; Butterworth, P. H. W.: Physical mapping of the human carbonic anhydrase gene cluster on chromosome 8. *Genomics* 10: 882-888, 1991.
- [33153] 6705.Marriq, C.; Gulian, J. M.; Laurent, G.: Cleavage by cyanogen bromide of carbonic anhydrase from human erythrocyte B. *Biochim. Biophys. Acta* 221: 662-664, 1970.
- [33154] 6706.Moore, M. J.; Deutsch, H. F.; Ellis, F. R.: Human carbonic anhydrase. IX. Inheritance of variant erythrocyte forms. *Am. J. Hum. Genet.* 25:29-35, 1973.
- [33155] 6707.Omoto, K.: Carbonic anhydrase-I polymorphism in a

Philippineaboriginal population. Am. J. Hum. Genet. 31: 747–750, 1979.

[33156] 6708.Omoto, K.; Ueda, S.; Goriki, K.; Takahashi, N.; Misawa, S.; Pagaran,I. G.: Population genetic studies of the Philippine Negritos. III.Identification of the carbonic anhydrase–1 variant with CA(1) Guam. Am.J. Hum. Genet. 33: 105–111, 1981.

[33157] 6709.Tashian, R. E.: The esterases and carbonic anhydrases of humanerythrocytes.In: Yunis, J. J.: Biochemical Methods in Red Cell Genetics. New York: Academic Press (pub.) 1969. Pp. 307–336.

[33158] 6710.Tashian, R. E.; Carter, N. D.: Biochemical genetics of carbonicanhydrase. Adv. Hum. Genet. 7: 1–56, 1976.

[33159] 6711.Tashian, R. E.; Goodman, M.; Headings, V. E.; Desimone, J.; Ward,R. H.: Genetic variation and evolution in the red cell carbonic anhydraseisozymes of Macaque monkeys. Biochem. Genet. 5: 183–200, 1971.

[33160] 6712.Tashian, R. E.; Plato, C. C.; Shows, T. B.: Inherited variantof erythrocyte carbonic anhydrase in Micronesians from Guam and Saipan. Science 140:53–54, 1963.

[33161] 6713.Wagner, L. E.; Venta, P. J.; Tashian, R. E.: A human carbonicanhydrase I deficiency appears to be caused by a destabilizing aminoacid substitution (246arg–to–his).

Isozyme Bull. 24: 35 only, 1991.

- [33162] 6714. Avramopoulos, D.; Cox, T.; Forrest, G. L.; Chakravarti, A.; Antonarakis, S. E.: Linkage mapping of the carbonyl reductase (CBR) gene on human chromosome 21 using a DNA polymorphism in the 3-prime untranslated region. *Genomics* 13: 447–448, 1992.
- [33163] 6715. Forrest, G. L.; Akman, S.; Krutzik, S.; Paxton, R. J.; Sparkes, R. S.; Doroshov, J.; Felsted, R. L.; Glover, C. J.; Mohandas, T.; Bachur, N. R.: Induction of a human carbonyl reductase gene located on chromosome 21. *Biochim. Biophys. Acta* 1048: 149–155, 1990.
- [33164] 6716. Lemieux, N.; Malfoy, B.; Forrest, G. L.: Human carbonyl reductase (CBR) localized to band 21q22.1 by high-resolution fluorescence in situ hybridization displays gene dosage effects in trisomy 21 cells. *Genomics* 15: 169–172, 1993.
- [33165] 6717. Watanabe, K.; Sugawara, C.; Ono, A.; Fukuzumi, Y.; Itakura, S.; Yamazaki, M.; Tashiro, H.; Osoegawa, K.; Soeda, E.; Nomura, T.: Mapping of a novel human carbonyl reductase, CBR3, and ribosomal pseudogene to human chromosome 21q22.2. *Genomics* 52: 95–100, 1998.
- [33166] 6718. Wei, J.; Dlouhy, S. R.; Hara, A.; Ghetti, B.; Hodes, M. E.: Cloning a cDNA for carbonyl reductase (Cbr) from

mouse cerebellum: murine genes that express Cbr map to chromosomes 16 and 11. *Genomics* 34:147–148, 1996.

[33167] 6719. Wermuth, B.; Bohren, K. M.; Heinemann, G.; von Wartburg, J.-P.; Gabbay, K. H.: Human carbonyl reductase: nucleotide sequence analysis of a cDNA and amino acid sequence of the encoded protein. *J. Biol. Chem.* 263: 16185–16188, 1988.

[33168] 6720. Yang-Feng, T. L.; Floyd-Smith, G.; Nemer, M.; Drouin, J.; Francke, U.: The pronatriodilatin gene is located on the distal short arm of human chromosome 1 and on mouse chromosome 4. *Am. J. Hum. Genet.* 37:1117–1128, 1985.

[33169] 6721. Ferrari, P.; Weidmann, P.; Ferrier, C.; Dietler, R.; Hollmann, R.; Pisoni, R. J.; Wey, J.; Shaw, S.: Dysregulation of atrial natriuretic factor in hypertension-prone man. *J. Clin. Endocr. Metab.* 71: 944–951, 1990.

[33170] 6722. Kishimoto, I.; Rossi, K.; Garbers, D. L.: A genetic model provides evidence that the receptor for atrial natriuretic peptide (guanylyl cyclase-A) inhibits cardiac ventricular myocyte hypertrophy. *Proc. Nat. Acad. Sci.* 98: 2703–2706, 2001.

[33171] 6723. Lowe, D. G.; Klisak, I.; Sparkes, R. S.; Mohandas, T.; Goeddel, D. V.: Chromosomal distribution of three mem-

bers of the human natriuretic peptide receptor/guanylyl cyclase gene family. *Genomics* 8: 304–312, 1990.

- [33172] 6724. AbdAlla, S.; Lothar, H.; Quitterer, U.: AT(1)–receptor heterodimers show enhanced G–protein activation and altered receptor sequestration. *Nature* 407:94–98, 2000.
- [33173] 6725. Bergsma, D. J.; Ellis, C.; Kumar, C.; Nuthulaganti, P.; Kersten, H.; Elshourbagy, N.; Griffin, E.; Stadel, J. M.; Aiyar, N.: Cloning and characterization of a human angiotensin II type 1 receptor. *Biochem. Biophys. Res. Commun.* 183: 989–995, 1992.
- [33174] 6726. Bonnardeaux, A.; Davies, E.; Jeunemaitre, X.; Fery, I.; Charru, A.; Clauser, E.; Tiret, L.; Cambien, F.; Corvol, P.; Soubrier, F.: Angiotensin II type 1 receptor gene polymorphisms in human essential hypertension. *Hypertension* 24: 63–69, 1994.
- [33175] 6727. Curnow, K. M.; Pascoe, L.; White, P. C.: Genetic analysis of the human type–1 angiotensin II receptor. *Molec. Endocr.* 6: 1113–1118, 1992.
- [33176] 6728. Elton, T. S.; Stephan, C. C.; Taylor, G. R.; Kimball, M. G.; Martin, M. M.; Durand, J. N.; Oparil, S.: Isolation of two distinct type I angiotensin II receptor genes. *Biochem. Biophys. Res. Commun.* 184:1067–1073, 1992.
- [33177] 6729. Furuta, H.; Guo, D.–F.; Inagami, T.: Molecular

cloning and sequencing of the gene encoding human angiotensin II type 1 receptor. *Biochem. Biophys. Res. Commun.* 183: 8–13, 1992.

[33178] 6730. Gemmill, R. M.; Drabkin, H. A.: Report of The Second International Workshop on Human Chromosome 3 Mapping. *Cytogenet. Cell Genet.* 57:162–166, 1991.

[33179] 6731. Guo, D.-F.; Furuta, H.; Mizukoshi, M.; Inagami, T.: The genomic organization of human angiotensin II type 1 receptor. *Biochem. Biophys. Res. Commun.* 200: 313–319, 1994.

[33180] 6732. Harada, K.; Komuro, I.; Hayashi, D.; Sugaya, T.; Murakami, K.; Yazaki, Y.: Angiotensin II type 1a receptor is involved in the occurrence of reperfusion arrhythmias. *Circulation* 97: 315–317, 1998.

[33181] 6733. Haywood, G. A.; Gullestad, L.; Katsuya, T.; Hutchinson, H. G.; Pratt, R. E.; Horiuchi, M.; Fowler, M. B.: AT(1) and AT(2) angiotensin receptor gene expression in human heart failure. *Circulation* 95:1201–1206, 1997.

[33182] 6734. Herzig, T. C.; Jobe, S. M.; Aoki, H.; Molkentin, J. D.; Cowley, A. W., Jr.; Izumo, S.; Markham, B. E.: Angiotensin II type-1a receptor gene expression in the heart: AP-1 and GATA-4 participate in the response to pressure overload. *Proc. Nat. Acad. Sci.* 94: 7543–7548, 1997.

- [33183] 6735.Ito, M.; Oliverio, M. I.; Mannon, P. J.; Best, C. F.; Maeda, N.;Smithies, O.; Coffman, T. M.: Regulation of blood pressure by type1A angiotensin II receptor gene. Proc. Nat. Acad. Sci. 92: 3521–3525,1995.
- [33184] 6736.Iyer, S. N.; Lu, D.; Katovich, M. J.; Raizada, M. K.: Chroniccontrol of high blood pressure in the sponta-
neously hypertensive ratby delivery of angiotensin type 1
receptor antisense. Proc. Nat.Acad. Sci. 93: 9960–9965,
1996.
- [33185] 6737.Konishi, H.; Kuroda, S.; Inada, Y.; Fujisawa, Y.: Novel
subtypeof human angiotensin II type 1 receptor: cDNA
cloning and expression. Biochem.Biophys. Res. Commun.
199: 467–474, 1994.
- [33186] 6738.Martens, J. R.; Reaves, P. Y.; Lu, D.; Katovich, M. J.;
Berecek,K. H.; Bishop, S. P.; Raizada, M. K.; Gelband, C. H.:
Preventionof renovascular and cardiac pathophysiological
changes in hypertensionby angiotensin II type 1 receptor
antisense gene therapy. Proc. Nat.Acad. Sci. 95:
2664–2669, 1998.
- [33187] 6739.Martin, M. M.; Willardson, B. M.; Burton, G. F.; White,
C. R.;McLaughlin, J. N.; Bray, S. M.; Ogilvie, J. W., Jr.; Elton,
T. S.: Human angiotensin II type 1 receptor isoforms en-
coded by messengerRNA splice variants are functionally

distinct. *Molec. Endocr.* 15:281–293, 2001.

- [33188] 6740. Mauzy, C. A.; Hwang, O.; Egloff, A. M.; Wu, L.-H.; Chung, F.-Z.: Cloning, expression, and characterization of a gene encoding the human angiotensin II type 1A receptor. *Biochem. Biophys. Res. Commun.* 186:277–284, 1992.
- [33189] 6741. Murphy, T. J.; Alexander, R. W.; Griendling, K. K.; Runge, M. S.; Bernstein, K. E.: Isolation of a cDNA encoding the vascular type-1 angiotensin II receptor. *Nature* 351: 233–236, 1991.
- [33190] 6742. Paradis, P.; Dali-Youcef, N.; Paradis, F. W.; Thibault, G.; Nemer, M.: Overexpression of angiotensin II type I receptor in cardiomyocytes induces cardiac hypertrophy and remodeling. *Proc. Nat. Acad. Sci.* 97:931–936, 2000.
- [33191] 6743. Sasaki, K.; Murohara, T.; Ikeda, H.; Sugaya, T.; Shimada, T.; Shintani, S.; Imaizumi, T.: Evidence for the importance of angiotensin II type 1 receptor in ischemia-induced angiogenesis. *J. Clin. Invest.* 109:603–611, 2002.
- [33192] 6744. Oliverio, M. I.; Kim, H.-S.; Ito, M.; Le, T.; Audoly, L.; Best, C. F.; Hiller, S.; Kluckman, K.; Maeda, N.; Smithies, O.; Coffman, T. M.: Reduced growth, abnormal kidney structure, and type 2 (AT₂) angiotensin receptor-mediated blood pressure regulation in mice lacking both AT_{1A} and

AT1B receptors for angiotensin II. Proc. Nat. Acad.Sci. 95: 15496–15501, 1998.

[33193] 6745.Sasaki, K.; Yamano, Y.; Bardhan, S.; Iwai, N.; Murray, J. J.;Hasegawa, M.; Matsuda, Y.; Inagami, T.: Cloning and expression of a complementary DNA encoding a bovine adrenal angiotensin II type-1receptor. Nature 351: 230–233, 1991.

[33194] 6746.Scott, A. F.: Personal Communication. Baltimore, Md. 3/20/2001.

[33195] 6747.Szpirer, C.; Riviere, M.; Szpirer, J.; Levan, G.; Guo, D. F.;Iwai, N.; Inagami, T.: Chromosomal assignment of human and rat hypertensioncandidate genes: type 1 angiotensin II receptor genes and the SA gene. J.Hypertension 11: 919–925, 1993.

[33196] 6748.Takayanagi, R.; Ohnaka, K.; Sakai, Y.; Nakao, R.; Yanase, T.;Haji, M.; Inagami, T.; Furuta, H.; Gou, D.-F.; Nakamura, M.; Nawata,H.: Molecular cloning, sequence analysis and expression of a cDNAencoding human type-1 angiotensin II receptor. Biochem. Biophys.Res. Commun. 183: 910–916, 1992.

[33197] 6749.Tsuchida, S.; Matsusaka, T.; Chen, X.; Okubo, S.; Nishimura, F.;Nishimura, H.; Fogo, A.; Utsunomiya, H.; Inagami, T.; Ichikawa, I.: Murine double nullizygotes of the

angiotensin type 1A and 1B receptor genes duplicate severe abnormal phenotypes of angiotensinogen nullizygotes. *J. Clin. Invest.* 101: 755–760, 1998.

[33198] 6750. Erba, H. P.; Eddy, R.; Shows, T.; Kedes, L.; Gunning, P.: Structure, chromosome location, and expression of the human gamma-actin gene: differential evolution, location, and expression of the cytoskeletal beta- and gamma-actin genes. *Molec. Cell. Biol.* 8: 1775–1789, 1988.

[33199] 6751. Ueyama, H.; Inazawa, J.; Nishino, H.; Ohkubo, I.; Miwa, T.: FISH localization of human cytoplasmic actin genes ACTB to 7p22 and ACTG1 to 17q25 and characterization of related pseudogenes. *Cytogenet. Cell Genet.* 74: 221–224, 1996.

[33200] 6752. Beggs, A. H.; Byers, T. J.; Knoll, J. H. M.; Boyce, F. M.; Bruns, G. A. P.; Kunkel, L. M.: Cloning and characterization of two human skeletal muscle alpha-actinin genes located on chromosomes 1 and 11. *J. Biol. Chem.* 267: 9281–9288, 1992.

[33201] 6753. Beggs, A. H.; Phillips, H. A.; Kozman, H.; Mulley, J. C.; Wilton, S. D.; Kunkel, L. M.; Laing, N. G.: A (CA)_n repeat polymorphism for the human skeletal muscle alpha-actinin gene ACTN2 and its localization on the linkage map of chromosome 1. *Genomics* 13: 1314–1315, 1992.

- [33202] 6754.Mills, M. A.; Yang, N.; Weinberger, R. P.; Vander Woude, D. L.;Beggs, A. H.; Easteal, S.; North, K. N.: Differential expressionof the actin-binding proteins, alpha-actinin-2 and -3, in differentspecies: implications for the evolution of functional redundancy. *Hum.Molec. Genet.* 10: 1335-1346, 2001.
- [33203] 6755.Attisano, L.; Carcamo, J.; Ventura, F.; Weis, F. M. B.; Massague,J.; Wrana, J. L.: Identification of human activin and TGF-beta typeI receptors that form heteromeric kinase complexes with type II receptors. *Cell* 75:671-680, 1993.
- [33204] 6756.Mathews, L. S.; Vale, W. W.: Expression cloning of an activinreceptor, a predicted transmembrane serine kinase. *Cell* 65: 973-982,1991.
- [33205] 6757.Matsuzaki, K.; Xu, J.; Wang, F.; McKeehan, W. L.; Krummen, L.;Kan, M.: A widely expressed transmembrane serine/threonine kinasethat does not bind activin, inhibin, transforming growth factor beta,or bone morphogenic factor. *J. Biol. Chem.* 268: 12719-12723, 1993.
- [33206] 6758.Roijer, E.; Miyazono, K.; Astrom, A.-K.; Geurts van Kessel, A.;ten Dijke, P.; Stenman, G.: Chromosomal localization of three humangen es encoding members of the TGF-beta superfamily of type I serine/threoninekinase re-

ceptors. *Mammalian Genome* 9: 266–268, 1998.

[33207] 6759.ten Dijke, P.; Ichijo, H.; Franzen, P.; Schulz, P.; Saras, J.; Toyoshima, H.; Heldin, C.–H.; Miyazono, K.: Activin receptor–like kinases: a novel subclass of cell–surface receptors with predicted serine/threonine kinase activity. *Oncogene* 8: 2879–2887, 1993.

[33208] 6760.Chen, M.; Pan, Z.–Q.; Hurwitz, J.: Studies of the cloned 37–kDa subunit of activator 1 (replication factor C) of HeLa cells. *Proc.Nat. Acad. Sci.* 89: 5211–5215, 1992.

[33209] 6761.Ferrari, B.; Pavia, A. A.: Blood group antigens: synthesis of Ss antigenic peptides related to human glycophorin B. *Int. J. Peptide Protein Res.* 28: 456–461, 1986.

[33210] 6762.Huang, C.–H.; Blumenfeld, O. O.: Characterization of a genomic hybrid specifying the human erythrocyte antigen Dantu: Dantu gene is duplicated and linked to a delta glycophorin gene deletion. *Proc.Nat. Acad. Sci.* 85: 9640–9644, 1988.

[33211] 6763.Huang, C.–H.; Johe, K.; Moulds, J. J.; Siebert, P. D.; Fukuda, M.; Blumenfeld, O. O.: Delta–glycophorin (glycophorin B) gene deletion in two individuals homozygous for the S–s–U– blood group phenotype. *Blood* 70:1830–1835, 1987.

[33212] 6764.Kudo, S.; Fukuda, M.: Structural organization of gly–

cophorin A and B genes: glycophorin B gene evolved by homologous recombination at Alu repeat sequences. *Proc. Nat. Acad. Sci.* 86: 4619–4623, 1989.

[33213] 6765. Marchesi, V. T.; Tillack, T. M.; Jackson, R. L.; Segrest, J. P.; Scott, R. E.: Chemical characterization and surface orientation of the major glycoprotein of the human erythrocyte membrane. *Proc. Nat. Acad. Sci.* 69: 1445–1449, 1972.

[33214] 6766. Buettner, R.; Schaffler, A.; Arndt, H.; Rogler, G.; Nusser, J.; Zietz, B.; Enger, I.; Hugl, S.; Cuk, A.; Scholmerich, J.; Palitzsch, K.-D.: The trp64arg polymorphism of the beta-3-adrenergic receptor gene is not associated with obesity or type 2 diabetes mellitus in a large population-based Caucasian cohort. *J. Clin. Endocr. Metab.* 83:2892–2897, 1998.

[33215] 6767. Clement, K.; Vaisse, C.; Manning, B. S. J.; Basdevant, A.; Guy-Grand, B.; Ruiz, J.; Silver, K. D.; Shuldiner, A. R.; Froguel, P.; Strosberg, A. D.: Genetic variation in the beta-3-adrenergic receptor and an increased capacity to gain weight in patients with morbid obesity. *New Eng. J. Med.* 333: 352–354, 1995.

[33216] 6768. Elbein, S. C.; Hoffman, M.; Barrett, K.; Wegner, K.; Miles, C.; Bachman, K.; Berkowitz, D.; Shuldiner, A. R.;

Leppert, M. F.; Hasstedt, S.: Role of the beta-3-adrenergic receptor locus in obesity and noninsulin-dependent diabetes among members of Caucasian families with a diabetic sibling pair. *J. Clin. Endocr. Metab.* 81: 4422-4427, 1996.

[33217] 6769. Emorine, L. J.; Marullo, S.; Briand-Sutren, M.-M.; Patey, G.; Tate, K.; Delavie-Klutchko, C.; Strosberg, A. D.: Molecular characterization of the human beta-3-adrenergic receptor. *Science* 245: 1118-1121, 1989.

[33218] 6770. Festa, A.; Krugluger, W.; Shnawa, N.; Hopmeier, P.; Haffner, S. M.; Schernthaner, G.: Trp64Arg polymorphism of the beta-3-adrenergic receptor gene in pregnancy: association with mild gestational diabetes mellitus. *J. Clin. Endocr. Metab.* 84: 1695-1699, 1999.

[33219] 6771. Gagnon, J.; Mauriege, P.; Roy, S.; Sjostrom, D.; Chagnon, Y. C.; Dionne, F. T.; Oppert, J.-M.; Perusse, L.; Sjostrom, L.; Bouchard, C.: The trp64arg mutation of the beta-3 adrenergic receptor gene has no effect on obesity phenotypes in the Quebec Family Study and Swedish Obese Subjects cohorts. *J. Clin. Invest.* 98: 2086-2093, 1996.

[33220] 6772. Garcia-Rubi, E.; Starling, R. D.; Tchernof, A.; Matthews, D. E.; Walston, J. D.; Shuldiner, A. R.; Silver, K.; Poehlman, E. T.; Calles-Escandon, J.: Trp64Arg variant of

the beta-3-adrenoceptor and insulin resistance in obese postmenopausal women. *J. Clin. Endocr. Metab.* 83: 4002-4005, 1998.

[33221] 6773. Hoffstedt, J.; Poirier, O.; Thorne, A.; Lonnqvist, F.; Herrmann, S. M.; Cambien, F.; Arner, P.: Polymorphism of the human beta-3-adrenoceptor gene forms a well-conserved haplotype that is associated with moderate obesity and altered receptor function. *Diabetes* 48: 203-205, 1999.

[33222] 6774. Kim-Motoyama, H.; Yasuda, K.; Yamaguchi, T.; Yamada, N.; Katakura, T.; Shuldiner, A. R.; Akanuma, Y.; Ohashi, Y.; Yazaki, Y.; Kadowaki, T.: A mutation of the beta-3-adrenergic receptor is associated with visceral obesity but decreased serum triglyceride. *Diabetologia* 40:469-472, 1997.

[33223] 6775. Mitchell, B. D.; Blangero, J.; Comuzzie, A. G.; Almasy, L. A.; Shuldiner, A. R.; Silver, K.; Stern, M. P.; McCluer, J. W.; Hixson, J. E.: A paired sibling analysis of the beta-3 adrenergic receptor and obesity in Mexican Americans. *J. Clin. Invest.* 101: 584-587, 1998.

[33224] 6776. Nagase, T.; Aoki, A.; Yamamoto, M.; Yasuda, H.; Kado, S.; Nishikawa, M.; Kugai, N.; Akatsu, T.; Nagata, N.: Lack of association between the trp64arg mutation in the

beta-3-adrenergic receptor gene and obesity in Japanese men: a longitudinal analysis. *J. Clin. Endocr. Metab.* 82:1284-1287, 1997.

- [33225] 6777. Shihara, N.; Yasuda, K.; Moritani, T.; Ue, H.; Adachi, T.; Tanaka, H.; Tsuda, K.; Seino, Y.: The association between Trp64Arg polymorphism of the beta-3-adrenergic receptor and autonomic nervous system activity. *J. Clin. Endocr. Metab.* 84: 1623-1627, 1999.
- [33226] 6778. Van Spronsen, A.; Nahmias, C.; Krief, S.; Briand-Sutren, M.-M.; Strosberg, A. D.; Emorine, L. J.: The promoter and intron/exon structure of the human and mouse beta-3-adrenergic-receptor genes. *Europ. J. Biochem.* 213: 1117-1124, 1993.
- [33227] 6779. Urhammer, S. A.; Hansen, T.; Borch-Johnsen, K.; Pedersen, O.: Studies of the synergistic effect of the trp/arg64 polymorphism of the beta-3-adrenergic receptor gene and the -3826 A-G variant of the uncoupling protein-1 gene on features of obesity and insulin resistance in a population-based sample of 379 young Danish subjects. *J. Clin. Endocr. Metab.* 85: 3151-3154, 2000.
- [33228] 6780. Walston, J.; Silver, K.; Bogardus, C.; Knowler, W. C.; Celi, F. S.; Austin, S.; Manning, B.; Strosberg, A. D.; Stern, M. P.; Raben, N.; Sorkin, J. D.; Roth, J.; Shuldiner, A. R.:

Time of onset of non-insulin-dependent diabetes mellitus and genetic variation in the beta-3-adrenergic-receptor gene. *New Eng. J. Med.* 333: 343-347, 1995.

[33229] 6781. Walston, J.; Silver, K.; Hilfiker, H.; Andersen, R. E.; Seibert, M.; Beamer, B.; Roth, J.; Poehlman, E.; Shuldiner, A. R.: Insulin response to glucose is lower in individuals homozygous for the arg64 variant of the beta-3-adrenergic receptor. *J. Clin. Endocr. Metab.* 85:4019-4022, 2000.

[33230] 6782. Hershfield, M. S.; Mitchell, B. S.: Immunodeficiency diseases caused by adenosine deaminase deficiency and purine nucleoside phosphorylase deficiency. In: Scriver, C. R.; Beaudet, A. L.; Sly, W. S.; Valle, D. (eds.): *The Metabolic and Molecular Bases of Inherited Disease*. Vol. 2. New York: McGraw-Hill (7th ed.): 1995. Pp. 1725-1768.

[33231] 6783. Hirschhorn, R.: Identification of two new missense mutations (R156C and S291L) in two ADA(-) SCID patients unusual for response to therapy with partial exchange transfusions. *Hum. Mutat.* 1: 166-168, 1992.

[33232] 6784. Hirschhorn, R.: Personal Communication. New York, N.Y. 1976.

[33233] 6785. Hirschhorn, R.; Borkowsky, W.; Jiang, C.-K.; Yang, D. R.; Jenkins, T.: Two newly identified mutations (Thr233Ile

and Leu152Met) in partiallyadenosine deaminase-deficient (ADA-) individuals that result in differingbiochemical and metabolic phenotypes. Hum. Genet. 100: 22-29, 1997.

[33234] 6786.Hirschhorn, R.; Chakravarti, V.; Puck, J.; Douglas, S. D.: Homozygosityfor a newly identified missense mutation in a patient with very severecombined immunodeficiency due to adenosine deaminase deficiency (ADA-SCID). Am.J. Hum. Genet. 49: 878-885, 1991.

[33235] 6787.Hirschhorn, R.; Ellenbogen, A.: Genetic heterogeneity in adenosinedeaminase (ADA) deficiency: five different mutations in five new patientswith partial ADA deficiency. Am. J. Hum. Genet. 38: 13-25, 1986.

[33236] 6788.Hirschhorn, R.; Ellenbogen, A.; Tzall, S.: Five missense mutationsat the adenosine deaminase locus (ADA) detected by altered restrictionfragments and their frequency in ADA-patients with severe combinedimmunodeficiency (ADA-SCID). Am. J. Med. Genet. 42: 201-207, 1992.

[33237] 6789.Hirschhorn, R.; Levytska, V.; Parkman, R.: A mutant form of adenosinedeaminase in severe combined immunodeficiency. (Abstract) J. Clin.Invest. 53: 33A, 1974.

[33238] 6790.Hirschhorn, R.; Martiniuk, F.; Roegner-Maniscalco, V.; Ellenbogen,A.; Perignon, J.-L.; Jenkins, T.: Genetic het-

erogeneity in partial adenosine deaminase deficiency. J. Clin. Invest. 71: 1887–1892, 1983.

- [33239] 6791. Hirschhorn, R.; Papageorgiou, P. S.; Kesarwala, H. H.; Taft, L. T.: Amelioration of neurologic abnormalities after 'enzyme replacement' in adenosine deaminase deficiency. New Eng. J. Med. 303: 377–380, 1980.
- [33240] 6792. Hirschhorn, R.; Roegner, V.; Jenkins, T.; Seaman, C.; Piomelli, S.; Borkowsky, W.: Erythrocyte adenosine deaminase deficiency without immunodeficiency: evidence for an unstable mutant enzyme. J. Clin. Invest. 64: 1130–1139, 1979.
- [33241] 6793. Hirschhorn, R.; Tzall, S.; Ellenbogen, A.: Hot spot mutations in adenosine deaminase deficiency. Proc. Nat. Acad. Sci. 87: 6171–6175, 1990.
- [33242] 6794. Hirschhorn, R.; Tzall, S.; Ellenbogen, A.; Orkin, S. H.: Identification of a point mutation resulting in a heat-labile adenosine deaminase (ADA) in two unrelated children with partial ADA deficiency. J. Clin. Invest. 83: 497–501, 1989.
- [33243] 6795. Hirschhorn, R.; Vawter, G. F.; Kirkpatrick, J. A., Jr.; Rosen, F. S.: Adenosine deaminase deficiency: frequency and comparative pathology in autosomally recessive severe combined immunodeficiency. Clin. Immun. Immunopath. 14: 107–120, 1979.

- [33244] 6796.Hirschhorn, R.; Yang, D. R.; Israni, A.: An asp8-to-asn substitution results in the adenosine deaminase (ADA) genetic polymorphism (ADA2 allozyme): occurrence on different chromosomal backgrounds and apparent intragenic crossover. *Ann. Hum. Genet.* 58: 1-9, 1994.
- [33245] 6797.Hirschhorn, R.; Yang, D. R.; Israni, A.; Huie, M. L.; Ownby, D.R.: Somatic mosaicism for a newly identified splice-site mutation in a patient with adenosine deaminase-deficient immunodeficiency and spontaneous clinical recovery. *Am. J. Hum. Genet.* 55: 59-68, 1994.
- [33246] 6798.Hirschhorn, R.; Yang, D. R.; Puck, J. M.; Huie, M. L.; Jiang, C.-K.; Kurlandsky, L. E.: Spontaneous in vivo reversion to normal of an inherited mutation in a patient with adenosine deaminase deficiency. *Nature Genet.* 13: 290-295, 1996.
- [33247] 6799.Hong, R.; Galti, R.; Rathbun, J. C.; Good, R. A.: Thymic hypoplasia and thyroid dysfunction. *New Eng. J. Med.* 282: 470-474, 1970.
- [33248] 6800.Honig, J.; Martiniuk, F.; D'Eustachio, P.; Zamfirescu, C.; Desnick, R.; Hirschhorn, K.; Hirschhorn, L. R.; Hirschhorn, R.: Confirmation of the regional localization of the genes for human acid alpha-glucosidase (GAA) and

adenosine deaminase (ADA) by somatic cell hybridization. Ann.Hum. Genet. 48: 49–56, 1984.

- [33249] 6801.Hopkinson, D. A.; Cook, P. J. L.; Harris, H.: Further data onthe adenosine deaminase (ADA) polymorphism and a report of a new phenotype. Ann.Hum. Genet. 32: 361–368, 1969.
- [33250] 6802.Hutton, J. J.; Wiginton, D. A.; Coleman, M. S.; Fuller, S. A.;Limouze, S.; Lampkin, B. C.: Biochemical and functional abnormalitiesin lymphocytes from an adenosine deaminase–deficient patient duringenzyme replacement therapy. J. Clin. Invest. 68: 413–421, 1981.
- [33251] 6803.Jenkins, T.; Rabson, A. R.; Nurse, G. T.; Lane, A. B.; Hopkinson,D. A.: Deficiency of adenosine deaminase not associated with severecombined immunodeficiency. J. Pediat. 89: 732–736, 1976.
- [33252] 6804.Jhanwar, S. C.; Berkvens, T. M.; Breukel, C.; van Ormondt, H.;van der Eb, A. J.; Meera Khan, P.: Localization of human adenosinedeaminase (ADA) gene sequences to the q12–q13.11 region of chromosome20 by in situ hybridization. Cytogenet. Cell Genet. 50: 168–171,1989.
- [33253] 6805.Jiang, C.–K.; Hong, R.; Horowitz, S. D.; Kong, X.–P.; Hirschhorn,R.: An adenosine deaminase (ADA) allele contains two newly identifieddeleterious mutations (Y97C and

L106V) that interact to abolish enzyme activity. *Hum. Molec. Genet.* 6: 2271–2278, 1997.

[33254] 6806. Kaitila, I.; Rimoin, D. L.; Cederbaum, S. D.; Stiehm, E. R.; Lechman, R. S.: Chondroosseous histopathology in adenosine deaminase deficient combined immunodeficiency disease. *Birth Defects Orig. Art. Ser.*

XII(6):115–121, 1976.

[33255] 6807. Kellems, R. E.; Yeung, C.-Y.; Ingolia, D. E.: Adenosine deaminase deficiency and severe combined immunodeficiencies. *Trends Genet.* 1:278–283, 1985.

[33256] 6808. Kenny, A. B.; Hitzig, W. H.: Bone marrow transplantation for severe combined immunodeficiency disease: reported from 1968–1977. *Europ. J. Pediatr.* 131: 155–176, 1979.

[33257] 6809. Koch, G.; Shows, T. B.: Somatic cell genetics of adenosine deaminase expression and severe combined immunodeficiency disease in humans. *Proc. Nat. Acad. Sci.* 77: 4211–4215, 1980.

[33258] 6810. Kredich, N. M.; Martin, D. W., Jr.: Role of 5-adenosylhomocysteine in adenosine-mediated toxicity in cultured mouse T-lymphoma cells. *Cell* 12:931–938, 1977.

[33259] 6811. Levy, Y.; Hershfield, M. S.; Fernandez-Mejia, C.; Pol-

mar, S. H.; Scudiero, D.; Berger, M.; Sorensen, R. U.: Adenosine deaminase deficiency with late onset of recurrent infections: response to treatment with polyethylene glycol-modified adenosine deaminase. *J. Pediat.* 113:312–317, 1988.

[33260] 6812. Chashchin, V. L.; Lapko, V. N.; Adamovich, T. B.; Kirillova, N. M.; Lapko, A. G.; Akhrem, A. A.: The primary structure of hepatoferredoxin from bovine liver mitochondria. *Bioorg. Khim.* 12: 1286–1289, 1986.

[33261] 6813. Jefcoate, C. R.; McNamara, B. C.; DiBartolomeis, M. J.: Control of steroid synthesis in adrenal fasciculata cells. *Endocr. Res.* 12:314–350, 1986.

[33262] 6814. Maruya, N.; Hiwatashi, A.; Ichikawa, Y.; Yamano, T.: Purification and characterization of renal ferredoxin from bovine renal mitochondria. *J. Biochem.* 93: 1239–1247, 1983.

[33263] 6815. Mittal, S.; Zhu, Y. Z.; Vickery, L. E.: Molecular cloning and sequence analysis of human placental ferredoxin. *Arch. Biochem. Biophys.* 264:383–391, 1988.

[33264] 6816. Morel, Y.; Picado-Leonard, J.; Mohandas, T. K.; Miller, W. L.: Two highly homologous genes for adrenodoxin lie on human chromosomes 11 and 20. (Abstract) *Am. J. Hum. Genet.* 41: A178 only, 1987.

- [33265] 6817. Morel, Y.; Picado-Leonard, J.; Wu, D.-A.; Chang, C.-Y.; Mohandas, T. K.; Chung, B.-C.; Miller, W. L.: Assignment of the functional gene for human adrenodoxin to chromosome 11q13-qter and of adrenodoxin pseudo-genes to chromosome 20cen-q13.1. *Am. J. Hum. Genet.* 43: 52-59, 1988.
- [33266] 6818. Okamura, T.; John, M. E.; Zuber, M. X.; Simpson, E. R.; Waterman, M. R.: Molecular cloning and amino acid sequence of the precursor form of bovine adrenodoxin: evidence for a previously unidentified COOH-terminal peptide. *Proc. Nat. Acad. Sci.* 82: 5705-5709, 1985.
- [33267] 6819. Picado-Leonard, J.; Voutilainen, R.; Kao, L.-C.; Chung, B.-C.; Strauss, J. F., III; Miller, W. L.: Human adrenodoxin: cloning of three cDNAs and cycloheximide enhancement in JEG-3 cells. *J. Biol. Chem.* 263: 3240-3244, 1988.
- [33268] 6820. Sparkes, R. S.; Klisak, I.; Miller, W. L.: Regional mapping of genes encoding human steroidogenic enzymes: P450scc to 15q23-q24; adrenodoxin to 11q22; adrenodoxin reductase to 17q24-q25; and P450c17 to 10q24-q25. *DNA Cell Biol.* 10: 359-365, 1991.
- [33269] 6821. Lin, D.; Shi, Y.; Miller, W. L.: Cloning and sequence of the human adrenodoxin reductase gene. *Proc. Nat.*

Acad. Sci. 87: 8516–8520,1990.

[33270] 6822.Solish, S. B.; Picado–Leonard, J.; Morel, Y.; Kuhn, R. W.; Mohandas,T. K.; Hanukoglu, I.; Miller, W. L.: Human adrenodoxin reductase:two mRNAs encoded by a single gene on chromosome 17cen–q25 are expressed in steroidogenic tissues. Proc. Nat. Acad. Sci. 85: 7104–7108,1988.

[33271] 6823.Caron, K. M.; Smithies, O.: Extreme hydrops fetalis and cardiovascular abnormalities in mice lacking a functional adrenomedullin gene. Proc.Nat. Acad. Sci. 98: 615–619, 2001.

[33272] 6824.Ishimitsu, T.; Kojima, M.; Kangawa, K.; Hino, J.; Matsuoka, H.;Kitamura, K.; Eto, T.; Matsuo, H.: Genomic structure of human adrenomedullin gene. Biochem. Biophys. Res. Commun. 203: 631–639, 1994.

[33273] 6825.Kitamura, K.; Sakata, J.; Kangawa, K.; Kojima, M.; Matsuo, H.;Eto, T.: Cloning and characterization of cDNA encoding a precursor for human adrenomedullin. Biochem. Biophys. Res. Commun. 194: 720–725,1993.

[33274] 6826.Makino, Y.; Shibata, K.; Makino, I.; Kangawa, K.; Kawarabayashi,T.: Alteration of the adrenomedullin receptor components gene expression associated with the blood pressure in pregnancy–induced hypertension. J.Clin. En–

docr. Metab. 86: 5079–5082, 2001.

- [33275] 6827. McLatchie, L. M.; Fraser, N. J.; Main, M. J.; Wise, A.; Brown, J.; Thompson, N.; Solari, R.; Lee, M. G.; Foord, S. M.: RAMPs regulate the transport and ligand specificity of the calcitonin–receptor–like receptor. *Nature* 393: 333–339, 1998.
- [33276] 6828. Okazaki, T.; Ogawa, Y.; Tamura, N.; Mori, K.; Isse, N.; Aoki, T.; Rochelle, J. M.; Taketo, M. M.; Seldin, M. F.; Nakao, K.: Genomic organization, expression, and chromosomal mapping of the mouse adrenomedullin gene. *Genomics* 37: 395–399, 1996.
- [33277] 6829. Richards, A. M.; Nicholls, M. G.; Lewis, L.; Lainchbury, J. G.: Adrenomedullin. *Clin. Sci.* 91: 3–16, 1996.
- [33278] 6830. Uono, T.; Takahashi, K.; Nakayama, M.; Yoshinoya, A.; Totsune, K.; Murakami, O.; Durlu, Y. K.; Tamai, M.; Shibahara, S.: Induction of adrenomedullin by hypoxia in cultured retinal pigment epithelial cells. *Invest. Ophthalm. Vis. Sci.* 42: 1080–1086, 2001.
- [33279] 6831. van Heyningen, V.; Jones, C.: Report of the committee on the genetic constitution of chromosome 11. In: Cuticchia, A. J.; Pearson, P. L.; Klinger, H. P. (eds.): Chromosome coordinating meeting, 1992. *Genome Priority Reports*, Vol 1. Basel: S. Karger (pub.) 1993. Pp. 365–401.

- [33280] 6832.Ferguson-Smith, A. C.; Cattanach, B. M.; Barton, S. C.; Beechey, C. V.; Surani, M. A.: Embryological and molecular investigation of parental imprinting on mouse chromosome 7. *Nature* 351: 667–670, 1991.
- [33281] 6833.Ohno, S.: Ancient linkage groups and frozen accidents. *Nature* 244:259–262, 1973.
- [33282] 6834.Tolan, D. R.; Niclas, J.; Bruce, B. D.; Lebo, R. V.: Evolutionary implications of the human aldolase-A, -B, -C, and -pseudogene chromosomal locations. *Am. J. Hum. Genet.* 41: 907–924, 1987.
- [33283] 6835.Buono, P.; Paoletta, G.; Mancini, F. P.; Izzo, P.; Salvatore, F.: The complete nucleotide sequence of the gene coding for the human aldolase C. *Nucleic Acids Res.* 16: 4733 only, 1988.
- [33284] 6836.Rocchi, M.; Vitale, E.; Covone, A.; Romeo, G.; Santamaria, R.; Buono, P.; Paoletta, G.; Salvatore, F.: Assignment of human aldolase C gene to chromosome 17, region centromere-q21.1. *Hum. Genet.* 82: 279–282, 1989.
- [33285] 6837.Rottmann, W. H.; Deselms, K. R.; Niclas, J.; Camerato, T.; Holman, P. S.; Green, C. J.; Tolan, D. R.: The complete amino acid sequence of the human aldolase C isozyme derived from genomic clones. *Biochimie* 69:137–145, 1987.

- [33286] 6838. Bateman, J. B.; Kojis, T.; Heinzmann, C.; Klisak, I.; Diep, A.; Carper, D.; Nishimura, C.; Mohandas, T.; Sparkes, R. S.: Mapping of aldose reductase gene sequences to human chromosomes 1, 3, 7, 9, 11, and 13. *Genomics* 17: 560–565, 1993.
- [33287] 6839. Brown, L.; Hedge, P. J.; Markham, A. F.; Graham, A.: A human aldehyde dehydrogenase (aldose reductase) pseudogene: nucleotide sequence analysis and assignment to chromosome 3. *Genomics* 13: 465–468, 1992.
- [33288] 6840. Chung, S.; LaMendola, J.: Cloning and sequence determination of human placental aldose reductase gene. *J. Biol. Chem.* 264: 14775–14777, 1989.
- [33289] 6841. Graham, A.; Brown, L.; Hedge, P. J.; Gammack, A. J.; Markham, A. F.: Structure of the human aldose reductase gene. *J. Biol. Chem.* 266: 6872–6877, 1991.
- [33290] 6842. Graham, A.; Heath, P.; Morten, J. E. N.; Markham, A. F.: The human aldose reductase gene maps to chromosome region 7q35. *Hum. Genet.* 86: 509–514, 1991.
- [33291] 6843. Nishimura, C.; Matsuura, Y.; Kokai, Y.; Akera, T.; Carper, D.; Morjana, N.; Lyons, C.; Flynn, T. G.: Cloning and expression of human aldose reductase. *J. Biol. Chem.* 265: 9788–9792, 1990.
- [33292] 6844. Shah, V. O.; Dorin, R. I.; Sun, Y.; Braun, M.; Zager, P.

G.: Aldosereductase gene expression is increased in diabetic nephropathy. *J.Clin. Endocr. Metab.* 82: 2294–2298, 1997.

[33293] 6845.Roberds, S. L.; Anderson, J.; Basi, G.; Bienkowski, M. J.; Branstetter, D. G.; Chen, K. S.; Freedman, S. B.; Frigon, N. L.; Games, D.; Hu, K.; Johnson–Wood, K.; Kappenman, K. E.; and 20 others: BACE knockout mice are healthy despite lacking the primary beta–secretase activity in brain: implications for Alzheimer's disease therapeutics. *Hum.Molec. Genet.* 10: 1317–1324, 2001.

[33294] 6846.Botto, M.; Hawkins, P. N.; Bickerstaff, M. C.; Herbert, J.; Bygrave, A. E.; McBride, A.; Hutchinson, W. L.; Tennent, G. A.; Walport, M.J.; Pepys, M. B.: Amyloid deposition is delayed in mice with targeted deletion of the serum amyloid P component gene. *Nature Med.* 3: 855–859, 1997.

[33295] 6847.Floyd–Smith, G. A.; Whitehead, A. S.; Colten, H. R.; Francke, U.: Human serum amyloid P component (SAP) is located on the proximal long arm of chromosome 1. (Abstract) *Cytogenet. Cell Genet.* 40:631 only, 1985.

[33296] 6848.Ionasescu, V.; Burns, T.; Searby, C.; Ionasescu, R.: Linkage between the loci for Duffy (FY) and serum amyloid P component (APCS) on human chromosome 1. *Cytogenet. Cell Genet.* 45: 240–241, 1987.

- [33297] 6849.Mantzouranis, E. C.; Dowton, S. B.; Whitehead, A. S.; Edge, M.D.; Bruns, G. A. P.; Colten, H. R.: Human serum amyloid P component:cDNA isolation, complete sequence of pre-serum amyloid P component,and localization of the gene to chromosome 1. *J. Biol. Chem.* 260:7752–7756, 1985.
- [33298] 6850.Mortensen, R. F.; Le, P. T.; Taylor, B. A.: Mouse serum amyloidP–component (SAP) levels controlled by a locus on chromosome 1. *Immunogenetics* 22:367–375, 1985.
- [33299] 6851.Pepys, M. B.; Herbert, J.; Hutchinson, W. L.; Tennent, G. A.; Lachmann,H. J.; Gallimore, J. R.; Lovat, L. B.; Bartfai, T.; Alanine, A.; Hertel,C.; Hoffmann, T.; Jakob–Roetne, R.; and 11 others: Targeted pharmacologicaldepletion of serum amyloid P component for treatment of human amyloidosis. *Nature* 417:254–259, 2002.
- [33300] 6852.Prelli, F.; Pras, M.; Frangione, B.: The primary structure ofhuman tissue amyloid P component from a patient with primary idiopathicamyloidosis. *J. Biol. Chem.* 260: 12895–12898, 1985.
- [33301] 6853.Whitehead, A. S.; Rits, M.; Michaelson, J.: Molecular geneticsof mouse serum amyloid P component (SAP): cloning and gene mapping. *Immunogenetics* 28:388–390,

1988.

- [33302] 6854.Woo, P.; O'Brien, J.; Robson, M.; Ansell, B. M.: A genetic marker for systemic amyloidosis in juvenile arthritis. *Lancet* I: 767–769, 1987.
- [33303] 6855.Lenkkeri, U.; Kestila, M.; Lamerdin, J.; McCready, P.; Adamson, A.; Olsen, A.; Tryggvason, K.: Structure of the human amyloid–precursor–like protein gene APLP1 at 19q13.1. *Hum. Genet.* 102: 192–196, 1998.
- [33304] 6856.Wasco, W.; Brook, J. D.; Tanzi, R. E.: The amyloid precursor–like protein (APLP) gene maps to the long arm of human chromosome 19. *Genomics* 15:237–239, 1993.
- [33305] 6857.Wasco, W.; Bupp, K.; Magendantz, M.; Gusella, J. F.; Tanzi, R.E.; Solomon, F.: Identification of a mouse brain cDNA that encodes a protein related to the Alzheimer–associated amyloid beta–protein precursor. *Proc. Nat. Acad. Sci.* 89: 10758–10762, 1992.
- [33306] 6858.Leach, R.; Ko, M.; Krawetz, S. A.: Assignment of amyloid–precursor–like protein 2 gene (APLP2) to 11q24 by fluorescent in situ hybridization. *Cytogenet. Cell Genet.* 87: 215–216, 1999.
- [33307] 6859.von der Kammer, H.; Loffler, C.; Hanes, J.; Klaudiny, J.; Scheit, K. H.; Hansmann, I.: The gene for the amyloid precursor–like protein APLP2 is assigned to human chro–

mosome 11q23–25. *Genomics* 10: 308–311,1994.

- [33308] 6860.von Koch, C. S.; Lahiri, D. K.; Mammen, A. L.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Sisodia, S. S.: The mouse APLP2 gene:chromosomal localization and promoter characterization. *J. Biol.Chem.* 270: 25475–25480, 1995.
- [33309] 6861.Yan, Y. C.; Bai, Y.; Wang, L.; Miao, S.; Koide, S. S.: Characterizationof cDNA encoding a human sperm membrane protein related to A4 amyloidprotein. *Proc. Nat. Acad. Sci.* 87: 2405–2408, 1990.
- [33310] 6862.Yang, Y.; Martin, L.; Cuzin, F.; Mattei, M.–G.; Rasoulzadegan,M.: Genomic structure and chromosomal localization of the mouse CDEI–bindingprotein CDEBP (APLP2) gene and promoter sequences. *Genomics* 35:24–29, 1996.
- [33311] 6863.Wallukat, G.; Homuth, V.; Fischer, T.; Lindschau, C.; Horstkamp,B.; Jupner, A.; Baur, E.; Nissen, E.; Vetter, K.; Neichel, D.; Dudenhausen,J. W.; Haller, H.; Luft, F. C.: Patients with preeclampsia developagonistic autoantibodies against the angiotensin AT–1 receptor. *J.Clin. Invest.* 103: 945–952, 1999.
- [33312] 6864.Wang, W. Y. S.; Zee, R. Y. L.; Morris, B. J.: Association ofangiotensin II type 1 receptor gene polymorphism with

essential hypertension. Clin.Genet. 51: 31–34, 1997.

- [33313] 6865.Wilson, G. N.; Dasouki, M.; Barr, M., Jr.: Further delineation of the dup(3q) syndrome. Am. J. Med. Genet. 22: 117–123, 1985.
- [33314] 6866.Amant, C.; Bauters, C.; Bodart, J.–C.; Lablanche, J.–M.; Grollier, G.; Danchin, N.; Hamon, M.; Richard, F.; Helbecque, N.; McFadden, E. P.; Amouyel, P.; Bertrand, M. E.: D allele of the angiotensin I–converting enzyme is a major risk factor for restenosis after coronary stenting. Circulation 96: 56–60, 1997.
- [33315] 6867.Arbustini, E.; Grasso, M.; Fasani, R.; Klersy, C.; Diegoli, M.; Porcu, E.; Banchieri, N.; Fortina, P.; Danesino, C.; Specchia, G.: Angiotensin converting enzyme gene deletion allele is independently and strongly associated with coronary atherosclerosis and myocardial infarction. Brit. Heart J. 74: 584–591, 1995.
- [33316] 6868.Berge, K. E.; Berg, K.: No effect of insertion/deletion polymorphism at the ACE locus on normal blood pressure level or variability. Clin.Genet. 45: 169–174, 1994.
- [33317] 6869.Bohn, M.; Berge, K. E.; Bakken, A.; Erikssen, J.; Berg, K.: Insertion/deletion(I/D) polymorphism at the locus for angiotensin I–converting enzyme and myocardial infarction. Clin. Genet. 44: 292–297, 1993.

- [33318] 6870.Bohn, M.; Berge, K. E.; Bakken, A.; Erikssen, J.; Berg, K.: Insertion/deletion(I/D) polymorphism at the locus for angiotensin I-converting enzymeand parental history of myocardial infarction. Clin. Genet. 44:298–301, 1993.
- [33319] 6871.Brown, N. J.; Ray, W. A.; Snowden, M.; Griffin, M. R.: Black Americanshave an increased rate of angiotensin converting enzyme inhibitor-associatedangioedema. Clin. Pharmacol. Therapeutics 60: 8–13, 1996.
- [33320] 6872.Cambien, F.; Alhenc-Gelas, F.; Herbeth, B.; Andre, J. L.; Rakotovao,R.; Gonzales, M. F.; Allegrini, J.; Bloch, C.: Familial resemblanceof plasma angiotensin-converting enzyme level: the Nancy study. Am.J. Hum. Genet. 43: 774–780, 1988.
- [33321] 6873.Cambien, F.; Poirier, O.; Lecerf, L.; Evans, A.; Cambou, J.-P.;Arveiler, D.; Luc, G.; Bard, J.-M.; Bara, L.; Ricard, S.; Tiret, L.;Amouyel, P.; Alhenc-Gelas, F.; Soubrier, F.: Deletion polymorphismin the gene for angiotensin-converting enzyme is a potent risk factorfor myocardial infarction. Nature 359: 641–644, 1992.
- [33322] 6874.Doria, A.; Warram, J. H.; Krolewski, A. S.: Genetic predispositionto diabetic nephropathy: evidence for a role of the angiotensin I-convertingenzyme gene. Diabetes 43: 690–695, 1994.

- [33323] 6875.Ehlers, M. R. W.; Fox, E. A.; Strydom, D. J.; Riordan, J. F.:Molecular cloning of human testicular angiotensin-converting enzyme:the testis isozyme is identical to the C-terminal half of endothelialangiotensin-converting enzyme. Proc. Nat. Acad. Sci. 86: 7741-7745,1989.
- [33324] 6876.Esther, C. R., Jr.; Marino, E. M.; Howard, T. E.; Machaud, A.;Corvol, P.; Capecchi, M. R.; Bernstein, K. E.: The critical roleof tissue angiotensin-converting enzyme as revealed by gene targetingin mice. J. Clin. Invest. 99: 2375-2385, 1997.
- [33325] 6877.Evans, A. E.; Poirier, O.; Kee, F.; Lecerf, L.; McCrum, E.; Falconer,T.; Crane, J.; O'Rourke, D. F.; Cambien, F.: Polymorphisms of theangiotensin-converting-enzyme gene in subjects who die from coronaryheart disease. Quart. J. Med. 87: 211-214, 1994.
- [33326] 6878.Exner, D. V.; Dries, D. L.; Domanski, M. J.; Cohn, J. N.: Lesserresponse to angiotensin-converting-enzyme inhibitor therapy in blacks compared with white patients with left ventricular dysfunction. NewEng. J. Med. 344: 1351-1357, 2001.
- [33327] 6879.Gardemann, A.; Fink, M.; Stricker, J.; Nguyen, Q. D.; Humme, J.;Katz, N.; Tillmanns, H.; Hehrlein, F. W.; Rau, M.; Haberbosch, W.: ACE I/D gene polymorphism: presence of

the ACE D allele increases the risk of coronary artery disease in younger individuals. *Atherosclerosis* 139:153–159, 1998.

- [33328] 6880. Gayagay, G.; Yu, B.; Hambly, B.; Boston, T.; Hahn, A.; Celermajer, D. S.; Trent, R. J.: Elite endurance athletes and the ACE I allele: the role of genes in athletic performance. *Hum. Genet.* 103: 48–50, 1998.
- [33329] 6881. Hagaman, J. R.; Moyer, J. S.; Bachman, E. S.; Sibony, M.; Magyar, P. L.; Welch, J. E.; Smithies, O.; Kregge, J. H.; O'Brien, D. A.: Angiotensin-converting enzyme and male fertility. *Proc. Nat. Acad. Sci.* 95: 2552–2557, 1998.
- [33330] 6882. Hamdi, H. K.; Reznik, J.; Castellon, R.; Atilano, S. R.; Ong, J. M.; Udar, N.; Tavis, J. H.; Aoki, A. M.; Nesburn, A. B.; Boyer, D. S.; Small, K. W.; Brown, D. J.; Kenney, M. C.: Alu DNA polymorphism in ACE gene is protective for age-related macular degeneration. *Biochem. Biophys. Res. Commun.* 295: 668–672, 2002.
- [33331] 6883. Howard, T. E.; Shai, S. Y.; Langford, K. G.; Martin, B. M.; Bernstein, K. E.: Transcription of testicular angiotensin-converting enzyme (ACE) is initiated within the 12th intron of the somatic ACE gene. *Molec. Cell. Biol.* 10: 4294–4302, 1990.
- [33332] 6884. Huang, W.; Gallois, Y.; Bouby, N.; Bruneval, P.;

Heudes, D.; Belair, M.-F.; Krege, J. H.; Menteton, P.; Marre, M.; Smithies, O.; Alhenc-Gelas, F.: Genetically increased angiotensin I-converting enzyme level and renal complications in the diabetic mouse. *Proc. Nat. Acad. Sci.* 98:13330–13334, 2001.

[33333] 6885. Jeffery, S.; Malik, A. K. S.; Crosby, A.; Bland, M.; Eastwood, J. B.; Amoah-Danquah, J.; Acheampong, J. W.; Plange-Rhule, J.: Adominant relationship between the ACE D allele and serum ACE levels in a Ghanaian population. (Letter) *J. Med. Genet.* 36: 869–870, 1999.

[33334] 6886. Jeunemaitre, X.; Lifton, R. P.; Hunt, S. C.; Williams, R. R.; Lalouel, J.-M.: Absence of linkage between the angiotensin converting enzyme locus and human essential hypertension. *Nature Genet.* 1:72–75, 1992.

[33335] 6887. Keavney, B.; McKenzie, C.; Parish, S.; Palmer, A.; Clark, S.; Youngman, L.; Delepine, M.; Lathrop, M.; Peto, R.; Collins, R.: Large-scale test of hypothesized associations between the angiotensin-converting-enzyme insertion/deletion polymorphism and myocardial infarction in about 5000 cases and 6000 controls. *Lancet* 355: 434–442, 2000.

[33336] 6888. Kehoe, P. G.; Russ, C.; McIlroy, S.; Williams, H.; Holmans, P.; Holmes, C.; Liolitsa, D.; Vahidassr, D.; Powell, J.;

McGleenon, B.;Liddell, M.; Plomin, R.; and 9 others: Variation in DCP1, encodingACE, is associated with susceptibility to Alzheimer disease. (Letter) NatureGenet. 21: 71–72, 1999.

- [33337] 6889.Keramatipour, M.; McConnell, R. S.; Kirkpatrick, P.; Tebbs, S.;Furlong, R. A.; Rubinsztein, D. C.: The ACE I allele is associatedwith increased risk for ruptured intracranial aneurysms. J. Med.Genet. 37: 498–500, 2000.
- [33338] 6890.Oliver, P. M.; Fox, J. E.; Kim, R.; Rockman, H. A.; Kim, H.–S.;Reddick, R. L.; Pandey, K. N.; Milgram, S. L.; Smithies, O.; Maeda,N.: Hypertension, cardiac hypertrophy, and sudden death in mice lackingnatriuretic peptide receptor A. Proc. Nat. Acad. Sci. 94: 14730–14735,1997.
- [33339] 6891.Oliver, P. M.; John, S. W. M.; Purdy, K. E.; Kim, R.; Maeda, N.;Goy, M. F.; Smithies, O.: Natriuretic peptide receptor 1 expressioninfluences blood pressures of mice in a dose–dependent manner. Proc.Nat. Acad. Sci. 95: 2547–2551, 1998.
- [33340] 6892.He, X.; Chow, D.; Martick, M. M.; Garcia, K. C.: Allosteric activationof a spring–loaded natriuretic peptide receptor dimer by hormone. Science 293:1657–1662, 2001.
- [33341] 6893.Jaubert, J.; Jaubert, F.; Martin, N.; Washburn, L. L.;

Lee, B.K.; Eicher, E. M.; Guenet, J.-L.: Three new allelic mouse mutations that cause skeletal overgrowth involve the natriuretic peptide receptor C gene (Npr3). *Proc. Nat. Acad. Sci.* 96: 10278–10283, 1999.

[33342] 6894. Lopez, M. J.; Wong, S. K.-F.; Kishimoto, I.; Dubois, S.; Mach, V.; Friesen, J.; Garbers, D. L.; Beuve, A.: Salt-resistant hypertension in mice lacking the guanylyl cyclase-A receptor for atrial natriuretic peptide. *Nature* 378: 65–68, 1995.

[33343] 6895. Tweardy, D. J.; Cannizzaro, L. A.; Palumbo, A. P.; Shane, S.; Huebner, K.; Vantuinen, P.; Ledbetter, D. H.; Finan, J. B.; Nowell, P. C.; Rovera, G.: Molecular cloning and characterization of a cDNA for human granulocyte colony-stimulating factor (G-CSF) from a glioblastoma multiforme cell line and localization of the G-CSF gene to chromosome band 17q21. *Oncogene Res.* 1: 209–220, 1987.

[33344] 6896. Dong, F.; Hoefsloot, L. H.; Schelen, A. M.; Broeders, L. C. A.M.; Meijer, Y.; Veerman, A. J. P.; Touw, I. P.; Lowenberg, B.: Identification of a nonsense mutation in the granulocyte-colony-stimulating factor receptor in severe congenital neutropenia. *Proc. Nat. Acad. Sci.* 91: 4480–4484, 1994.

[33345] 6897. Dong, F.; Qiu, Y.; Yi, T.; Touw, I. P.; Larner, A. C.:

The carboxylterminus of the granulocyte colony-stimulating factor receptor, truncated in patients with severe congenital neutropenia/acute myeloid leukemia, is required for SH2-containing phosphatase-1 suppression of Stat activation. *J. Immun.* 167: 6447-6452, 2001.

[33346] 6898. Fukunaga, R.; Seto, Y.; Mizushima, S.; Nagata, S.: Three different mRNAs encoding human granulocyte colony-stimulating factor receptor. *Proc. Nat. Acad. Sci.* 87: 8702-8706, 1990.

[33347] 6899. Inazawa, J.; Fukunaga, R.; Seto, Y.; Nakagawa, H.; Misawa, S.; Abe, T.; Nagata, S.: Assignment of the human granulocyte colony-stimulating factor receptor gene (CSF3R) to chromosome 1 at region p35-p34.3. *Genomics* 10:1075-1078, 1991.

[33348] 6900. Kostmann, R.: Infantile Genetic Agranulocytosis (agranulocytosis infantilis hereditaria): a new recessive lethal disease in man. Uppsala: Almqvist and Wiksells Boktryckeri (pub.) 1956.

[33349] 6901. McLemore, M. L.; Poursine-Laurent, J.; Link, D. C.: Increased granulocyte colony-stimulating factor responsiveness but normal resting granulopoiesis in mice carrying a targeted granulocyte colony-stimulating factor receptor mutation derived from a patient with severe con-

genitalneutropenia. J. Clin. Invest. 102: 483–492, 1998.

[33350] 6902.Seto, Y.; Fukunaga, R.; Nagata, S.: Chromosomal gene organization of the human granulocyte colony-stimulating factor receptor. J. Immun. 148:259–266, 1992.

[33351] 6903.Tidow, N.; Pilz, C.; Teichmann, B.; Muller-Brechlin, A.; Germeshausen, M.; Kasper, B.; Rauprich, P.; Sykora, K.-W.; Welte, K.: Clinical relevance of point mutations in the cytoplasmic domain of the granulocyte colony-stimulating factor receptor gene in patients with severe congenitalneutropenia. Blood 89: 2369–2375, 1997.

[33352] 6904.Tweardy, D. J.; Anderson, K.; Cannizzaro, L. A.; Steinman, R.A.; Croce, C. M.; Huebner, K.: Molecular cloning of cDNAs for the human granulocyte colony-stimulating factor receptor from HL-60 and mapping of the gene to chromosome region 1p32–34. Blood 79: 1148–1154, 1992.

[33353] 6905.Wong, W.-Y.; Williams, D.; Slovak, M. L.; Charak, B.; Mazumder, A.; Snyder, D.; Powars, D. R.; Byrnes, R. K.: Terminal acute myelogenous leukemia in a patient with congenital agranulocytosis. Am. J. Hemat. 43:133–138, 1993.

[33354] 6906.D'Andrea, R.; Rayner, J.; Moretti, P.; Lopez, A.;

Goodall, G. J.;Gonda, T. J.; Vadas, M.: A mutation of the common receptor subunitfor interleukin-3 (IL-3), granulocyte-macrophage colony-stimulatingfactor, and IL-5 that leads to ligand independence and tumorigenicity. Blood 83:2802-2808, 1994.

[33355] 6907.Dirksen, U.; Hattenhorst, U.; Schneider, P.; Schroten, H.; Gobel,U.; Bocking, A.; Muller, K.-M.; Murray, R.; Burdach, S.: Defectiveexpression of granulocyte-macrophage colony-stimulating factor/interleukin-3/interleukin-5receptor common beta chain in children with acute myeloid leukemiaassociated with respiratory failure. Blood 92: 1097-1103, 1998.

[33356] 6908.Dirksen, U.; Nishinakamura, R.; Groneck, P.; Hattenhorst, U.; Noguee,L.; Murray, R.; Burdach, S.: Human pulmonary alveolar proteinosisassociated with a defect in GM-CSF/IL-3/IL-5 receptor common betachain expression. J. Clin. Invest. 100: 2211-2217, 1997.

[33357] 6909.Wada, K.; Yokotani, N.; Hunter, C.; Doi, K.; Wentholt, R. J.; Shimasaki,S.: Differential expression of two distinct forms of mRNA encodingmembers of a dipeptidyl aminopeptidase family. Proc. Nat. Acad. Sci. 89:197-201, 1992.

[33358] 6910.Wada, K.; Zimmerman, K. L.; Adamson, M. C.;

Yokotani, N.; Wenthold, R. J.; Kozak, C. A.: Genetic mapping of the mouse gene encoding dipeptidylaminopeptidase-like proteins. *Mammalian Genome* 4: 234–237, 1993.

[33359] 6911. Yokotani, N.; Doi, K.; Wenthold, R. J.; Wada, K.: Non-conservation of a catalytic residue in a dipeptidyl aminopeptidase IV-related protein encoded by a gene on human chromosome 7. *Hum. Molec. Genet.* 2: 1037–1039, 1993.

[33360] 6912. Chang, T.-M.; Neville, D. M., Jr.: Demonstration of diphtheria toxin receptors on surface membranes from both toxin-sensitive and toxin-resistant species. *J. Biol. Chem.* 253: 6866–6871, 1978.

[33361] 6913. Creagan, R. P.; Chen, S.-H.; Ruddle, F. H.: Genetic analysis of the cell surface: association of human chromosome 5 with sensitivity to diphtheria toxin in mouse-human somatic cell hybrids. *Proc. Nat. Acad. Sci.* 72: 2237–2241, 1975.

[33362] 6914. Fen, Z.; Dhadly, M. S.; Yoshizumi, M.; Hilkert, R. J.; Quertermous, T.; Eddy, R. L.; Shows, T. B.; Lee, M.-E.: Structural organization and chromosomal assignment of the gene encoding the human heparin-binding epidermal growth factor-like growth factor/diphtheria toxin recep-

tor. *Biochemistry* 32:7932–7938, 1993.

- [33363] 6915. George, D. L.; Francke, U.: Regional mapping of human genes for hexosaminidase B and diphtheria toxin sensitivity on chromosome 5 using mouse X human hybrid cells. *Somat. Cell Genet.* 3: 629–638, 1977.
- [33364] 6916. Gupta, R. S.; Siminovitch, L.: Isolation and characterization of mutants of human diploid fibroblasts resistant to diphtheria toxin. *Proc. Nat. Acad. Sci.* 75: 3337–3340, 1978.
- [33365] 6917. Hayes, H.; Kaneda, Y.; Uchida, T.; Okada, Y.: Regional assignment of the gene for diphtheria toxin sensitivity using subchromosomal fragments in microcell hybrids. *Chromosoma* 96: 26–32, 1987.
- [33366] 6918. Higashiyama, S.; Abraham, J. A.; Miller, J.; Fiddes, J. C.; Klagsbrun, M.: A heparin-binding growth factor secreted by macrophage-like cells that is related to EGF. *Science* 251: 936–939, 1991.
- [33367] 6919. Wang, Q.; Curren, M. E.; Splawski, I.; Burn, T. C.; Millholland, J. M.; VanRaay, T. J.; Shen, J.; Timothy, K. W.; Vincent, G. M.; deJager, T.; Schwartz, P. J.; Towbin, J. A.; Moss, A. J.; Atkinson, D. L.; Landes, G. M.; Connors, T. D.; Keating, M. T.: Positional cloning of a novel potassium channel gene: KVLQT1 mutations cause cardiac arrhyth-

mias. *Nature Genet.* 12: 17–23, 1996.

- [33368] 6920. Gorman, D. M.; Itoh, N.; Jenkins, N. A.; Gilbert, D. J.; Copeland, N. G.; Miyajima, A.: Chromosomal localization and organization of the murine genes encoding the beta subunits (AIC2A and AIC2B) of the interleukin 3, granulocyte/macrophage colony-stimulating factor, and interleukin 5 receptors. *J. Biol. Chem.* 267: 15842–15848, 1992.
- [33369] 6921. Jenkins, B. J.; D'Andrea, R.; Gonda, T. J.: Activating point mutations in the common beta subunit of the human GM-CSF, IL-3 and IL-5 receptors suggest the involvement of beta subunit dimerization and cell type-specific molecules in signalling. *EMBO J.* 14: 4276–4287, 1995.
- [33370] 6922. Kitamura, T.; Sato, N.; Arai, K.; Miyajima, A.: Expression cloning of the human IL-3 receptor cDNA reveals a shared beta subunit for the human IL-3 and GM-CSF receptors. *Cell* 66: 1165–1174, 1991.
- [33371] 6923. Kondo, M.; Scherer, D. C.; Miyamoto, T.; King, A. G.; Akashi, K.; Sugamura, K.; Weissman, I. L.: Cell-fate conversion of lymphoid-committed progenitors by instructive actions of cytokines. *Nature* 407: 383–386, 2000.
- [33372] 6924. Robb, L.; Drinkwater, C. C.; Metcalf, D.; Li, R.; Kont-

gen, F.; Nicola, N. A.; Begley, C. G.: Hematopoietic and lung abnormalities in mice with a null mutation of the common beta subunit of the receptors for granulocyte-macrophage colony-stimulating factor and interleukins 3 and 5. *Proc. Nat. Acad. Sci.* 92: 9565–9569, 1995.

[33373] 6925. Shen, Y.; Baker, E.; Callen, D. F.; Sutherland, G. R.; Willson, T. A.; Rakar, S.; Gough, N. M.: Localization of the human GM-CSF receptor beta chain gene (CSF2RB) to chromosome 22q12.2–q13.1. *Cytogenet. Cell Genet.* 61: 175–177, 1992.

[33374] 6926. Tavernier, J.; Devos, R.; Cornelis, S.; Tuypens, T.; Van der Heyden, J.; Fiers, W.; Plaetinck, G.: A human high affinity interleukin-5 receptor (IL5R) is composed of an IL5-specific alpha chain and a beta chain shared with the receptor for GM-CSF. *Cell* 66: 1175–1184, 1991.

[33375] 6927. Prohl, C.; Pelzer, W.; Diekert, K.; Kmita, H.; Bedekovics, T.; Kispal, G.; Lill, R.: The yeast mitochondrial carrier Leu5p and its human homologue Graves' disease protein are required for accumulation of coenzyme A in the matrix. *Molec. Cell Biol.* 21: 1089–1097, 2001.

[33376] 6928. Rossi, E.; Zarrilli, R.; Zuffardi, O.: Regional assignment of the gene coding for a human Graves' disease autoantigen to 10q21.3–q22.1. *Hum. Genet.* 90: 653–654,

1993.

- [33377] 6929.Zarrilli, R.; Oates, E. L.; McBride, O. W.; Lerman, M. I.; Chan,J. Y.; Santisteban, P.; Ursini, M. V.; Notkins, A. L.; Kohn, L. D.: Sequence and chromosomal assignment of a novel cDNA identified by immunoscreening of a thyroid expression library: similarity to a family of mitochondrial solute carrier proteins. *Molec. Endocr.* 3: 1498–1508, 1989.
- [33378] 6930.Douglas, A. J.; Fox, M. F.; Abbott, C. M.; Hinks, L. J.; Sharpe,G.; Povey, S.; Thompson, R. J.: Structure and chromosomal localization of the human 2–prime,3–prime–cyclic nucleotide 3–prime–phosphodiesterase gene. *Ann. Hum. Genet.* 56: 243–254, 1992.
- [33379] 6931.Douglas, A. J.; Fox, M. F.; Hinks, L. J.; Povey, S.; Thompson,R. J.: Localization of the myelin specific enzyme 2–prime,3–prime–cyclic nucleotide–3–prime–phosphohydrolase to 17q21. (Abstract) *Cytogenet.Cell Genet.* 58: 2004 only, 1991.
- [33380] 6932.Kurihara, T.; Monoh, K.; Sakimura, K.; Takahashi, Y.: Alternativesplicing of mouse brain 2–prime,3–prime–cyclic nucleotide

3-prime-phosphodiesterase mRNA. Biochem. Biophys. Res. Commun. 170: 1074–1081, 1990.

[33381] 6933. Monoh, K.; Kurihara, T.; Takahashi, Y.; Ichikawa, T.; Kumanishi, T.; Hayashi, S.; Minoshima, S.; Shimizu, N.: Structure, expression and chromosomal localization of the gene encoding human 2-prime,3-prime-cyclic-nucleotide 3-prime-phosphodiesterase. Gene 129: 297–301, 1993.

[33382] 6934. Bernier, L.; Colman, D. R.; D'Eustachio, P.: Chromosomal locations of genes encoding 2-prime,3-prime cyclic nucleotide 3-prime-phosphodiesterase and glial fibrillary acidic protein in the mouse. J. Neurosci. Res. 20:497–504, 1988.

[33383] 6935. Bifulco, M.; Laezza, C.; Stingo, S.; Wolff, J.: 2-prime,3-prime-cyclic nucleotide 3-prime-phosphodiesterase: a membrane-bound, microtubule-associated protein and membrane anchor for tubulin. Proc. Nat. Acad. Sci. 99:1807–1812, 2002.

[33384] 6936. Sprinkle, T. J.; Kouri, R. E.; Fain, P. D.; Stoming, T. A.; Whitney, J. B., III: Chromosomal mapping of the human CNP gene using a meiotic crossover DNA panel, PCR, and allele-specific probes. Genomics 16:542–545, 1993.

[33385] 6937. Sprinkle, T. J.; Lanclos, K. D.; Lapp, D. F.: Assign-

ment of the human 2'-prime,3'-prime-cyclic nucleotide 3'-prime-phosphohydrolase gene to chromosome 17. *Genomics* 13: 877-880, 1992.

- [33386] 6938. Vogel, U. S.; Thompson, R. J.: Molecular structure, localization, and possible functions of the myelin-associated enzyme 2'-prime,3'-prime-cyclic nucleotide 3'-prime-phosphodiesterase. *J. Neurochem.* 50: 1667-1677, 1988.
- [33387] 6939. Bibb, J. A.; Chen, J.; Taylor, J. R.; Svenningsson, P.; Nishi, A.; Snyder, G. L.; Yan, Z.; Sagawa, Z. K.; Ouimet, C. C.; Nairn, A. C.; Nestler, E. J.; Greengard, P.: Effects of chronic exposure to cocaine are regulated by the neuronal protein Cdk5. *Nature* 410:376-380, 2001.
- [33388] 6940. Bibb, J. A.; Snyder, G. L.; Nishi, A.; Yan, Z.; Meijer, L.; Fienberg, A. A.; Tsai, L.-H.; Kwon, Y. T.; Girault, J.-A.; Czernik, A. J.; Huganir, R. L.; Hemmings, H. C., Jr.; Nairn, A. C.; Greengard, P.: Phosphorylation of DARPP-32 by Cdk5 modulates dopamine signalling in neurons. *Nature* 402:669-671, 1999.
- [33389] 6941. Ohshima, T.; Nagle, J. W.; Pant, H. C.; Joshi, J. B.; Kozak, C. A.; Brady, R. O.; Kulkarni, A. B.: Molecular cloning and chromosomal mapping of the mouse cyclin-dependent kinase 5 gene. *Genomics* 28:585-588, 1995.

- [33390] 6942.Ohshima, T.; Ward, J. M.; Huh, C.-G.; Longenecker, G.; Veeranna;Pant, H. C.; Brady, R. O.; Martin, L. J.; Kulka-rni, A. B.: Targeteddisruption of the cyclin-dependent ki-nase 5 gene results in abnormalcorticogenesis, neuronal pathology and perinatal death. *Proc. Nat.Acad. Sci.* 93: 11173–11178, 1996.
- [33391] 6943.Patrick, G. N.; Zukerberg, L.; Nikolic, M.; de la Monte, S.; Dikkes,P.; Tsai, L.-H.: Conversion of p35 to p25 deregulates Cdk5 activityand promotes neurodegenera-tion. *Nature* 402: 615–622, 1999.
- [33392] 6944.Demetrick, D. J.; Matsumoto, S.; Hannon, G. J.; Okamoto, K.; Xiong,Y.; Zhang, H.; Beach, D. H.: Chromo-somal mapping of the genes forthe human cell cycle pro-teins cyclin C (CCNC), cyclin E (CCNE), p21(CDKN1) and KAP (CDKN3). *Cytogenet. Cell Genet.* 69: 190–192, 1995.
- [33393] 6945.Inaba, T.; Matsushime, H.; Valentine, M.; Roussel, M. F.; Sherr,C. J.; Look, A. T.: Genomic organization, chromo-somal localization,and independent expression of human cyclin D genes. *Genomics* 13:565–574, 1992.
- [33394] 6946.Kim, H. A.; Pomeroy, S. L.; Whoriskey, W.; Pawlitzky, I.; Benowitz,L. I.; Sicinski, P.; Stiles, C. D.; Roberts, T. M.: A developmentallyregulated switch directs regenerative growth of Schwann cells throughcyclin D1. *Neuron* 26:

405–416, 2000.

- [33395] 6947.Xiong, Y.; Menninger, J.; Beach, D.; Ward, D. C.: Molecular cloning and chromosomal mapping of CCND genes encoding human D-type cyclins. *Genomics* 13:575–584, 1992.
- [33396] 6948.Motokura, T.; Keyomarsi, K.; Kronenberg, H. M.; Arnold, A.: Cloning and characterization of human cyclin D3, a cDNA closely related in sequence to the PRAD1/cyclin D1 proto-oncogene. *J. Biol. Chem.* 267:20412–20415, 1992.
- [33397] 6949.Motokura, T.; Yi, H. F.; Kronenberg, H. M.; McBride, O. W.; Arnold, A.: Assignment of the human cyclin D3 gene (CCND3) to chromosome 6p–q13. *Cytogenet. Cell Genet.* 61: 5–7, 1992.
- [33398] 6950.Wang, Z.; Sicinski, P.; Weinberg, R. A.; Zhang, Y.; Ravid, K.: Characterization of the mouse cyclin D3 gene: exon/intron organization and promoter activity. *Genomics* 35: 156–163, 1996.
- [33399] 6951.Schwartz, J.–C. D.; Zhang, X.; Fedorov, A. A.; Nathenson, S. G.; Almo, S. C.: Structural basis for co-stimulation by the human CTLA–4/B7–2 complex. *Nature* 410: 604–608, 2001.
- [33400] 6952.Stamper, C. C.; Zhang, Y.; Tobin, J. F.; Erbe, D. V.;

Ikemizu,S.; Davis, S. J.; Stahl, M. L.; Seehra, J.; Somers, W. S.; Mosyak,L.: Crystal structure of the B7–1/CTLA–4 complex that inhibits human immune responses. *Nature* 410: 608–611, 2001. Note: Erratum: *Nature*411: 617 only, 2001.

[33401] 6953.Allenspach, E. J.; Cullinan, P.; Tong, J.; Tang, Q.; Tesciuba,A. G.; Cannon, J. L.; Takahashi, S. M.; Morgan, R.; Burkhardt, J.K.; Sperling, A. I.: ERM–dependent movement of CD43 defines a novel protein complex distal to the immunological synapse. *Immunity* 15:739–750, 2001.

[33402] 6954.Bonilha, V. L.; Rodriguez–Boulan, E.: Polarity and developmental regulation of two PDZ proteins in the retinal pigment epithelium. *Invest.Ophthalm. Vis. Sci.* 42: 3274–3282, 2001.

[33403] 6955.Gould, K. L.; Bretscher, A.; Esch, F. S.; Hunter, T.: cDNA cloning and sequencing of the protein–tyrosine kinase substrate, ezrin, reveals homology to band 4.1. *EMBO J.* 8: 4133–4142, 1989.

[33404] 6956.Majander–Nordenswan, P.; Sainio, M.; Turunen, O.; Jaaskelainen,J.; Carpen, O.; Kere, J.; Vaheri, A.: Genomic structure of the human ezrin gene. *Hum. Genet.* 103: 662–665, 1998.

[33405] 6957.Pakkanen, R.; Vaheri, A.: Cytovillin and other mi–

crovillar proteins of human choriocarcinoma cells. J. Cell. Biochem. 41: 1–12, 1989.

- [33406] 6958. Rao, P. H.; Murty, V. V. V. S.; Gaidano, G.; Hauptschein, R.; Dalla-Favera, R.; Chaganti, R. S. K.: Sub-regional mapping of 8 single copy loci on chromosome 6 by fluorescence in situ hybridization. Cytogenet. Cell Genet. 66: 272–273, 1994.
- [33407] 6959. Roumier, A.; Olivo-Marin, J. C.; Arpin, M.; Michel, F.; Martin, M.; Mangeat, P.; Acuto, O.; Dautry-Varsat, A.; Alcover, A.: The membrane-microfilament linker ezrin is involved in the formation of the immunological synapse and in T cell activation. Immunity 15: 715–728, 2001.
- [33408] 6960. Turunen, O.; Winqvist, R.; Pakkanen, R.; Grzeschik, K.-H.; Wahlstrom, T.; Vaheri, A.: Cytovillin, a microvillar Mr 75,000 protein: cDNA sequence, prokaryotic expression, and chromosomal localization. J. Biol. Chem. 264: 16727–16732, 1989.
- [33409] 6961. Winqvist, R.; Turunen, O.; Pakkanen, R.; Grzeschik, K.-H.; Wahlstrom, T. and Vaheri, A.: Localization of the cytovillin gene to region q22–q27 of human chromosome 6. (Abstract) Cytogenet. Cell Genet. 51: 1108–1109, 1989.
- [33410] 6962. Brunet, J.-F.; Dosseto, M.; Denizot, F.; Mattei, M.-G.; Clark, W. R.; Haqqi, T. M.; Ferrier, P.; Nabholz, M.; Schmitt-

Verhulst, A.-M.; Luciani, M.-F.; Golstein, P.: The inducible cytotoxic T-lymphocyte-associated gene transcript CTLA-1 sequence and gene localization to mouse chromosome 14. *Nature* 322: 268-271, 1986.

[33411] 6963. Crosby, J. L.; Bleackley, R. C.; Nadeau, J. H.: A complex of serine protease genes expressed preferentially in cytotoxic T-lymphocytes closely linked to the T-cell receptor alpha- and delta-chain genes on mouse chromosome 14. *Genomics* 6: 252-259, 1990.

[33412] 6964. Dahl, C. A.; Bach, F. H.; Chan, W.; Huebner, K.; Russo, G.; Croce, C. M.; Herfurth, T.; Cairns, J. S.: Isolation of a cDNA clone encoding a novel form of granzyme B from human NK cells and mapping to chromosome 14. *Hum. Genet.* 84: 465-470, 1990.

[33413] 6965. Sheffield, V. C.; Stone, E. M.; Alward, W. L. M.; Drack, A. V.; Johnson, A. T.; Streb, L. M.; Nichols, B. E.: Genetic linkage of familial open angle glaucoma to chromosome 1q21-q31. *Nature Genet.* 4:47-50, 1993.

[33414] 6966. Stone, E. M.; Fingert, J. H.; Alward, W. L. M.; Nguyen, T. D.; Polansky, J. R.; Sunden, S. L. F.; Nishimura, D.; Clark, A. F.; Nystuen, A.; Nichols, B. E.; Mackey, D. A.; Ritch, R.; Kalenak, J. W.; Craven, E. R.; Sheffield, V. C.: Identification of a gene that causes primary open angle glaucoma. *Sci-*

ence 275: 668–670, 1997.

[33415] 6967. Wiggs, J. L.; Vollrath, D.: Molecular and clinical evaluation of a patient hemizygous for TIGR/MYOC. *Arch. Ophthalmol.* 119: 1674–1678, 2001.

[33416] 6968. Yoon, S.-J. K.; Kim, H.-S.; Moon, J.-I.; Lim, J. M.; Joo, C.-K.: Mutations of the TIGR/MYOC gene in primary open-angle glaucoma in Korea. (Letter) *Am. J. Hum. Genet.* 64: 1775–1778, 1999.

[33417] 6969. Muragaki, Y.; Mattei, M.-G.; Yamaguchi, N.; Olsen, B. R.; Ninomiya, Y.: The complete primary structure of the human alpha-1(VIII) chain and assignment of its gene (COL8A1) to chromosome 3. *Europ. J. Biochem.* 197: 615–622, 1991.

[33418] 6970. Muragaki, Y.; Shiota, C.; Inoue, M.; Ooshima, A.; Olsen, B. R.; Ninomiya, Y.: Alpha-1(VIII)-collagen gene transcripts encode a short-chain collagen polypeptide and are expressed by various epithelial, endothelial and mesenchymal cells in newborn mouse tissues. *Europ. J. Biochem.* 207: 895–902, 1992.

[33419] 6971. Odink, K.; Cerletti, N.; Bruggen, J.; Clerc, R. G.; Tarcay, L.; Zwadlo, G.; Gerhards, G.; Schlegel, R.; Sorg, C.: Two calcium-binding proteins in infiltrate macrophages of rheumatoid arthritis. *Nature* 330: 80–82, 1987.

- [33420] 6972.Schafer, B. W.; Wicki, R.; Engelkamp, D.; Mattei, M.-G.; Heizmann,C. W.: Isolation of a YAC clone covering a cluster of nine S100 geneson human chromosome 1q21: rationale for a new nomenclature of theS100 calcium-binding protein family. *Genomics* 25: 638–643, 1995.
- [33421] 6973.van Heyningen, V.; Emslie, E.; Dorin, J. R.: Related calcium bindingproteins map to the same sub-region of chromosome 1q and to an extendedregion of synteny on mouse chromosome 3. (Abstract) *Cytogenet.Cell Genet.* 51: 1095, 1989.
- [33422] 6974.van Heyningen, V.; Hayward, C.; Fletcher, J.; McAuley, C.: Tissuelocalization and chromosomal assignment of a serum protein which tracksthe cystic fibrosis gene. *Nature* 315: 513–515, 1985.
- [33423] 6975.Welsh, M. J.; Liedtke, C. M.: Chloride and potassium channelsin cystic fibrosis airway epithelia. *Nature* 322: 467–470, 1986.
- [33424] 6976.Wilkinson, M. M.; Busuttil, A.; Hayward, C.; Brock, D. J. H.;Dorin, J. R.; van Heyningen, V.: Expression pattern of two relatedcystic fibrosis-associated calcium-binding proteins in normal andabnormal tissues. *J. Cell Sci.* 91: 221–230, 1988.
- [33425] 6977.Wilson, G. B.; Fudenberg, H. H.; Jahn, T. L.: Studies

on cysticfibrosis using isoelectric focusing. I. An assay for detection ofcystic fibrosis homozygotes and heterozygote carriers from serum.Pediat. Res. 9: 635–640, 1975.

- [33426] 6978.van Heyningen, V.; Emslie, E.; Dorin, J. R.: Related calcium bindingproteins map to the same sub–region of chromosome 1q and to an extendedregion of synteny on mouse chromosome 3. (Abstract) Cytogenet. CellGenet. 51: 1095 only, 1989.
- [33427] 6979.Buonavista, N.; Balzano, C.; Pontarotti, P.; Le Paslier, D.; Golstein,P.: Molecular linkage of the human CTLA4 and CD28 Ig–superfamilygenes in yeast artificial chromo–somes. Genomics 13: 856–861, 1992.
- [33428] 6980.Howard, T. A.; Rochelle, J. M.; Seldin, M. F.: Cd28 and Ctla–4,two related members of the Ig supergene fam–ily, are tightly linkedon proximal mouse chromosome 1. Immunogenetics 33: 74–76, 1991.
- [33429] 6981.Albertson, D. G.; Ylstra, B.; Segraves, R.; Collins, C.; Dairkee,S. H.; Kowbel, D.; Kuo, W.– L.; Gray, J. W.; Pinkel, D.: Quantitativemapping of amplicon structure by array CGH identifies CYP24 as a candidateoncogene. Nature Genet. 25: 144–146, 2000.
- [33430] 6982.Chen, K.–S.; Prahl, J. M.; DeLuca, H. F.: Isolation and expressionof human 1,25–dihydroxyvitamin D3

24-hydroxylase cDNA. Proc. Nat.Acad. Sci. 90:
4543–4547, 1993.

[33431] 6983.Hahn, C. N.; Baker, E.; Laslo, P.; May, B. K.; Omdahl, J. L.; Sutherland,G. R.: Localization of the human vitamin D 24-hydroxylase gene (CYP24)to chromosome 20q13.2–q13.3. Cytogenet. Cell Genet. 62: 192–193,1993.

[33432] 6984.Labuda, M.; Lemieux, N.; Tihiy, F.; Prinster, C.; Glo-rioux, F. H.: Human 25-hydroxyvitamin D 24-hydroxylase cytochrome P450 subunitmaps to a different chromoso-mal location than that of pseudovitaminD-deficient rick-ets. J. Bone Miner. Res. 8: 1397–1406, 1993.

[33433] 6985.Malas, S.; Peters, J.; Abbott, C.: The genes for en-dothelin 3,vitamin D 24-hydroxylase, and melanocortin 3 receptor map to distalmouse chromosome 2, in the region of conserved syntenry with humanchromosome 20. Mam-malian Genome 5: 577–579, 1994.

[33434] 6986.Ohyama, Y.; Noshiro, M.; Okuda, K.: Cloning and expression ofcDNA encoding 25-hydroxyvitamin D(3) 24-hydroxylase. FEBS Lett. 278:195–198, 1991.

[33435] 6987.Mendel, D. B.; Khavari, P. A.; Conley, P. B.; Graves, M. K.; Hansen,L. P.; Admon, A.; Crabtree, G. R.: Character-ization of a cofactorthat regulates dimerization of a mam-

malian homeodomain protein. *Science* 254:1762–1767, 1991.

[33436] 6988. Hogenesch, J. B.; Chan, W. K.; Jackiw, V. H.; Brown, R. C.; Gu, Y.-Z.; Pray-Grant, M.; Perdew, G. H.; Bradfield, C. A.: Characterization of a subset of the basic-helix-loop-helix-PAS superfamily that interacts with components of the dioxin signaling pathway. *J. Biol. Chem.* 272:8581–8593, 1997.

[33437] 6989. Wang, G. L.; Jiang, B.-H.; Rue, E. A.; Semenza, G. L.: Hypoxia-inducible factor 1 is a basic-helix-loop-helix-PAS heterodimer regulated by cellular O(2) tension. *Proc. Nat. Acad. Sci.* 92: 5510–5514, 1995.

[33438] 6990. Le Beau, M. M.; Lemons, R. S.; Carrino, J. J.; Pette-nati, M. J.; Souza, L. M.; Diaz, M. O.; Rowley, J. D.: Chromosomal localization of the human G-CSF gene to 17q11 proximal to the breakpoint of the t(15;17) in acute promyelocytic leukemia. *Leukemia* 1: 795–799, 1987.

[33439] 6991. Haaparanta, T.; Uitto, J.; Ruoslahti, E.; Engvall, E.: Molecular cloning of the cDNA encoding human laminin A chain. *Matrix* 11: 151–160, 1991.

[33440] 6992. Gallagher, P. G.; Forget, B. G.: Structure, organization, and expression of the band 7.2b gene, a candidate gene for hereditary hydrocytosis. *J. Biol. Chem.* 270:

26358–26363, 1995.

- [33441] 6993. Gallagher, P. G.; Romana, M.; Lieman, J. H.; Ward, D. C.: cDNA structure, tissue-specific expression, and chromosomal localization of the murine band 7.2b gene. *Blood* 86: 359–365, 1995.
- [33442] 6994. Gallagher, P. G.; Upender, M.; Ward, D. C.; Forget, B. G.: The gene for human erythrocyte membrane protein band 7.2 (EPB72) maps to 9q33–q34 centromeric to the Philadelphia chromosome translocation breakpoint region. *Genomics* 18: 167–169, 1993.
- [33443] 6995. Hiebl–Dirschmied, C. M.; Entler, B.; Glotzmann, C.; Maurer–Fogy, I.; Stratowa, C.; Prohaska, R.: Cloning and nucleotide sequence of cDNA encoding human erythrocyte band 7 integral membrane protein. *Biochim. Biophys. Acta* 1090: 123–124, 1991.
- [33444] 6996. Pilz, A.; Prohaska, R.; Peters, J.; Abbott, C.: Genetic linkage analysis of the Ak1, Col5a1, Epb7.2, Fpgs, Grp78, Pbx3, and Notch1 genes in the region of mouse chromosome 2 homologous to human chromosome 9q. *Genomics* 21: 104–109, 1994.
- [33445] 6997. Unfried, I.; Entler, B.; Prohaska, R.: The organization of the gene (EPB72) encoding the human erythrocyte band 7 integral membrane protein (protein 7.2b). *Genomics* 30:

521–528, 1995.

- [33446] 6998. Westberg, J. A.; Entler, B.; Prohaska, R.; Schroder, J. P.: The gene coding for erythrocyte protein band 7.2b (EPB72) is located in band q34.1 of human chromosome 9. *Cytogenet. Cell Genet.* 63: 241–243, 1993.
- [33447] 6999. Zhu, Y.; Paszty, C.; Turetsky, T.; Tsai, S.; Kuypers, F. A.; Lee, G.; Cooper, P.; Gallagher, P. G.; Stevens, M. E.; Rubin, E.; Mohandas, N.; Mentzer, W. C.: Stomatocytosis is absent in 'stomatin'-deficient murine red blood cells. *Blood* 93: 2404–2410, 1999.
- [33448] 7000. de la Chapelle, A.; Sistonen, P.; Lehvaslaiho, H.; Ikkala, E.; Juvonen, E.: Familial erythrocytosis genetically linked to erythropoietin receptor gene. *Lancet* 341: 82–84, 1993.
- [33449] 7001. Juvonen, E.; Ikkala, E.; Fyhrquist, F.; Ruutu, T.: Autosomal dominant erythrocytosis caused by increased sensitivity to erythropoietin. *Blood* 78: 3066–3069, 1991.
- [33450] 7002. Craig, J. E.; Baird, P. N.; Healey, D. L.; McNaught, A. I.; McCartney, P. J.; Rait, J. L.; Dickinson, J. L.; Roe, L.; Fingert, J. H.; Stone, E. M.; Mackey, D. A.: Evidence for genetic heterogeneity within eight glaucoma families, with the GLC1A Gln368STOP mutation being an important phenotypic modifier. *Ophthalmology* 108: 1607–1620, 2001.

- [33451] 7003.Crombie, A. L.; Cullen, J. F.: Hereditary glaucoma: occurrence in five generations of an Edinburgh family. *Brit. J. Ophthalmol.* 48:143–147, 1964.
- [33452] 7004.Metcalf, D.: The granulocyte–macrophage colony–stimulating factors. *Science* 229:16–22, 1985.
- [33453] 7005.Nagata, S.; Tsuchiya, M.; Asano, S.; Kaziro, Y.; Yamazaki, T.; Yamamoto, O.; Hirata, Y.; Kubota, N.; Oheda, M.; Nomura, H.; Ono, M.: Molecular cloning and expression of cDNA for the human granulocyte colony–stimulating factor. *Nature* 319: 415–418, 1986.
- [33454] 7006.Nagata, S.; Tsuchiya, M.; Asano, S.; Yamamoto, O.; Hirata, Y.; Kubota, N.; Oheda, M.; Nomura, H.; Yamazaki, T.: The chromosomal gene structure and two mRNAs for human granulocyte colony–stimulating factor. *EMBO J.* 5: 575–581, 1986.
- [33455] 7007.Petit, I.; Szyper–Kravitz, M.; Nagler, A.; Lahav, M.; Peled, A.; Habler, L.; Ponomaryov, T.; Taichman, R. S.; Arenzana–Seisdedos, F.; Fujii, N.; Sandbank, J.; Zipori, D.; Lapidot, T.: G–CSF induces stem cell mobilization by decreasing bone marrow SDF–1 and up–regulating CXCR4. *Nature Immunol.* 3: 687–694, 2002.
- [33456] 7008.Simmers, R. N.; Smith, J.; Shannon, M. F.; Wong, G.; Lopez, A.F.; Baker, E.; Sutherland, G. R.; Vadas, M. A.: Lo–

calization of the human G-CSF gene to the region of a breakpoint in the translocation typical of acute promyelocytic leukemia. Hum. Genet. 78: 134–136, 1988.

[33457] 7009. Souza, L. M.; Boone, T. C.; Gahrilove, J.; Lai, P. H.; Zsebo, K. M.; Murdock, D. C.; Chazin, V. R.; Bruszewski, J.; Lee, H.; Chen, K. K.; Barendt, J.; Platzer, E.; Moore, M. A. S.; Mertelsmann, R.; Welte, K.: Recombinant human granulocyte colony-stimulating factor: effects on normal and leukemic myeloid cells. Science 232: 61–65, 1986.

[33458] 7010. Sudo, S.; Yamada, H.; Kikuchi, K.; Sumie, A.; Yamashita, Y.; Tumura, N.; Kawaguchi, I.; Fujimoto, S.; Kato, A.; Yamaguchi, J.: A case of ovarian carcinoma with production of granulocyte colony-stimulating factor. Brit. J. Haemat. 92: 137–139, 1996.

[33459] 7011. Dong, F.; Brynes, R. K.; Tidow, N.; Welte, K.; Lowenberg, B.; Touw, I. P.: Mutations in the gene for the granulocyte colony-stimulating factor receptor in patients with acute myeloid leukemia preceded by severe congenital neutropenia. New Eng. J. Med. 333: 487–493, 1995.

[33460] 7012. Scott, M. P.: Vertebrate homeobox gene nomenclature. Cell 71: 551–553, 1992.

[33461] 7013. Carr, D. W.; Hausken, Z. E.; Fraser, I. D.; Stofko-Hahn, R. E.; Scott, J. D.: Association of the type II cAMP-

dependent protein kinase with a human thyroid RII-anchoring protein: cloning and characterization of the RII-binding domain. *J. Biol. Chem.* 267: 13376–13382, 1992.

[33462] 7014. Carr, D. W.; Stofko-Hahn, R. E.; Fraser, I. D.; Bishop, S. M.; Acott, T. S.; Brennan, R. G.; Scott, J. D.: Interaction of the regulatory subunit (RII) of cAMP-dependent protein kinase with RII-anchoring proteins occurs through an amphipathic helix binding motif. *J. Biol. Chem.* 266: 14188–14192, 1991.

[33463] 7015. Rubino, D.; Driggers, P.; Arbit, D.; Kemp, L.; Miller, B.; Coso, O.; Pagliai, K.; Gray, K.; Gutkind, S.; Segars, J.: Characterization of Brx, a novel Dbl family member that modulates estrogen receptor action. *Oncogene* 16: 2513–2526, 1998.

[33464] 7016. Sterpetti, P.; Hack, A. A.; Bashar, M. P.; Park, B.; Cheng, S.-D.; Knoll, J. H. M.; Urano, T.; Feig, L. A.; Toksoz, D.: Activation of the Lbc Rho exchange factor proto-oncogene by truncation of an extended C terminus that regulates transformation and targeting. *Molec. Cell. Biol.* 19: 1334–1345, 1999.

[33465] 7017. Toksoz, D.; Williams, D. A.: Novel human oncogene lbc detected by transfection with distinct homology regions to signal transduction products. *Oncogene* 9:

621–628, 1994.

- [33466] 7018.Hirai, H.; Tanaka, K.; Yoshie, O.; Ogawa, K.; Kenmotsu, K.; Takamori,Y.; Ichimasa, M.; Sugamura, K.; Nakamura, M.; Takano, S.; Nagata,K.: Prostaglandin D2 selectively induces chemotaxis in T helper type2 cells, eosinophils, and basophils via seven-transmembrane receptorCRTH2. *J. Exp. Med.* 193: 255–261, 2001.
- [33467] 7019.Margolis, R. L.; O'Hearn, E.; Rosenblatt, A.; Willour, V.; Holmes,S. E.; Franz, M. L.; Callahan, C.; Hwang, H. S.; Troncoso, J. C.;Ross, C. A.: A disorder similar to Huntington's disease is associatedwith a novel CAG repeat expansion. *Ann. Neurol.* 50: 373–380, 2001.
- [33468] 7020.Lacy, S. E.; Bonnemann, C. G.; Buzney, E. A.; Kunkel, L. M.: Identificationof FLRT1, FLRT2, and FLRT3: a novel family of transmembrane leucine-richrepeat proteins. *Genomics* 62: 417–426, 1999.
- [33469] 7021.Andre, P.; Biassoni, R.; Colonna, M.; Cosman, D.; Lanier, L. L.;Long, E. O.; Lopez–Botet, M.; Moretta, A.; Moretta, L.; Parham, P.;Trowsdale, J.; Vivier, E.; Wagtmann, N.; Wilson, M. J.: New nomenclaturefor MHC receptors. *Nature Immun.* 2: 661 only, 2001.
- [33470] 7022.Fenster, S. D.; Chung, W. J.; Zhai, R.; Cases–Langhoff, C.; Voss,B.; Garner, A. M.; Kaempfer, U.; Kindler,

S.; Gundelfinger, E. D.; Garner, C. C.: Piccolo, a presynaptic zinc finger protein structurally related to Bassoon. *Neuron* 25: 203–214, 2000.

[33471] 7023. Cowan, C. A.; Henkemeyer, M.: The SH2/SH3 adaptor Grb4 transduces B–ephrin reverse signals. *Nature* 413: 174–179, 2001.

[33472] 7024. Hayashi, T.; Huang, J.; Deeb, S. S.: RINX(VSX1), a novel homeobox gene expressed in the inner nuclear layer of the adult retina. *Genomics* 67:128–139, 2000.

[33473] 7025. Ohtoshi, A.; Justice, M. J.; Behringer, R. R.: Isolation and characterization of Vsx1, a novel mouse CVC paired–like homeobox gene expressed during embryogenesis and in the retina. *Biochem. Biophys. Res. Commun.* 286:133–140, 2001.

[33474] 7026. Semina, E. V.; Mintz–Hittner, H. A.; Murray, J. C.: Isolation and characterization of a novel human paired–like homeodomain–containing transcription factor gene, VSX1, expressed in ocular tissues. *Genomics* 63:289–293, 2000.

[33475] 7027. Jones, J. M.; Morrell, J. C.; Gould, S. J.: Identification and characterization of HAOX1, HAOX2, and HAOX3, three human peroxisomal α –hydroxy acid oxidases. *J. Biol. Chem.* 275: 12590–12597, 2000.

[33476] 7028. Kohler, S. A.; Menotti, E.; Kuhn, L. C.: Molecular

cloning of mouse glycolate oxidase: high evolutionary conservation and presence of an iron-responsive element-like sequence in the mRNA. *J. Biol. Chem.* 274: 2401–2407, 1999.

- [33477] 7029. Williams, E.; Cregeen, D.; Rumsby, G.: Identification and expression of a cDNA for human glycolate oxidase. *Biochim. Biophys. Acta* 1493:246–248, 2000.
- [33478] 7030. Horikoshi, N.; Cong, J.; Kley, N.; Shenk, T.: Isolation of differentially expressed cDNAs from p53-dependent apoptotic cells: activation of the human homologue of the *Drosophila* peroxidase gene. *Biochem. Biophys. Res. Commun.* 261: 864–869, 1999.
- [33479] 7031. Weiler, S. R.; Taylor, S. M.; Deans, R. J.; Kan-Mitchell, J.; Mitchell, M. S.; Trent, J. M.: Assignment of a human melanoma associated gene MG50 (D2S448) to chromosome 2p25.3 by fluorescence in situ hybridization. *Genomics* 22:243–244, 1994.
- [33480] 7032. Deng, H.; Unutmaz, D.; KewalRamani, V. N.; Littman, D. R.: Expression cloning of new receptors used by simian and human immunodeficiency viruses. *Nature* 388: 296–300, 1997.
- [33481] 7033. Liao, F.; Alkhatib, G.; Peden, K. W. C.; Sharma, G.; Berger, E. A.; Farber, J. M.: STRL33, a novel chemokine re-

ceptor-like protein, functions as a fusion cofactor for both macrophage-tropic and T cellline-tropic HIV-1. *J. Exp. Med.* 185: 2015–2023, 1997.

- [33482] 7034. Matloubian, M.; David, A.; Engel, S.; Ryan, J. E.; Cyster, J. G.: A transmembrane CXC chemokine is a ligand for HIV-coreceptor Bonzo. *Nature Immun.* 1: 298–304, 2000.
- [33483] 7035. Betz, R.; Gray, S. G.; Ekstrom, C.; Larsson, C.; Ekstrom, T. J.: Human histone deacetylase 2, HDAC2 (human RPD3), is localized to 6q21 by radiation hybrid mapping. *Genomics* 52: 245–246, 1998.
- [33484] 7036. Inouye, C. J.; Seto, E.: Relief of YY1-induced transcriptional repression by protein-protein interaction with the nuclear phosphoprotein B23. *J. Biol. Chem.* 269: 6506–6510, 1994.
- [33485] 7037. Randhawa, G. S.; Bell, D. W.; Testa, J. R.; Feinberg, A. P.: Identification and mapping of human histone acetylation modifier gene homologues. *Genomics* 51: 262–269, 1998.
- [33486] 7038. Yang, W.-M.; Inouye, C.; Zeng, Y.; Bearss, D.; Seto, E.: Transcriptional repression by YY1 is mediated by interaction with a mammalian homolog of the yeast global regulator RPD3. *Proc. Nat. Acad. Sci.* 93: 12845–12850, 1996.
- [33487] 7039. Yarden, R. I.; Brody, L. C.: BRCA1 interacts with

components of the histone deacetylase complex. *Proc. Nat. Acad. Sci.* 96: 4983–4988, 1999.

- [33488] 7040. Fedele, M.; Benvenuto, G.; Pero, R.; Majello, B.; Battista, S.; Lembo, F.; Vollono, E.; Day, P. M.; Santoro, M.; Lania, L.; Bruni, C. B.; Fusco, A.; Chiariotti, L.: A novel member of the BTB/POZ family, PATZ, associates with the RNF4 RING finger protein and acts as a transcriptional repressor. *J. Biol. Chem.* 275: 7894–7901, 2000.
- [33489] 7041. Kobayashi, A.; Yamagiwa, H.; Hoshino, H.; Muto, A.; Sato, K.; Morita, M.; Hayashi, N.; Yamamoto, M.; Igarashi, K.: A combinatorial code for gene expression generated by transcription factor Bach2 and MAZR (MAZ-related factor) through the BTB/POZ domain. *Molec. Cell. Biol.* 20: 1733–1746, 2000.
- [33490] 7042. McAleer, M. A.; Breen, M. A.; White, N. L.; Matthews, N.: pABC11 (also known as MOAT-C and MRP5), a member of the ABC family of proteins, has anion transporter activity but does not confer multidrug resistance when overexpressed in human embryonic kidney 293 cells. *J. Biol. Chem.* 274: 23541–23548, 1999.
- [33491] 7043. Oguri, T.; Isobe, T.; Suzuki, T.; Nishio, K.; Fujiwara, Y.; Katoh, O.; Yamakido, M.: Increased expression of the MRP5 gene is associated with exposure to platinum drugs

in lung cancer. *Int. J. Cancer* 86:95–100, 2000.

- [33492] 7044. Suzuki, T.; Nishio, K.; Sasaki, H.; Kurokawa, H.; Saito–Ohara, F.; Ikeuchi, T.; Tanabe, S.; Terada, M.; Saijo, N.: cDNA cloning of a short type of multidrug resistance protein homologue, SMRP, from a human lung cancer cell line. *Biochem. Biophys. Res. Commun.* 238:790–794, 1997.
- [33493] 7045. Suzuki, T.; Sasaki, H.; Kuh, H.–J.; Agui, M.; Tatsumi, Y.; Tanabe, S.; Terada, M.; Saijo, N.; Nishio, K.: Detailed structural analysis on both human MRP5 and mouse mrp5 transcripts. *Gene* 242: 167–173, 2000.
- [33494] 7046. Wijnholds, J.; Mol, C. A. A. M.; van Deemter, L.; de Haas, M.; Scheffer, G. L.; Baas, F.; Beijnen, J. H.; Scheper, R. J.; Hatse, S.; De Clercq, E.; Balzarini, J.; Borst, P.: Multidrug–resistance protein 5 is a multispecific organic anion transporter able to transport nucleotide analogs. *Proc. Nat. Acad. Sci.* 97: 7476–7481, 2000.
- [33495] 7047. Bebenek, K.; Tissier, A.; Frank, E. G.; McDonald, J. P.; Prasad, R.; Wilson, S. H.; Woodgate, R.; Kunkel, T. A.: 5–prime–deoxyribosephosphate lyase activity of human DNA polymerase ι in vitro. *Science* 291:2156–2159, 2001.
- [33496] 7048. Johnson, R. E.; Washington, M. T.; Haracska, L.;

Prakash, S.; Prakash, L.: Eukaryotic polymerases ϵ and ζ act sequentially to bypass DNA lesions. *Nature* 406: 1015–1019, 2000.

[33497] 7049. McDonald, J. P.; Rasic-Otrin, V.; Epstein, J. A.; Broughton, B.C.; Wang, X.; Lehmann, A. R.; Wolgemuth, D. J.; Woodgate, R.: Novel human and mouse homologs of *Saccharomyces cerevisiae* DNA polymerase ϵ . *Genomics* 60: 20–30, 1999.

[33498] 7050. Dermaut, B.; Theuns, J.; Sleegers, K.; Hasegawa, H.; Van den Broeck, M.; Vennekens, K.; Corsmit, E.; St. George-Hyslop, P.; Cruts, M.; van Duijn, C. M.; Van Broeckhoven, C.: The gene encoding nicastrin, a major gamma-secretase component, modifies risk for familial early-onset Alzheimer disease in a Dutch population-based sample. *Am. J. Hum. Genet.* 70: 1568–1574, 2002.

[33499] 7051. Feldman, R. G.; Chandler, K. A.; Levy, L. L.; Glaser, G. H.: Familial Alzheimer's disease. *Neurology* 13: 811–824, 1963.

[33500] 7052. Foncin, J.-F.; Salmon, D.; Supino-Viterbo, V.; Feldman, R. G.; Macchi, G.; Mariotti, P.; Scoppetta, C.; Caruso, G.; Bruni, A. C.: Dementia presenile d'Alzheimer transmise dans une famille étendue. *Rev. Neurol. (Paris)* 141: 194–202, 1985.

- [33501] 7053.Hiltunen, M.; Mannermaa, A.; Thompson, D.; Easton, D.; Pirskanen,M.; Helisalmi, S.; Koivisto, A. M.; Lehtovirta, M.; Ryyananen, M.;Soininen, H.: Genome-wide linkage disequilibrium mapping of late-onsetAlzheimer's disease in Finland. *Neurology* 57: 1663–1668, 2001.
- [33502] 7054.Kehoe, P.; Wavrant–De Vrieze, F.; Crook, R.; Wu, W. S.; Holmans,P.; Fenton, I.; Spurlock, G.; Norton, N.; Williams, H.; Williams,N.; Lovestone, S.; Perez–Tur, J.; Hutton, J.; and 10 others: Afull genome scan for late onset Alzheimer disease. *Hum. Molec. Genet.* 8:237–245, 1999.
- [33503] 7055.Kopan, R.; Goate, A.: Aph–2/nicastrin: an essential componentof gamma–secretase and regulator of Notch signaling and presenilinlocalization. *Neuron* 33: 321–324, 2002.
- [33504] 7056.Yu, G.; Nishimura, M.; Arawaka, S.; Levitan, D.; Zhang, L.; Tandon,A.; Song, Y.–Q.; Rogaeva, E.; Chen, F.; Kawarai, T.; Supala, A.; Levesque,L.; and 18 others: Nicas–trin modulates presenilin–mediated notch/glp–1signal transduction and beta–APP processing. *Nature* 407: 48–54,2000.
- [33505] 7057.Zubenko, G. S.; Hughes, H. B.; Stiffler, J. S.; Hurtt, M. R.; Kaplan,B. B.: A genome survey for novel Alzheimer disease risk loci: resultsat 10–cM resolution. *Genomics* 50:

121–128, 1998.

- [33506] 7058. Tateishi, S.; Sakuraba, Y.; Masuyama, S.; Inoue, H.; Yamaizumi, M.: Dysfunction of human Rad18 results in defective postreplication repair and hypersensitivity to multiple mutagens. *Proc. Nat. Acad. Sci.* 97: 7927–7932, 2000.
- [33507] 7059. Arakawa, H.; Hauschild, J.; Buerstedde, J.-M.: Requirement of the activation-induced deaminase (AID) gene for immunoglobulin gene conversion. *Science* 295: 1301–1306, 2002.
- [33508] 7060. Fagarasan, S.; Kinoshita, K.; Muramatsu, M.; Ikuta, K.; Honjo, T.: In situ class switching and differentiation to IgA producing cells in the gut lamina propria. *Nature* 413: 639–643, 2001.
- [33509] 7061. Muramatsu, M.; Kinoshita, K.; Fagarasan, S.; Yamada, S.; Shinkai, Y.; Honjo, T.: Class switch recombination and hypermutation require activation-induced cytidine deaminase (AID), a potential RNA editing enzyme. *Cell* 102: 553–563, 2000.
- [33510] 7062. Muramatsu, M.; Sankaranand, V. S.; Anant, S.; Sugai, M.; Kinoshita, K.; Davidson, N. O.; Honjo, T.: Specific expression of activation-induced cytidine deaminase (AID), a novel member of the RNA-editing deaminase family in

germinal center B cells. *J. Biol. Chem.* 274:
18470–18476,1999.

- [33511] 7063.Rowley, D. R.; Tindall, D. J.: Responses of NBT-II bladder carcinomacells to conditioned medium from normal fetal urogenital sinus. *CancerRes.* 47: 2955–2960, 1987.
- [33512] 7064.Kasaian, M. T.; Whitters, M. J.; Carter, L. L.; Lowe, L. D.; Jussif,J. M.; Deng, B.; Johnson, K. A.; Witek, J. S.; Senices, M.; Konz,R. F.; Wurster, A. L.; Donaldson, D. D.; Collins, M.; Young, D. A.;Grusby, M. J.: IL-21 limits NK cell responses and promotes antigen-specificT cell activation: a mediator of the transition from innate to adaptiveimmunity. *Immunity* 16: 559–569, 2002.
- [33513] 7065.Pashmforoush, M.; Pomies, P.; Peterson, K. L.; Kubalak, S.; Ross,J., Jr.; Hefti, A.; Aebi, U.; Beckerle, M. C.; Chien, K. R.: Adultmice deficient in actinin-associated LIM-domain protein reveal a developmentalpathway for right ventricular cardiomyopathy. *Nature Med.* 7: 591–597,2001.
- [33514] 7066.Zheng, P.; Eastman, J.; Vande Pol, S.; Pimplikar, S. W.: PAT1,a microtubule-interacting protein, recognizes the basolateral sortingsignal of amyloid precursor protein. *Proc. Nat. Acad. Sci.* 95: 14745–14750,1998.

- [33515] 7067. Aoyama, T.; Yamano, S.; Waxman, D. J.; Lapenson, D. P.; Meyer, U. A.; Fischer, V.; Tyndale, R.; Inaba, T.; Kalow, W.; Gelboin, H. V.; Gonzalez, F. J.: Cytochrome P-450 hPCN3, a novel cytochrome P-450 IIIA product that is differentially expressed in adult human liver: cDNA and deduced amino acid sequence and distinct specificities of cDNA-expressed hPCN1 and hPCN3 for the metabolism of steroid hormones and cyclosporine. *J. Biol. Chem.* 264: 10388–10395, 1989.
- [33516] 7068. Jounaidi, Y.; Guzelian, P. S.; Maurel, P.; Vilarem, M.-J.: Sequence of the 5'-prime-flanking region of CYP3A5: comparative analysis with CYP3A4 and CYP3A7. *Biochem. Biophys. Res. Commun.* 205: 1741–1747, 1994.
- [33517] 7069. Kuehl, P.; Zhang, J.; Lin, Y.; Lamba, J.; Assem, M.; Schuetz, J.; Watkins, P. B.; Daly, A.; Wrighton, S. A.; Hall, S. D.; Maurel, P.; Relling, M.; Brimer, C.; Yasuda, K.; Venkataramanan, R.; Strom, S.; Thummel, K.; Boguski, M. S.; Schuetz, E.: Sequence diversity in CYP3A promoters and characterization of the genetic basis of polymorphic-CYP3A5 expression. *Nature Genet.* 27: 383–391, 2001.
- [33518] 7070. Paulussen, A.; Lavrijsen, K.; Bohets, H.; Hendrickx, J.; Verhasselt, P.; Luyten, W.; Konings, F.; Armstrong, M.: Two linked mutations in transcriptional regulatory ele-

ments of the CYP3A5 gene constitute the major genetic determinant of polymorphic activity in humans. *Pharmacogenetics* 10:415–424, 2000.

- [33519] 7071. Yamakoshi, Y.; Kishimoto, T.; Sugimura, K.; Kawashima, H.: Human prostate CYP3A5: identification of a unique 5-prime-untranslated sequence and characterization of purified recombinant protein. *Biochem. Biophys. Res. Commun.* 260: 676–681, 1999.
- [33520] 7072. De Valck, D.; Jin, D. Y.; Heyninck, K.; Van de Craen, M.; Contreras, R.; Fiers, W.; Jeang, K. T.; Beyaert, R.: The zinc finger protein A20 interacts with a novel anti-apoptotic protein which is cleaved by specific caspases. *Oncogene* 18: 4182–4190, 1999.
- [33521] 7073. Gachon, F.; Peleraux, A.; Thebault, S.; Dick, J.; Lemasson, I.; Devaux, C.; Mesnard, J.-M.: CREB-2, a cellular CRE-dependent transcription repressor, functions in association with tax as an activator of the human T-cell leukemia virus type 1 promoter. *J. Virol.* 72: 8332–8337, 1998.
- [33522] 7074. Ling, L.; Goeddel, D. V.: T6BP, a TRAF6-interacting protein involved in IL-1 signaling. *Proc. Nat. Acad. Sci.* 97: 9567–9572, 2000.
- [33523] 7075. Ozaki, K.; Kikly, K.; Michalovich, D.; Young, P. R.;

Leonard, W.J.: Cloning of a type I cytokine receptor most related to the IL-2 receptor beta chain. *Proc. Nat. Acad. Sci.* 97: 11439–11444, 2000.

- [33524] 7076. Parrish–Novak, J.; Dillon, S. R.; Nelson, A.; Hammond, A.; Sprecher, C.; Gross, J. A.; Johnston, J.; Madden, K.; Xu, W.; West, J.; Schrader, S.; Burkhead, S.; and 26 others: Interleukin 21 and its receptor are involved in NK cell expansion and regulation of lymphocyte function. *Nature* 408:57–63, 2000.
- [33525] 7077. Boyartchuk, V. L.; Ashby, M. N.; Rine, J.: Modulation of Ras and Raf function by carboxyl-terminal proteolysis. *Science* 275:1796–1800, 1997.
- [33526] 7078. Freije, J. M. P.; Blay, P.; Pendas, A. M.; Cadinanos, J.; Crespo, P.; Lopez–Otin, C.: Identification and chromosomal location of two human genes encoding enzymes potentially involved in proteolytic maturation of farnesylated proteins. *Genomics* 58: 270–280, 1999.
- [33527] 7079. Otto, J. C.; Kim, E.; Young, S. G.; Casey, P. J.: Cloning and characterization of a mammalian prenyl protein-specific protease. *J. Biol. Chem.* 274: 8379–8382, 1999.
- [33528] 7080. Reither, A.; Hehlmann, R.; Goldman, J. M.; Cross, N. C.: The 8p11 myeloproliferative syndrome. *Med. Klin.* 94: 207–210, 1999.

- [33529] 7081. Bartholin, L.; Maguer-Satta, V.; Hayette, S.; Martel, S.; Gadoux, M.; Bertrand, S.; Corbo, L.; Lamadon, C.; Morera, A.-M.; Magaud, J.-P.; Rimokh, R.: FLRG, an activin-binding protein, is a new target of TGF-beta transcription activation through Smad proteins. *Oncogene* 20:5409-5419, 2001.
- [33530] 7082. Rimokh, R.; Berger, F.; Delsol, G.; Charrin, C.; Bertheas, M. F.; Ffrench, M.; Garoscio, M.; Felman, P.; Coiffier, C.; Bryon, P. A.: Rearrangement and overexpression of the BCL-1/PRAD-1 gene in intermediatelymphocytic lymphomas and in t(11q13)-bearing leukemias. *Blood* 81:3063-3067, 1993.
- [33531] 7083. Bahou, W. F.; Campbell, A. D.; Wicha, M. S.: cDNA cloning and molecular characterization of MSE55, a novel human serum constituent protein that displays bone marrow stromal/endothelial cell-specific expression. *J. Biol. Chem.* 267: 13986-13992, 1992.
- [33532] 7084. Burbelo, P. D.; Drechsel, D.; Hall, A.: A conserved binding motif defines numerous candidate target proteins for both Cdc42 and Rac GTPases. *J. Biol. Chem.* 270: 29071-29074, 1995.
- [33533] 7085. Bak, M.; Hansen, C.; Henriksen, K. F.; Tommerup, N.: The human hedgehog-interacting protein gene: structure

and chromosome mapping to 4q31.21–q31.3. *Cytogenet. Cell Genet.* 92: 300–303, 2001.

- [33534] 7086. Chuang, P.-T.; McMahon, A. P.: Vertebrate hedgehog signalling modulated by induction of a hedgehog-binding protein. *Nature* 397:617–621, 1999.
- [33535] 7087. Bacher, N.; Zisman, Y.; Berent, E.; Livneh, E.: Isolation and characterization of PKC-L, a new member of the protein kinase C-related gene family specifically expressed in lung, skin, and heart. *Molec. Cell. Biol.* 11: 126–133, 1991.
- [33536] 7088. Quan, T.; Fisher, G. J.: Cloning and characterization of the human protein kinase C- ϵ promoter. *J. Biol. Chem.* 274: 28566–28574, 1999.
- [33537] 7089. Gong, T.-W. L.; Burmeister, M.; Lomax, M. I.: The novel gene D4Mit1 maps to mouse chromosome 4 and human chromosome 1p36. *Mammalian Genome* 7:790–791, 1996.
- [33538] 7090. Nangaku, M.; Sato-Yoshitake, R.; Okada, Y.; Noda, Y.; Takemura, R.; Yamazaki, H.; Hirokawa, N.: KIF1B, a novel microtubule plus end-directed monomeric motor protein for transport of mitochondria. *Cell* 79:1209–1220, 1994.
- [33539] 7091. Saito, M.; Hayashi, Y.; Suzuki, T.; Tanaka, H.;

Hozumi, I.; Tsuji, S.: Linkage mapping of the gene for Charcot-Marie-Tooth disease type 2 to chromosome 1p (CMT2A) and the clinical features of CMT2A. *Neurology* 49:1630-1635, 1997.

[33540] 7092. Yang, H. W.; Chen, Y. Z.; Takita, J.; Soeda, E.; Piao, H. Y.; Hayashi, Y.: Genomic structure and mutational analysis of the human KIF1B gene which is homozygously deleted in neuroblastoma at chromosome 1p36.2. *Oncogene* 20: 5075-5083, 2001.

[33541] 7093. Suzuki, Y.; Demoliere, C.; Kitamura, D.; Takeshita, H.; Deuschle, U.; Watanabe, T.: HAX-1, a novel intracellular protein, localized on mitochondria, directly associates with HS1, a substrate of Src family tyrosine kinases. *J. Immun.* 158: 2736-2744, 1997.

[33542] 7094. Dell'Angelica, E. C.; Puertollano, R.; Mullins, C.; Aguilar, R. C.; Vargas, J. D.; Hartnell, L. M.; Bonifacino, J. S.: GGAs: a family of ADP ribosylation factor-binding proteins related to adaptors and associated with the Golgi complex. *J. Cell Biol.* 149: 81-93, 2000.

[33543] 7095. Hirst, J.; Lui, W. W. Y.; Bright, N. A.; Totty, N.; Seaman, M. N. J.; Robinson, M. S.: A family of proteins with gamma-adaptin and VHS domains that facilitate trafficking between the trans-Golgi network and the vacuole/lyso-

some. *J. Cell Biol.* 149: 67–69, 2000.

- [33544] 7096. Ma, Z.; Morris, S. W.; Valentine, V.; Li, M.; Herbrick, J.-A.; Cui, X.; Bouman, D.; Li, Y.; Mehta, P. K.; Nizetic, D.; Kaneko, Y.; Chan, G. C. F.; Chan, L. C.; Squire, J.; Scherer, S. W.; Hitzler, J. K.: Fusion of two novel genes, RBM15 and MKL1, in the t(1;22)(p13;q13) of acute megakaryoblastic leukemia. *Nature Genet.* 28: 220–221, 2001.
- [33545] 7097. Mercher, T.; Coniat, M. B.-L.; Monni, R.; Mauchauffe, M.; Khac, F. N.; Gressin, L.; Mugneret, F.; Leblanc, T.; Dastugue, N.; Berger, R.; Bernard, O. A.: Involvement of a human gene related to the *Drosophila* *spen* gene in the recurrent t(1;22) translocation of acute megakaryocytic leukemia. *Proc. Nat. Acad. Sci.* 98: 5776–5779, 2001.
- [33546] 7098. Wang, D.-Z.; Chang, P. S.; Wang, Z.; Sutherland, L.; Richardson, J. A.; Small, E.; Krieg, P. A.; Olson, E. N.: Activation of cardiac gene expression by myocardin, a transcriptional cofactor for serum response factor. *Cell* 105: 851–862, 2001.
- [33547] 7099. Blumberg, H.; Conklin, D.; Xu, W.; Grossmann, A.; Brender, T.; Carollo, S.; Eagan, M.; Foster, D.; Haldeman, B. A.; Hammond, A.; Haugen, H.; Jelinek, L.; and 14 others: Interleukin 20: discovery, receptor identification, and role in epidermal function. *Cell* 104: 9–19, 2001.

- [33548] 7100.Huang, E. Y.; Madireddi, M. T.; Gopalkrishnan, R. V.; Leszczyniecka,M.; Su, Z.; Lebedeva, I. V.; Kang, D.; Jiang, H.; Lin, J. J.; Alexandre,D.; Chen, Y.; Vozhilla, N.: and 9 others: Genomic structure, chromosomallocalization and expression profile of a novel melanoma differentiationas–sociated (mda–7) gene with cancer specific growth sup–pressing andapoptosis inducing properties. *Oncogene* 20: 7051–7063, 2001.
- [33549] 7101.Jiang, H.; Lin, J. J.; Su, Z.–Z.; Goldstein, N. I.; Fisher, P.B.: Subtraction hybridization identifies a novel melanoma differentiationassociated gene, mda–7, modu–lated during human melanoma differentiation,growth and progression. *Oncogene* 11: 2477–2486, 1995.
- [33550] 7102.Jiang, H.; Su, Z.–Z.; Lin, J. J.; Goldstein, N. I.; Young, C. S.H.; Fisher, P. B.: The melanoma differentiation associ–ated gene mda–7suppresses cancer cell growth. *Proc. Nat. Acad. Sci.* 93: 9160–9165,1996.
- [33551] 7103.Su, Z.; Madireddi, M. T.; Lin, J. J.; Young, C. S. H.; Ki–tada,S.; Reed, J. C.; Goldstein, N. I.; Fisher, P. B.: The can–cer growthsuppressor gene mda–7 selectively induces apoptosis in human breastcancer cells and inhibits tumor growth in nude mice. *Proc. Nat. Acad.Sci.* 95: 14400–14405, 1998.

- [33552] 7104.Hsieh, S.-L.; Campbell, R. D.: Evidence that gene G7a in the humanmajor histocompatibility complex encodes valyl-tRNA synthetase. *Biochem.J.* 278: 809–816, 1991. Note: Erratum: *Biochem. J.* 281: 879 only,1992.
- [33553] 7105.Hsieh, S.-L.; Kendall, E.; Milner, C.; Cross, S.; Cheng, J.; Khanna,A.; Olaversen, M.; Campbell, R. D.: Cloning of the human valyl-tRNA synthetase gene and its location in the major histocompatibility complex on chromosome 6. (Abstract) *Cytogenet. Cell Genet.* 58: 1912 only,1991.
- [33554] 7106.Mansour, S. J.; Skaug, J.; Zhao, X.-H.; Giordano, J.; Scherer,S. W.; Melancon, P.: p200 ARF-GEP1: a Golgi-localized guanine nucleotideexchange protein whose Sec7 domain is targeted by the drug brefeldinA. *Proc. Nat. Acad. Sci.* 96: 7968–7973, 1999.
- [33555] 7107.Morinaga, N.; Moss, J.; Vaughan, M.: Cloning and expression of a cDNA encoding a bovine brain brefeldin A-sensitive guanine nucleotide-exchange protein for ADP-ribosylation factor. *Proc. Nat. Acad. Sci.* 94: 12926–12931,1997.
- [33556] 7108.Morinaga, N.; Tsai, S.-C.; Moss, J.; Vaughan, M.: Isolation of a brefeldin A-inhibited guanine nucleotide-exchange protein for ADPribosylation factor (ARF) 1 and ARF3 that contains a Sec7-like domain. *Proc.Nat. Acad.*

Sci. 93: 12856–12860, 1996.

- [33557] 7109. Togawa, A.; Morinaga, N.; Ogasawara, M.; Moss, J.; Vaughan, M.: Purification and cloning of a brefeldin A–inhibited guanine nucleotide–exchange protein for ADP–ribosylation factors. *J. Biol. Chem.* 274: 12308–12315, 1999.
- [33558] 7110. Mitsui, S.; Tsuruoka, N.; Yamashiro, K.; Nakazato, H.; Yamaguchi, N.: A novel form of human neuropsin, a brain–related serine protease, is generated by alternative splicing and is expressed preferentially in human adult brain. *Europ. J. Biochem.* 260: 627–634, 1999.
- [33559] 7111. Underwood, L. J.; Tanimoto, H.; Wang, Y.; Shigemasa, K.; Parmley, T. H.; O'Brien, T. J.: Cloning of tumor–associated differentially expressed gene–14, a novel serine protease overexpressed by ovarian carcinoma. *Cancer Res.* 59: 4435–4439, 1999.
- [33560] 7112. Yoshida, S.; Taniguchi, M.; Hirata, A.; Shiosaka, S.: Sequence analysis and expression of human neuropsin cDNA and gene. *Gene* 213: 9–16, 1998.
- [33561] 7113. Markert, M. L.; Hershfield, M. S.; Schiff, R. I.; Buckley, R. H.: Adenosine deaminase and purine nucleoside phosphorylase deficiencies: evaluation of therapeutic interventions in eight patients. *J. Clin. Immun.* 7: 389–399,

1987.

- [33562] 7114.Ewen, M. E.; Xing, Y.; Lawrence, J. B.; Livingston, D. M.: Molecularcloning, chromosomal mapping, and expression of the cDNA for p107,a retinoblastoma gene product-related protein. Cell 66: 1155–1164,1991.
- [33563] 7115.Huppi, K.; Siwarski, D.; Mock, B. A.; Dosik, J.; Hamel, P. A.:Molecular cloning, chromosomal mapping, and expression of the mousep107 gene. Mammalian Genome 7: 353–355, 1996.
- [33564] 7116.Kim, K. K.; Soonpaa, M. H.; Wang, H.; Field, L. J.: Developmentalexpression of p107 mRNA and evidence for alternative splicing of thep107 (RBL1) gene product. Genomics 28: 520–529, 1995.
- [33565] 7117.LeCouter, J. E.; Kablar, B.; Hardy, W. R.; Ying, C.; Megeney, L.A.; May, L. L.; Rudnicki, M. A.: Strain-dependent myeloid hyperplasia,growth deficiency, and accelerated cell cycle in mice lacking theRb-related p107 gene. Molec. Cell. Biol. 18: 7455–7465, 1998.
- [33566] 7118.Morohashi, K.; Sogawa, K.; Omura, T.; Fujii-Kuriyama, Y.: Genestructure of human cytochrome P-450(SCC), cholesterol desmolase. J.Biochem. 101: 879–887, 1987.
- [33567] 7119.Youngblood, G. L.; Nesbitt, M. N.; Payne, A. H.: The

structural genes encoding P450SCC and P450AROM are closely linked on mouse chromosome 9. *Endocrinology* 125: 2784–2786, 1989.

- [33568] 7120. Barrard, B. A.; Lottspeich, F.; Braun, A.; Barde, Y. A.; Mallet, J.: cDNA cloning and complete sequence of porcine choline acetyltransferase: in vitro translation of the corresponding RNA yields an active protein. *Proc. Nat. Acad. Sci.* 84: 9280–9284, 1987.
- [33569] 7121. Chireux, M. A.; Le Van Thai, A.; Weber, M. J.: Human choline acetyltransferase gene: localization of alternative first exons. *J. Neurosci. Res.* 40: 427–438, 1995.
- [33570] 7122. Cohen-Haguenauer, O.; Brice, A.; Berrard, S.; Van Cong, N.; Mallet, J.; Frezal, J.: Localization of the choline acetyltransferase (CHAT) gene to human chromosome 10. *Genomics* 6: 374–378, 1990.
- [33571] 7123. Misawa, H.; Ishii, K.; Deguchi, T.: Gene expression of mouse choline acetyltransferase: alternative splicing and identification of a highly active promoter region. *J. Biol. Chem.* 267: 20392–20399, 1992.
- [33572] 7124. Ohno, K.; Tsujino, A.; Brengman, J. M.; Harper, C. M.; Bajzer, Z.; Udd, B.; Beyring, R.; Robb, S.; Kirkham, F. J.; Engel, A. G.: Choline acetyltransferase mutations cause myasthenic syndrome associated with episodic apnea in

humans. Proc. Nat. Acad. Sci. 98: 2017–2022,2001.

[33573] 7125.Strauss, W. L.; Kemper, R. R.; Jayakar, P.; Kong, C. F.; Hersh,L. B.; Hilt, D. C.; Rabin, M.: Human choline acetyltransferase genemaps to region 10q11–q22.2 by in situ hybridization. Genomics 9:396–398, 1991.

[33574] 7126.Toussaint, J. L.; Geoffroy, V.; Schmitt, M.; Werner, A.; Garnier,J. M.; Simoni, P.; Kempf, J.: Human choline acetyltransferase (CHAT):partial gene sequence and potential control regions. Genomics 12:412–416, 1992.

[33575] 7127.Viegas–Pequignot, E.; Berrard, S.; Brice, A.; Apiou, F.; Mallet,J.: Localization of a 900–bp–long fragment of the human choline acetyltransferasegene to 10q11.2 by non-radioactive in situ hybridization. Genomics 9:210–212, 1991.

[33576] 7128.Bonner, T. I.; Modi, W. S.; Seuanez, H. N.; O'Brien, S. J.: Chromosomalmapping of five human genes encoding muscarinic acetylcholine receptors.(Abstract) Cytogenet. Cell Genet. 58: 1850–1851, 1991.

[33577] 7129.Anand, R.; Lindstrom, J.: Chromosomal localization of seven neuronalnicotinic acetylcholine receptor subunit genes in humans. Genomics 13:962–967, 1992.

[33578] 7130.Bessis, A.; Simon–Chazottes, D.; Devillers–Thiery, A.; Guenet,J.–L.; Changeux, J.–P.: Chromosomal localiza–

tion of the mouse genes coding for alpha-2, alpha-3, alpha-4 and beta-2 subunits of neuronal nicotinic acetylcholine receptor. FEBS Lett. 264: 48-52, 1990.

- [33579] 7131. Boulter, J.; O'Shea-Greenfield, A.; Duvoisin, R. M.; Connolly, J. G.; Wada, E.; Jensen, A.; Gardner, P. D.; Ballivet, M.; Deneris, E. S.; McKinnon, D.; Heinemann, S.; Patrick, J.: Alpha3, alpha5 and beta4: three members of the rat neuronal nicotinic acetylcholine receptor-related gene family form a gene cluster. J. Biol. Chem. 265: 4472-4482, 1990.
- [33580] 7132. Duga, S.; Solda, G.; Asselta, R.; Bonati, M. T.; Dalpra, L.; Malcovati, M.; Tenchini, M. L.: Characterization of the genomic structure of the human neuronal nicotinic acetylcholine receptor CHRNA5/A3/B4 gene cluster and identification of novel intragenic polymorphisms. J. Hum. Genet. 46: 640-648, 2001.
- [33581] 7133. Elliott, K. J.; Ellis, S. B.; Berckhan, K. J.; Urrutia, A.; Chavez-Noriega, L. E.; Johnson, E. C.; Velicelebi, G.; Harpold, M. M.: Comparative structure of human neuronal alpha-2-alpha-7 and beta-2-beta-4 nicotinic acetylcholine receptor subunits and functional expression of the alpha-2, alpha-3, alpha-4, alpha-7, beta-2, and beta-4 subunits. J. Molec. Neurosci. 7: 217-228, 1996.
- [33582] 7134. Armstrong, E.; Partanen, J.; Cannizzaro, L.; Huebner,

K.; Alitalo, K.: Localization of the fibroblast growth factor receptor-4 gene to chromosome region 5q33-qter. *Genes Chromosomes Cancer* 4: 94–98, 1992.

[33583] 7135. Bange, J.; Prechtel, D.; Cheburkin, Y.; Specht, K.; Harbeck, N.; Schmitt, M.; Knyazeva, T.; Muller, S.; Gartner, S.; Sures, I.; Wang, H.; Imyanitov, E.; Haring, H.-U.; Knayzev, P.; Iacobelli, S.; Hofler, H.; Ullrich, A.: Cancer progression and tumor cell motility are associated with the FGFR4 Arg388 allele. *Cancer Res.* 62: 840–847, 2002.

[33584] 7136. Holtrich, U.; Brauninger, A.; Strebhardt, K.; Rubsamens-Waigmann, H.: Two additional protein-tyrosine kinases expressed in human lung: fourth member of the fibroblast growth factor receptor family and an intracellular protein-tyrosine kinase. *Proc. Nat. Acad. Sci.* 88: 10411–10415, 1991.

[33585] 7137. Kostrzewa, M.; Muller, U.: Genomic structure and complete sequence of the human FGFR4 gene. *Mammalian Genome* 9: 131–135, 1998.

[33586] 7138. Partanen, J.; Makela, T. P.; Eerola, E.; Korhonen, J.; Hirvonen, H.; Claesson-Welsh, L.; Alitalo, K.: FGFR-4, a novel acidic fibroblast growth factor receptor with a distinct expression pattern. *EMBO J.* 10: 1347–1354, 1991.

[33587] 7139. Scott, A. F.: Personal Communication. Baltimore, Md.

10/12/1999.

- [33588] 7140.Vainikka, S.; Partanen, J.; Bellosta, P.; Coulier, F.; Basilico,C.; Jaye, M.; Alitalo, K.: Fibroblast growth factor receptor-4 shows novel features in genomic structure, ligand binding and signal transduction. *EMBOJ.* 11: 4273-4280, 1992.
- [33589] 7141.Warrington, J. A.; Bailey, S. K.; Armstrong, E.; Aprelikova, O.;Alitalo, K.; Dolganov, G. M.; Wilcox, A. S.; Sikela, J. M.; Wolfe,S. F.; Lovett, M.; Wasmuth, J. J.: A radiation hybrid map of 18 growthfactor, growth factor receptor, hormone receptor, or neurotransmitterreceptor genes on the distal region of the long arm of chromosome5. *Genomics* 13: 803-808, 1992.
- [33590] 7142.Becker, P. E.; Von Knorre, G. V.: Myositis ossificans progressiva. *Ergeb.Inn. Med. Kinderheilk.* 27: 1-31, 1968.
- [33591] 7143.Beratis, N. G.; Kaffe, S.; Aron, A. M.; Hirschhorn, K.: Alkalinephosphatase activity in cultured skin fibroblasts from fibrodysplasiaossificans progressiva. *J. Med. Genet.* 13: 307-309, 1976.
- [33592] 7144.Cohen, R. B.; Hahn, G. V.; Tabas, J. A.; Peeper, J.; Levitz, C.L.; Sando, A.; Sando, N.; Zasloff, M.; Kaplan, F. S.: The naturalhistory of heterotopic ossification in patients who have fibrodysplasiaossificans progressiva: a study of

forty-four patients. J. Bone Joint Surg. 75A: 215–219, 1993.

[33593] 7145. Connor, J. M.: Soft Tissue Ossification. New York: Springer-Verlag(pub.) 1983.

[33594] 7146. Connor, J. M.; Evans, D. A. P.: Genetic aspects of fibrodysplasia ossificans progressiva. J. Med. Genet. 19: 35–39, 1982.

[33595] 7147. Connor, J. M.; Evans, D. A. P.: Fibrodysplasia ossificans progressiva: the clinical features and natural history of 34 patients. J. Bone Joint Surg. 64: 76–83, 1982.

[33596] 7148. Connor, J. M.; Skirton, H.; Lunt, P. W.: A three generation family with fibrodysplasia ossificans progressiva. J. Med. Genet. 30: 687–689, 1993.

[33597] 7149. Daltroff, G.; Lutz, P.; Bellocq, P.; Christmann, D.; Flamant, F.; Lenoir, G.; Clavert, J. M.; Levy, J. M.: Fibromatose et fibrodysplasie ossifiante progressive: une erreur diagnostique evitable. Arch. Fr. Pediat. 49: 441–444, 1992.

[33598] 7150. Feldman, G.; Li, M.; Martin, S.; Urbanek, M.; Urtizberea, J. A.; Fardeau, M.; LeMerrer, M.; Connor, J. M.; Triffitt, J.; Smith, R.; Muenke, M.; Kaplan, F. S.; Shore, E. M.: Fibrodysplasia ossificans progressiva, a heritable disorder of severe heterotopic ossification, maps to human chro-

mosome 4q27–31. *Am. J. Hum. Genet.* 66: 128–135,2000.

[33599] 7151.Janoff, H. B.; Muenke, M.; Johnson, L. O.; Rosenberg, A.; Shore,E. M.; Okereke, E.; Zasloff, M.; Kaplan, F. S.: Fibrodysplasia ossificansprogressiva in two half-sisters: evidence for maternal mosaicism. *Am.J. Med. Genet.* 61: 320–324, 1996.

[33600] 7152.Digicaylioglu, M.; Lipton, S. A.: Erythropoietin-mediated neuroprotectioninvolves cross-talk between Jak2 and NF-kappa-B signalling cascades. *Nature* 412:641–647, 2001.

[33601] 7153.Clancy, D. J.; Gems, D.; Harshman, L. G.; Oldham, S.; Stocker,H.; Hafen, E.; Leivers, S. J.; Partridge, L.: Extension of life-spanby loss of CHICO, a Drosophila insulin receptor substrate protein. *Science* 292:104–106, 2001.

[33602] 7154.Chai, K. X.; Chen, L.-M.; Chao, J.; Chao, L.: Kallistatin: a novelhuman serine proteinase inhibitor: molecular cloning, tissue distribution,and expression in *Escherichia coli*. *J. Biol. Chem.* 268: 24498–24505,1993.

[33603] 7155.Chai, K. X.; Ward, D. C.; Chao, J.; Chao, L.: Molecular cloning,sequence analysis, and chromosomal localization of the human proteaseinhibitor 4 (kallistatin) gene (PI4). *Genomics* 23: 370–378, 1994.

[33604] 7156.Zhou, G. X.; Chao, L.; Chao, J.: Kallistatin: a novel

human tissue kallikrein inhibitor; purification, characterization, and reactive center sequence. *J. Biol. Chem.* 267: 25873–25880, 1992.

[33605] 7157. Christmanson, L.; Rorsman, F.; Stenman, G.; Westermark, P.; Betsholtz, C.: The human islet amyloid polypeptide (IAPP) gene: organization, chromosomal localization and functional identification of a promoter region. *FEBS Lett.* 267: 160–166, 1990.

[33606] 7158. Cockburn, D. C.; Holt, S. M.; Roberts, A. N.; Cooper, G. J. S.; Reid, K. B. M.; Boyd, Y.: Localization of the amylin locus to chromosome 12. (Abstract) *Cytogenet. Cell Genet.* 51: 977, 1989.

[33607] 7159. Cooper, G. J.; Willis, A. C.; Clark, A.; Turner, R. C.; Sim, R. B.; Reid, K. B.: Purification and characterization of a peptide from amyloid-rich pancreases of type 2 diabetic patients. *Proc. Nat. Acad. Sci.* 84: 8628–8632, 1987.

[33608] 7160. Cooper, G. J. S.; Leighton, B.; Dimitriadis, G. D.; Parry-Billings, M.; Kowalchuk, J. M.; Howland, K.; Rothbard, J. B.; Willis, A. C.; Reid, K. B. M.: Amylin found in amyloid deposits in human type 2 diabetes mellitus may be a hormone that regulates glycogen metabolism in skeletal muscle. *Proc. Nat. Acad. Sci.* 85: 7763–7766, 1988.

[33609] 7161. Fan, Y.-S.; Eddy, R. L.; Byers, M. G.; Haley, L. L.;

Henry, W.M.; Sanke, T.; Steiner, D. F.; Shows, T. B.; Bell, G. I.: Localization of the human islet amyloid polypeptide gene (IAP) to chromosome 12p12.3.(Abstract) Cytogenet. Cell Genet. 51: 997, 1989.

- [33610] 7162. Harvey, A. M.: Medical students on the march: Brown, MacCallum and Opie. Johns Hopkins Med. J. 134: 330–345, 1974.
- [33611] 7163. Hoovers, J. M. N.; Redeker, E.; Speleman, F.; Hoppener, J. W. M.; Bhola, S.; Blik, J.; van Roy, N.; Leschot, N. J.; Westerveld, A.; Mannens, M.: High-resolution chromosomal localization of the human calcitonin/CGRP/IAPP gene family members. Genomics 15: 525–529, 1993.
- [33612] 7164. Hoppener, J. W. M.; Oosterwijk, C.; van Hulst, K. L.; Verbeek, J. S.; Capel, P. J. A.; de Koning, E. J. P.; Clark, A.; Jansz, H. S.; Lips, C. J. M.: Molecular physiology of the islet amyloid polypeptide (IAPP)/amylin gene in man, rat, and transgenic mice. J. Cell. Biochem. 55S:39–53, 1994.
- [33613] 7165. Jaikaran, E. T. A. S.; Clark, A.: Islet amyloid and type 2 diabetes: from molecular misfolding to islet pathophysiology. Biochim. Biophys. Acta 1537: 179–203, 2001.
- [33614] 7166. Janson, J.; Soeller, W. C.; Roche, P. C.; Nelson, R. T.; Torchia, A. J.; Kreutter, D. K.; Butler, P. C.: Spontaneous diabetes mellitus in transgenic mice expressing human islet

amyloid polypeptide. *Proc.Nat. Acad. Sci.* 93: 7283–7288, 1996.

[33615] 7167.Johnson, K. H.; O'Brien, T. D.; Betsholtz, C.; Westermark, P.: Islet amyloid, islet-amyloid polypeptide, and diabetes mellitus. *NewEng. J. Med.* 321: 513–518, 1989.

[33616] 7168.Lorenzo, A.; Razzaboni, B.; Weir, G. C.; Yankner, B. A.: Pancreatic islet cell toxicity of amylin associated with type-2 diabetes mellitus. *Nature* 368:756–760, 1994.

[33617] 7169.Mosselman, S.; Hoppener, J. W. M.; Lips, C. J. M.; Jansz, H. S.: The complete islet amyloid polypeptide precursor is encoded by two exons. *FEBS Lett.* 247: 154–158, 1989.

[33618] 7170.Mosselman, S.; Hoppener, J. W. M.; Zandberg, J.; van Mansfeld, A. D. M.; Geurts van Kessel, A. H. M.; Lips, C. J. M.; Jansz, H. S.: Islet amyloid polypeptide: identification and chromosomal localization of the human gene. *FEBS Lett.* 239: 227–232, 1988.

[33619] 7171.Nishi, M.; Sanke, T.; Seino, S.; Eddy, R. L.; Fan, Y.-S.; Byers, M. G.; Shows, T. B.; Bell, G. I.; Steiner, D. F.: Human islet amyloid polypeptide gene: complete nucleotide sequence, chromosomal localization, and evolutionary history. *Molec. Endocr.* 3: 1775–1781, 1989.

[33620] 7172.Opie, E. L.: On the relation of the chronic interstitial

pancreatitis to the islands of Langerhans and to diabetes mellitus. *J. Exp. Med.* 5:397–428, 1900.

- [33621] 7173. Bergsagel, P. L.; Chesi, M.; Nardini, E.; Brents, L. A.; Kirby, S. L.; Kuehl, W. M.: Promiscuous translocations into immunoglobulin heavy chain switch regions in multiple myeloma. *Proc. Nat. Acad. Sci.* 93: 13931–13936, 1996.
- [33622] 7174. Chen, L.; Li, C.; Qiao, W.; Xu, X.; Deng, C.: A ser365–cys mutation of fibroblast growth factor receptor 3 in mouse downregulates Ihh/PTHrP signals and causes severe achondroplasia. *Hum. Molec. Genet.* 10:457–465, 2001.
- [33623] 7175. Clausen, J. O.; Hansen, T.; Bjorbaek, C.; Echwald, S. M.; Urhammer, S. A.; Rasmussen, S.; Andersen, C. B.; Hansen, L.; Almind, K.; Winther, K.; Haraldsdottir, J.; Borch-Johnsen, K.; Pedersen, O.: Insulin resistance: interactions between obesity and a common variant of insulin receptor substrate–1. *Lancet* 346: 397–402, 1995.
- [33624] 7176. Esposito, D. L.; Mammarella, S.; Ranieri, A.; Loggia, F. D.; Capani, F.; Consoli, A.; Mariani–Costantini, R.; Caramia, F. G.; Cama, A.; Battista, P.: Deletion of gly723 in the insulin receptor substrate–1 of a patient with noninsulin–dependent diabetes mellitus. *Hum. Mutat.*

7:364–366, 1996.

- [33625] 7177.Hribal, M. L.; Federici, M.; Porzio, O.; Lauro, D.; Borboni, P.; Accili, D.; Lauro, R.; Sesti, G.: The gly-to-arg(972) amino acid polymorphism in insulin receptor substrate-1 affects glucose metabolism in skeletal muscle cells. *J. Clin. Endocr. Metab.* 85: 2004–2013, 2000.
- [33626] 7178.Kido, Y.; Burks, D. J.; Withers, D.; Bruning, J. C.; Kahn, C.R.; White, M. F.; Accili, D.: Tissue-specific insulin resistance in mice with mutations in the insulin receptor, IRS-1, and IRS-2. *J. Clin. Invest.* 105: 199–205, 2000.
- [33627] 7179.Kulkarni, R. N.; Winnay, J. N.; Daniels, M.; Bruning, J. C.; Flier, S. N.; Hanahan, D.; Kahn, C. R.: Altered function of insulin receptor substrate-1-deficient mouse islets and cultured beta-cell lines. *J. Clin. Invest.* 104: R69–R75, 1999.
- [33628] 7180.Laakso, M.; Malkki, M.; Kekalainen, P.; Kuusisto, J.; Deeb, S.S.: Insulin receptor substrate-1 variants in non-insulin-dependent diabetes. *J. Clin. Invest.* 94: 1141–1146, 1994.
- [33629] 7181.Myers, M. G., Jr.; Sun, X. J.; White, M. F.: The IRS-1 signaling system. *Trends Biochem. Sci.* 19: 289–293, 1994.
- [33630] 7182.Nishiyama, M.; Inazawa, J.; Ariyama, T.; Nakamura,

Y.; Matsufuji, S.; Furusaka, A.; Tanaka, T.; Hayashi, S.;

Wands, J. R.: The human insulin receptor substrate-1 gene (IRS1) is localized on 2q36. *Genomics* 20:139–141, 1994.

[33631] 7183. Nishiyama, M.; Wands, J. R.: Cloning and increased expression of an insulin receptor substrate-1-like gene in human hepatocellular carcinoma. *Biochem. Biophys. Res. Commun.* 183: 280–285, 1992.

[33632] 7184. Ogata, N.; Chikazu, D.; Kubota, N.; Terauchi, Y.; Tobe, K.; Azuma, Y.; Ohta, T.; Kadowaki, T.; Nakamura, K.; Kawaguchi, H.: Insulin receptor substrate-1 in osteoblast is indispensable for maintaining bone turnover. *J. Clin. Invest.* 105: 935–943, 2000.

[33633] 7185. Ogihara, T.; Isobe, T.; Ichimura, T.; Taoka, M.; Funaki, M.; Sakoda, H.; Onishi, Y.; Inukai, K.; Anai, M.; Fukushima, Y.; Kikuchi, M.; Yazaki, Y.; Oka, Y.; Asano, T.: 14-3-3 protein binds to insulin receptor substrate-1, one of the binding sites of which is in the phosphotyrosine binding domain. *J. Biol. Chem.* 272: 25267–25274, 1997.

[33634] 7186. Porzio, O.; Federici, M.; Hribal, M. L.; Lauro, D.; Accili, D.; Lauro, R.; Borboni, P.; Sesti, G.: The gly972-to-arginine amino acid polymorphism in IRS-1 impairs insulin secretion in pancreatic beta cells. *J. Clin. Invest.* 104: 357–364,

1999.

- [33635] 7187.Stoffel, M.; Espinosa, R., III; Keller, S. R.; Lienhard, G. E.;Le Beau, M. M.; Bell, G. I.: Human insulin receptor substrate-1 gene(IRS1): chromosomal localization to 2q35-q36.1 and identificationof a simple tandem repeat DNA polymorphism. *Diabetologia* 36: 335-337,1993.
- [33636] 7188.Sun, X. J.; Rothenberg, P.; Kahn, C. R.; Backer, J. M.; Araki,E.; Wilden, P. A.; Cahill, D. A.; Goldstein, B. J.; White, M. F.:Structure of the insulin receptor substrate IRS-1 defines a unique signal transduction protein. *Nature* 352: 73-77, 1991.
- [33637] 7189.Diaz, M. O.; Bohlander, S.: Nomenclature of the human interferongenes. *J. Interferon Res.* 13: 443-444, 1993.
- [33638] 7190.Henco, K.; Brosius, J.; Fujisawa, A.; Fujisawa, J.-I.; Haynes,J. R.; Hochstadt, J.; Kovacic, T.; Pasek, M.; Schambock, A.; Schmid,J.; Todokoro, K.; Walchli, M.; Nagata, S.; Weissmann, C.: Structuralrelationship of human interferon alpha genes and pseudogenes. *J.Molec. Biol.* 185: 227-260, 1985.
- [33639] 7191.Olopade, O. I.; Bohlander, S. K.; Pomykala, H.; Maltepe, E.; VanMelle, E.; Le Beau, M. M.; Diaz, M. O.: Mapping of the shortest regionof overlap of deletions of the

short arm of chromosome 9 associated with human neoplasia. *Genomics* 14: 437–443, 1992.

[33640] 7192. Hogervorst, F.; Kuikman, I.; Geurts van Kessel, A.; Sonnenberg, A.: Molecular cloning of the human alpha-6 integrin subunit: alternative splicing of alpha-6 mRNA and chromosomal localization of the alpha-6 and beta-4 genes. *Europ. J. Biochem.* 199: 425–433, 1991.

[33641] 7193. Chavanas, S.; Gache, Y.; Vailly, J.; Kanitakis, J.; Pulkkinen, L.; Uitto, J.; Ortonne, J.-P.; Meneguzzi, G.: Splicing modulation of integrin beta-4 pre-mRNA carrying a branch point mutation underlies epidermolysis bullosa with pyloric atresia undergoing spontaneous amelioration with ageing. *Hum. Molec. Genet.* 8: 2097–2105, 1999.

[33642] 7194. Inoue, M.; Tamai, K.; Shimizu, H.; Owaribe, K.; Nakama, T.; Hashimoto, T.; McGrath, J. A.: A homozygous missense mutation in the cytoplasmic tail of beta-4 integrin, G931D, that disrupts hemidesmosome assembly and underlies non-Herlitz junctional epidermolysis bullosa without pyloric atresia? *J. Invest. Derm.* 114: 1061–1064, 2000.

[33643] 7195. Mellerio, J. E.; Pulkkinen, L.; McMillan, J. R.; Lake, B. D.; Horn, H. M.; Tidman, M. J.; Harper, J. I.; McGrath, J. A.; Uitto, J.; Eady, R. A. J.: Pyloric atresia–junctional epider–

molysis bullosa syndrome: mutations in the integrin beta-4 gene (ITGB4) in two unrelated patients with mild disease. *Brit. J. Derm.* 139: 862–871, 1998.

- [33644] 7196. Carayannopoulos, M. O.; Chi, M. M.-Y.; Cui, Y.; Pingsterhaus, J. M.; McKnight, R. A.; Mueckler, M.; Devaskar, S. U.; Moley, K. H.: GLUT8 is a glucose transporter responsible for insulin-stimulated glucose uptake in the blastocyst. *Proc. Nat. Acad. Sci.* 97: 7313–7318, 2000.
- [33645] 7197. Doege, H.; Schurmann, A.; Bahrenberg, G.; Brauers, A.; Joost, H. G.: GLUT8, a novel member of the sugar transport facilitator family with glucose transport activity. *J. Biol. Chem.* 275: 16275–16280, 2000.
- [33646] 7198. Ibberson, M.; Uldry, M.; Thorens, B.: GLUTX1, a novel mammalian glucose transporter expressed in the central nervous system and insulin-sensitive tissues. *J. Biol. Chem.* 275: 4607–4612, 2000.
- [33647] 7199. Kool, M.; de Haas, M.; Scheffer, G. L.; Scheper, R. J.; van Eijk, M. J.; Juijn, J. A.; Baas, F.; Borst, P.: Analysis of expression of cMOAT (MRP2), MRP3, MRP4, and MRP5, homologues of the multidrug resistance-associated protein gene (MRP1), in human cancer cell lines. *Cancer Res.* 57: 3537–3547, 1997.
- [33648] 7200. Lee, K.; Belinsky, M. G.; Bell, D. W.; Testa, J. R.; Kruh,

G. D.: Isolation of MOAT-B, a widely expressed multidrug resistance-associated protein/canalicular multispecific organic anion transporter-related transporter. *Cancer Res.* 58: 2741–2747, 1998.

[33649] 7201. Schuetz, J. D.; Connelly, M. C.; Sun, D.; Paibir, S. G.; Flynn, P. M.; Srinivas, R. V.; Kumar, A.; Fridland, A.: MRP4: a previously unidentified factor in resistance to nucleoside-based antiviral drugs. *Nature Med.* 5: 1048–1051, 1999.

[33650] 7202. Mahlknecht, U.; Schnittger, S.; Ottmann, O. G.; Schoch, C.; Mosebach, M.; Hiddemann, W.; Hoelzer, D.: Chromosomal organization and localization of the human histone deacetylase 5 gene (HDAC5). *Biochim. Biophys. Acta* 1493: 342–348, 2000.

[33651] 7203. McKinsey, T. A.; Zhang, C.-L.; Lu, J.; Olson, E. N.: Signal-dependent nuclear export of a histone deacetylase regulates muscle differentiation. *Nature* 408: 106–111, 2000.

[33652] 7204. Bruce, H. A.; Margolis, R. L.: FOXP2: novel exons, splice variants, and CAG repeat length stability. *Hum. Genet.* 111: 136–144, 2002.

[33653] 7205. Enard, W.; Przeworski, M.; Fisher, S. E.; Lai, C. S. L.; Wiebe, V.; Kitano, T.; Monaco, A. P.; Paabo, S.: Molecular

evolution of FOXP2, a gene involved in speech and language. *Nature* 418: 869–872, 2002.

- [33654] 7206.Chesney, J.; Mitchell, R.; Benigni, F.; Bacher, M.; Spiegel, L.;Al-Abed, Y.; Han, J. H.; Metz, C.; Bucala, R.: An inducible geneproduct for 6-phosphofructo-2-kinase with an AU-rich instability element:role in tumor cell glycolysis and the Warburg effect. *Proc. Nat.Acad. Sci.* 96: 3047-3052, 1999.
- [33655] 7207.Manzano, A.; Rosa, J. L.; Ventura, F.; Perez, J. X.; Nadal, M.;Estivill, X.; Ambrosio, S.; Gil, J.; Bartrons, R.: Molecular cloning,expression, and chromosomal localization of a ubiquitously expressedhuman 6-phosphofructo-2-kinase/fructose-2,6-bisphosphatase gene (PFKFB3). *Cytogenet.Cell Genet.* 83: 214-217, 1998.
- [33656] 7208.Nicholl, J.; Hamilton, J. A.; Sutherland, G. R.; Sutherland, R.L.; Watts, C. K. W.: The third human isoform of 6-phosphofructo-2-kinase/fructose-2,6-bisphosphatase(PFKFB3) map position 10p14-p15. *Chromosome Res.* 5: 150 only, 1997.
- [33657] 7209.Sakai, A.; Kato, M.; Fukasawa, M.; Ishiguro, M.; Furuya, E.; Sakakibara,R.: Cloning of cDNA encoding for a novel isozyme of fructose 6-phosphate,2-kinase/fructose2,6-bisphosphatase from human placenta. *J. Biochem.* 119: 506-511,1996.
- [33658] 7210.Newbury, D. F.; Bonora, E.; Lamb, J. A.; Fisher, S. E.;

Lai, C.S. L.; Baird, G.; Jannoun, L.; Slonims, V.; Stott, C. M.; Merricks, M. J.; Bolton, P. F.; Bailey, A. J.; Monaco, A. P.; International Molecular Genetic Study of Autism Consortium: FOXP2 is not a major susceptibility gene for autism or specific language impairment. *Am.J. Hum. Genet.* 70: 1318–1327, 2002.

[33659] 7211. Sakakibara, R.; Kato, M.; Okamura, N.; Nakagawa, T.; Komada, Y.; Tominaga, N.; Shimojo, M.; Fukasawa, M.: Characterization of a human placental fructose-6-phosphate, 2-kinase/fructose-2,6-bisphosphatase. *J. Biochem.* 122: 122–128, 1997.

[33660] 7212. Manzano, A.; Perez, J. X.; Nadal, M.; Estivill, X.; Lange, A.; Bartrons, R.: Cloning, expression and chromosomal localization of a human testis 6-phosphofructo-2-kinase/fructose-2,6-bisphosphatase gene. *Gene* 229: 83–89, 1999.

[33661] 7213. Larsen, M.; Ressler, S. J.; Gerdes, M. J.; Lu, B.; Byron, M.; Lawrence, J. B.; Rowley, D. R.: The WFDC1 gene encoding p50 localizes to 16q24, a region of LOH in multiple cancers. *Mammalian Genome* 11: 767–773, 2000.

[33662] 7214. Larsen, M.; Ressler, S. J.; Lu, B.; Gerdes, M. J.; McBride, L.; Dang, T. D.; Rowley, D. R.: Molecular cloning and expression of p50 growth inhibitor: a novel WAP-type

'four-disulfide core' domain protein expressed in smooth muscle. *J. Biol. Chem.* 273: 4574–4584, 1998.

[33663] 7215. Rowley, D. R.: Characterization of a fetal urogenital sinus mesenchymal cell line U4F: secretion of a negative growth factor regulatory activity. *In Vitro Cell. Dev. Biol.* 28A: 29–38, 1992.

[33664] 7216. Rowley, D. R.; Dang, T. D.; Larsen, M.; Gerdes, M. J.; McBride, L.; Lu, B.: Purification of a novel protein (ps20) from urogenital sinus mesenchymal cells with growth inhibitory properties in vitro. *J. Biol. Chem.* 270: 22058–22065, 1995.

[33665] 7217. Chen, G.; Cizeau, J.; Vande Velde, C.; Park, J. H.; Bozek, G.; Bolton, J.; Shi, L.; Dubik, D.; Greenberg, A.: Nix and nip3 form a subfamily of pro-apoptotic mitochondrial proteins. *J. Biol. Chem.* 274: 7–10, 1999.

[33666] 7218. Matsushima, M.; Fujiwara, T.; Takahashi, E.; Minaguchi, T.; Eguchi, Y.; Tsujimoto, Y.; Suzumori, K.; Nakamura, Y.: Isolation, mapping, and functional analysis of a novel human cDNA (BNIP3L) encoding a protein homologous to human NIP3. *Genes Chromosomes Cancer* 21: 230–235, 1998.

[33667] 7219. Yasuda, M.; Han, J.; Dionne, C. A.; Boyd, J. M.; Chinadurai, G.: BNIP3- α : a human homolog of mitochon-

drial proapoptotic proteinBNIP3. Cancer Res. 59: 533–537, 1999.

[33668] 7220.Hildebrand, J. D.; Taylor, J. M.; Parsons, T. J.: An SH3 domain–containingGTPase–activating protein for Rho and Cdc42 associates with focaladhesion kinase. Molec. Cell. Biol. 16: 3169–3178, 1996.

[33669] 7221.Smith, D. E.; Renshaw, B. R.; Ketchem, R. R.; Kubin, M.; Garka,K. E.; Sims, J. E.: Four new members expand the interleukin–1 superfamily. J.Biol. Chem. 275: 1169–1175, 2000.

[33670] 7222.Kumar, S.; McDonnell, P. C.; Lehr, R.; Tierney, L.; Tzimas, M.N.; Griswold, D. E.; Capper, E. A.; Tal–Singer, R.; Wells, G. I.;Doyle, M. L.; Young, P. R.: Identification and initial characterizationof four novel members of the interleukin–1 family. J. Biol. Chem. 275:10308–10314, 2000.

[33671] 7223.Borkhardt, A.; Bojesen, S.; Haas, O. A.; Fuchs, U.; Bartelheimer,D.; Loncarevic, I. F.; Bohle, R. M.; Harbott, J.; Repp, R.; Jaeger,U.; Viehmann, S.; Henn, T.; Korth, P.; Scharr, D.; Lampert, F.: Thehuman GRAF gene is fused to MLL in a unique t(5;11)(q31;q23) and bothalleles are disrupted in three cases of myelodysplastic syndrome/ acutemyeloid leukemia with a deletion 5q. Proc. Nat. Acad. Sci. 97: 9168–9173,2000.

- [33672] 7224. Krystal, G. W.; Armstrong, B. C.; Battey, J. F.: N-myc mRNA forms an RNA-RNA duplex with endogenous anti-sense transcripts. *Molec. Cell Biol.* 10: 4180-4191, 1990.
- [33673] 7225. Torrents, D.; Estevez, R.; Pineda, M.; Fernandez, E.; Lloberas, J.; Shi, Y.-B.; Zorzano, A.; Palacin, M.: Identification and characterization of a membrane protein ($\gamma(+)$ L amino acid transporter-1) that associates with 4F2hc to encode the amino acid transport activity $\gamma(+)$ L: a candidate gene for lysinuric protein intolerance. *J. Biol. Chem.* 273: 32437-32445, 1998.
- [33674] 7226. Cohen-Salmon, M.; Frenz, D.; Liu, W.; Verpy, E.; Voegelings, S.; Petit, C.: Fdp, a new fibrocyte-derived protein related to MIA/CD-RAP, has an in vitro effect on the early differentiation of the inner ear mesenchyme. *J. Biol. Chem.* 275: 40036-40041, 2000.
- [33675] 7227. Rendtorff, N. D.; Frodin, M.; Attie-Bitach, T.; Veke-mans, M.; Tommerup, N.: Identification and characterization of an inner ear-expressed human melanoma inhibitory activity (MIA)-like gene (MIAL) with a frequent polymorphism that abolishes translation. *Genomics* 71: 40-52, 2001.
- [33676] 7228. Robertson, N. G.; Heller, S.; Lin, J. S.; Resendes, B. L.; Weremowicz, S.; Denis, C. S.; Bell, A. M.; Hudspeth, A. J.;

Morton, C. C.: A novel conserved cochlear gene, OTOR: identification, expression analysis, and chromosomal mapping. *Genomics* 66: 242–248, 2000.

[33677] 7229. Haft, C. R.; de la Luz Sierra, M.; Bafford, R.; Lesniak, M. A.; Barr, V. A.; Taylor, S. I.: Human orthologs of yeast vacuolar protein sorting proteins Vps26, 29, and 35: assembly into multimeric complexes. *Molec. Biol. Cell* 11: 4105–4116, 2000.

[33678] 7230. Busfield, S. J.; Comrack, C. A.; Yu, G.; Chickering, T. W.; Smutko, J. S.; Zhou, H.; Leiby, K. R.; Holmgren, L. M.; Gearing, D. P.; Pan, Y.: Identification and gene organization of three novel members of the IL-1 family on human chromosome 2. *Genomics* 66: 213–216, 2000.

[33679] 7231. Mulero, J. J.; Pace, A. M.; Nelken, S. T.; Loeb, D. B.; Correa, T. R.; Drmanac, R.; Ford, J. E.: IL1HY1: a novel interleukin-1 receptor antagonist gene. *Biochem. Biophys. Res. Commun.* 263: 702–706, 1999.

[33680] 7232. Pratt, W. S.; Crawley, S.; Hicks, J.; Ho, J.; Nash, M.; Kim, Y. S.; Gum, J. R.; Swallow, D. M.: Multiple transcripts of MUC3: evidence for two genes, MUC3A and MUC3B. *Biochem. Biophys. Res. Commun.* 275: 916–923, 2000.

[33681] 7233. Ishida, N.; Ito, M.; Yoshioka, S.; Sun-Wada, G.-H.; Kawakita, M.: Functional expression of human Golgi CMP-

sialic acid transporter in the Golgi complex of a transporter-deficient Chinese hamster ovary cell mutant. *J. Biochem.* 124: 171–178, 1998.

[33682] 7234. Kasof, G. M.; Gomes, B. C.: Livin, a novel inhibitor of apoptosis protein family member. *J. Biol. Chem.* 276: 3238–3246, 2001.

[33683] 7235. Lin, J.-H.; Deng, G.; Huang, Q.; Morser, J.: KIAP, a novel member of the inhibitor of apoptosis protein family. *Biochem. Biophys. Res. Commun.* 279: 820–831, 2000.

[33684] 7236. Vucic, D.; Stennicke, H. R.; Pisabarro, M. T.; Salvesen, G. S.; Dixit, V. M.: ML-IAP, a novel inhibitor of apoptosis that is preferentially expressed in human melanomas. *Curr. Biol.* 10: 1359–1366, 2000.

[33685] 7237. Kobayashi, S.; Akiyama, T.; Nata, K.; Abe, M.; Tajima, M.; Shervani, N. J.; Unno, M.; Matsuno, S.; Sasaki, H.; Takasawa, S.; Okamoto, H.: Identification of a receptor for Reg (regenerating gene) protein, a pancreatic beta-cell regeneration factor. *J. Biol. Chem.* 275: 10723–10726, 2000.

[33686] 7238. Van Hul, W.; Wuyts, W.; Hendrickx, J.; Speleman, F.; Wauters, J.; De Boulle, K.; Van Roy, N.; Bossuyt, P.; Willems, P. J.: Identification of a third EXT-like gene (EXTL3) belonging to the EXT gene family. *Genomics* 47: 230–237,

1998.

- [33687] 7239.Dowler, S.; Currie, R. A.; Downes, C. P.; Alessi, D. R.: DAPP1:a dual adaptor for phosphotyrosine and 3-phosphoinositides. *Biochem.J.* 342: 7–12, 1999.
- [33688] 7240.Marshall, A. J.; Niiro, H.; Lerner, C. G.; Yun, T. J.; Thomas,S.; Disteché, C. M.; Clark, E. A.: A novel B lymphocyte-associated adaptor protein, Bam32, regulates antigen receptor signaling downstream of phosphatidylinositol 3-kinase. *J. Exp. Med.* 191: 1319–1331, 2000.
- [33689] 7241.Di Fiore, P. P.; Pelicci, P. G.; Sorkin, A.: EH: a novel protein-protein interaction domain potentially involved in intracellular sorting. *Trends Biochem. Sci.* 22: 411–413, 1997.
- [33690] 7242.Haider, N. B.; Searby, C.; Galperin, E.; Mintz, L.; Horowitz, M.; Stone, E. M.; Sheffield, V. C.: Evaluation and molecular characterization of EHD1, a candidate gene for Bardet-Biedl syndrome 1 (BBS1). *Gene* 240:227–232, 1999.
- [33691] 7243.Mintz, L.; Galperin, E.; Pasmanik-Chor, M.; Tulzinsky, S.; Bromberg, Y.; Kozak, C. A.; Joyner, A.; Fein, A.; Horowitz, M.: EHD1--an EH-domain-containing protein with a specific expression pattern. *Genomics* 59: 66–76, 1999.

- [33692] 7244. Bouju, S.; Pietu, G.; Le Cunff, M.; Cros, N.; Malzac, P.; Pellissier, J.-F.; Pons, F.; Leger, J.-J.; Auffray, C.; Dechesne, C. A.: Exclusion of muscle specific actinin-associated LIM protein (ALP) gene from 4q35 facioscapulohumeral muscular dystrophy (FSHD) candidate genes. *Neuro-musc. Disord.* 9: 3–10, 1999.
- [33693] 7245. Black, B. E.; Levesque, L.; Holaska, J. M.; Wood, T. C.; Paschal, B. M.: Identification of an NTF2-related factor that binds Ran-GTP and regulates nuclear protein export. *Molec. Cell. Biol.* 19: 8616–8624, 1999.
- [33694] 7246. Katahira, J.; Straber, K.; Podtelejnikov, A.; Mann, M.; Jung, J. U.; Hurt, E.: The Mex67p-mediated nuclear mRNA export pathway is conserved from yeast to human. *EMBO J.* 18: 2593–2609, 1999.
- [33695] 7247. Ossareh-Nazari, B.; Maison, C.; Black, B. E.; Levesque, L.; Paschal, B. M.; Dargemont, C.: RanGTP-binding protein NXT1 facilitates nuclear export of different classes of RNA in vitro. *Molec. Cell. Biol.* 20: 4562–4571, 2000.
- [33696] 7248. Scott, A. F.: Personal Communication. Baltimore, Md. 4/3/2001.
- [33697] 7249. Pati, D.; Meistrich, M. L.; Plon, S. E.: Human Cdc34 and Rad6 Ubiquitin-conjugating enzymes target repres-

sors of cyclic AMP–induced transcription for proteolysis.

Molec. Cell. Biol. 19: 5001–5013, 1999.

- [33698] 7250. Courjal, F.; Louason, G.; Speiser, P.; Katsaros, D.; Zeillinger, R.; Theillet, C.: Cyclin gene amplification and overexpression in breast and ovarian cancers: evidence for the selection of cyclin D1 in breast and cyclin E in ovarian tumors. *Int. J. Cancer* 69: 247–253, 1996.
- [33699] 7251. Strohmaier, H.; Spruck, C. H.; Kaiser, P.; Won, K.-A.; Sangfelt, O.; Reed, S. I.: Human F-box protein hCdc4 targets cyclin E for proteolysis and is mutated in a breast cancer cell line. *Nature* 413: 316–322, 2001.
- [33700] 7252. Matsuda, S.; Iriyama, C.; Yokozaki, S.; Ichigotani, Y.; Shirafuji, N.; Yamaki, K.; Hayakawa, T.; Hamaguchi, M.: Cloning and sequencing of a novel human gene that encodes a putative target protein of Nesh-SH3. *J. Hum. Genet.* 46: 483–486, 2001.
- [33701] 7253. Jager, D.; Stockert, E.; Jager, E.; Gure, A. O.; Scanlan, M. J.; Knuth, A.; Old, L. J.; Chen, Y.-T.: Serological cloning of a melanocyte-associated guanosine 5-prime-triphosphate-binding protein and a chromosome condensation protein from a melanoma complementary DNA library. *Cancer Res.* 60: 3584–3591, 2000.
- [33702] 7254. Loftus, S. K.; Larson, D. M.; Baxter, L. L.; Antonellis,

A.; Chen,Y.; Wu, X.; Jiang, Y.; Bittner, M.; Hammer, J. A., III; Pavan, W.J.: Mutation of melanosome protein RAB38 in chocolate mice. *Proc.Nat. Acad. Sci.* 99: 4471–4476, 2002.

[33703] 7255.Matsuyoshi, N.; Tanaka, T.; Toda, K.; Imamura, S.: Identification of novel cadherins expressed in human melanoma cells. *J. Invest.Derm.* 108: 908–913, 1997.

[33704] 7256.Chen, K.-S.; DeLuca, H. F.: Isolation and characterization of a novel cDNA from HL-60 cells treated with 1,25-dihydroxyvitamin D-3. *Biochim.Biophys. Acta* 1219: 26–32, 1994.

[33705] 7257.Ludwig, D. L.; Kotanides, H.; Le, T.; Chavkin, D.; Bohlen, P.;Witte, L.: Cloning, genetic characterization, and chromosomal mapping of the mouse VDUP1 gene. *Gene* 269: 103–112, 2001.

[33706] 7258.Yun, C. H. C.; Oh, S.; Zizak, M.; Steplock, D.; Tsao, S.; Tse,C.-M.; Weinman, E. J.; Donowitz, M.: cAMP-mediated inhibition of the epithelial brush border Na(+)/H(+) exchanger, NHE3, requires an associated regulatory protein. *Proc. Nat. Acad. Sci.* 94: 3010–3015,1997.

[33707] 7259.Li, Y.; Chin, L.-S.; Weigel, C.; Li, L.: Spring, a novel RINGfinger protein that regulates synaptic vesicle exocytosis. *J. Biol.Chem.* 276: 40824–40833, 2001.

- [33708] 7260.Reymond, A.; Meroni, G.; Fantozzi, A.; Merla, G.; Cairo, S.; Luzi,L.; Riganelli, D.; Zanaria, E.; Messali, S.; Cainarca, S.; Guffanti,A.; Minucci, S.; Pelicci, P. G.; Bal-labio, A.: The tripartite motiffamily identifies cell com-partments. *EMBO J.* 20: 2140–2151, 2001.
- [33709] 7261.Scardigli, R.; Schuurmans, C.; Gradwohl, G.; Guille-mot, F.: Crossregulationbetween neurogenin2 and path-ways specifying neuronal identity in thespinal cord. *Neu-ron* 31: 203–217, 2001.
- [33710] 7262.Boles, K. S.; Mathew, P. A.: Molecular cloning of CS1, a novelhuman natural killer cell receptor belonging to the CD2 subset ofthe immunoglobulin superfamily. *Immuno-genetics* 52: 302–307, 2001.
- [33711] 7263.Bouchon, A.; Cella, M.; Grierson, H. L.; Cohen, J. I.; Colonna,M.: Cutting edge: activation of NK cell-mediated cytotoxicity bya SAP-independent receptor of the CD2 family. *J. Immun.* 167: 5517–5521,2001.
- [33712] 7264.Habas, R.; Kato, Y.; He, X.: Wnt/Frizzled activation of Rho regulatesvertebrate gastrulation and requires a novel Formin homology proteinDaam1. *Cell* 107: 843–854, 2001.
- [33713] 7265.Peters, C. S.; Liang, X.; Li, S.; Kannan, S.; Peng, Y.; Taub, R.;Diamond, R. H.: ATF-7, a novel bZIP protein, in-

teracts with the PRL-1 protein-tyrosine phosphatase. J. Biol. Chem. 276: 13718–13726, 2001.

[33714] 7266. White, J. H.; McIlhinney, R. A. J.; Wise, A.; Ciruela, F.; Chan, W.-Y.; Emson, P. C.; Billinton, A.; Marshall, F. H.: The GABA-B receptor interacts directly with the related transcription factors CREB2 and ATFx. Proc. Nat. Acad. Sci. 97: 13967–13972, 2000.

[33715] 7267. Futai, E.; Kubo, T.; Sorimachi, H.; Suzuki, K.; Maeda, T.: Molecular cloning of PalBH, a mammalian homologue of the *Aspergillus* atypical calpain PalB. Biochim. Biophys. Acta 1517: 316–319, 2001.

[33716] 7268. Huang, Y.; Wang, K. K. W.: The calpain family and human disease. Trends Molec. Med. 7: 355–362, 2001.

[33717] 7269. Lee, H.-J.; Sorimachi, H.; Jeong, S.-Y.; Ishiura, S.; Suzuki, K.: Molecular cloning and characterization of a novel tissue-specific calpain predominantly expressed in digestive tract. Biol. Chem. 379: 175–183, 1998.

[33718] 7270. Yoshikawa, Y.; Mukai, H.; Hino, F.; Asada, K.; Kato, I.: Isolation of two novel genes, down-regulated in gastric cancer. Jpn. J. Cancer Res. 91: 459–463, 2000.

[33719] 7271. Burgess, D. L.; Davis, C. F.; Gefrides, L. A.; Noebels, J. L.: Identification of three novel Ca(2+) channel gamma subunit genes reveals molecular diversification by tandem

and chromosome duplication. *GenomeRes.* 9: 1204–1213, 1999.

- [33720] 7272. Burgess, D. L.; Gefrides, L. A.; Foreman, P. J.; Noebels, J. L.: A cluster of three novel Ca(2+) channel gamma subunit genes on chromosome 19q13.4: evolution and expression profile of the gamma subunit gene family. *Genomics* 71: 339–350, 2001.
- [33721] 7273. Chu, P.-J.; Robertson, H. M.; Best, P. M.: Calcium channel gamma subunits provide insights into the evolution of this gene family. *Gene* 280:37–48, 2001.
- [33722] 7274. Chen, X.; Wen, S.; Fukuda, M. N.; Gava, N. R.; Hsu, D.; Akama, T. O.; Yang-Feng, T.; Shen, C. K. J.: Human ITCH is a coregulator of the hematopoietic transcription factor NF-E2. *Genomics* 73: 238–241, 2001.
- [33723] 7275. D'Andrea, A. D.; Serhan, C. N.: Relieving the Itch. *Nature Genet.* 18:97–99, 1998.
- [33724] 7276. Perry, W. L.; Hustad, C. M.; Swing, D. A.; O'Sullivan, T. N.; Jenkins, N. A.; Copeland, N. G.: The itchy locus encodes a novel ubiquitin protein ligase that is disrupted in a18H mice. *Nature Genet.* 18:143–146, 1998.
- [33725] 7277. Qiu, L.; Joazeiro, C.; Fang, N.; Wang, H.-Y.; Elly, C.; Altman, Y.; Fang, D.; Hunter, T.; Liu, Y.-C.: Recognition and ubiquitination of Notch by Itch, a Hect-type E3 ubiqui-

tin ligase. J. Biol. Chem. 275:35734–35737, 2000.

- [33726] 7278. Winberg, G.; Matskova, L.; Chen, F.; Plant, P.; Rotin, D.; Gish, G.; Ingham, R.; Ernberg, I.; Pawson, T.: Latent membrane protein 2A of Epstein–Barr virus binds WW domain E3 protein–ubiquitin ligase that ubiquitinates B-cell tyrosine kinases. Molec. Cell. Biol. 20:8526–8535, 2000.
- [33727] 7279. Bradley, K. A.; Mogridge, J.; Mourez, M.; Collier, R. J.; Young, J. A. T.: Identification of the cellular receptor for anthrax toxin. Nature 414:160–161, 2001.
- [33728] 7280. Dragon, F.; Pogacic, V.; Filipowicz, W.: In vitro assembly of human H/ACA small nucleolar RNPs reveals unique features of U17 and telomerase RNAs. Molec. Cell. Biol. 20: 3037–3048, 2000.
- [33729] 7281. Tollervey, D.; Kiss, T.: Function and synthesis of small nucleolar RNAs. Curr. Opin. Cell Biol. 9: 337–342, 1997.
- [33730] 7282. Pogacic, V.; Dragon, F.; Filipowicz, W.: Human H/ACA small nucleolar RNPs and telomerase share evolutionarily conserved proteins NHP2 and NOP10. Molec. Cell. Biol. 20: 9028–9040, 2000.
- [33731] 7283. Ichtchenko, K.; Nguyen, T.; Sudhof, T. C.: Structures, alternative splicing, and neurexin binding of multiple neuroligins. J. Biol. Chem. 271: 2676–2682, 1996.

- [33732] 7284.Hammarsund, M.; Wilson, W.; Corcoran, M.; Merup, M.; Einhorn, S.;Grander, D.; Sangfelt, O.: Identification and characterization oftwo novel human mitochondrial elongation factor genes, hEFG2 and hEFG1,phylogenetically conserved through evolution. Hum. Genet. 109: 542–550,2001.
- [33733] 7285.Ishibashi, K.; Suzuki, M.; Sasaki, S.; Imai, M.: Identificationof a new multigene four-transmembrane family (MS4A) related to CD20,HTm4 and beta subunit of the high-affinity IgE receptor. Gene 264:87–93, 2001.
- [33734] 7286.Liang, Y.; Tedder, T. F.: Identification of a CD20–, Fc-epsilon-RI-beta-relatedgene family: sixteen new MS4A family members expressed in human andmouse. Genomics 72: 119–127, 2001.
- [33735] 7287.Ishii, H.; Vecchione, A.; Murakumo, Y.; Baldassarre, G.; Numata,S.; Trapasso, F.; Alder, H.; Baffa, R.; Croce, C. M.: FEZ1/LZTS1gene at 8p22 suppresses cancer cell growth and regulates mitosis. Proc.Nat. Acad. Sci. 98: 10374–10379, 2001.
- [33736] 7288.Mahon, M. J.; Donowitz, M.; Yun, C. C.; Segre, G. V.: Na⁺/H⁺ exchangerregulatory factor 2 directs parathyroid hormone 1 receptor signalling. Nature 417:858–861, 2002.

- [33737] 7289. Poulat, F.; de Santa Barbara, P.; Desclozeaux, M.; Soullier, S.; Moniot, B.; Bonneaud, N.; Boizet, B.; Berta, P. The human testis-determining factor SRY binds a nuclear factor containing PDZ protein interaction domains. *J. Biol. Chem.* 272: 7167–7172, 1997.
- [33738] 7290. Yun, C.-H. C.; Lamprecht, G.; Forster, D. V.; Sidor, A.: NHE3 kinase A regulatory protein E3KARP binds the epithelial brush border Na(+)/H(+) exchanger NHE3 and the cytoskeletal protein ezrin. *J. Biol. Chem.* 273: 25856–25863, 1998.
- [33739] 7291. Hall, R. A.; Ostedgaard, L. S.; Premont, R. T.; Blitzer, J. T.; Rahman, N.; Welsh, M. J.; Lefkowitz, R. J.: A C-terminal motif found in the beta-2-adrenergic receptor, P2Y1 receptor and cystic fibrosis transmembrane conductance regulator determines binding to the Na(+)/H(+) exchanger regulatory factor family of PDZ proteins. *Proc. Nat. Acad. Sci.* 95: 8496–8501, 1998.
- [33740] 7292. Druck, T.; Podolski, J.; Byrski, T.; Wyrwicz, L.; Zajaczek, S.; Kata, G.; Borowka, A.; Lubinski, J.; Huebner, K.: The DIRC1 gene at chromosome 2q33 spans a familial RCC-associated t(2;3)(q33;q21) chromosome translocation. *J. Hum. Genet.* 46: 583–589, 2001.
- [33741] 7293. Podolski, J.; Zajaczek, S.; Byrski, T.; Druck, T.; Zi-

monjic, D.B.; Popescu, N. C.; Lubinski, J.; Huebner, K.:
Characterization of a familial RCC-associated
t(2;3)(q33;q21) chromosome translocation. J. Hum. Genet.
in-press, 2001.

[33742] 7294. Bruick, R. K.; McKnight, S. L.: A conserved family of
prolyl-4-hydroxylases that modify HIF. Science 294:
1337–1340, 2001.

[33743] 7295. Epstein, A. C. R.; Gleadle, J. M.; McNeill, L. A.; Hewit-
son, K.S.; O'Rourke, J.; Mole, D. R.; Mukherji, M.; Metzen,
E.; Wilson, M.I.; Dhanda, A.; Tian, Y.-M.; Masson, N.;
Hamilton, D. L.; Jaakkola, P.; Barstead, R.; Hodgkin, J.;
Maxwell, P. H.; Pugh, C. W.; Schofield, C. J.; Ratcliffe, P. J.:
C. elegans EGL-9 and mammalian homologs define a fam-
ily of dioxygenases that regulate HIF by prolyl hydroxyla-
tion. Cell 107:43–54, 2001.

[33744] 7296. Dupuy, D.; Aubert, I.; Duperat, V. G.; Petit, J.; Taine,
L.; Stef, M.; Bloch, B.; Arveiler, B.: Mapping, characteriza-
tion, and expression analysis of the SM-20 human homo-
logue, C1orf12, and identification of a novel related gene,
SCAND2. Genomics 69: 348–354, 2000.

[33745] 7297. Ritter, J. K.; Crawford, J. M.; Owens, I. S.: Cloning of
two human liver bilirubin UDP-glucuronosyltransferase
cDNAs with expression in COS-1 cells. J. Biol. Chem. 266:

1043–1047, 1991.

[33746] 7298.Rheaume, E.; Leblanc, J. F.; Lachance, Y.; Labrie, F.; Simard, J.: Detection of frequent BglII polymorphism by polymerase chain reaction and TaqI restriction fragment length polymorphism for 3-beta-hydroxysteroid dehydrogenase/delta-5-delta-4 isomerase at the human HSDB3 locus (1p11-p13). Hum. Genet. 87: 753–754, 1991.

[33747] 7299.Reynolds, D. S.; Gurley, D. S.; Austen, K. F.: Cloning and characterization of the novel gene for mast cell carboxypeptidase A. J. Clin. Invest. 89:273–282, 1992.

[33748] 7300.Yamaguchi, N.; Benya, P. D.; van der Rest, M.; Nishimura, Y.: The cloning and sequencing of alpha-1(VIII) collagen cDNAs demonstrate that type VIII collagen is a short chain collagen and contains triple-helical and carboxyl-terminal non-triple-helical domains similar to those of type X collagen. J. Biol. Chem. 264: 16022–16029, 1989.

[33749] 7301.Bleul, C. C.; Farzan, M.; Choe, H.; Parolin, C.; Clark-Lewis, I.; Soderstrom, J.; Springer, T. A.: The lymphocyte chemoattractant SDF-1 is a ligand for LESTR/fusin and blocks HIV-1 entry. Nature 382:829–833, 1996.

[33750] 7302.Caruz, A.; Samsom, M.; Alonso, J. M.; Alcami, J.;

Baleux, F.; Virelizier, J. L.; Parmentier, M.; Arenzana-Seisdedos, F.: Genomic organization and promoter characterization of human CXCR4 gene. *FEBS Lett.* 426:271–278, 1998.

[33751] 7303. Clapham, P. R.; Blanc, D.; Weiss, R. A.: Specific cell surface requirements for the infection of CD4-positive cells by human immunodeficiency virus types 1 and 2 and by Simian immunodeficiency virus. *Virology* 181:703–715, 1991.

[33752] 7304. Dragic, T.; Alizon, M.: Different requirements for membrane fusion mediated by the envelopes of human immunodeficiency virus types 1 and 2. *J. Virol.* 67: 2355–2359, 1993.

[33753] 7305. Federspiel, B.; Melhado, I. G.; Duncan, A. M. V.; Delaney, A.; Schappert, K.; Clark-Lewis, I.; Jirik, F. R.: Molecular cloning of the cDNA and chromosomal localization of the gene for a putative seven-transmembrane segment (7-TMS) receptor isolated from human spleen. *Genomics* 16:707–712, 1993.

[33754] 7306. Feng, Y.; Broder, C. C.; Kennedy, P. E.; Berger, E. A.: HIV-1 entry cofactor: functional cDNA cloning of a seven-transmembrane, G protein-coupled receptor. *Science* 272: 872–876, 1996.

- [33755] 7307.Hendrix, C. W.; Flexner, C.; MacFarland, R. T.; Giandomenico, C.;Fuchs, E. J.; Redpath, E.; Bridger, G.; Henson, G. W.: Pharmacokineticsand safety of AMD-3100, a novel antagonist of the CXCR-4 chemokinereceptor, in human volunteers. *Antimicrob. Agents Chemother.* 44:1667-1673, 2000.
- [33756] 7308.Herzog, H.; Hort, Y. J.; Shine, J.; Selbie, L. A.: Molecular cloning,characterization, and localization of the human homolog to the reportedbovine NPY Y3 receptor: lack of NPY binding and activation. *DNA CellBiol.* 12: 465-471, 1993.
- [33757] 7309.Jazin, E. E.; Yoo, H.; Blomqvist, A. G.; Yee, F.; Weng, G.; Walker,M. W.; Salon, J.; Larhammar, D.; Wahlestedt, C.: A proposed bovineneuropeptide Y (NPY) receptor cDNA clone, or its human homologue,confers neither NPY binding sites nor NPY responsiveness on transfectedcells. *Regul. Pept.* 47: 247-258, 1993.
- [33758] 7310.Liotta, L. A.: An attractive force in metastasis. *Nature* 410:24-25, 2001.
- [33759] 7311.Loetscher, M.; Geiser, T.; O'Reilly, T.; Zwahlen, R.; Baggiolini,M.; Moser, B.: Cloning of a human seven-transmembrane domain receptor,LESTR, that is highly expressed in leukocytes. *J. Biol. Chem.* 269:232-237, 1994.

- [33760] 7312.Lu, M.; Grove, E. A.; Miller, R. J.: Abnormal development of the hippocampal dentate gyrus in mice lacking the CXCR4 chemokine receptor. *Proc. Nat. Acad. Sci.* 99: 7090–7095, 2002.
- [33761] 7313.Ma, Q.; Jones, D.; Borghesani, P. R.; Segal, R. A.; Nagasawa, T.; Kishimoto, T.; Bronson, R. T.; Springer, T. A.: Impaired B-lymphopoiesis, myelopoiesis, and derailed cerebellar neuron migration in CXCR4- and SDF-1-deficient mice. *Proc. Nat. Acad. Sci.* 95: 9448–9453, 1998.
- [33762] 7314.Muller, A.; Homey, B.; Soto, H.; Ge, N.; Catron, D.; Buchanan, M. E.; McClanahan, T.; Murphy, E.; Yuan, W.; Wagner, S. N.; Barrera, J. L.; Mohar, A.; Verastegui, E.; Zlotnik, A.: Involvement of chemokine receptors in breast cancer metastasis. *Nature* 410: 50–56, 2001.
- [33763] 7315.Nagasawa, T.; Hirota, S.; Tachibana, K.; Takakura, N.; Nishikawa, S.; Kitamura, Y.; Yoshida, N.; Kikutani, H.; Kishimoto, T.: Defects of B-cell lymphopoiesis and bone-marrow myelopoiesis in mice lacking the CXC chemokine PBSF/SDF-1. *Nature* 382: 635–638, 1996.
- [33764] 7316.Oberlin, E.; Amara, A.; Bachelier, F.; Bessia, C.; Virelizier, J.-L.; Arenzana-Seisdedos, F.; Schwartz, O.; Heard, J.-M.; Clark-Lewis, I.; Legler, D. F.; Loetscher, M.;

Baggiolini, M.; Moser, B.: The CXC chemokine SDF-1 is the ligand for LESTR/fusin and prevents infection by T-cell-line-adapted HIV-1. *Nature* 382: 833-835, 1996.

[33765] 7317. Peled, A.; Petit, I.; Kollet, O.; Magid, M.; Ponomarev, T.; Byk, T.; Nagler, A.; Ben-Hur, H.; Many, A.; Shultz, L.; Lider, O.; Alon, R.; Zipori, D.; Lapidot, T.: Dependence of human stem cell engraftment and repopulation of NOD/SCID mice on CXCR4. *Science* 283: 845-848, 1999.

[33766] 7318. Rimland, J.; Xin, W.; Sweetnam, P.; Saijoh, K.; Nestler, E. J.; Duman, R. S.: Sequence and expression of a neuropeptide Y receptor cDNA. *Molec. Pharm.* 40: 869-875, 1991.

[33767] 7319. Tachibana, K.; Hirota, S.; Iizasa, H.; Yoshida, H.; Kawabata, K.; Kataoka, Y.; Kitamura, Y.; Matsushima, K.; Yoshida, N.; Nishikawa, S.; Kishimoto, T.; Nagasawa, T.: The chemokine receptor CXCR4 is essential for vascularization of the gastrointestinal tract. *Nature* 393: 591-594, 1998.

[33768] 7320. Wegner, S. A.; Ehrenberg, P. K.; Chang, G.; Dayhoff, D. E.; Sleeker, A. L.; Michael, N. L.: Genomic organization and functional characterization of the chemokine receptor CXCR4, a major entry co-receptor for human immunodeficiency virus type 1. *J. Biol. Chem.* 273: 4754-4760, 1998.

- [33769] 7321.Weiner, D. B.; Huebner, K.; Williams, W. V.; Greene, M. I.: Humangen es other than CD4 facilitate HIV-1 infection of murine cells. *Pathobiology* 59:361-371, 1991.
- [33770] 7322.Szostecki, C.; Guldner, H. H.; Netter, H. J.; Will, H.: Isolationand characterization of cDNA encoding a human nuclear antigen predominantlyrecognized by autoantibodies from patients with primary biliary cirrhosis. *J.Immun.* 145: 4338-4347, 1990.
- [33771] 7323.Freeman, G. J.; Disteche, C. M.; Gribben, J. G.; Adler, D. A.;Freedman, A. S.; Dougery, J.; Nadler, L. M.: The gene for B7, a costimulatorysignal for T-cell activation, maps to chromosomal region 3q13.3-3q21. *Blood* 79:489-494, 1992.
- [33772] 7324.Reeves, R. H.; Patch, D.; Sharpe, A. H.; Borriello, F.; Freeman,G. J.; Edelhoff, S.; Disteche, C.: The costimulatory genes Cd80 andCd86 are linked on mouse chromosome 16 and human chromosome 3. *MammalianGenome* 8: 581-582, 1997.
- [33773] 7325.Selvakumar, A.; Mohanraj, B. K.; Eddy, R. L.; Shows, T. B.; White,P. C.; Dupont, B.: Genomic organization and chromosomal locationof the human gene encoding the B-lymphocyte activation antigen B7. *Immunogenetics* 36:175-181, 1992.

- [33774] 7326.Ha, H.; Kubagawa, H.; Burrows, P. D.: Molecular cloning and expression pattern of a human gene homologous to the murine mb-1 gene. J. Immun. 148:1526-1531, 1992.
- [33775] 7327.Hashimoto, S.; Mohrenweiser, H. W.; Gregersen, P. K.; Chiorazzi, N.: Chromosomal localization, genomic structure, and allelic polymorphism of the human CD79a (Ig-alpha/mb-1) gene. Immunogenetics 40: 287-295, 1994.
- [33776] 7328.Reth, M.: Antigen receptors on B lymphocytes. Annu. Rev. Immun. 10:97-121, 1992.
- [33777] 7329.Liang, Y.; Buckley, T. R.; Tu, L.; Langdon, S. D.; Tedder, T. F.: Structural organization of the human MS4A gene cluster on chromosome 11q12. Immunogenetics 53: 357-368, 2001.
- [33778] 7330.Tedder, T. F.; Disteche, C. M.; Louie, E.; Adler, D. A.; Croce, C. M.; Schlossman, S. F.; Saito, H.: The gene that encodes the human CD20 (B1) differentiation antigen is located on chromosome 11 near the t(11;14)(q13;q32) translocation site. J. Immun. 142: 2555-2559, 1989.
- [33779] 7331.Tedder, T. F.; Klejman, G.; Schlossman, S. F.; Saito, H.: Structure of the gene encoding the human B lymphocyte differentiation antigen CD20 (B1). J. Immun. 142:

2560–2568, 1989.

- [33780] 7332.Tedder, T. F.; Streuli, M.; Schlossman, S. F.; Saito, H.: Isolation and structure of a cDNA encoding the B1 (CD20) cell-surface antigen of human B lymphocytes. *Proc. Nat. Acad. Sci.* 85: 208–212, 1988.
- [33781] 7333.Kaplan, F. S.; Tabas, J. A.; Zasloff, M. A.: Fibrodysplasia ossificans progressiva: a clue from the fly? *Calcif. Tissue Int.* 47: 117–125, 1990.
- [33782] 7334.Hopken, U. E.; Lu, D.; Gerard, N. P.; Gerard, C.: The C5a chemoattractant receptor mediates mucosal defense to infection. *Nature* 383: 86–89, 1996.
- [33783] 7335.Chen, H.; Jawahar, S.; Qian, Y.; Duong, Q.; Chan, G.; Parker, A.; Meyer, J. M.; Moore, K. J.; Chayen, S.; Gross, D. J.; Glaser, B.; Permutt, M. A.; Fricker, L. D.: Missense polymorphism in the human carboxypeptidase E gene alters enzymatic activity. *Hum. Mutat.* 18:120–131, 2001.
- [33784] 7336.Hall, C.; Manser, E.; Spurr, N. K.; Lim, L.: Assignment of the human carboxypeptidase E (CPE) gene to chromosome 4. *Genomics* 15:461–463, 1993.
- [33785] 7337.Manser, E.; Fernandez, D.; Loo, L.; Goh, P. Y.; Monfries, C.; Hall, C.; Lim, L.: Human carboxypeptidase E: isolation and characterization of the cDNA, sequence conservation, expression and processing in vitro. *Biochem.J.*

267: 517–525, 1990.

- [33786] 7338.Kas, K.; Schoenmakers, E. F. P. M.; Van de Ven, W. J. M.: Physicalmap location of the human carboxypeptidase M gene (CPM) distal toD12S375 and proximal to D12S8 at chromosome 12q15. *Genomics* 30:403–405, 1995.
- [33787] 7339.Rehli, M.; Krause, S. W.; Kreutz, M.; Andreesen, R.: CarboxypeptidaseM is identical to the MAX.1 antigen and its expression is associatedwith monocyte to macrophage differentiation. *J. Biol. Chem.* 270:15644–15649, 1995.
- [33788] 7340.Tan, F.; Chan, S. J.; Steiner, D. F.; Schilling, J. W.; Skidgel,R. A.: Molecular cloning and sequencing of the cDNA for human membrane–boundcarboxypeptidase M: comparison with carboxypeptidases A, B, H, andN. *J. Biol. Chem.* 264: 13165–13170, 1989.
- [33789] 7341.Gold, P.; Freedman, S. O.: Demonstration of tumor–specific antigensin human colonic carcinomata by immunological tolerance and absorptiontechniques. *J. Exp. Med.* 121: 439–462, 1965.
- [33790] 7342.Kamarck, M. E.; Elting, J. J.; Hart, J. T.; Goebel, S. J.; Rae,P. M. M.; Nothdurft, M. A.; Nedwin, J. J.; Barnett, T. R.: Carcinoembryonicantigen family: expression in a mouse L–cell transfectant and characterizationof a partial cDNA in bacteriophage lambda–gt11. *Proc. Nat. Acad.Sci.* 84:

5350–5354, 1987.

- [33791] 7343.Nishi, M.; Inazawa, J.; Inoue, K.; Nakagawa, H.; Taniwaki, M.; Misawa, S.; Oikawa, S.; Nakazato, H.; Abe, T.: Regional chromosomal assignment of carcinoembryonic antigen gene (CEA) to chromosome 19 at band q13.2. *Cancer Genet. Cytogenet.* 54: 77–81, 1991.
- [33792] 7344.Oikawa, S.; Nakazato, H.; Kosaki, G.: Primary structure of human carcinoembryonic antigen (CEA) deduced from cDNA sequence. *Biochem. Biophys. Res. Commun.* 142: 511–528, 1987.
- [33793] 7345.Thompson, J.; Zimmermann, W.: The carcinoembryonic antigen gene family: structure, expression and evolution. *Tumor Biol.* 9: 63–83, 1988.
- [33794] 7346.Thompson, J. A.; Pande, H.; Paxton, R. J.; Shively, L.; Padma, A.; Simmer, R. L.; Todd, C. W.; Riggs, A. D.; Shively, J. E.: Molecular cloning of a gene belonging to the carcinoembryonic antigen gene family and discussion of a domain model. *Proc. Nat. Acad. Sci.* 84: 2965–2969, 1987.
- [33795] 7347.Willcocks, T. C.; Craig, I. W.: Characterization of the genomic organization of human carcinoembryonic antigen (CEA): comparison with other family members and sequence analysis of 5-prime controlling region. *Genomics* 8: 492–500, 1990.

- [33796] 7348. Willcocks, T. C.; Craig, S. P.; Coates, D.; Craig, I. W.: Coding sequences for carcinoembryonic antigen (CEA) assigned to human chromosome 19q13. (Abstract) Cytogenet. Cell Genet. 46: 716 only, 1987.
- [33797] 7349. Zimmer, R.; Thomas, P.: Mutations in the carcinoembryonic antigen gene in colorectal cancer patients: implications on liver metastasis. Cancer Res. 61: 2822–2826, 2001.
- [33798] 7350. Zimmermann, W.; Ortlieb, B.; Friedrich, R.; von Kleist, S.: Isolation and characterization of cDNA clones encoding the human carcinoembryonic antigen reveal a highly conserved repeating structure. Proc. Nat. Acad. Sci. 84: 2960–2964, 1987.
- [33799] 7351. Zimmermann, W.; Weber, B.; Ortlieb, B.; Rudert, F.; Schempp, W.; Fiebig, H.-H.; Shively, J. E.; von Kleist, S.; Thompson, J. A.: Chromosomal localization of the carcinoembryonic antigen gene family and differential expression in various tumors. Cancer Res. 48: 2550–2554, 1988.
- [33800] 7352. Walker, E. S.: Familial paroxysmal dystonic choreoathetosis: a neurologic disorder simulating psychiatric illness. Johns Hopkins Med. J. 148: 108–113, 1981.
- [33801] 7353. Schuback, D.; Kramer, P.; Ozelius, L.; Holmgren, G.; Forsgren, L.; Kyllerman, M.; Wahlstrom, J.; Craft, C. M.; Ny-

gaard, T.; Brin, M.; de Leon, D.; Bressman, S.; Moskowitz, C. B.; Burke, R. E.; Sanner, G.; Drugge, U.; Gusella, J. F.; Fahn, S.; Breakefield, X. O.: Dopamine beta-hydroxylase gene excluded in four subtypes of hereditary dystonia. Hum. Genet. 87: 311–316, 1991.

[33802] 7354. Arahata, K.; Hayashi, Y. K.; Mizuno, Y.; Yoshida, M.; Ozawa, E.: Dystrophin-associated glycoprotein and dystrophin co-localisation at sarcolemma in Fukuyama congenital muscular dystrophy. (Letter) Lancet 342: 623–624, 1993.

[33803] 7355. Campanelli, J. T.; Roberds, S. L.; Campbell, K. P.; Scheller, R. H.: A role for dystrophin-associated glycoproteins and utrophin in agrin-induced AChR clustering. Cell 77: 663–674, 1994.

[33804] 7356. Cao, W.; Henry, M. D.; Borrow, P.; Yamada, H.; Elder, J. H.; Ravkov, E. V.; Nichol, S. T.; Compans, R. W.; Campbell, K. P.; Oldstone, M. B. A.: Identification of alpha-dystroglycan as a receptor for lymphocytic choriomeningitis virus and Lassa fever virus. Science 282: 2079–2081, 1998.

[33805] 7357. Ottman, R.; Risch, N.; Hauser, W. A.; Pedley, T. A.; Lee, J. H.; Barker-Cummings, C.; Lustenberger, A.; Nagle, K. J.; Lee, K. S.; Scheuer, M. L.; Neystat, M.; Susser, M.; Wil-

helmsen, K. C.: Localization of a gene for partial epilepsy to chromosome 10q. *Nature Genet.* 10:56–60, 1995.

[33806] 7358. Stogmann, E.; Zimprich, A.; Baumgartner, C.; Aull-Watschinger, S.; Holtt, V.; Zimprich, F.: A functional polymorphism in the prodynorphin gene promoter is associated with temporal lobe epilepsy. *Ann. Neurol.* 51:260–263, 2002.

[33807] 7359. Summar, M. L.; Phillips, J. A., III; Battey, J.; Castiglione, C. M.; Kidd, K. K.; Maness, K. J.; Weiffenbach, B.; Gravius, T. C.: Linkage relationships of human arginine vasopressin-neurophysin-II and oxytocin-neurophysin-I to prodynorphin and other loci on chromosome 20. *Molec. Endocr.* 4: 947–950, 1990.

[33808] 7360. Zimprich, A.; Kraus, J.; Woltje, M.; Mayer, P.; Rauch, E.; Holtt, V.: An allelic variation in the human prodynorphin gene promoter alters stimulus-induced expression. *J. Neurochem.* 74: 472–477, 2000.

[33809] 7361. Craig, S. P.; Day, I. N. M.; Thompson, R. J.; Craig, I. W.: Localization of human neurone-specific enolase to chromosome 12p13. (Abstract) *Cytogenet. Cell Genet.* 51: 980 only, 1989.

[33810] 7362. Craig, S. P.; Day, I. N. M.; Thompson, R. J.; Craig, I. W.: Localisation of neurone-specific enolase (ENO2) to

12p13. Cytogenet. Cell Genet. 54:71–73, 1990.

[33811] 7363.Feo, S.; Oliva, D.; Barbieri, G.; Xu, W.; Fried, M.; Giallongo, A.: The gene for the muscle-specific enolase is on the short arm of human chromosome 17. Genomics 6: 192–194, 1990.

[33812] 7364.Grzeschik, K.-H.: Assignment of human genes: beta-glucuronidase to chromosome 7, adenylate kinase-1 to 9, a second enzyme with enolase activity to 12, and mitochondrial IDH to 15. Cytogenet. Cell Genet. 16:142–148, 1976. Note: Alternate: Birth Defects Orig. Art. Ser. 12(7):142–148, 1976.

[33813] 7365.Herbschleb-Voogt, E.; Monteba-van Heuvel, M.; Wijnen, L. M. M.; Westerveld, A.; Pearson, P. L.; Meera Khan, P.: Chromosomal assignment and regional localization of CS, ENO-2, GAPDH, LDH-B, PEP-B and TPI in man-rodent cell hybrids. Cytogenet. Cell Genet. 22: 482–486, 1978.

[33814] 7366.Hinks, L. J.; Day, I. N. M.: Further studies of enolase loci. (Abstract) Cytogenet. Cell Genet. 58: 1854 only, 1991.

[33815] 7367.Law, M. L.; Kao, F.: Regional mapping of the gene coding for enolase-2 on human chromosome 12. J. Cell Sci. 53: 245–254, 1982.

[33816] 7368.Mattei, J. F.; Baeteman, M. A.; Mattei, M. G.; Ardisson, J. P.; Giraud, F.: Regional assignments of CS and

ENO2 on chromosome 12.(Abstract) Cytogenet. Cell Genet. 32: 297 only, 1982.

[33817] 7369.Oliva, D.; Cali, L.; Feo, S.; Giallongo, A.: Complete structure of the human gene encoding neuron-specific enolase. Genomics 10:157–165, 1991.

[33818] 7370.Jenkins, N. A.; Justice, M. J.; Gilbert, D. J.; Chu, M.-L.; Copeland, N. G.: Nidogen/entactin (Nid) maps to the proximal end of mouse chromosome 13 linked to beige (bg) and identifies a new region of homology between mouse and human chromosomes. Genomics 9: 401–403, 1991.

[33819] 7371.Olavesen, M. G.; Bentley, E.; Mason, R. V. F.; Stephens, R. J.; Ragoussis, J.: Fine mapping of 39 ESTs on human chromosome 6p23–p25. Genomics 46:303–306, 1997.

[33820] 7372.Boyd, C. D.; Toth-Fejel, S.; Gadi, I. K.; Litt, M.; Condon, M.R.; Kolbe, M.; Hagen, I. K.; Kurkinen, M.; Mackenzie, J. W.; Magenis, E.: The genes coding for human pro alpha-1(IV) collagen and pro alpha-2(IV) collagen are both located at the end of the long arm of chromosome 13. Am. J. Hum. Genet. 42: 309–314, 1988.

[33821] 7373.Brazel, D.; Pollner, R.; Oberbaumer, I.; Kuhn, K.: Human basement membrane collagen (type IV): the amino

acid sequence of the alpha-2(IV) chain and its comparison with the alpha-1(IV) chain reveals deletions in the alpha-1(IV) chain. *Europ. J. Biochem.* 172: 35–42, 1988.

[33822] 7374. Griffin, C. A.; Emanuel, B. S.; Hansen, J. R.; Cavenee, W. K.; Myers, J. C.: Human collagen genes encoding basement membrane alpha-1(IV) and alpha-2(IV) chains map to the distal long arm of chromosome 13. *Proc. Nat. Acad. Sci.* 84: 512–516, 1987.

[33823] 7375. Haniel, A.; Welge-Lussen, U.; Kuhn, K.; Poschl, E.: Identification and characterization of a novel transcriptional silencer in the human collagen type IV gene COL4A2. *J. Biol. Chem.* 270: 11209–11215, 1995.

[33824] 7376. Killen, P. D.; Francomano, C. A.; Yamada, Y.; Modi, W. S.; O'Brien, S. J.: Partial structure of the human alpha-2(IV) collagen chain and chromosomal localization of the gene (COL4A2). *Hum. Genet.* 77: 318–324, 1987.

[33825] 7377. Solomon, E.; Hall, V.; Kurkinen, M.: The human alpha-2(IV) collagen gene, COL4A2, is syntenic with the alpha-1(IV) gene, COL4A1, on chromosome 13. *Ann. Hum. Genet.* 51: 125–127, 1987.

[33826] 7378. Davis, R. C.; Xia, Y.; Mohandas, T.; Schotz, M. C.; Lysis, A. J.: Assignment of the human pancreatic colipase gene to chromosome 6p21.1 to pter. *Genomics* 10:

262–265, 1991.

- [33827] 7379.Sims, H. F.; Lowe, M. E.: The human colipase gene: isolation, chromosomal location, and tissue-specific expression. *Biochemistry* 31:7120–7125, 1992.
- [33828] 7380.Alper, C. A.; Marcus, D.; Raum, D.; Petersen, B. H.; Spira, T.J.: Genetic polymorphism in C8 beta-chains: evidence for two unlinked genetic loci for the eighth component of human complement (C8). *J.Clin. Invest.* 72: 1526–1531, 1983.
- [33829] 7381.Densen, P.; Brown, E. J.; O'Neill, G. J.; Tedesco, F.; Clark, R.A.; Frank, M. M.; Webb, D.; Myers, J.: Inherited deficiency of C8 in a patient with recurrent meningococcal infections: further evidence for a dysfunctional C8 molecule and nonlinkage to the HLA system. *J.Clin. Immun.* 3: 90–99, 1983.
- [33830] 7382.Giraldo, G.; Degos, L.; Beth, E.; Sasportes, M.; Marcelli, A.; Gharbi, R.; Day, N. K.: C8 deficiency in a family with xeroderma pigmentosum: lack of linkage to HLA region. *Clin. Immun. Immunopath.* 8:377–384, 1977.
- [33831] 7383.Jasin, H. E.: Absence of the eighth component of complement in association with systemic lupus erythematosus-like disease. *J. Clin. Invest.* 60: 709–715, 1977.
- [33832] 7384.Kolb, W. P.; Muller-Eberhard, H. J.: The membrane

attack mechanism of complement: the three polypeptide chain structure of the eighth component (C8). *J. Exp. Med.* 143: 1131–1139, 1976.

- [33833] 7385. Komatsu, M.; Imaoka, K.; Satoh, M.; Mikami, H.: Hereditary C8- α - γ deficiency associated with dwarfism in the rabbit. *J. Hered.* 81:413–417, 1990.
- [33834] 7386. Komatsu, M.; Yamamoto, K.; Kawashima, T.; Migita, S.: Genetic deficiency of the α - γ -subunit of the eighth complement component in the rabbit. *J. Immun.* 134: 2607–2609, 1985.
- [33835] 7387. Marcus, D.; Spira, T. J.; Petersen, B. H.; Raum, D.; Alper, C. A.: There are two unlinked genetic loci for human C8. (Abstract) *Molec. Immun.* 19: 1385 only, 1982.
- [33836] 7388. Matthews, N.; Stark, J. M.; Harper, P. S.; Doran, J.; Jones, D. M.: Recurrent meningococcal infections associated with a functional deficiency of the C8 component of human complement. *Clin. Exp. Immun.* 39:53–59, 1980.
- [33837] 7389. Merritt, A. D.; Petersen, B. H.; Biegel, A. A.; Meyers, D. A.; Brooks, G. F.; Hodes, M. E.: Chromosome 6: linkage of the eighth component of complement (C8) to the histocompatibility region (HLA). *Cytogenet. Cell Genet.* 16: 331–334, 1976.
- [33838] 7390. Michelotti, G. A.; Snider, J. V.; Sodetz, J. M.: Genomic

organization of human complement protein C8-alpha and further examination of its linkage to C8-beta. Hum. Genet. 95: 513-518, 1995.

- [33839] 7391. Nakamura, S.; Ohue, O.; Abe, K.: Genetic polymorphism of human complement component C81 in the Japanese population. Hum. Genet. 72:344-347, 1986.
- [33840] 7392. Pericak-Vance, M. A.; Elston, R. C.; Spira, T. J.; Band, J.: Segregation and linkage analysis of immunochemical C8 levels in a family with C8 beta-chain deficiency. (Abstract) Am. J. Hum. Genet. 34:109A only, 1982.
- [33841] 7393. Petersen, B. H.; Graham, J. A.; Brooks, G. F.: Human deficiency of the eighth component of complement: the requirement of C8 for serum *Neisseria gonorrhoeae* bactericidal activity. J. Clin. Invest. 57:283-290, 1976.
- [33842] 7394. Pickering, R. J.; Rynes, R. I.; Lo Cascio, N.; Monahan, J. B.; Sodetz, J. M.: Identification of the alpha-gamma subunit of the eighth component of complement (C8) in a patient with systemic lupus erythematosus and absent C8 activity: patient and family studies. Clin. Immun. Immunopath. 23: 323-334, 1982.
- [33843] 7395. Rittner, C.; Hargesheimer, W.; Mollenhauer, E.: Population and formal genetics of the human C81(alpha-gamma) polymorphism. Hum. Genet. 67:

166–169, 1984.

- [33844] 7396. Rittner, C.; Hargesheimer, W.; Stradmann, B.; Bertrams, J.; Baur, M. P.; Petersen, B. H.: Human C81 (alpha-gamma) polymorphism: detection in the alpha-gamma subunit on SDS-PAGE, formal genetics and linkage relationship. *Am. J. Hum. Genet.* 38: 482–491, 1986.
- [33845] 7397. Rogde, S.; Gedde-Dahl, T., Jr.; Teisberg, P.; Jonassen, R.; Hoyheim, B.; Olaisen, B.: Linkage and association studies with C8A and C8B RFLPs on chromosome 1. *Ann. Hum. Genet.* 56: 233–242, 1992.
- [33846] 7398. Rogde, S.; Mevag, B.; Olaisen, B.; Gedde-Dahl, T., Jr.; Teisberg, P.: Structural genes for complement factor C8 on chromosome 1. (Abstract) *Cytogenet. Cell Genet.* 37: 571 only, 1984.
- [33847] 7399. Rogde, S.; Mevag, B.; Teisberg, P.; Gedde-Dahl, T., Jr.; Tedesco, F.; Olaisen, B.: Genetic polymorphism of complement component C8. *Hum. Genet.* 70: 211–216, 1985.
- [33848] 7400. Rogde, S.; Olaisen, B.; Gedde-Dahl, T., Jr.; Teisberg, P.: Two complement component C8 loci are localized between PGM1 and Rh on chromosome 1. (Abstract) *Cytogenet. Cell Genet.* 40: 734–735, 1985.
- [33849] 7401. Rogde, S.; Olaisen, B.; Gedde-Dahl, T., Jr.; Teisberg, P.: The C8A and C8B loci are closely linked on chromo-

some 1. Ann. Hum. Genet. 50:139–144, 1986.

- [33850] 7402. Tedesco, F.; Densen, P.; Villa, M. A.; Petersen, B. H.; Sirchia, G.: Two types of dysfunctional eighth component of complement (C8) molecules in C8 deficiency in man: reconstitution of normal C8 from the mixture of two abnormal C8 molecules. J. Clin. Invest. 71: 183–191, 1983.
- [33851] 7403. Quax, W.; Meera Khan, P.; Quax-Jeuken, Y.; Bloemendal, H.: The human desmin and vimentin genes are located on different chromosomes. Gene 38:189–196, 1985.
- [33852] 7404. Reynolds, F. H., Jr.; Todaro, G. J.; Fryling, C.; Stephenson, J. R.: Human transforming growth factors induce tyrosine phosphorylation of EGF receptors. Nature 292: 259–262, 1981.
- [33853] 7405. Shimizu, N.; Behzadian, M. A.; Shimizu, Y.: Genetics of cell surface receptors for bioactive polypeptides: binding of epidermal growth factor is associated with the presence of human chromosome 7 in human–mouse cell hybrids. Proc. Nat. Acad. Sci. 77: 3600–3604, 1980.
- [33854] 7406. Sibilio, M.; Fleischmann, A.; Behrens, A.; Stingl, L.; Carroll, J.; Watt, F. M.; Schlessinger, J.; Wagner, E. F.: The EGF receptor provides an essential survival signal for SOS-dependent skin tumor development. Cell 102: 211–220,

2000.

- [33855] 7407. Silver, J.; Whitney, J. B., III; Kozak, C.; Hollis, G.; Kirsch, I.: Erbb is linked to the alpha-globin locus on mouse chromosome 11. *Molec. Cell. Biol.* 5: 1784–1786, 1985.
- [33856] 7408. Spurr, N. K.; Goodfellow, P. N.; Solomon, E.; Parkar, M.; Vennstrom, B.; Bodmer, W. F.: Mapping of cellular oncogenes; erb B on chromosome 7. (Abstract) *Cytogenet. Cell Genet.* 37: 590 only, 1984.
- [33857] 7409. Spurr, N. K.; Solomon, E.; Jansson, M.; Sheer, D.; Goodfellow, P. N.; Bodmer, W. F.; Vennstrom, B.: Chromosomal localisation of the human homologues to the oncogenes erbA and B. *EMBO J.* 3: 159–163, 1984.
- [33858] 7410. Thaung, C.; West, K.; Clark, B. J.; McKie, L.; Morgan, J. E.; Arnold, K.; Nolan, P. M.; Peters, J.; Hunter, A. J.; Brown, S. D. M.; Jackson, I. J.; Cross, S. H.: Novel ENU-induced eye mutations in the mouse: models for human eye disease. *Hum. Molec. Genet.* 11: 755–767, 2002.
- [33859] 7411. Ullrich, A.; Coussens, L.; Hayflick, J. S.; Dull, T. J.; Gray, A.; Tam, A. W.; Lee, J.; Yarden, Y.; Libermann, T. A.; Schlessinger, J.; Downward, J.; Mayes, E. L. V.; Whittle, N.; Waterfield, M. D.; Seeburg, P. H.: Human epidermal growth factor receptor cDNA sequence and aberrant expression of

the amplified gene in A431 epidermoid carcinomacells.

Nature 309: 418–425, 1984.

[33860] 7412.Verveer, P. J.; Wouters, F. S.; Reynolds, A. R.; Bastiaens, P.I. H.: Quantitative imaging of lateral ErbB1 receptor signal propagationin the plasma membrane. Science 290: 1567–1570, 2000.

[33861] 7413.Wakeling, E. L.; Abu–Amero, S. N.; Stanier, P.; Preece, M. A.;Moore, G. E.: Human EGFR, a candidate gene for the Silver–Russellsyndrome, is biallelically expressed in a wide range of fetal tissues. Europ.J. Hum. Genet. 6: 158–164, 1998.

[33862] 7414.Yang, E.–B.; Wang, D.–F.; Mack, P.; Cheng, L.–Y.: Genistein,a tyrosine kinase inhibitor, reduces EGF–induced EGF receptor internalizationand degradation in human hepatoma HepG2 cells. Biochem. Biophys.Res. Commun. 224: 309–317, 1996.

[33863] 7415.Baumann, C. A.; Ribon, V.; Kanzaki, M.; Thurmond, D. C.; Mora,S.; Shigematsu, S.; Bickel, P. E.; Pessin, J. E.; Saltiel, A. R.:CAP defines a second signalling pathway required for insulin–stimulatedglucose transport. Nature 407: 202–207, 2000.

[33864] 7416.Bickel, P. E.; Scherer, P. E.; Schnitzer, J. E.; Oh, P.; Lisanti,M. P.; Lodish, H. F.: Flotillin and epidermal surface

antigen define a new family of caveolae-associated integral membrane proteins. J.Biol. Chem. 272: 13793–13802, 1997.

- [33865] 7417.Cho, Y.-J.; Chema, D.; Moskow, J. J.; Cho, M.; Schroeder, W. T.; Overbeek, P.; Buchberg, A. M.; Duvic, M.: Epidermal surface antigen(MS17S1) is highly conserved between mouse and human. Genomics 27:251–258, 1995.
- [33866] 7418.Kayes, L. M.; Schroeder, W. T.; Marchuk, D. A.; Collins, F. S.; Riccardi, V. M.; Duvic, M.; Stephens, K.: The gene for a novel epidermal antigen maps near the neurofibromatosis 1 gene. Genomics 14: 369–376, 1992.
- [33867] 7419.Schroeder, W. T.; Siciliano, M. J.; Stewart-Galetka, S. L.; Duvic, M.: The human gene for an epidermal surface antigen (M17S1) is located at 17q11–12. Genomics 11: 481–482, 1991.
- [33868] 7420.Schroeder, W. T.; Stephens, K.; Stewart-Galetka, S.; Riccardi, V.; Duvic, M.: The gene for an epidermal surface antigen is in close proximity to the locus for von Recklinghausen neurofibromatosis. (Abstract) Clin.Res. 39: 323A only, 1991.
- [33869] 7421.Cohn, R. D.; Henry, M. D.; Michele, D. E.; Barresi, R.; Saito, F.; Moore, S. A.; Flanagan, J. D.; Skwarchuk, M. W.; Robbins, M. E.; Mendell, J. R.; Williamson, R. A.; Campbell,

K. P.: Disruption ofDag1 in differentiated skeletal muscle reveals a role for dystroglycanin muscle regeneration. Cell 110: 639–648, 2002.

[33870] 7422.Matsuoka, R.; Yoshida, M. C.; Furutani, Y.; Imamura, S.; Kanda,N.; Yanagisawa, M.; Masaki, T.; Takao, A.: Human smooth muscle myosinheavy chain gene mapped to chromosomal region 16q12. Am. J. Med.Genet. 46: 61–67, 1993.

[33871] 7423.Apperley, J. F.; Gardembas, M.; Melo, J. V.; Russell-Jones, R.;Bain, B. J.; Baxter, E. J.; Chase, A.; Chessells, J. M.; Colombat,M.; Dearden, C. E.; Dimitrijevic, S.; Mahon, F.-X.; Marin, D.; Nikolova,Z.; Olavarria, E.; Silberman, S.; Schultheis, B.; Cross, N. C. P.;Goldman, J. M.: Response to imatinib mesylate in patients with chronicmyeloproliferative diseases with rearrangements of the platelet-derivedgrowth factor receptor beta. New Eng. J. Med. 347: 481–487, 2002.

[33872] 7424.Matsuoka, R.; Yoshida, M. C.; Kanda, N.; Furutani, Y.; Bruns, G.;Yanagisawa, M.; Masaki, T.; Takao, A.: Human smooth muscle myosinheavy-chain gene mapped to chromosomal region 16q12.1–q12.2. (Abstract) Cyto-genet.Cell Genet. 58: 2000–2001, 1991.

[33873] 7425.Spear, P. G.: A welcome mat for leprosy and Lassa

fever. *Science* 282:1999–2000, 1998.

- [33874] 7426. Tinsley, J. M.; Blake, D. J.; Zuellig, R. A.; Davies, K. E.: Increasing complexity of the dystrophin-associated protein complex. *Proc. Nat. Acad. Sci.* 91: 8307–8313, 1994.
- [33875] 7427. Williamson, R. A.; Henry, M. D.; Daniels, K. J.; Hrstka, R. F.; Lee, J. C.; Sunada, Y.; Ibraghimov-Beskrovnaya, O.; Campbell, K. P.: Dystroglycan is essential for early embryonic development: disruption of Reichert's membrane in *Dag1*-null mice. *Hum. Molec. Genet.* 6:831–841, 1997.
- [33876] 7428. Yamada, H.; Denzer, A. J.; Hori, H.; Tanaka, T.; Anderson, L. V. B.; Fujita, S.; Fukuta-Ohi, H.; Shimizu, T.; Ruegg, M. A.; Matsumura, K.: Dystroglycan is a dual receptor for agrin and laminin-2 in Schwann cell membrane. *J. Biol. Chem.* 271: 23418–23423, 1996.
- [33877] 7429. Cote, P. D.; Moukhles, H.; Lindenbaum, M.; Carbonetto, S.: Chimeric mice deficient in dystroglycans develop muscular dystrophy and have disrupted myoneural synapses. *Nature Genet.* 23: 338–342, 1999.
- [33878] 7430. Gee, S. H.; Montanaro, F.; Lindenbaum, M. H.; Carbonetto, S.: Dystroglycan- α , a dystrophin-associated glycoprotein, is a functional agrin receptor. *Cell* 77: 675–686, 1994.

- [33879] 7431.Gorecki, D. C.; Derry, J. M. J.; Barnard, E. A.: Dystroglycan:brain localisation and chromosome mapping in the mouse. *Hum. Molec.Genet.* 3: 1589–1597, 1994.
- [33880] 7432.Hayashi, Y. K.; Ogawa, M.; Tagawa, K.; Noguchi, S.; Ishihara, T.;Nonaka, I.; Arahata, K.: Selective deficiency of alpha–dystroglycanin Fukuyama–type congenital muscular dystrophy. *Neurology* 57: 115–121,2001.
- [33881] 7433.Henry, M. D.; Campbell, K. P.: A role for dystroglycan in basementmembrane assembly. *Cell* 95: 859–970, 1998.
- [33882] 7434.Ibraghimov–Beskrovnaya, O.; Ervasti, J. M.; Leveille, C. J.; Slaughter,C. A.; Sernett, S. W.; Campbell, K. P.: Primary structure of dystrophin–associatedglycoproteins linking dystrophin to the extracellular matrix. *Nature* 355:696–702, 1992.
- [33883] 7435.Ibraghimov–Beskrovnaya, O.; Milatovich, A.; Ozcelik, T.; Yang,B.; Francke, U.; Campbell, K. P.: Dystroglycan: tissue distribution,human muscle cDNA, genomic structure and chromosome mapping. (Abstract) *Am.J. Hum. Genet.* 51 (suppl.): A130 only, 1992.
- [33884] 7436.Ibraghimov–Beskrovnaya, O.; Milatovich, A.; Ozcelik, T.; Yang,B.; Koepnick, K.; Francke, U.; Campbell, K. P.: Human dystroglycan:skeletal muscle cDNA, genomic struc–

ture, origin of tissue specific isoforms and chromosomal localization. Hum. Molec. Genet. 2: 1651–1657, 1993.

- [33885] 7437. Ma, J.; Nastuk, M. A.; McKechnie, B. A.; Fallon, J. R.: The agrin receptor: localization in the postsynaptic membrane, interaction with agrin, and relationship to the acetylcholine receptor. J. Biol. Chem. 268:25108–25117, 1993.
- [33886] 7438. Matsumura, K.; Nonaka, I.; Campbell, K. P.: Abnormal expression of dystrophin-associated proteins in Fukuyama-type congenital muscular dystrophy. Lancet 341: 521–522, 1993.
- [33887] 7439. Matsumura, K.; Tome, F. M. S.; Collin, H.; Azibi, K.; Chaouch, M.; Kaplan, J.-C.; Fardeau, M.; Campbell, K. P.: Deficiency of the 50K dystrophin-associated glycoprotein in severe childhood autosomal recessive muscular dystrophy. Nature 359: 320–322, 1992.
- [33888] 7440. Matsumura, K.; Tome, F. M. S.; Ionasescu, V.; Ervasti, J. M.; Anderson, R. D.; Romero, N. B.; Simon, D.; Recan, D.; Kaplan, J.-C.; Fardeau, M.; Campbell, K. P.: Deficiency of dystrophin-associated proteins in Duchenne muscular dystrophy patients lacking COOH-terminal domains of dystrophin. J. Clin. Invest. 92: 866–871, 1993.

- [33889] 7441.Michele, D. E.; Barresi, R.; Kanagawa, M.; Saito, F.; Cohn, R.D.; Satz, J. S.; Dollar, J.; Nishino, I.; Kelley, R. I.; Somer, H.;Straub, V.; Mathews, K. D.; Moore, S. A.; Campbell, K. P.: Post-translational disruption of dystroglycan-ligand interactions in congenital muscular dystrophies. *Nature* 418: 417-422, 2002.
- [33890] 7442.Moore, S. A.; Saito, F.; Chen, J.; Michele, D. E.; Henry, M. D.;Messing, A.; Cohn, R. D.; Ross-Barta, S. E.; Westra, S.; Williamson,R. A.; Hoshi, T.; Campbell, K. P.: Deletion of brain dystroglycan recapitulates aspects of congenital muscular dystrophy. *Nature* 418:422-425, 2002.
- [33891] 7443.Rambukkana, A.; Yamada, H.; Zanazzi, G.; Mathus, T.; Salzer, J.L.; Yurchenco, P. D.; Campbell, K. P.; Fischetti, V. A.: Role of alpha-dystroglycan as a Schwann cell receptor for *Mycobacterium leprae*. *Science* 282:2076-2078, 1998.
- [33892] 7444.Sealock, R.; Froehner, S. C.: Dystrophin-associated proteins and synapse formation: is alpha-dystroglycan the agrin receptor? *Cell* 77:617-619, 1994.
- [33893] 7445.Yamada, H.; Saito, F.; Fukuta-Ohi, H.; Zhong, D.; Hase, A.; Arai,K.; Okuyama, A.; Maekawa, R.; Shimizu, T.; Matsumura, K.: Processing of beta-dystroglycan by matrix

metalloproteinase disrupts the link between the extracellular matrix and cell membrane via the dystroglycan complex. *Hum. Molec. Genet.* 10: 1563–1569, 2001.

- [33894] 7446. Blake, D. J.; Love, D. R.; Tinsley, J.; Morris, G. E.; Turley, H.; Gatter, K.; Dickson, G.; Edwards, Y. H.; Davies, K. E.: Characterization of a 4.8kb transcript from the Duchenne muscular dystrophy locus expressed in schwannoma cells. *Hum. Molec. Genet.* 1: 103–109, 1992.
- [33895] 7447. Golub, T. R.; Barker, G. F.; Lovett, M.; Gilliland, D. G.: Fusion of PDGF receptor beta to a novel ets-like gene, tel, in chronic myelomonocytic leukemia with t(5;12) chromosomal translocation. *Cell* 77: 307–316, 1994.
- [33896] 7448. Brissenden, J. E.; Ullrich, A.; Francke, U.: Chromosomal mapping of loci for insulin-like growth factors I and II and for epidermal growth factor in man. (Abstract) *Am. J. Hum. Genet.* 36: 133S only, 1984.
- [33897] 7449. Brissenden, J. E.; Ullrich, A.; Francke, U.: Human chromosomal mapping of genes for insulin-like growth factors I and II and epidermal growth factor. *Nature* 310: 781–784, 1984.
- [33898] 7450. Carpenter, G.; Cohen, S.: Epidermal growth factor. *Ann. Rev. Biochem.* 48: 193–216, 1979.
- [33899] 7451. Cohen, S.: Isolation of a mouse submaxillary gland

protein accelerating incisor eruption and eyelid opening in the new-born animal. J. Biol.Chem. 237: 1555–1562, 1962.

[33900] 7452.Gray, A.; Dull, T. J.; Ullrich, A.: Nucleotide sequence of epidermal growth factor cDNA predicts a 128,000–molecular weight protein precursor. Nature 303:722–725, 1983.

[33901] 7453.Morton, C. C.; Byers, M. G.; Nakai, H.; Bell, G. I.; Shows, T.B.: Human genes for insulin–like growth factors I and II and epidermal growth factor are located on 12q22–q24.1, 11p15, and 4q25–q27, respectively. Cytogenet.Cell Genet. 41: 245–249, 1986.

[33902] 7454.Sassone–Corsi, P.; Mizzen, C. A.; Cheung, P.; Crosjo, C.; Monaco,L.; Jacquot, S.; Hanauer, A.; Allis, C. D.: Requirement of Rsk–2 for epidermal growth factor–activated phosphorylation of histone H3. Science 285:886–891, 1999.

[33903] 7455.Scott, A. F.: Personal Communication. Baltimore, Md. 10/11/1999.

[33904] 7456.Shahbazi, M.; Pravica, V.; Nasreen, N.; Fakhoury, H.; Fryer, A.A.; Strange, R. C.; Hutchinson, P. E.; Osborne, J. E.; Lear, J. T.;Smith, A. G.; Hutchinson, I. V.: Association between functional polymorphisms in EGF gene and malig–

nant melanoma. Lancet 359: 397–401, 2002.

- [33905] 7457. Smith, J.; Cook, E.; Fotheringham, I.; Pheby, S.; Derbyshire, R.; Eaton, M. A. W.; Doel, M.; Lilley, D. M. J.; Pardon, J. F.; Patel, T.; Lewis, H.; Bell, L. D.: Chemical synthesis and cloning of a gene for human beta-urogastrone. Nucleic Acids Res. 10: 4467–4482, 1982.
- [33906] 7458. Sudhof, T. C.; Russell, D. W.; Goldstein, J. L.; Brown, M. S.; Sanchez-Pescador, R.; Bell, G. I.: Cassette of eight exons shared by genes for LDL receptor and EGF precursor. Science 228: 893–895, 1985.
- [33907] 7459. Tsutsumi, O.; Kurachi, H.; Oka, T.: A physiological role of epidermal growth factor in male reproductive function. Science 233: 975–977, 1986.
- [33908] 7460. Urdea, M. S.; Merryweather, J. P.; Mullenbach, G. T.; Coit, D.; Heberlein, U.; Valenzuela, P.; Barr, P. J.: Chemical synthesis of a gene for human epidermal growth factor urogastrone and its expression in yeast. Proc. Nat. Acad. Sci. 80: 7461–7465, 1983.
- [33909] 7461. Zabel, B. U.; Eddy, R. L.; Lalley, P. A.; Scott, J.; Bell, G. I.; Shows, T. B.: Chromosomal locations of the human and mouse genes for precursors of epidermal growth factor and the beta subunit of nerve growth factor. Proc. Nat. Acad. Sci. 82: 469–473, 1985.

- [33910] 7462. Aden, D. P.; Knowles, B. B.: Cell surface antigens coded for by the human chromosome 7. *Immunogenetics* 3: 209–211, 1976.
- [33911] 7463. Carlin, C. R.; Aden, D. P.; Knowles, B. B.: S6 is the human receptor for epidermal growth factor (EGF). (Abstract) *Cytogenet. Cell Genet.* 32:256 only, 1982.
- [33912] 7464. Carlin, C. R.; Knowles, B. B.: Identity of human epidermal growth factor (EGF) receptor with glycoprotein SA-7: evidence for differential phosphorylation of the two components of the EGF receptor from A431 cells. *Proc. Nat. Acad. Sci.* 79: 5026–5030, 1982.
- [33913] 7465. Carpenter, G.: Properties of the receptor for epidermal growth factor. *Cell* 37: 357–358, 1984.
- [33914] 7466. Chen, B.; Bronson, R. T.; Klamann, L. D.; Hampton, T. G.; Wang, J.; Green, P. J.; Magnuson, T.; Douglas, P. S.; Morgan, J. P.; Neel, B. G.: Mice mutant for *Egfr* and *Shp2* have defective cardiac semilunar valvulogenesis. *Nature Genet.* 24: 296–299, 2000.
- [33915] 7467. Ishii, H.; Baffa, R.; Numata, S.-I.; Murakumo, Y.; Rattan, S.; Inoue, H.; Mori, M.; Fidanza, V.; Alder, H.; Croce, C. M.: The FEZ1 gene at chromosome 8p22 encodes a leucine-zipper protein, and its expression is altered in multiple human tumors. *Proc. Nat. Acad. Sci.* 96:

3928–3933, 1999.

- [33916] 7468.Kuroki, T.; Trapasso, F.; Shiraishi, T.; Alder, H.; Mimi, K.;Mori, M.; Croce, C. M.: Genetic alterations of the tumor suppressorgene WWOX in esophageal squamous cell carcinoma. *Cancer Res.* 62:2258–2260, 2002.
- [33917] 7469.Li, G.; Hu, N.; Goldstein, A. M.; Tang, Z.–Z.; Roth, M. J.; Wang,Q.–H.; Dawsey, S. M.; Han, X.–Y.; Ding, T.; Huang, J.; Giffen, C.;Taylor, P. R.; Emmert–Buck, M. R.: Allelic loss on chromosome bands13q11–q13 in esophageal squamous cell carcinoma. *Genes ChromosomesCancer* 31: 390–397, 2001.
- [33918] 7470.Lo, H. S.; Hu, N.; Gere, S.; Lu, N.; Su, H.; Goldstein, A. M.;Taylor, P. R.; Lee, M. P.: Identification of somatic mutations ofthe RNF6 gene in human esophageal squamous cell carcinoma. *CancerRes.* 62: 4191–4193, 2002.
- [33919] 7471.Su, P.–H.; Hou, J.–W.; Hwu, W.–L.; Wu, M.–H.; Wang, J.–K.; Wang,T.–R.: Congenital contractural arachnodactyly (Beals syndrome). *ActaPaediat.* 41: 59–62, 2000.
- [33920] 7472.Viljoen, D.: Congenital contractural arachnodactyly (Beals syndrome). *J.Med. Genet.* 31: 640–643, 1994.
- [33921] 7473.Viljoen, D.; Ramesar, R.; Behari, D.: Beals syndrome: clinicaland molecular investigations in a kindred of Indian descent. *Clin.Genet.* 39: 181–188, 1991.

- [33922] 7474.Wang, M.; Clericuzio, C. L.; Godfrey, M.: Familial occurrence of typical and severe lethal congenital contractural arachnodactyly caused by missplicing of exon 34 of fibrillin-2. *Am. J. Hum. Genet.* 59:1027-1034, 1996.
- [33923] 7475.Wang, M.; Price, C. E.; Han, J.; Cisler, J.; Imaizumi, K.; VanThienen, M. N.; DePaepe, A.; Godfrey, M.: Recurrent mis-splicing of fibrillin exon 32 in two patients with neonatal Marfan syndrome. *Hum.Molec. Genet.* 4: 607-613, 1995.
- [33924] 7476.Wang, M.; Tsipouras, P.; Godfrey, M.: Fibrillin-2 (FBN2) mutation in congenital contractural arachnodactyly. (Abstract) *Am. J. Hum.Genet.* 57: A231, 1995.
- [33925] 7477.Zhang, H.; Apfelroth, S. D.; Hu, W.; Davis, E. C.; Sanguineti, C.; Bonadio, J.; Mecham, R. P.; Ramirez, F.: Structure and expression of fibrillin-2, a novel microfibrillar component preferentially located in elastic matrices. *J. Cell Biol.* 124: 855-863, 1994.
- [33926] 7478.Zhang, H.; Hu, W.; Ramirez, F.: Developmental expression of fibrillin genes suggests heterogeneity of extracellular microfibrils. *J. Cell Biol.* 129: 1165-1176, 1995.
- [33927] 7479.Beck, C.; Moulard, B.; Steinlein, O.; Guipponi, M.; Vallee, L.; Montpied, P.; Baldy-Moulinier, M.; Malafosse, A.: A nonsense mutation in the alpha-4 subunit of the nico-

tinic acetylcholine receptor (CHRNA4) cosegregates with 20q-linked benign neonatal familial convulsions (EBN1). *Neurobiol. Dis.* 1: 95–99, 1994.

- [33928] 7480. Singh, N. A.; Charlier, C.; Stauffer, D.; DuPont, B. R.; Leach, R. J.; Melis, R.; Ronen, G. M.; Bjerre, I.; Quattlebaum, T.; Murphy, J. V.; McHarg, M. L.; Gagnon, D.; Rosales, T. O.; Peiffer, A.; Anderson, V. E.; Leppert, M.: A novel potassium channel gene, KCNQ2, is mutated in an inherited epilepsy of newborns. *Nature Genet.* 18: 25–29, 1998.
- [33929] 7481. Charlier, C.; Singh, N. A.; Ryan, S. G.; Lewis, T. B.; Reus, B. E.; Leach, R. J.; Leppert, M.: A pore mutation in a novel KQT-like potassium channel gene in an idiopathic epilepsy family. *Nature Genet.* 18: 53–55, 1998.
- [33930] 7482. Ryan, S. G.; Wiznitzer, M.; Hollman, C. H.; Torres, M. C.; Szekeresova, M.; Schneider, S.: Benign familial neonatal convulsions: evidence for clinical and genetic heterogeneity. *Ann. Neurol.* 29: 469–473, 1991.
- [33931] 7483. Earnshaw, W. C.: When is a centromere not a kinetochore? *J. Cell Sci.* 99: 1–4, 1991.
- [33932] 7484. Fowler, K. J.; Newson, A. J.; MacDonald, A. C.; Kalitsis, P.; Lyu, M. S.; Kozak, C. A.; Choo, K. H. A.: Chromosomal localization of mouse Cenpa gene. *Cytogenet. Cell Genet.* 79: 298–301, 1997.

- [33933] 7485.Howman, E. V.; Fowler, K. J.; Newson, A. J.; Redward, S.; MacDonald,A. C.; Kalitsis, P.; Choo, K. H. A.: Early disruption of centromericchromatin organization in centromere protein A (Cenpa) null mice. *Proc.Nat. Acad. Sci.* 97: 1148–1153, 2000.
- [33934] 7486.Palmer, D. K.; O'Day, K.; Trong, H. L.; Charbonneau, H.; Margolis,R. L.: Purification of the centromere–specific protein CENP–A anddemonstration that it is a distinctive histone. *Proc. Nat. Acad.Sci.* 88: 3734–3738, 1991.
- [33935] 7487.Sullivan, K. F.; Hechenberger, M.; Masri, K.: Human CENP–A containsa histone H3 related histone fold domain that is required for targetingto the centromere. *J. Cell Biol.* 127: 581–592, 1994.
- [33936] 7488.Earnshaw, W. C.; Sullivan, K. F.; Machlin, P. S.; Cooke, C. A.;Kaiser, D. A.; Pollard, T. D.; Rothfield, N. F.; Cleveland, D. W.: Molecular cloning of cDNA for CENP–B, the major human centromereautoantigen. *J. Cell Biol.* 104: 817–829, 1987.
- [33937] 7489.Seki, N.; Saito, T.; Kitagawa, K.; Masumoto, H.; Okazaki, T.; Hori,T.–A.: Mapping of the human centromere protein B gene (CENPB) tochromosome 20p13 by fluorescence in situ hybridization. *Genomics* 24:187–188, 1994.

- [33938] 7490.Sugimoto, K.; Yata, H.; Himeno, M.: Mapping of the human CENP-B gene to chromosome 20 and the CENP-C gene to chromosome 12 by a rapidcycle DNA amplification procedure. *Genomics* 17: 240–242, 1993.
- [33939] 7491.Earnshaw, W.; Bordwell, B.; Marino, C.; Rothfield, N.: Three human chromosomal autoantigens are recognized by sera from patients with anti-centromere antibodies. *J. Clin. Invest.* 77: 426–430, 1986.
- [33940] 7492.Earnshaw, W. C.; Ratrie, H., III; Stetten, G.: Visualization of centromere proteins CENP-B and CENP-C on a stable dicentric chromosome in cytological spreads. *Chromosoma* 98: 1–12, 1989.
- [33941] 7493.Earnshaw, W. C.; Rothfield, N.: Identification of a family of human centromere proteins using autoimmune sera from patients with scleroderma. *Chromosoma* 91: 313–321, 1985.
- [33942] 7494.Jones, C.; Nguyen, L.; Burkin, D.; McGrew, J.; Tomkiel, J.; Earnshaw, W.: Localization of centromere autoantigen C (CENPC) to human chromosome 12. (Abstract) Human Genome Mapping Workshop 93 25 only, 1993.
- [33943] 7495.McKay, S.; Thomson, E.; Cooke, H.: Sequence homologies and linkage group conservation of the human and mouse Cenpc genes. *Genomics* 22:36–40, 1994.

- [33944] 7496. Page, S. L.; Earnshaw, W. C.; Choo, K. H. A.; Shaffer, L. G.: Further evidence that CENP-C is a necessary component of active centromeres: studies of a dic(X;15) with simultaneous immunofluorescence and FISH. *Hum. Molec. Genet.* 4: 289-294, 1995.
- [33945] 7497. Saitoh, H.; Tomkiel, J.; Cooke, C. A.; Ratrie, H., III; Maurer, M.; Rothfield, N. F.; Earnshaw, W. C.: CENP-C, an autoantigen in scleroderma, is a component of the human inner kinetochore plate. *Cell* 70:115-125, 1992.
- [33946] 7498. Gaynor, R. B.; Muchardt, C.; Xia, Y.; Klisak, I.; Mohandas, T.; Sparkes, R. S.; Lusk, A. J.: Localization of the gene for the DNA-binding protein AP-2 to human chromosome 6p22.3-pter. *Genomics* 10: 1100-1102, 1991.
- [33947] 7499. Schorle, H.; Meier, P.; Buchert, M.; Jaenisch, R.; Mitchell, P. J.: Transcription factor AP-2 essential for cranial closure and craniofacial development. *Nature* 381: 235-238, 1996.
- [33948] 7500. Warren, G.; Gordon, M.; Siracusa, L. D.; Buchberg, A. M.; Williams, T.: Physical and genetic localization of the gene encoding the AP-2 transcription factor to mouse chromosome 13. *Genomics* 31: 234-237, 1996.
- [33949] 7501. Williams, T.; Admon, A.; Lüscher, B.; Tjian, R.: Cloning and expression of AP-2, a cell-type-specific tran-

scription factor that activates inducible enhancer elements. *Genes Dev.* 2: 1557–1569, 1988.

- [33950] 7502. Williamson, J. A.; Bosher, J. M.; Skinner, A.; Sheer, D.; Williams, T.; Hurst, H. C.: Chromosomal mapping of the human and mouse homologues of two new members of the AP-2 family of transcription factors. *Genomics* 35:262–264, 1996.
- [33951] 7503. Zhang, J.; Hagopian–Donaldson, S.; Serbedzija, G.; Elsemore, J.; Plehn–Dujowich, D.; McMahon, A. P.; Flavell, R. A.; Williams, T.: Neural tube, skeletal and body wall defects in mice lacking transcription factor AP-2. *Nature* 381: 238–241, 1996.
- [33952] 7504. Agarwal, V. R.; Ashanullah, C. I.; Simpson, E. R.; Bulun, S. E.: Alternatively spliced transcripts of the aromatase cytochrome P450 (CYP19) gene in adipose tissue of women. *J. Clin. Endocr. Metab.* 82:70–74, 1997.
- [33953] 7505. Nalabolu, S. R.; Shukla, H.; Nallur, G.; Parimoo, S.; Weissman, S. M.: Genes in a 220–kb region spanning the TNF cluster in human MHC. *Genomics* 31: 215–222, 1996.
- [33954] 7506. Gao, B.; Guo, J.; She, C.; Shu, A.; Yang, M.; Tan, Z.; Yang, X.; Guo, S.; Feng, G.; He, L.: Mutations in IHH, encoding Indian hedgehog, cause brachydactyly type A-1. *Nature Genet.* 28: 386–388, 2001.

- [33955] 7507.DeChiara, T. M.; Kimble, R. B.; Poueymirou, W. T.; Rojas, J.; Masiakowski,P.; Valenzuela, D. M.; Yancopoulos, G. D.: Ror2, encoding a receptor–liketyrosine kinase, is required for cartilage and growth plate development. *NatureGenet.* 24: 271–274, 2000.
- [33956] 7508.Oldridge, M.; Fortuna, A. M.; Maringa, M.; Propping, P.; Mansour,S.; Pollitt, C.; DeChiara, T. M.; Kimble, R. B.; Valenzuela, D. M.;Yancopoulos, G. D.; Wilkie, A. O. M.: Dominant mutations in ROR2,encoding an orphan receptor tyrosine kinase, cause brachydactyly typeB. *Nature Genet.* 24: 375–378, 2000.
- [33957] 7509.De Windt, L. J.; Lim, H. W.; Bueno, O. F.; Liang, Q.; Delling,U.; Braz, J. C.; Glascock, B. J.; Kimball, T. F.; del Monte, F.; Hajjar,R. J.; Molkentin, J. D.: Targeted inhibition of calcineurin attenuatescardiac hypertrophy in vivo. *Proc. Nat. Acad. Sci.* 98: 3322–3327,2001.
- [33958] 7510.Fuentes, J. J.; Genesca, L.; Kingsbury, T. J.; Cunningham, K. W.;Perez–Riba, M.; Estivill, X.; de la Luna, S.: DSCR1, overexpressedin Down syndrome, is an inhibitor of calcineurin–mediated signalingpathways. *Hum. Molec. Genet.* 9: 1681–1690, 2000.
- [33959] 7511.Giri, P.; Higuchi, S.; Kincaid, R. L.: Chromosomal mapping ofthe human genes for the calmodulin–depen–

dent protein phosphatase (calcineurin)catalytic subunit.

Biochem. Biophys. Res. Commun. 181: 252–258,1991.

[33960] 7512.Guerini, D.; Klee, C. B.: Cloning of human calcineurin A: evidencefor two isozymes and identification of a polyproline structural domain. Proc.Nat. Acad. Sci. 86: 9183–9187, 1989.

[33961] 7513.Leinwand, L. A.: Calcineurin inhibition and cardiac hypertrophy:a matter of balance. Proc. Nat. Acad. Sci. 98: 2947–2949, 2001.

[33962] 7514.Malleret, G.; Haditsch, U.; Genoux, D.; Jones, M. W.; Bliss, T.V. P.; Vanhose, A. M.; Weitlauf, C.; Kandel, E. R.; Winder, D. G.;Mansuy, I. M.: Inducible and reversible enhancement of learning,memory, and long-term potentiation by genetic inhibition of calcineurin. Cell 104:675–686, 2001.

[33963] 7515.Mansuy, I. M.; Mayford, M.; Jacob, B.; Kandel, E. R.; Bach, M.E.: Restricted and regulated overexpression reveals calcineurin asa key component in the transition from short-term to long-term memory. Cell 92:39–49, 1998.

[33964] 7516.Rothermel, B. A.; McKinsey, T. A.; Vega, R. B.; Nicol, R. L.; Mammen,P.; Yang, J.; Antos, C. L.; Shelton, J. M.; Bassel-Duby, R.; Olson,E. N.; Williams, R. S.: Myocyte-enriched calcineurin-interactingprotein, MCIP1, inhibits car-

diac hypertrophy in vivo. Proc. Nat.Acad. Sci. 98:
3328–3333, 2001.

[33965] 7517.Seitz, D. P.; Pasha, M. K.; Singh, B.; Chu, A.; Sharma, R. K.: Localization and characterization of calcineurin in bovine eye. Invest.Ophthal. Vis. Sci. 43: 15–21, 2002.

[33966] 7518.Wang, M. G.; Yi, H.; Guerini, D.; Klee, C. B.; McBride, O. W.: Calcineurin A alpha (PPP3CA), calcineurin A beta (PPP3CB) and calcineurinB (PPP3R1) are located on human chromosomes 4, 10q21–q22 and 2p16–p15respectively. Cytogenet. Cell Genet. 72: 236–241, 1996.

[33967] 7519.Winder, D. G.; Mansuy, I. M.; Osman, M.; Moallem, T. M.; Kandel,E. R.: Genetic and pharmacological evidence for a novel, intermediatephase of long–term potentiation suppressed by calcineurin. Cell 92:25–37, 1998.

[33968] 7520.Bueno, O. F.; Wilkins, B. J.; Tymitz, K. M.; Glascock, B. J.; Kimball,T. F.; Lorenz, J. N.; Molkentin, J. D.: Impaired cardiac hypertrophicresponse in calcineurin A–beta–deficient mice. Proc. Nat. Acad. Sci. 99:4586–4591, 2002.

[33969] 7521.Guerini, D.; Krinks, M. H.; Sikela, J. M.; Hahn, W. E.; Klee, C.B.: Isolation and sequence of a cDNA clone for human calcineurinB, the Ca(2+)–binding subunit of the Ca(2+)/calmodulin–stimulatedprotein phosphatase. DNA

8: 675–682, 1989.

- [33970] 7522.Wang, M. G.; Yi, H.; Guerini, D.; Klee, C. B.; McBride, O. W.: Calcineurin A alpha (PPP3CA), calcineurin A beta (PPP3CB) and calcineurinB (PPP3R1) are located on human chromosomes 4, 10q21–q22 and 2p16–p15 respectively. *Cytogenet. Cell Genet.* 72: 236–241, 1996.
- [33971] 7523.Ferrari, S.; Calabretta, B.; deRiel, J. K.; Battini, R.; Ghezzi, F.; Lauret, E.; Griffin, C.; Emanuel, B. S.; Gurrieri, F.; Baserga, R.: Structural and functional analysis of a growth–regulated gene, the human calcyclin. *J. Biol. Chem.* 262: 8325–8332, 1987.
- [33972] 7524.Alevizaki, M.; Stevenson, J. C.; Girgis, S. I.; MacIntyre, I.; Legon, S.: Altered calcitonin gene in a young patient with osteoporosis. *Brit. Med. J.* 298: 1215–1216, 1989.
- [33973] 7525.Amara, S. G.; Jonas, V.; Rosenfeld, M. G.; Ong, E. S.; Evans, R.M.: Alternative RNA processing in calcitonin gene expression generates mRNAs encoding different polypeptide products. *Nature* 298: 240–244, 1982.
- [33974] 7526.Aiyar, N.; Rand, K.; Elshourbagy, N. A.; Zeng, Z.; Adamou, J. E.; Bergsma, D. J.; Li, Y.: A cDNA encoding the calcitonin gene–related peptide type 1 receptor. *J. Biol. Chem.* 271: 11325–11329, 1996.

- [33975] 7527.Fluhmann, B.; Muff, R.; Hunziker, W.; Fischer, J. A.; Born, W.: A human orphan calcitonin receptor-like structure. *Biochem. Biophys. Res. Commun.* 206: 341–347, 1995.
- [33976] 7528.Foord, S. M.; Craig, R. K.: Isolation and characterisation of a human calcitonin-gene-related-peptide receptor. *Europ. J. Biochem.* 170:373–379, 1987.
- [33977] 7529.Foord, S. M.; Marshall, F. H.: RAMPs: accessory proteins for seven transmembrane domain receptors. *Trends Pharm. Sci.* 20: 184–187, 1999.
- [33978] 7530.Kamitani, S.; Asakawa, M.; Shimekake, Y.; Kuwasako, K.; Nakahara, K.; Sakata, T.: The RAMP2/CRLR complex is a functional adrenomedullin receptor in human endothelial and vascular smooth muscle cells. *FEBS Lett.* 448: 111–114, 1999.
- [33979] 7531.Nakazawa, I.; Nakajima, T.; Harada, H.; Ishigami, T.; Umemura, S.; Emi, M.: Human calcitonin receptor-like receptor for adrenomedullin: genomic structure, eight single-nucleotide polymorphisms, and haplotype analysis. *J. Hum. Genet.* 46: 132–136, 2001.
- [33980] 7532.Dolmetsch, R. E.; Pajvani, U.; Fife, K.; Spotts, J. M.; Greenberg, M. E.: Signaling to the nucleus by an L-type calcium channel-calmodulin complex through the MAP ki-

nase pathway. Science 294: 333–339, 2001.

- [33981] 7533. McAlpine, P. J.: Personal Communication. Winnipeg, Manitoba, Canada 2/14/1992.
- [33982] 7534. Powers, P. A.; Gregg, R. G.; Hogan, K.: Linkage mapping of the human gene for the alpha-1 subunit of the cardiac DHP-sensitive Ca^{2+} channel (CACNL1A1) to chromosome 12p13.2–pter using a dinucleotide repeat. Genomics 14: 206–207, 1992.
- [33983] 7535. Powers, P. A.; Gregg, R. G.; Lalley, P. A.; Liao, M.; Hogan, K.: Assignment of the human gene for the alpha-1 subunit of the cardiac DHP-sensitive Ca^{2+} channel (CCHL1A1) to chromosome 12p12–pter. Genomics 10: 835–839, 1991.
- [33984] 7536. Yang, R.; Morosetti, R.; Koeffler, H. P.: Characterization of a second human cyclin A that is highly expressed in testis and in several leukemic cell lines. Cancer Res. 57: 913–920, 1997.
- [33985] 7537. Sartor, H.; Ehlert, F.; Grzeschik, K.-H.; Muller, R.; Adolph, S.: Assignment of two human cell cycle genes, CDC25C and CCNB1, to 5q31 and 5q12, respectively. Genomics 13: 911–912, 1992.
- [33986] 7538. Geng, Y.; Whoriskey, W.; Park, M. Y.; Bronson, R. T.; Medema, R. H.; Li, T.; Weinberg, R. A.; Sicinski, P.: Rescue

of cyclin D1 deficiency by knockin cyclin E. *Cell* 97:
767–777, 1999.

- [33987] 7539. Hinchcliffe, E. H.; Li, C.; Thompson, E. A.; Maller, J. L.; Sluder, G.: Requirement of Cdk2–cyclin E activity for repeated centrosome reproduction in *Xenopus* egg extracts. *Science* 283: 851–854, 1999.
- [33988] 7540. Lew, D. J.; Dulic, V.; Reed, S. I.: Isolation of three novel human cyclins by rescue of G1 cyclin (cln) function in yeast. *Cell* 66:1197–1206, 1991.
- [33989] 7541. Li, H.; Lahti, J. M.; Valentine, M.; Saito, M.; Reed, S. I.; Look, A. T.; Kidd, V. J.: Molecular cloning and chromosomal localization of the human cyclin C (CCNC) and cyclin E (CCNE) genes: deletion of the CCNC gene in human tumors. *Genomics* 32: 253–259, 1996.
- [33990] 7542. Moberg, K. H.; Bell, D. W.; Wahrer, D. C. R.; Haber, D. A.; Hariharan, I. K.: Archipelago regulates cyclin E levels in *Drosophila* and is mutated in human cancer cell lines. *Nature* 413: 311–316, 2001.
- [33991] 7543. Sheaff, R. J.; Groudine, M.; Gordon, M.; Roberts, J. M.; Clurman, B. E.: Cyclin E–CDK2 is a regulator of p27(Kip1). *Genes Dev.* 11:1464–1478, 1997.
- [33992] 7544. Akoulitchiev, S.; Chuikov, S.; Reinberg, D.: TFIIF is negatively regulated by cdk8–containing mediator com–

plexes. *Nature* 407: 102–106,2000.

[33993] 7545.Hanson, R. D.; Hohn, P. A.; Popescu, N. C.; Ley, T. J.: A cluster of hematopoietic serine protease genes is found on the same chromosomal band as the human alpha/delta T-cell receptor locus. *Proc. Nat. Acad.Sci.* 87: 960–963, 1990.

[33994] 7546.Harper, K.; Mattei, M.–G.; Simon, D.; Suzan, M.; Guenet, J.–L.; Haddad, P.; Sasportes, M.; Golstein, P.: Proximity of the CTLA–1 serine esterase and Tcr(alpha) loci in mouse and man. *Immunogenetics* 28:439–444, 1988.

[33995] 7547.Klein, J. L.; Shows, T. B.; Dupont, B.; Trapani, J. A.: Genomic organization and chromosomal assignment for a serine protease gene(CSP–B), expressed by human cytotoxic lymphocytes. *Genomics* 5: 110–117,1989.

[33996] 7548.Lin, C. C.; Meier, M.; Sorensen, O.; Sasi, R.; Tainaka, T.; Bleackley, R. C.: Chromosome localization of two human serine protease genes to region 14q11.2–q12 by in situ hybridization. *Cytogenet. Cell Genet.* 53:169–171, 1990.

[33997] 7549.Motyka, B.; Korbitt, G.; Pinkoski, M. J.; Heibin, J. A.; Caputo, A.; Hobman, M.; Barry, M.; Shostak, I.; Sawchuk, T.; Holmes, C. F.B.; Gauldie, J.; Bleackley, R. C.: Mannose 6–phosphate/insulin–like growth factor II receptor is a

death receptor for granzyme B during cytotoxic T cell-induced apoptosis. *Cell* 103: 491–500, 2000.

- [33998] 7550. Miles, J. S.; Spurr, N. K.; Gough, A. C.; Jowett, T.; McLaren, A. W.; Brook, J. D.; Wolf, C. R.: A novel human cytochrome P450 gene (P450IIB): chromosomal localization and evidence for alternative splicing. *Nucleic Acids Res.* 16: 5783–5795, 1988.
- [33999] 7551. Santisteban, I.; Povey, S.; Shephard, E. A.; Phillips, I. R.: The major phenobarbital-inducible cytochrome P-450 gene subfamily (P450IIB) mapped to the long arm of human chromosome 19. *Ann. Hum. Genet.* 52: 129–135, 1988.
- [34000] 7552. Trask, B.; Fertitta, A.; Christensen, M.; Youngblom, J.; Bergmann, A.; Copeland, A.; de Jong, P.; Mohrenweiser, H.; Olsen, A.; Carrano, A.; Tynan, K.: Fluorescence in situ hybridization mapping of human chromosome 19: cytogenetic band location of 540 cosmids and 70 genes or DNA markers. *Genomics* 15: 133–145, 1993.
- [34001] 7553. Barletta, C.; Batticane, N.; Ragusa, R. M.; Leube, R.; Peschle, C.; Romano, V.: Subchromosomal localization of two human cytochrome genes (KRT4 and KRT15) by in situ hybridization. *Cytogenet. Cell Genet.* 54: 148–150, 1990.
- [34002] 7554. Barletta, C.; Batticane, N.; Ragusa, R. M.; Leube, R.

E.; Franke, W. W.; Peschle, C.; Romano, V.: Sub-chromosomal localization of human cytokeratin 4, 15, and 19 genes. (Abstract) *Cytogenet. Cell Genet.* 51:958 only, 1989.

[34003] 7555. Romano, V.; Bosco, P.; Costa, G.; Leube, R.; Franke, W. W.; Rocchi, M.; Romeo, G.: Chromosomal assignment of cytokeratin genes. (Abstract) *Cytogenet. Cell Genet.* 46: 683 only, 1987.

[34004] 7556. Romano, V.; Bosco, P.; Rocchi, M.; Costa, G.; Leube, R. E.; Franke, W. W.; Romeo, G.: Chromosomal assignments of human type I and type II cytokeratin genes to different chromosomes. *Cytogenet. Cell Genet.* 48:148–151, 1988.

[34005] 7557. Schrijver, H. M.; Crusius, J. B. A.; Uitdehaag, B. M. J.; GarciaGonzalez, M. A.; Kostense, P. J.; Polman, C. H.; Pena, A. S.: Association of interleukin-1-beta and interleukin-1 receptor antagonist genes with disease severity in MS. *Neurology* 52: 595–599, 1999.

[34006] 7558. Terasaki, P. I.; Park, M. S.; Opelz, G.; Ting, A.: Multiple sclerosis and high incidence of a B-lymphocyte antigen. *Science* 193: 1245–1247, 1976.

[34007] 7559. Feldman, R. D.; Hegele, R. A.: G-protein polymorphisms and maternal/neonatal metabolism: still a weight for the answer. (Commentary) *Lancet* 355:1201–1202,

2000.

- [34008] 7560. Shitoh, K.; Konishi, F.; Iijima, T.; Ohdaira, T.; Sakai, K.; Kanazawa, K.; Miyaki, M.: A novel case of a sporadic desmoid tumour with mutation of the beta catenin gene. *J. Clin. Path.* 52: 695–696, 1999.
- [34009] 7561. Cartron, P.-F.; Oliver, L.; Martin, S.; Moreau, C.; LeCabellec, M.-T.; Jezequel, P.; Meflah, K.; Vallette, F. M.: The expression of a new variant of the pro-apoptotic molecule Bax, Bax-psi, is correlated with an increased survival of glioblastoma multiforme patients. *Hum. Molec. Genet.* 11: 675–687, 2002.
- [34010] 7562. Holland, E. C.; Celestino, J.; Dai, C.; Schaefer, L.; Sawaya, R. E.; Fuller, G. N.: Combined activation of Ras and Akt in neural progenitors induces glioblastoma formation in mice. *Nature Genet.* 25: 55–57, 2000.
- [34011] 7563. Mollenhauer, J.; Wiemann, S.; Scheurlen, W.; Korn, B.; Hayashi, Y.; Wilgenbus, K. K.; van Deimling, A.; Poustka, A.: DMBT1, a new member of the SRCR superfamily, on chromosome 10q25.3–26.1 is deleted in malignant brain tumours. *Nature Genet.* 17: 32–39, 1997.
- [34012] 7564. Efstratiadis, A.; Posakony, J. W.; Maniatis, T.; Lawn, R. M.; O'Connell, C.; Spritz, R. A.; DeRiel, J. K.; Forget, B. G.; Weissman, S. M.; Slightom, J. L.; Blechl, A. E.; Smithies,

O.; Baralle, F. E.; Shoulders, C. C.; Proudfoot, N. J.: The structure and evolution of the human beta-globin gene family. *Cell* 21: 653–668, 1980.

[34013] 7565. Gutersohn, A.; Naber, C.; Muller, N.; Erbel, R.; Siffert, W.: G protein beta-3 subunit 825 TT genotype and post-pregnancy weight retention. *Lancet* 355: 1240–1241, 2000.

[34014] 7566. Hegele, R. A.; Anderson, C.; Young, T. K.; Connelly, P. W.: G-protein beta-3 subunit gene splice variant and body fat distribution in Nunavut Inuit. *Genome Res.* 9: 972–977, 1999.

[34015] 7567. Hoher, B.; Slowinski, T.; Stolze, T.; Pleschka, A.; Neumayer, H.-H.; Halle, H.: Association of maternal G protein beta-3 subunit 825T allele with low birthweight. *Lancet* 355: 1241–1242, 2000.

[34016] 7568. Jin, Y.; Dietz, H. C.; Montgomery, R. A.; Bell, W. R.; McIntosh, I.; Coller, B.; Bray, P. F.: Glanzmann thrombasthenia: cooperation between sequence variants in *Cis* during splice site selection. *J. Clin. Invest.* 98: 1745–1754, 1996.

[34017] 7569. Levine, M. A.; Modi, W. S.; O'Brien, S. J.: Chromosomal localization of the genes encoding two forms of the G-protein beta polypeptide, beta-1 and beta-3, in man. *Ge-*

nomics 8: 380–386, 1990.

- [34018] 7570. Levine, M. A.; Smallwood, P. M.; Moen, P. T., Jr.; Helman, L. J.; Ahn, T. G.: Molecular cloning of beta-3 subunit, a third form of the G protein beta-subunit polypeptide. *Proc. Nat. Acad. Sci.* 87:2329–2333, 1990.
- [34019] 7571. Liu, W.; Qian, C.; Francke, U.: Silent mutation induces exon skipping of fibrillin-1 gene in Marfan syndrome. (Letter) *Nature Genet.* 16:328–329, 1997.
- [34020] 7572. Modi, W. S.; Levine, M. A.; Seuanez, H.; O'Brien, S. J.: Chromosomal localization of the gene encoding a third form of the beta subunit of GTP-binding regulatory proteins. (Abstract) *Cytogenet. Cell Genet.* 51:1046 only, 1989.
- [34021] 7573. Pietruck, F.; Moritz, A.; Montemurro, M.; Sell, A.; Busch, S.; Roskopf, D.; Virchow, S.; Esche, H.; Brockmeyer, N.; Jakobs, K. H.; Siffert, W.: Selectively enhanced cellular signaling by G(i) proteins in essential hypertension: G-alpha(i2), G-alpha(i3), G-beta(2) are not mutated. *Circ. Res.* 79: 974–983, 1996.
- [34022] 7574. Siffert, W.; Forster, P.; Jockel, K.-H.; Mvere, D. A.; Brinkmann, B.; Naber, C.; Crookes, R.; Du P. Heyns, A.; Eppelen, J. T.; Fridley, J.; Freedman, B. I.; Muller, N.; and 15 others: Worldwide ethnic distribution of the G protein

beta-3 subunit 825T allele and its association with obesity in Caucasian, Chinese, and black African individuals. *J. Am. Soc. Nephrol.* 10: 1921-1030, 1999.

- [34023] 7575. Siffert, W.; Roszkopf, D.; Moritz, A.; Wieland, T.; Kaldenberg-Stasch, S.; Kettler, N.; Hartung, K.; Beckmann, S.; Jakobs, K. H.: Enhanced G protein activation in immortalized lymphoblasts from patients with essential hypertension. *J. Clin. Invest.* 96: 759-766, 1995.
- [34024] 7576. Siffert, W.; Roszkopf, D.; Siffert, G.; Busch, S.; Moritz, A.; Erbel, R.; Sharma, A. M.; Ritz, E.; Wichmann, H.-E.; Jakobs, K. H.; Horsthemke, B.: Association of a human G-protein beta-3 subunit variant with hypertension. *Nature Genet.* 18: 45-48, 1998.
- [34025] 7577. Stallings-Mann, M. L.; Ludwiczak, R. L.; Klinger, K. W.; Rottman, F.: Alternative splicing of exon 3 of the human growth hormone receptor is the result of an unusual genetic polymorphism. *Proc. Nat. Acad. Sci.* 93: 12394-12399, 1996.
- [34026] 7578. Chang, C.; Kokontis, J.; Liao, S. S.; Chang, Y.: Isolation and characterization of human TR3 receptor: a member of steroid receptor superfamily. *J. Steroid Biochem.* 34: 391-395, 1989.
- [34027] 7579. DeGroot, L. J.: Personal Communication. Chicago, Ill.

12/19/1991.

- [34028] 7580. Forman, B. M.; Umesono, K.; Chen, J.; Evans, R. M.: Unique response pathways are established by allosteric interactions among nuclear hormone receptors. *Cell* 81: 541–550, 1995.
- [34029] 7581. Labelle, Y.; Bussieres, J.; Courjal, F.; Goldring, M. B.: The EWS/TEC fusion protein encoded by the t(9;22) chromosomal translocation in human chondrosarcomas is a highly potent transcriptional activator. *Oncogene* 18:3303–3308, 1999.
- [34030] 7582. Li, H.; Kolluri, S. K.; Gu, J.; Dawson, M. I.; Cao, X.; Hobbs, P. D.; Lin, B.; Chen, G.; Lu, J.; Lin, F.; Xie, Z.; Fontana, J. A.; Reed, J. C.; Zhang, X.: Cytochrome c release and apoptosis induced by mitochondrial targeting of orphan receptor TR3 nuclear. *Science* 289:1159–1164, 2000.
- [34031] 7583. Nakai, A.; Kartha, S.; Sakurai, A.; Toback, F. G.; DeGroot, L. J.: A human early response gene homologous to murine nur77 and rat NGFI-B, and related to the nuclear receptor superfamily. *Molec. Endocr.* 4: 1438–1443, 1990.
- [34032] 7584. Perlmann, T.; Jansson, L.: A novel pathway for vitamin A signaling mediated by RXR heterodimerization with NGFI-B and NURR1. *Genes Dev.* 9:769–782, 1995.

- [34033] 7585. Ryseck, R.-P.; Macdonald-Bravo, H.; Mattei, M. G.; Siegfried, R.L.; Bravo, R.: Structure, mapping and expression of a growth factor-inducible gene encoding a putative nuclear hormonal binding receptor. *EMBO J.* 8: 3327-3335, 1989.
- [34034] 7586. Wu, Q.; Dawson, M. I.; Zheng, Y.; Hobbs, P. D.; Agadir, A.; Jong, L.; Li, Y.; Liu, R.; Lin, B.; Zhang, X. K.: Inhibition of trans-retinoic acid-resistant human breast cancer cell growth by retinoid X receptor-selective retinoids. *Molec. Cell. Biol.* 17: 6598-6608, 1997.
- [34035] 7587. Youn, H.-D.; Sun, L.; Prywes, R.; Liu, J. O.: Apoptosis of T cells mediated by Ca^{2+} -induced release of the transcription factor MEF2. *Science* 286: 790-793, 1999.
- [34036] 7588. Friedman, E.; Gejman, P. V.; Martin, G. A.; McCormick, F.: Nonsense mutations in the C-terminal SH2 region of the GTPase activating protein (GAP) gene in human tumours. *Nature Genet.* 5: 242-247, 1993.
- [34037] 7589. Hsieh, C. L.; Francke, U.: The gene for GTPase activating protein (GAP) is on human chromosome 5q and mouse chromosome 13. (Abstract) *Cytogenet. Cell Genet.* 51: 1016 only, 1989.
- [34038] 7590. Hsieh, C. L.; Vogel, U. S.; Dixon, R. A.; Francke, U.: Chromosome localization and cDNA sequence of murine

and human genes for ras p21GTPase activating protein (GAP). *Somat. Cell Molec. Genet.* 15: 579–590, 1989.

- [34039] 7591. Lemons, R. S.; Espinosa, R., III; Rebentisch, M.; McCormick, F.; Ladner, M.; Le Beau, M. M.: Chromosomal localization of the gene encoding GTPase-activating protein (RASA) to human chromosome 5, bands q13–q15. *Genomics* 6: 383–385, 1990.
- [34040] 7592. Mitsudomi, T.; Friedman, E.; Gejman, P. V.; McCormick, F.; Gazdar, A. F.: Genetic analysis of the catalytic domain of the GAP gene in human lung cancer cell lines. *Hum. Genet.* 93: 27–31, 1994.
- [34041] 7593. Trahey, M.; Wong, G.; Halenbeck, R.; Rubinfeld, B.; Martin, G. A.; Ladner, M.; Long, C. M.; Crosier, W. J.; Watt, K.; Kohts, K.; McCormick, F.: Molecular cloning of two types of GAP complementary DNA from human placenta. *Science* 242: 1697–1700, 1988.
- [34042] 7594. Blatt, C.; Eversole-Cire, P.; Cohn, V. H.; Zollman, S.; Fournier, R. E. K.; Mohandas, L. T.; Nesbitt, M.; Lugo, T.; Jones, D. T.; Reed, R. R.; Weiner, L. P.; Sparkes, R. S.; Simon, M. I.: Chromosomal localization of genes encoding guanine nucleotide-binding protein subunits in mouse and human. *Proc. Nat. Acad. Sci.* 85: 7642–7646, 1988.
- [34043] 7595. Allen, B.; Ostrer, H.; Stein, J.; Stein, G.: Histone gene

clustersmap to chromosomes 1 and 6. (Abstract) Cytogenet. Cell Genet. 51:950 only, 1989.

- [34044] 7596. Carozzi, N.; Marashi, F.; Plumb, M.; Zimmerman, S.; Zimmerman, A.; Coles, L. S.; Wells, J. R. E.; Stein, G.; Stein, J.: Clustering of human H1 and core histone genes. Science 224: 1115–1117, 1984.
- [34045] 7597. Chandler, M. E.; Kedes, L. H.; Cohn, R. H.; Yunis, J. J.: Genes coding for histone proteins in man are located on the distal end of chromosome 7. Science 205: 908–910, 1979.
- [34046] 7598. Clark, S. J.; Krieg, P. A.; Wells, J. R. E.: Isolation of a clone containing human histone genes. Nucleic Acids Res. 9: 1583–1597, 1981.
- [34047] 7599. Delange, R. J.; Smith, E. L.: Histones: structure and function. Ann. Rev. Biochem. 40: 279–314, 1971.
- [34048] 7600. Felsenfeld, G.: Chromatin as an essential part of the transcriptional mechanism. Nature 355: 219–224, 1992.
- [34049] 7601. Green, L.; Van Antwerpen, R.; Stein, J.; Stein, G.; Tripputi, P.; Emanuel, B.; Selden, J.; Croce, C.: A major human histone gene cluster on the long arm of chromosome 1. Science 226: 838–840, 1984.
- [34050] 7602. Heintz, N.; Zernik, M.; Roeder, R. G.: The structure of the human histone genes: clustered but not tandemly

repeated. Cell 24: 661–668,1981.

[34051] 7603.Hentschel, C. C.; Birnstiel, M. L.: The organization and expression of histone gene families. Cell 25: 301–313, 1981.

[34052] 7604.Kedes, L.; Maxson, R.: Histone gene organization: paradigm lost. Nature 294:11–12, 1981.

[34053] 7605.Lichtler, A. C.; Sierra, F.; Clark, S.; Wells, J. R. E.; Stein, J. L.; Stein, G. S.: Multiple H4 histone mRNAs of HeLa cells are encoded in different genes. Nature 298: 195–198, 1982.

[34054] 7606.McAlpine, P. J.: Personal Communication. Winnipeg, Manitoba, Canada 1989.

[34055] 7607.Sierra, F.; Lichtler, A.; Marashi, F.; Rickles, R.; Van Dyke, T.; Clark, S.; Wells, J.; Stein, G.; Stein, J.: Organization of human histone genes. Proc. Nat. Acad. Sci. 79: 1795–1799, 1982.

[34056] 7608.Steffensen, D. M.: Human histone genes mapped to chromosome 7.(Abstract) Cytogenet. Cell Genet. 25: 211 only, 1979.

[34057] 7609.Szabo, P.; Yu, L. C.; Borun, T.; Varicchio, F.; Siniscalco, M.; Prenskey, W.: Localization of the histone genes in man. Cytogenet. Cell Genet. 22: 359–363, 1978.

[34058] 7610.Tripputi, P.; Emanuel, B. S.; Croce, C. M.; Green, L.

G.; Stein, G. S.; Stein, J. L.: Human histone genes map to multiple chromosomes. *Proc. Nat. Acad. Sci.* 83: 3185–3188, 1986.

- [34059] 7611. Yunis, J. J.; Chandler, M. E.: Localization of histone genes to bands 7q32–36 in man and the homologous chromosome segments in chimpanzee, gorilla, and orangutan. (Abstract) *Cytogenet. Cell Genet.* 25:220 only, 1979.
- [34060] 7612. Doyle, J.; Hoffman, S.; Ucla, C.; Reith, W.; Mach, B.; Stubbs, L.: Locations of human and mouse genes encoding the RFX1 and RFX2 transcription factor proteins. *Genomics* 35: 227–230, 1996.
- [34061] 7613. Pugliatti, L.; Derre, J.; Berger, R.; Ucla, C.; Reith, W.; Mach, B.: The genes for MHC class II regulatory factors RFX1 and RFX2 are located on the short arm of chromosome 19. *Genomics* 13: 1307–1310, 1992.
- [34062] 7614. Reith, W.; Ucla, C.; Barras, E.; Gaud, A.; Durand, B.; Herrero-Sanchez, C.; Kobr, M.; Mach, B.: RFX1, a transactivator of hepatitis B virus enhancer I, belongs to a novel family of homodimeric and heterodimeric DNA-binding proteins. *Molec. Cell. Biol.* 14: 1230–1244, 1994.
- [34063] 7615. Claesson, L.; Larhammar, D.; Rask, L.; Peterson, P. A.: cDNA clone for the human invariant gamma chain of

class II histocompatibility antigens and its implications for the protein structure. *Proc. Nat. Acad. Sci.* 80: 7395–7399, 1983.

- [34064] 7616. Claesson–Welsh, L.; Barker, P. E.; Larhammar, D.; Rask, L.; Ruddle, F. H.; Peterson, P. A.: The gene encoding the human class II antigen-associated gamma chain is located on chromosome 5. *Immunogenetics* 20: 89–93, 1984.
- [34065] 7617. Driessen, C.; Bryant, R. A.; Lennon–Dumenil, A. M.; Villadangos, J. A.; Bryant, P. W.; Shi, G. P.; Chapman, H. A.; Ploegh, H. L.: Cathepsin S controls the trafficking and maturation of MHC class II molecules in dendritic cells. *J. Cell Biol.* 147: 775–790, 1999.
- [34066] 7618. Genuardi, M.; Saunders, G. F.: Localization of the HLA class II-associated invariant chain gene to human chromosome band 5q32. *Immunogenetics* 28: 53–56, 1988.
- [34067] 7619. Bergsma, D. J.; Eder, C.; Gross, M.; Kersten, H.; Sylvester, D.; Appelbaum, E.; Cusimano, D.; Livi, G. P.; McLaughlin, M. M.; Kasyan, K.; Porter, T. G.; Silverman, C.; Dunnington, D.; Hand, A.; Prichett, W. P.; Bossard, M. J.; Brandt, M.; Levy, M. A.: The cyclophilin multigene family of peptidyl–prolyl isomerases: characterization of three sep–

aratehuman isoforms. J. Biol. Chem. 266: 23204–23214, 1991.

- [34068] 7620.Bowles, K. R.; Zintz, C.; Abraham, S. E.; Brandon, L.; Bowles,N. E.; Towbin, J. A.: Genomic characterization of the human peptidyl–pro–lyl–cis–trans–isomerase,mitochondrial precursor gene: assessment of its role in familial dilatedcardiomyopathy. Hum. Genet. 105: 582–586, 1999.
- [34069] 7621.Bachmaier, K.; Krawczyk, C.; Kozieradzki, I.; Kong, Y.–Y.; Sasaki,T.; Oliveira–dos–Santos, A.; Mariathasan, S.; Bouchard, D.; Wakeham,A.; Itie, A.; Le, J.; Ohashi, P. S.; Sarosi, I.; Nishina, H.; Lipkowitz,S.; Penninger, J. M.: Negative regulation of lymphocyte activationand autoimmunity by the molecular adaptor Cbl–b. Nature 403: 211–216,2000.
- [34070] 7622.Chiang, Y. J.; Kole, H. K.; Brown, K.; Naramura, M.; Fukuhara,S.; Hu, R.–J.; Jang, I. K.; Gutkind, J. S.; Shevach, E.; Gu, H.:Cbl–b regulates the CD28 dependence of T–cell activation. Nature 403:216–220, 2000.
- [34071] 7623.Keane, M. M.; Rivero–Lezcano, O. M.; Mitchell, J. A.; Robbins,K. C.; Lipkowitz, S.: Cloning and characterization of cbl–b: a SH3binding protein with homology to the c–cbl proto–oncogene. Oncogene 10:2367–2377, 1995.

- [34072] 7624.Komeda, K.; et al; et al: Establishment of two sub-
strains, diabetes-prone and nondiabetic, from Long-Evans
Tokushima Lean (LETL) rats. *Endocr.J.* 45: 737–744, 1998.
- [34073] 7625.Yokoi, N.; Komeda, K.; Wang, H.–Y.; Yano, H.; Ki-
tada, K.; Saitoh, Y.; Seino, Y.; Yasuda, K.; Serikawa, T.;
Seino, S.: Cblb is a major susceptibility gene for rat type 1
diabetes mellitus. *Nature Genet.* 31:391–394, 2002.
- [34074] 7626.Bathori, G.; Parolini, I.; Tombola, F.; Szabo, I.;
Messina, A.; Oliva, M.; De Pinto, V.; Lisanti, M.; Sargia-
como, M.; Zoratti, M.: Porin is present in the plasma
membrane where it is concentrated in caveolae and caveo-
lae-related domains. *J. Biol. Chem.* 274:
29607–29612, 1999.
- [34075] 7627.Huizing, M.; Ruitenbeek, W.; Thinnen, F. P.; DePinto,
V.; Wendel, U.; Trijbels, F. J. M.; Smit, L. M. E.; Ter Laak, H.
J.; Van Den Heuvel, L. P.: Deficiency of the voltage-de-
pendent anion channel: a novel cause of mitochondriopa-
thy. *Pediat. Res.* 39: 760–765, 1996.
- [34076] 7628.Chang, P. W. G.; Tsui, S. K. W.; Liew, C.; Lee, C.;
Waye, M. M.Y.; Fung, K.: Isolation, characterization, and
chromosomal mapping of a novel cDNA clone encoding
human selenium binding protein. *J. Cell. Biochem.* 64:
217–224, 1997.

- [34077] 7629.Lanfear, J.; Fleming, J.; Walker, M.; Harrison, P.: Different patterns of regulation of the genes encoding the closely related 56kDa selenium- and acetaminophen-binding proteins in normal tissues and during carcinogenesis. *Carcinogenesis* 14: 335-340, 1993.
- [34078] 7630.Creutz, C. E.; Tomsig, J. L.; Snyder, S. L.; Gautier, M.-C.; Skouri, F.; Beisson, J.; Cohen, J.: The copines, a novel class of C2 domain-containing, calcium-dependent, phospholipid-binding proteins conserved from Paramecium to humans. *J. Biol. Chem.* 273: 1393-1402, 1998.
- [34079] 7631.Caudell, E. G.; Caudell, J. J.; Tang, C.-H.; Yu, T.-K.; Frederick, M. J.; Grimm, E. A.: Characterization of human copine III as a phosphoprotein with associated kinase activity. *Biochemistry* 39: 13034-13043, 2000.
- [34080] 7632.Ito, M.; Yuan, C.-X.; Okano, H. J.; Darnell, R. B.; Roeder, R.G.: Involvement of the TRAP220 component of the TRAP/SMCC coactivator complex in embryonic development and thyroid hormone action. *Molec.Cell* 5: 683-693, 2000.
- [34081] 7633.Yuan, C.-X.; Ito, M.; Fondell, J. D.; Fu, Z.-Y.; Roeder, R. G.: The TRAP220 component of a thyroid hormone receptor-associated protein (TRAP) coactivator complex interacts directly with nuclear receptors in a ligand-de-

pendent fashion. Proc. Nat. Acad. Sci. 95:

7939–7944, 1998. Note: Erratum: Proc. Nat. Acad. Sci. 95: 14584 only, 1998.

[34082] 7634. Zhu, Y.; Qi, C.; Jain, S.; Le Beau, M. M.; Espinosa, R., III; Atkins, G. B.; Lazar, M. A.; Yeldandi, A. V.; Rao, M. S.; Reddy, J. K.: Amplification and overexpression of peroxisome proliferator-activated receptor binding protein (PBP/PPARBP) gene in breast cancer. Proc. Nat. Acad. Sci. 96:10848–10853, 1999.

[34083] 7635. Zhu, Y.; Qi, C.; Jain, S.; Rao, M. S.; Reddy, J. K.: Isolation and characterization of PBP, a protein that interacts with peroxisome proliferator-activated receptor. J. Biol. Chem. 272: 25500–25506, 1997.

[34084] 7636. Abrahamson, M.; Olafsson, I.; Palsdottir, A.; Ulvsback, M.; Lundwall, A.; Jensson, O.; Grubb, A.: Structure and expression of the human cystatin C gene. Biochem. J. 268: 287–294, 1990.

[34085] 7637. Balbin, M.; Abrahamson, M.: SstII polymorphic sites in the promoter region of the human cystatin C gene. Hum. Genet. 87: 751–752, 1991.

[34086] 7638. Barrett, A. J.; Davies, M. E.; Grubb, A.: The place of human gamma-trace (cystatin C) amongst the cysteine proteinase inhibitors. Biochem. Biophys. Res. Commun.

120: 631–636, 1984.

- [34087] 7639.Gopal Rao, V. V.; Schnittger, S.; Abrahamson, M.; Hansmann, I.: Cystatin–C (CST3), the candidate gene for the hereditary cystatin–Camyloid angiopathy (HCCAA) maps to or close to human chromosome 20p11.22.(Abstract) Cytogenet. Cell Genet. 58: 2029, 1991.
- [34088] 7640.Grubb, A.; Lofberg, H.: Human gamma–trace, a basic microprotein:amino acid sequence and presence in the adenohipophysis. Proc. Nat.Acad. Sci. 79: 3024–3027, 1982.
- [34089] 7641.Huh, C.; Nagle, J. W.; Kozak, C. A.; Abrahamson, M.; Karlsson,S.: Structural organization, expression and chromosomal mapping ofthe mouse cystatin–C–encoding gene (Cst3). Gene 152: 221–226, 1995.
- [34090] 7642.Lofberg, H.; Grubb, A.; Davidsson, L.; Kjellander, B.; Stromblad,L.–G.; Tibblin, S.; Olsson, S.–O.: Occurrence of gamma–trace in thecalcitonin–producing C–cells of simian thyroid gland. Acta Endocr. 104:69–76, 1983.
- [34091] 7643.Lofberg, H.; Grubb, A. O.: Quantitation of gamma–trace in humanbiological fluids: indications for production in the central nervoussystem. Scand. J. Clin. Lab. Invest. 39: 619–626, 1979.

- [34092] 7644.Galarneau, L.; Drouin, R.; Belanger, L.: Assignment of the fetoproteintranscription factor gene (FTF) to human chromosome band 1q32.11 byin situ hybridization. Cytogenet. Cell Genet. 82: 269–270, 1998.
- [34093] 7645.Li, M.; Xie, Y.–H.; Kong,Y.–Y.; Wu, X.; Zhu, L.; Wang, Y.: Cloningand characterization of a novel human hepatocyte transcription factor,hB1F, which binds and activates enhancer II of hepatitis B virus. J.Biol. Chem. 273: 29022–29031, 1998.
- [34094] 7646.Kadereit, S.; Gewert, D. R.; Galabru, J.; Hovanessian, A. G.; Meurs,E. F.: Molecular cloning of two new interferon–induced, highly relatednuclear phosphoproteins. J. Biol. Chem. 268: 24432–24441, 1993.
- [34095] 7647.Welsh, G. I.; Kadereit, S.; Coccia, E. M.; Hovanessian, A. G.;Meurs, E. F.: Colocalization within the nucleolus of two highly relatedIFN–induced human nuclear phosphoproteins with nucleolin. Exp. CellRes. 250: 62–74, 1999.
- [34096] 7648.Pietu, G.; Alibert, O.; Guichard, V.; Lamy, B.; Bois, F.; Leroy,E.; Mariage–Samson, R.; Houlgatte, R.; Soularue, P.; Auffray, C.:Novel gene transcripts preferentially expressed in human muscles revealedby quantitative hybridization of a high density cDNA array. GenomeRes. 6: 492–503, 1996.

- [34097] 7649.Xia, H.; Winokur, S. T.; Kuo, W.-L.; Altherr, M. R.; Bredt, D.S.: Actinin-associated LIM protein: identification of a domain interaction between PDZ and spectrin-like repeat motifs. *J. Cell Biol.* 139:507-515, 1997.
- [34098] 7650.Carim, L.; Sumoy, L.; Andreu, N.; Estivill, X.; Escarceller, M.: Identification and expression analysis of C15orf3, a novel gene on chromosome 15q21.1-q21.2. *Cytogenet. Cell Genet.* 88: 330-332, 2000.
- [34099] 7651.MacLellan, W. R.; Xiao, G.; Abdellatif, M.; Schneider, M. D.: A novel Rb- and p300-binding protein inhibits transactivation by MyoD. *Molec. Cell. Biol.* 20: 8903-8915, 2000.
- [34100] 7652.Miyake, S.; Sellers, W. R.; Safran, M.; Li, X.; Zhao, W.; Grossman, S. R.; Gan, J.; DeCaprio, J. A.; Adams, P. D.; Kaelin, W. G., Jr.: Cells degrade a novel inhibitor of differentiation with E1A-like properties upon exiting the cell cycle. *Molec. Cell. Biol.* 20: 8889-8902, 2000.
- [34101] 7653.Schweizer, A.; Valdenaire, O.; Koster, A.; Lang, Y.; Schmitt, G.; Lenz, B.; Bluethmann, H.; Rohrer, J.: Neonatal lethality in mice deficient in XCE, a novel member of the endothelin-converting enzyme and neutral endopeptidase family. *J. Biol. Chem.* 274: 20450-20456, 1999.
- [34102] 7654.Valdenaire, O.; Richards, J. G.; Faull, R. L. M.;

Schweizer, A.: XCE, a new member of the endothelin-converting enzyme and neutral endopeptidase family, is preferentially expressed in the CNS. *Molec. Brain Res.* 64: 211–221, 1999.

- [34103] 7655. Valdenaire, O.; Rohrbacher, E.; Langeveld, A.; Schweizer, A.; Meijers, C.: Organization and chromosomal localization of the human ECEL1(XCE) gene encoding a zinc metallopeptidase involved in the nervous control of respiration. *Biochem. J.* 346: 611–616, 2000.
- [34104] 7656. Hengst, U.; Albrecht, H.; Hess, D.; Monard, D.: The phosphatidylethanolamine-binding protein is the prototype of a novel family of serine protease inhibitors. *J. Biol. Chem.* 276: 535–540, 2001.
- [34105] 7657. Hori, N.; Chae, K.; Murakawa, K.; Matoba, R.; Fukushima, A.; Okubo, K.; Matsubara, K.: A human cDNA sequence homologue of bovine phosphatidylethanolamine-binding protein. *Gene* 140: 293–294, 1994.
- [34106] 7658. Moore, C.; Perry, A. C. F.; Love, S.; Hall, L.: Sequence analysis and immunolocalisation of phosphatidylethanolamine binding protein (PBP) in human brain tissue. *Molec. Brain Res.* 37: 74–78, 1996.
- [34107] 7659. Schoentgen, F.; Jolles, P.: From structure to function:

possible biological roles of a new widespread protein family binding hydrophobic ligands and displaying a nucleotide binding site. FEBS Lett. 369:22–26, 1995.

- [34108] 7660. Seddiqui, N.; Bollengier, F.; Alliel, P. M.; Perin, J.-P.; Bonnet, F.; Bucquoy, S.; Jolles, P.; Schoentgen, F.: Amino acid sequence of the Homo sapiens brain 21–23-kDa protein (neuropolypeptide h3), comparison with its counterparts from Rattus norvegicus and Bos taurus species, and expression of its mRNA in different tissues. J. Molec. Evol. 39: 655–660, 1994.
- [34109] 7661. Tohdoh, N.; Tojo, S.; Agui, H.; Ojika, K.: Sequence homology of rat and human HCNP precursor proteins, bovine phosphatidylethanolamine-binding protein and rat 23-kDa protein associated with the opioid-binding protein. Molec. Brain Res. 30: 381–384, 1995.
- [34110] 7662. Yeung, K.; Seitz, T.; Li, S.; Janosch, P.; McFerran, B.; Kaiser, C.; Fee, F.; Katsanakis, K. D.; Rose, D. W.; Mischak, H.; Sedivy, J. M.; Kolch, W.: Suppression of Raf-1 kinase activity and MAP kinase signalling by RKIP. Nature 401: 173–177, 1999.
- [34111] 7663. Heinemann, T.; Bulwin, G. C.; Randall, J.; Schnieders, B.; Sandhoff, K.; Volk, H. D.; Milford, E.; Gullans, S. R.; Utku, N.: Genomic organization of the gene coding for

TIRC7, a novel membrane protein essential for T cell activation. *Genomics* 57: 398–406, 1999.

- [34112] 7664. Li, Y.-P.; Chen, W.; Liang, Y.; Li, E.; Stashenko, P.: ATP6i-deficient mice exhibit severe osteopetrosis due to loss of osteoclast-mediated extracellular acidification. *Nature Genet.* 23: 447–451, 1999.
- [34113] 7665. Li, Y.-P.; Chen, W.; Stashenko, P.: Molecular cloning and characterization of a putative novel human osteoclast-specific 116-kDa vacuolar proton pump subunit. *Biochem. Biophys. Res. Commun.* 218: 813–821, 1996.
- [34114] 7666. Scimeca, J.-C.; Franchi, A.; Trojani, C.; Parrinello, H.; Grosgeorge, J.; Robert, C.; Jaillon, O.; Poirier, C.; Gaudray, P.; Carle, G. F.: The gene encoding the mouse homologue of the human osteoclast-specific 116-kDa V-ATPase subunit bears a deletion in osteosclerotic (oc/oc) mutants. *Bone* 26: 207–213, 2000.
- [34115] 7667. Sobacchi, C.; Frattini, A.; Orchard, P.; Porras, O.; Tezcan, I.; Andolina, M.; Babul-Hirji, R.; Baric, I.; Canham, N.; Chitayat, D.; Dupuis-Girod, S.; Ellis, I.; and 21 others: The mutational spectrum of human malignant autosomal recessive osteopetrosis. *Hum. Molec. Genet.* 10: 1767–1773, 2001.
- [34116] 7668. Utku, N.; Heinemann, T.; Tullius, S. G.; Bulwin, G. C.;

Beinke,S.; Blumberg, R. S.; Beato, F.; Randall, J.; Kojima, R.; Busconi,L.; Robertson, E. S.; Schulein, R.; Volk, H. D.; Milford, E. L.; Gullans,S. R.: Prevention of acute allograft rejection by antibody targeting of TIRC7, a novel T cell membrane protein. *Immunity* 9: 509–518,1998.

[34117] 7669.Blum, R.; Feick, P.; Puype, M.; Vandekerckhove, J.; Klengel, R.;Nastainczyk, W.; Schulz, I.: Tmp21 and p24A, two type I proteinsenriched in pancreatic microsomal membranes, are members of a proteinfamily involved in vesicular trafficking. *J. Biol. Chem.* 271: 17183–17189,1996.

[34118] 7670.Horer, J.; Blum, R.; Feick, P.; Nastainczyk, W.; Schulz, I.: Acomparative study of rat and human Tmp21 (p23) reveals the pseudogene-likefeatures of human Tmp21-II. *DNA Seq.* 10: 121–126, 1999.

[34119] 7671.Sherrington, R.; Rogaev, E. I.; Liang, Y.; Rogaeva, E. A.; Levesque,G.; Ikeda, M.; Chi, H.; Lin, C.; Li, G.; Holman, K.; Tsuda, T.; Mar,L.; Foncin, J.–F.; Bruni, A. C.; Montesi, M. P.; Sorbi, S.; Rainero,I.; Pinessi, L.; Nee, L.; Chumakov, I.; Pollen, D.; Brookes, A.; Sanseau,P.; Polinsky, R. J.; Wasco, W.; Da Silva, H. A. R.; Haines, J. L.;Pericak–Vance, M. A.; Tanzi, R. E.; Roses, A. D.; Fraser, P. E.; Rommens,J. M.; St George–Hyslop, P. H.: Cloning of a gene bearing

missense mutations in early-onset familial Alzheimer's disease. *Nature* 375:754–760, 1995.

- [34120] 7672. Bohne, J.; Cole, S. E.; Sune, C.; Lindman, B. R.; Ko, V. D.; Vogt, T. F.; Garcia-Blanco, M. A.: Expression analysis and mapping of the mouse and human transcriptional regulator CA150. *Mammalian Genome* 11:930–933, 2000.
- [34121] 7673. Sune, C.; Garcia-Blanco, M. A.: Transcriptional co-factor CA150 regulates RNA polymerase II elongation in a TATA-box-dependent manner. *Molec. Cell. Biol.* 19: 4719–4728, 1999.
- [34122] 7674. Sune, C.; Garcia-Blanco, M. A.: Transcriptional trans activation by human immunodeficiency virus type 1 Tat requires specific coactivators that are not basal factors. *J. Virol.* 69: 3098–3107, 1995.
- [34123] 7675. Sune, C.; Hayashi, T.; Liu, Y.; Lane, W. S.; Young, R. A.; Garcia-Blanco, M. A.: CA150, a nuclear protein associated with the RNA polymerase II holoenzyme, is involved in Tat-activated human immunodeficiency virus type 1 transcription. *Molec. Cell. Biol.* 17: 6029–6039, 1997.
- [34124] 7676. Postma, A. V.; Bezzina, C. R.; de Vries, J. F.; Wilde, A. A. M.; Moorman, A. F. M.; Mannens, M. M. A. M.: Genomic organisation and chromosomal localisation of two members of the KCND ion channel family, KCND2 and KCND3.

Hum. Genet. 106: 614–619, 2000.

- [34125] 7677. Zhu, X.-R.; Wulf, A.; Schwarz, M.; Isbrandt, D.; Pongs, O.: Characterization of human Kv4.2 mediating a rapidly-inactivating transient voltage-sensitive K⁺ current. *Receptors Channels* 6: 387–400, 1999.
- [34126] 7678. Dilks, D.; Ling, H.-P.; Cockett, M.; Sokol, P.; Numan, R.: Cloning and expression of the human Kv4.3 potassium channel. *J. Neurophysiol.* 81:1974–1977, 1999.
- [34127] 7679. Dixon, J. E.; Shi, W.; Wang, H.-S.; McDonald, C.; Yu, H.; Wymore, R. S.; Cohen, I. S.; McKinnon, D.: Role of the Kv4.3 K⁺ channel in ventricular muscle: a molecular correlate for the transient outward current. *Circ. Res.* 79: 659–668, 1996.
- [34128] 7680. Kong, W.; Po, S.; Yamagishi, T.; Ashen, M. D.; Stetten, G.; Tomaselli, G. F.: Isolation and characterization of the human gene encoding I_{to}: further diversity by alternative mRNA splicing. *Am. J. Physiol.* 275:H1963–H1970, 1998.
- [34129] 7681. Tseng, G.-N.: Molecular structure of cardiac I_{to} channels: Kv4.2, Kv4.3, and other possibilities? *Cardiovasc. Res.* 41: 16–18, 1999.
- [34130] 7682. Zhu, X. R.; Wulf, A.; Schwarz, M.; Isbrandt, D.; Pongs, O.: Characterization of human Kv4.2 mediating a

rapidly-inactivating transient voltage-sensitiveK⁺ current.
Receptors Channels 6: 387–400, 1999.

- [34131] 7683.van den Eynde, B. J.; Gaugler, B.; Probst–Kepper, M.; Michaux,L.; Devuyst, O.; Lorge, F.; Weynants, P.; Boon, T.: A new antigenrecognized by cytolytic T lymphocytes on a human kidney tumor resultsfrom reverse strand transcrip-
tion. J. Exp. Med. 190: 1793–1799,1999.
- [34132] 7684.Lai, C.–H.; Chou, C.–Y.; Ch'ang, L.–Y.; Liu, C.–S.; Lin, W.: Identificationof novel human genes evolutionarily con-
served in Caenorhabditis elegansby comparative pro-
teomics. Genome Res. 10: 703–713, 2000.
- [34133] 7685.Shamsadin, R.; Adham, I. M.; von Beust, G.; Engel, W.: Molecularcloning, expression and chromosome loca-
tion of the human pelota genePELO. Cytogenet. Cell
Genet. 90: 75–78, 2000.
- [34134] 7686.Kile, B. T.; Viney, E. M.; Willson, T. A.; Brodnicki, T. C.; Cancilla,M. R.; Herlihy, A. S.; Croker, B. A.; Baca, M.; Nicola, N. A.; Hilton,D. J.; Alexander, W. S.: Cloning and
characterization of the genesencoding the ankyrin repeat
and SOCS box-containing proteins Asb–1,Asb–2, Asb–3,
and Asb–4. Gene 258: 31–41, 2000.
- [34135] 7687.Gaugler, B.; Brouwenstijn, N.; Vantomme, V.; Szikora, J.–P.; Vander Spek, C. W.; Patard, J.–J.; Boon, T.;

Schrier, P.; Van den Eynde, B. J.: A new gene coding for an antigen recognized by autologous cytolytic T lymphocytes on a human renal carcinoma. *Immunogenetics* 44:323–330, 1996.

[34136] 7688. Miyata, Y.; Akashi, M.; Nishida, E.: Molecular cloning and characterization of a novel member of the MAP kinase superfamily. *Genes Cells* 4:299–309, 1999.

[34137] 7689. Liu, Y.; Corcoran, M.; Rasool, O.; Ivanova, G.; Ibbotson, R.; Grander, D.; Iyengar, A.; Baranova, A.; Kashuba, V.; Merup, M.; Wu, X.; Gardiner, A.; and 12 others: Cloning of two candidate tumor suppressor genes within a 10 kb region on chromosome 13q14, frequently deleted in chronic lymphocytic leukemia. *Oncogene* 15: 2463–2473, 1997.

[34138] 7690. Wolf, S.; Mertens, D.; Schaffner, C.; Korz, C.; Dohner, H.; Stilgenbauer, S.; Lichter, P.: B-cell neoplasia associated gene with multiple splicing (BCMS): the candidate B-CLL gene on 13q14 comprises more than 560 kb covering all critical regions. *Hum. Molec. Genet.* 10: 1275–1285, 2001.

[34139] 7691. Agarwal, A. K.; Garg, A.: A novel heterozygous mutation in peroxisome proliferator-activated receptor- γ gene in a patient with familial partial lipodystrophy. *J. Clin. Endocr. Metab.* 87: 408–411, 2002.

[34140] 7692. Muto, T.; Muramatsu, M.; Taniwaki, M.; Kinoshita,

K.; Honjo, T.: Isolation, tissue distribution, and chromosomal localization of the human activation-induced cytidine deaminase (AID) gene. *Genomics* 68:85–88, 2000.

- [34141] 7693. Geck, P.; Maffini, M. V.; Szelei, J.; Sonnenschein, C.; Soto, A.M.: Androgen-induced proliferative quiescence in prostate cancer cells: the role of AS3 as its mediator. *Proc. Nat. Acad. Sci.* 97:10185–10190, 2000.
- [34142] 7694. Kas, K.; Finger, E.; Grall, F.; Gu, X.; Akbarali, Y.; Boltax, J.; Weiss, A.; Oettgen, P.; Kapeller, R.; Libermann, T. A.: ESE-3, a novel member of an epithelium-specific Ets transcription factor subfamily, demonstrates different target gene specificity from ESE-1. *J. Biol. Chem.* 275: 2986–2998, 2000.
- [34143] 7695. Kleinbaum, L. A.; Duggan, C.; Ferreira, E.; Coffey, G. P.; Buttice, G.; Burton, F. H.: Human chromosomal localization, tissue/tumor expression, and regulatory function of the ets family gene EHF. *Biochem. Biophys. Res. Commun.* 264: 119–126, 1999.
- [34144] 7696. Davidson, J. D.; Riley, B.; Burrigh, E.N.; Duvick, L.A.; Zoghbi, H.Y.; Orr, H. T.: Identification and characterization of an ataxin-1-interacting protein: A1Up, a ubiquitin-like nuclear protein. *Hum. Molec. Genet.* 9:2305–2312, 2000.
- [34145] 7697. Zhao, L.; Gregoire, F.; Sul, H. S.: Transient induction

of ENC-1, a kelch-related actin-binding protein, is required for adipocyte differentiation. *J. Biol. Chem.* 275: 16845–16850, 2000.

[34146] 7698. Hansen, L. L.; Jorgensen, R.; Justesen, J.: Assignment of the human mitochondrial translational release factor 1 (MTRF1) to chromosome 13q14.1–q14.3 and of the human mitochondrial ribosome recycling factor (MRRF) to chromosome 9q32–q34.1 with radiation hybrid mapping. *Cytogenet. Cell Genet.* 88: 91–92, 2000.

[34147] 7699. Zhang, Y.; Spremulli, L. L.: Identification and cloning of human mitochondrial translational release factor 1 and the ribosome recycling factor. *Biochim. Biophys. Acta* 1443: 245–250, 1998.

[34148] 7700. Stelnicki, E. J.; Arbeit, J.; Cass, D. L.; Saner, C.; Harrison, M.; Largman, C.: Modulation of the human homeobox genes PRX-2 and HOXB13 in scarless fetal wounds. *J. Invest. Derm.* 111: 57–63, 1998.

[34149] 7701. Zeltser, L.; Desplan, C.; Heintz, N.: Hoxb-13: a new Hox gene in a distant region of the HOXB cluster maintains colinearity. *Development* 122: 2475–2484, 1996.

[34150] 7702. Rajagopalan, S.; Fu, J.; Long, E. O.: Cutting edge: induction of IFN- γ production but not cytotoxicity by the killer cell Ig-like receptor KIR2DL4 (CD158d) in resting

NK cells. *J. Immun.* 167: 1877–1881,2001.

- [34151] 7703.Selvakumar, A.; Steffens, U.; Dupont, B.: NK cell receptor gene of the KIR family with two IG domains but highest homology to KIR receptors with three IG domains. *Tissue Antigens* 48: 285–294, 1996.
- [34152] 7704.Pende, D.; Biassoni, R.; Cantoni, C.; Verdiani, S.; Falco, M.; di Donato, C.; Accame, L.; Bottino, C.; Moretta, A.; Moretta, L.: The natural killer cell receptor specific for HLA-A allotypes: a novel member of the p58/p70 family of inhibitory receptors that is characterized by three immunoglobulin-like domains and is expressed as a 140-kD disulphide-linked dimer. *J. Exp. Med.* 184: 505–518, 1996.
- [34153] 7705.Beadling, C.; Johnson, K. W.; Smith, K. A.: Isolation of interleukin2-induced immediate-early genes. *Proc. Nat. Acad. Sci.* 90: 2719–2723,1993.
- [34154] 7706.Gong, R.; Yu, L.; Zhang, H.; Tu, Q.; Zhao, Y.; Yang, J.; Xu, Y.; Zhao, S.: Assignment of human GADD45G to chromosome 9q22.1–q22.3 by radiation hybrid mapping. *Cytogenet. Cell Genet.* 88: 95–96, 2000.
- [34155] 7707.Nakayama, K.; Hara, T.; Hibi, M.; Hirano, T.; Miyajima, A.: A novel oncostatin M-inducible gene OIG37 forms a gene family with MyD118 and GADD45 and negatively

regulates cell growth. *J. Biol. Chem.* 274:24766–24772, 1999.

[34156] 7708. Suzuki, M.; Watanabe, T. K.; Fujiwara, T.; Nakamura, Y.; Takahashi, E.; Tanigami, A.: Molecular cloning, expression, and mapping of a novel human cDNA, GRP17, highly homologous to human gadd45 and murine MyD118. *J. Hum. Genet.* 44: 300–303, 1999.

[34157] 7709. Zhang, X.; Sun, H.; Danila, D. C.; Johnson, S. R.; Zhou, Y.; Swearingen, B.; Klibanski, A.: Loss of expression of GADD45–gamma, a growth inhibitory gene, in human pituitary adenomas: implications for tumorigenesis. *J. Clin. Endocr. Metab.* 87: 1262–1267, 2002.

[34158] 7710. Jones, S. D.; van der Flier, A.; Sonnenberg, A.: Genomic organization of the human alpha-3 integrin subunit gene. *Biochem. Biophys. Res. Commun.* 248: 896–898, 1998.

[34159] 7711. Takada, Y.; Murphy, E.; Pil, P.; Chen, C.; Ginsberg, M. H.; Hemler, M. E.: Molecular cloning and expression of the cDNA for alpha-3 subunit of human alpha-3/beta-1 (VLA-3), an integrin receptor for fibronectin, laminin, and collagen. *J. Cell Biol.* 115: 257–266, 1991.

[34160] 7712. Tsuji, T.; Hakomori, S.; Osawa, T.: Identification of human galactoprotein b3, an oncogenic transformation-in-

duced membrane glycoprotein, asVLA-3 alpha subunit:
the primary structure of human integrin alpha-3.

J.Biochem. 109: 659-665, 1991.

[34161] 7713.Hudson, J. W.; Chen, L.; Fode, C.; Binkert, C.; Dennis, J. W.: Sak kinase gene structure and transcriptional regulation. Gene 241:65-73, 2000.

[34162] 7714.Karn, T.; Holtrich, U.; Wolf, G.; Hock, B.; Strebhardt, K.; Rubsamen-Waigmann, H.: Human SAK related to the PLK/polo family of cell cycle kinases shows high mRNA expression in testis. Oncol. Reports 4: 505-510, 1997.

[34163] 7715.McMahon, H. T.; Missler, M.; Li, C.; Sudhof, T. C.: Complexins: cytosolic proteins that regulate SNAP receptor function. Cell 83:111-119, 1995.

[34164] 7716.Reim, K.; Mansour, M.; Varoqueaux, F.; McMahon, H. T.; Sudhof, T. C.; Brose, N.; Rosenmund, C.: Complexins regulate a late step in Ca^{2+} -dependent neurotransmitter release. Cell 104: 71-81, 2001.

[34165] 7717.Tokumaru, H.; Umayahara, K.; Pellegrini, L. L.; Ishizuka, T.; Saisu, H.; Betz, H.; Augustine, G. J.; Abe, T.: SNARE complex oligomerization by synaphin/complexin is essential for synaptic vesicle exocytosis. Cell 104:421-432, 2001.

[34166] 7718.Donzeau, M.; Kaldi, K.; Adam, A.; Paschen, S.; Wan-

ner, G.; Guiard, B.; Bauer, M. F.; Neupert, W.; Brunner, M.: Tim23 links the inner and outer mitochondrial membranes. *Cell* 101: 401–412, 2000.

[34167] 7719. Machesky, L. M.; Insall, R. H.: Scar1 and the related Wiskott–Aldrich syndrome protein, WASP, regulate the actin cytoskeleton through the Arp2/3 complex. *Curr. Biol.* 8: 1347–1356, 1998.

[34168] 7720. Miki, H.; Suetsugu, S.; Takenawa, T.: WAVE, a novel WASP–family protein involved in actin reorganization induced by Rac. *EMBO J.* 17: 6932–6941, 1998.

[34169] 7721. Scott, A. F.: Personal Communication. Baltimore, Md. 10/23/2001.

[34170] 7722. McGaughan, J. M.; Ward, H. B.; Evans, D. G.: WAGR syndrome and multiple exostoses in a patient with del(11)(p11.2p14.2). *J. Med. Genet.* 32: 823–824, 1995.

[34171] 7723. Wuyts, W.; Van Hul, W.; Wauters, J.; Nemtsova, M.; Reyniers, E.; Van Hul, E. V.; De Boulle, K.; de Vries, B. B.; Hendrickx, J.; Herrygers, I.; Bossuyt, P.; Balemans, W.; Fransen, E.; Vits, L.; Coucke, P.; Nowak, N. J.; Shows, T. B.; Mallet, L.; van den Ouweland, A. M.; McGaughan, J.; Hall, D. J.; Willems, P. J.: Positional cloning of a gene involved in hereditary multiple exostoses. *Hum. Molec. Genet.* 5: 1547–1557, 1996.

- [34172] 7724.Clark, M. E.; Kelner, G. S.; Turbeville, L. A.; Boyer, A.; Arden,K. C.; Maki, R. A.: ADAMTS9, a novel member of the ADAM-TS/metallospodingene family. *Genomics* 67: 343-350, 2000.
- [34173] 7725.Marie-Cardine, A.; Kirchgessner, H.; Bruyns, E.; Shevchenko, A.;Mann, M.; Autschbach, F.; Ratnofsky, S.; Meuer, S.; Schraven, B.:SHP2-interacting transmembrane adaptor protein (SIT), a novel disulfide-linkeddimer regulating human T cell activation. *J. Exp. Med.* 189: 1181-1194,1999.
- [34174] 7726.Creasy, C. L.; Chernoff, J.: Cloning and characterization of a human protein kinase with homology to Ste20. *J. Biol. Chem.* 270:21695-21700, 1995.
- [34175] 7727.Taylor, L. K.; Wang, H.-C. R.; Erikson, R. L.: Newly identifiedstress-responsive protein kinases, Krs-1 and Krs-2. *Proc. Nat. Acad.Sci.* 93: 10099-10104, 1996.
- [34176] 7728.Lennard, A.; Gorman, P.; Carrier, M.; Griffiths, S.; Scotney, H.;Sheer, D.; Solari, R.: Cloning and chromosome mapping of the humaninterleukin-1 receptor antagonist gene. *Cytokine* 4: 83-89, 1992.
- [34177] 7729.Mansfield, J. C.; Holden, H.; Tarlow, J. K.; Di Giovine, F. S.;McDowell, T. L.; Wilson, A. G.; Holdsworth, C. D.; Duff, G. W.: Novelgenetic association between ulcerative

colitis and the anti-inflammatory cytokine interleukin-1 receptor antagonist. *Gastroenterology* 106:637-642, 1994.

- [34178] 7730. Patterson, D.; Jones, C.; Hart, I.; Bleskan, J.; Berger, R.; Geyer, D.; Eisenberg, S. P.; Smith, M. F., Jr.; Arend, W. P.: The human interleukin-1 receptor antagonist (IL1RN) gene is located in the chromosome 2q14 region. *Genomics* 15: 173-176, 1993.
- [34179] 7731. Steinkasserer, A.; Spurr, N. K.; Cox, S.; Jeggo, P.; Sim, R. B.: The human IL-1 receptor antagonist gene (IL1RN) maps to chromosome 2q14-q21, in the region of the IL-1-alpha and IL-1-beta loci. *Genomics* 13:654-657, 1992.
- [34180] 7732. Tarlow, J. K.; Blakemore, A. I. F.; Lennard, A.; Solari, R.; Hughes, H. N.; Steinkasserer, A.; Duff, G. W.: Polymorphism in human IL-1 receptor antagonist gene intron 2 is caused by variable numbers of an 86-bp tandem repeat. *Hum. Genet.* 91: 403-404, 1993.
- [34181] 7733. Tarlow, J. K.; Clay, F. E.; Cork, M. J.; Blakemore, A. I. F.; McDonagh, A. J. G.; Messenger, A. G.; Duff, G. W.: Severity of alopecia areata is associated with a polymorphism in the interleukin-1 receptor antagonist gene. *J. Invest. Dermatol.* 103: 387-390, 1994.

- [34182] 7734.Schild, D.; Brake, A. J.; Kiefer, M. C.; Young, D.; Barr, P. J.: Cloning of three human multifunctional de novo purine biosynthetic genes by functional complementation of yeast mutations. *Proc. Nat. Acad. Sci.* 87: 2916–2920, 1990.
- [34183] 7735.Garrow, T. A.; Brenner, A. A.; Whitehead, V. M.; Chen, X.–N.; Duncan, R. G.; Korenberg, J. R.; Shane, B.: Cloning of human cDNAs encoding mitochondrial and cytosolic serine hydroxymethyltransferases and chromosomal localization. *J. Biol. Chem.* 268: 11910–11916, 1993.
- [34184] 7736.Kao, F.–T.; Chasin, L. A.; Puck, T. T.: Genetics of somatic mammalian cells. X. Complementation analysis of glycine–requiring mutants. *Proc. Nat. Acad. Sci.* 64: 1284–1291, 1969.
- [34185] 7737.Stover, P. J.; Chen, L. H.; Suh, J. R.; Stover, D. M.; Keyomarsi, K.; Shane, B.: Molecular cloning, characterization, and regulation of the human mitochondrial serine hydroxymethyltransferase gene. *J. Biol. Chem.* 272: 1842–1848, 1997.
- [34186] 7738.Abbal, M.; Moennarid, C.; Cambon–Thomsen, A.; Tkaczuk, J.; Ohayon, E.; Mauff, G.: A new BF variant (BF S11) with information for orientation of MHC class III genes. *Immunogenetics* 26: 320–322, 1987.

- [34187] 7739. Agarwal, D. P.; Goedde, H. W.; Benkmann, H.-G.; Flatz, G.; Rahimi, A. G.; Kaifie, A.; Delbruck, H.: Genetic polymorphism of C3 and serum levels of immunoglobulins, C3, C4 components of complement and C3-proactivator in four different populations of Afghanistan. *Hum. Genet.* 33: 67-72, 1976.
- [34188] 7740. Albert, E. D.; Rittner, C.; Grosse-Wilde, H.; Netzel, B.; Scholz, S.: Recombination frequency and linkage disequilibrium between HL-A and Bf. In: Kissmeyer-Nielsen, F.: *Histocompatibility Testing 1975*. Copenhagen: Munksgaard (pub.) 1975. Pp. 941-944.
- [34189] 7741. Allen, F. H., Jr.: Linkage of HL-A and GBG. *Vox Sang.* 27: 382-384, 1974.
- [34190] 7742. Alper, C. A.: Inherited structural polymorphism in human C2: evidence for genetic linkage between C2 and Bf. *J. Exp. Med.* 144: 1111-1115, 1976.
- [34191] 7743. Alper, C. A.; Boenisch, T.; Watson, L.: Genetic polymorphism in human glycine-rich-beta glycoprotein. *J. Exp. Med.* 135: 68-80, 1972.
- [34192] 7744. Alper, C. A.; Goodkofsky, I.; Lepow, I. H.: The relationship of glycine-rich beta-glycoprotein to factor B in the properdin system and to cobra-binding protein of human serum. *J. Exp. Med.* 137: 424-437, 1973.

- [34193] 7745. Arnason, A.; Larsen, B.; Marshall, W. H.; Edwards, J. H.; Mackintosh, P.; Olaisen, B.; Teisberg, P.: Very close linkage between HLA-B and Bf inferred from allelic association. *Nature* 268: 527–528, 1977.
- [34194] 7746. Bender, K.; Mayerova, A.; Frank, R.; Hiller, C.; Wienker, T.: Haplotype analysis of the linkage group HLA-A: HLA-B: Bf and its bearing on the interpretation of the linkage disequilibrium. *Hum. Genet.* 36: 191–196, 1977.
- [34195] 7747. Benkmann, H. G.; Goedde, H. W.; Agarwal, D. P.; Flatz, G.; Rahimi, A.; Kaif, R. S.; Delbruck, H.: Properdin factor B polymorphism in Afghanistan. *Hum. Hered.* 30: 39–43, 1980.
- [34196] 7748. Bertrams, J.; Mauff, G.: Another family with a silent allele of properdin factor B polymorphism (BF*QO). *Hum. Genet.* 70: 321–323, 1985.
- [34197] 7749. Campbell, R. D.: The molecular genetics and polymorphism of C2 and factor B. *Brit. Med. Bull.* 43: 37–49, 1987.
- [34198] 7750. Board, P. G.: Biochemical genetics of glutathione-S-transferase in man. *Am. J. Hum. Genet.* 33: 36–43, 1981.
- [34199] 7751. Rochelle, J. M.; Watson, M. L.; Oakey, R. J.; Seldin, M. F.: A linkage map of mouse chromosome 19: definition of

comparative mappingrelationships with human chromo-
somes 10 and 11 including the MEN1locus. Genomics 14:
26–31, 1992.

[34200] 7752.Seldin, M. F.; Saunders, A. M.; Rochelle, J. M.;
Howard, T. A.: A proximal mouse chromosome 9 linkage
map that further defines linkagegroups homologous with
segments of human chromosomes 11, 15, and 19. Ge-
nomics 9:678–685, 1991.

[34201] 7753.Baulac, S.; Huberfeld, G.; Gourfinkel-An, I.;
Mitropoulou, G.;Beranger, A.; Prud'homme, J.-F.; Baulac,
M.; Brice, A.; Bruzzone,R.; LeGuern, E.: First genetic evi-
dence of GABA(A) receptor dysfunctionin epilepsy: a mu-
tation in the gamma-2-subunit gene. Nature Genet.
28:46–48, 2001.

[34202] 7754.Buckwalter, M. S.; Lossie, A. C.; Scarlett, L. M.;
Camper, S. A.: Localization of the human chromosome 5q
genes Gabra-1, Gabrg-2,l1-4, l1-5, and lrf-1 on mouse
chromosome 11. Mammalian Genome 3:604–607, 1992.

[34203] 7755.Harkin, L. A.; Bowser, D. N.; Dibbens, L. M.; Singh,
R.; Phillips,F.; Wallace, R. H.; Richards, M. C.; Williams, D.
A.; Mulley, J. C.;Berkovic, S. F.; Scheffer, I. E.; Petrou, S.:
Truncation of the GABA-A-receptorgamma-2 subunit in a
family with generalized epilepsy with febrileseizures plus.

Am. J. Hum. Genet. 70: 530–536, 2002.

- [34204] 7756. Italian League Against Epilepsy Genetic Collaborative Group: Concordance of clinical forms of epilepsy in families with several affected members. *Epilepsia* 34: 819–826, 1993.
- [34205] 7757. Jensen, K. B.; Dredge, B. K.; Stefani, G.; Zhong, R.; Buckanovich, R. J.; Okano, H. J.; Yang, Y. Y. L.; Darnell, R. B.: Nova-1 regulates neuron-specific alternative splicing and is essential for neuronal viability. *Neuron* 25: 359–371, 2000.
- [34206] 7758. Kananura, C.; Haug, K.; Sander, T.; Runge, U.; Gu, W.; Hallmann, K.; Rebstock, J.; Heils, A.; Steinlein, O. K.: A splice-site mutation in GABRG2 associated with childhood absence epilepsy and febrile convulsions. *Arch. Neurol.* 59: 1137–1141, 2002.
- [34207] 7759. Olsen, R. W.; DeLorey, T. M.; Gordey, M.; Kang, M.-H.: GABA receptor function and epilepsy. *Adv. Neurol.* 79: 499–510, 1999.
- [34208] 7760. Wallace, R. H.; Marini, C.; Petrou, S.; Harkin, L. A.; Bowser, D. N.; Panchal, R. G.; Williams, D. A.; Sutherland, G. R.; Mulley, J. C.; Scheffer, I. E.; Berkovic, S. F.: Mutant GABA(A) receptor gamma-2-subunit in childhood absence epilepsy and febrile seizures. *Nature Genet.* 28: 49–52,

2001.

- [34209] 7761.Ozcelik, T.; Suedhof, T. C.; Francke, U.: The genes for inositol1,4,5-triphosphate receptors 1 (ITPR1) and 3 (ITPR3) are localized on human chromosomes 3p and 6pter-p21, respectively. (Abstract) Cytogenet.Cell Genet. 58: 1880, 1991.
- [34210] 7762.Aschauer, H.; Sanguanserm, S.; Braunitzer, G.: Embryonale Haemoglobine des Menschen: Die Primärstruktur der zeta-Ketten (Human embryonichemoglobins: the primary structure of the zeta chains). Hoppe Seylers Z. Physiol. Chem. 362: 1159-1162, 1981.
- [34211] 7763.Black, J. A.: Human zeta hemoglobin chain. (Letter) Nature 261:348 only, 1976.
- [34212] 7764.Capp, G. L.; Rigas, D. A.; Jones, R. T.: Evidence for a new hemoglobin chain (zeta chain). Nature 228: 278-280, 1970.
- [34213] 7765.Capp, G. L.; Rigas, D. A.; Jones, R. T.: Hemoglobin Portland 1: a new human hemoglobin unique in structure. Science 157: 65-66, 1967.
- [34214] 7766.Chung, S.-W.; Wong, S. C.; Clarke, B. J.; Patterson, M.; Walker, W. H. C.; Chui, D. H. K.: Human embryonic zeta-globin chains in adult patients with alpha-

thalassemias. Proc. Nat. Acad. Sci. 81: 6188–6191,1984.

- [34215] 7767.Clegg, J. B.; Gagnon, J.: Structure of the zeta chain of humanembryonic hemoglobin. Proc. Nat. Acad. Sci. 78: 6076–6080, 1981.
- [34216] 7768.Felice, A. E.; Cleek, M. P.; Marino, E. M.; McKie, K. M.; McKie,V. C.; Chang, B. K.; Huisman, T. H. J.: Different zeta globin genedeletions among black Americans. Hum. Genet. 73: 221–224, 1986.
- [34217] 7769.Goodbourn, S. E. Y.; Higgs, D. R.; Clegg, J. B.; Weatherall, D.J.: Molecular basis of length polymorphism in the human zeta–globingene complex. Proc. Nat. Acad. Sci. 80: 5022–5026, 1983.
- [34218] 7770.Hecht, F.; Jones, R. T.; Koler, R. D.: Newborn infants with HbPortland 1, an indicator of alpha–chain deficiency. Ann. Hum. Genet. 31:215–218, 1968.
- [34219] 7771.Higgs, D. R.; Pressley, L.; Aldridge, B.; Clegg, J. B.; Weatherall,D. J.; Cao, A.; Hadjiminias, M. G.; Kattamis, C.; Metaxatou–Mavromati,A.; Rachmilewitz, E. A.; Sophocleous, T.: Genetic and molecular diversityin nondeletion Hb H disease. Proc. Nat. Acad. Sci. 78: 5833–5837,1981.
- [34220] 7772.Woodroffe, M. N.; Tunnacliffe, A.; Pym, B.; Goodfellow, P. N.;Walsh, F. S.: Human muscle cell surface antigen 16–3A5 is encodedby a gene on chromosome 11. Somat.

Cell Molec. Genet. 10: 535–540,1984.

- [34221] 7773.Hasegawa, M.; Fujimoto, M.; Poe, J. C.; Steeber, D. A.; Lowell,C. A.; Tedder, T. F.: A CD19–dependent signaling pathway regulatesautoimmunity in Lyn–deficient mice. J. Immun. 167: 2469–2478, 2001.
- [34222] 7774.Hasegawa, T.; Hasegawa, Y.; Aso, T.; Koto, S.; Nagai, T.; Tsuchiya,Y.; Kim, K. C.; Ohashi, H.; Wakui, K.; Fukushima, Y.: HDR syndrome(hypoparathyroidism, sensorineural deafness, renal dysplasia) associatedwith del(10)(p13). Am. J. Med. Genet. 73: 416–418, 1997.
- [34223] 7775.Yamada, N.; Makino, Y.; Clark, R. A.; Pearson, D. W.; Mattei, M.–G.;Guenet, J.–L.; Ohama, E.; Fujino, I.; Miyawaki, A.; Furuichi, T.;Mikoshiba, K.: Human inositol 1,4,5–triphosphate type–1 receptor,InsP3R1: structure, function, regulation of expression and chromosomallocalization. Biochem. J. 302: 781–790, 1994.
- [34224] 7776.Maranto, A. R.: Primary structure, ligand binding, and localizationof the human type 3 inositol 1,4,5–trisphosphate receptor expressedin intestinal epithelium. J. Biol. Chem. 269: 1222–1230, 1994.
- [34225] 7777.Ozcelik, T.; Suedhof, T. C.; Francke, U.: The genes for inositol1,4,5–triphosphate receptors 1 (ITPR1) and 3 (ITPR3) are localizedon human chromosomes 3p and

6pter-p21, respectively. (Abstract) Cytogenet.Cell Genet. 58: 1880 only, 1991.

- [34226] 7778.Yamamoto-Hino, M.; Sugiyama, T.; Hikichi, K.; Mattei, M. G.; Hasegawa,K.; Sekine, S.; Sakurada, K.; Miyawaki, A.; Furuichi, T.; Hasegawa,M.; Mikoshiba, K.: Cloning and characterization of human type 2 and type 3 inositol 1,4,5-triphosphate receptors. Receptors Channels 2:9-22, 1994.
- [34227] 7779.Acquati, F.; Margaretti, N.; Hauptschein, R.; Rao, P.; Gaidano,G.; Taramelli, R.: A 2-Mb YAC contig linking the plasminogen-apoprotein(a)gene family to the insulin-like growth factor 2 receptor (IGF2R) gene on the telomeric region of chromosome 6 (6q26-q27). Genomics 22:664-666, 1994.
- [34228] 7780.Barlow, D. P.; Stoger, R.; Herrmann, B. G.; Saito, K.; Schweifer,N.: The mouse insulin-like growth factor type-2 receptor is imprinted and closely linked to the Tme locus. Nature 349: 84-87, 1991.
- [34229] 7781.DeChiara, T. M.; Robertson, E. J.; Efstratiadis, A.: Parental imprinting of the mouse insulin-like growth factor II gene. Cell 64:849-859, 1991.
- [34230] 7782.De Souza, A. T.; Hankins, G. R.; Washington, M. K.; Fine, R. L.;Orton, T. C.; Jirtle, R. L.: Frequent loss of het-

erozygosity on 6q at the mannose

6-phosphate/insulin-like growth factor II receptor locus in human hepatocellular tumors. *Oncogene* 10: 1725–1729, 1995.

[34231] 7783. De Souza, A. T.; Hankins, G. R.; Washington, M. K.; Orton, T. C.; Jirtle, R. L.: M6P/IGF2R gene is mutated in human hepatocellular carcinomas with loss of heterozygosity. *Nature Genet.* 11: 447–449, 1995.

[34232] 7784. Feinberg, A. P.: Genomic imprinting and gene activation in cancer. *Nature Genet.* 4: 110–113, 1993.

[34233] 7785. Haig, D.; Graham, C.: Genomic imprinting and the strange case of the insulin-like growth factor II receptor. *Cell* 64: 1045–1046, 1991.

[34234] 7786. Haig, D.; Westoby, M.: Parent-specific gene expression and the triploid endosperm. *Am. Nat.* 134: 147–155, 1989.

[34235] 7787. Kalscheuer, V. M.; Mariman, E. C.; Schepens, M. T.; Rehder, H.; Ropers, H.-H.: The insulin-like-growth factor type-2 receptor gene is imprinted in the mouse but not in humans. *Nature Genet.* 5: 74–78, 1993.

[34236] 7788. Kiess, W.; Blickenstaff, G. D.; Sklar, M. M.; Thomas, C. L.; Nissley, S. P.; Sahagian, G. G.: Biochemical evidence that the type II insulin-like growth factor receptor is iden-

tical to the cation-independent mannose 6-phosphate receptor. *J. Biol. Chem.* 263: 9339–9344, 1988.

- [34237] 7789. Killian, J. K.; Oka, Y.; Jang, H.-S.; Fu, X.; Waterland, R. A.; Sohda, T.; Sakaguchi, S.; Jirtle, R. L.: Mannose 6-phosphate/insulin-like growth factor 2 receptor (M6P/IGF2R) variants in American and Japanese populations. *Hum. Mutat.* 18: 25–31, 2001.
- [34238] 7790. Kornfeld, S.; Mellman, I.: The biogenesis of lysosomes. *Annu. Rev. Cell Biol.* 5: 483–525, 1989.
- [34239] 7791. Lau, M. M. H.; Stewart, C. E. H.; Liu, Z.; Bhatt, H.; Rotwein, P.; Stewart, C. L.: Loss of the imprinted IGF2/cation-independent mannose 6-phosphate receptor results in fetal overgrowth and perinatal lethality. *Genes Dev.* 8: 2953–2963, 1994.
- [34240] 7792. Laureys, G.; Barton, D. E.; Ullrich, A.; Francke, U.: Chromosomal mapping of the gene for the type II insulin-like growth factor receptor/cation-independent mannose 6-phosphate receptor in man and mouse. *Genomics* 3: 224–229, 1988.
- [34241] 7793. MacDonald, R. G.; Pfeffer, S. R.; Coussens, L.; Tepper, M. A.; Brocklebank, C. M.; Mole, J. E.; Anderson, J. K.: A single receptor binds both insulin-like growth factor II and mannose-6-phosphate. *Science* 239: 1134–1137,

1988.

- [34242] 7794. Morgan, D. O.; Edman, J. D.; Standring, D. N.; Fried, V. A.; Smith, M. C.; Roth, R. A.; Rutter, W. J.: Insulin-like growth factor II receptor as a multifunctional binding protein. *Nature* 329: 301–307, 1987.
- [34243] 7795. Morris, S. W.; Kirstein, M. N.; Valentine, M. B.; Dittmer, K. G.; Shapiro, D. N.; Saltman, D. L.; Look, A. T.: Fusion of a kinase gene, ALK, to a nucleolar protein gene, NPM, in non-Hodgkin's lymphoma. *Science* 263: 1281–1284, 1994.
- [34244] 7796. Tait, J. F.; Frankenberry, D. A.; Miao, C. H.; Killary, A. M.; Adler, D. A.; Distech, C. M.: Chromosomal localization of the human annexin III (ANX3) gene. *Genomics* 10: 441–448, 1991.
- [34245] 7797. Tait, J. F.; Smith, C.; Xu, L.; Cookson, B. T.: Structure and polymorphisms of the human annexin III (ANX3) gene. *Genomics* 18: 79–86, 1993.
- [34246] 7798. Grundmann, U.; Amann, E.; Abel, K.-J.; Kupper, H. A.: Isolation and expression of cDNA coding for a new member of the phospholipase A2 inhibitor family. *Behring Inst. Mitt.* 82: 59–67, 1988.
- [34247] 7799. Hauptmann, R.; Maurer-Fogy, I.; Krystek, E.; Bodo, G.; Andree, H.; Reutelingsperger, C. P. M.: Vascular antico-

agulant beta, a novel human Ca^{2+} /phospholipid binding protein that inhibits coagulation and phospholipase A-2 activity: its molecular cloning, expression and comparison with VAC-alpha. *Europ. J. Biochem.* 185: 63-71, 1989.

[34248] 7800. Tait, J. F.; Smith, C.; Frankenberry, D. A.; Miao, C. H.; Adler, D. A.; Disteché, C. M.: Chromosomal mapping of the human annexin IV (ANX4) gene. *Genomics* 12: 313-318, 1992.

[34249] 7801. Bilezikian, J. P.; Morishima, A.; Bell, J.; Grumbach, M. M.: Increased bone mass as a result of estrogen therapy in a man with aromatase deficiency. *New Eng. J. Med.* 339: 599-603, 1998.

[34250] 7802. Oldridge, M.; Temple, I. K.; Santos, H. G.; Gibbons, R. J.; Mustafa, Z.; Chapman, K. E.; Loughlin, J.; Wilkie, A. O. M.: Brachydactyly type B: linkage to chromosome 9q22 and evidence for genetic heterogeneity. *Am. J. Hum. Genet.* 64: 578-585, 1999.

[34251] 7803. Breimer, L. H.; MacIntyre, I.; Zaidi, M.: Peptides from the calcitonin genes: molecular genetics, structure and function. *Biochem. J.* 255: 377-390, 1988.

[34252] 7804. Schultz, D.; Mikala, G.; Yatani, A.; Engle, D. B.; Iles, D. E.; Segers, B.; Sinke, R. J.; Weghuis, D. O.; Klockner, U.; Wakamori, M.; Wang, J.-J.; Melvin, D.; Varadi, G.; Schwartz,

A.: Cloning, chromosomal localization, and functional expression of the α -1 subunit of the L-type voltage-dependent calcium channel from normal human heart. *Proc. Nat. Acad. Sci.* 90: 6228–6232, 1993.

[34253] 7805. Soldatov, N. M.: Genomic structure of human L-type $\text{Ca}(2+)$ channel. *Genomics* 22:77–87, 1994.

[34254] 7806. Sun, W.; McPherson, J. D.; Hoang, D. Q.; Wasmuth, J. J.; Evans, G. A.; Montal, M.: Mapping of a human brain voltage-gated calcium channel to human chromosome 12p13-pter. *Genomics* 14: 1092–1094, 1992.

[34255] 7807. Tsien, R. W.; Ellinor, P. T.; Horne, W. A.: Molecular diversity of voltage-dependent $\text{Ca}(2+)$ channels. *Trends Pharm. Sci.* 12: 349–354, 1991.

[34256] 7808. Chin, H.; Kozak, C. A.; Kim, H.-L.; Mock, B.; McBride, O. W.: A brain L-type calcium channel α -1 subunit gene (CCHL1A2) maps to mouse chromosome 14 and human chromosome 3. *Genomics* 11: 914–919, 1991.

[34257] 7809. Jinnah, H. A.; Yitta, S.; Drew, T.; Kim, B. S.; Visser, J. E.; Rothstein, J. D.: Calcium channel activation and self-biting in mice. *Proc. Nat. Acad. Sci.* 96: 15228–15232, 1999.

[34258] 7810. Mori, Y.; Friedrich, T.; Kim, M.-S.; Mikami, A.; Nakai, J.; Ruth, P.; Bosse, E.; Hofmann, F.; Flockerzi, V.; Furuichi,

T.; Mikoshiba, K.; Imoto, K.; Tanabe, T.; Numa, S.: Primary structure and functional expression from complementary DNA of a brain calcium channel. *Nature* 350:398–402, 1991.

[34259] 7811. Pennartz, C. M. A.; de Jeu, M. T. G.; Bos, N. P. A.; Schaap, J.; Geurtsen, A. M. S.: Diurnal modulation of pacemaker potentials and calcium current in the mammalian circadian clock. *Nature* 416: 286–290, 2002.

[34260] 7812. Platzer, J.; Engel, J.; Schrott-Fischer, A.; Stephan, K.; Bova, S.; Chen, H.; Zheng, H.; Striessnig, J.: Congenital deafness and sinoatrial node dysfunction in mice lacking class D L-type Ca^{2+} channels. *Cell* 102: 89–97, 2000.

[34261] 7813. Seino, S.; Chen, L.; Seino, M.; Blondel, O.; Takeda, J.; Johnson, J. H.; Bell, G. I.: Cloning of the α -1 subunit of a voltage-dependent calcium channel expressed in pancreatic beta-cells. *Proc. Nat. Acad. Sci.* 89: 584–588, 1992.

[34262] 7814. Seino, S.; Yamada, Y.; Espinosa, R., III; Le Beau, M. M.; Bell, G. I.: Assignment of the gene encoding the α -1 subunit of the neuroendocrine/brain-type calcium channel (CACNL1A2) to human chromosome 3, band p14.3. *Genomics* 13: 1375–1377, 1992.

[34263] 7815. Powers, P. A.; Liu, S.; Hogan, K.; Gregg, R. G.: Skeletal muscle and brain isoforms of a beta-subunit of human

voltage-dependent calcium channels are encoded by a single gene. *J. Biol. Chem.* 267: 22967–22972, 1992.

[34264] 7816. Gregg, R. G.; Messing, A.; Strube, C.; Beurg, M.; Moss, R.; Behan, M.; Sukhareva, M.; Haynes, S.; Powell, J. A.; Coronado, R.; Powers, P. A.: Absence of the beta subunit (cchb1) of the skeletal muscle dihydropyridine receptor alters expression of the alpha-1 subunit and eliminates excitation-contraction coupling. *Proc. Nat. Acad. Sci.* 93: 13961–13966, 1996.

[34265] 7817. Iles, D. E.; Segers, B.; Sengers, R. C. A.; Monsieurs, K.; Heytens, L.; Halsall, P. J.; Hopkins, P. M.; Ellis, F. R.; Hall-Curran, J. L.; Stewart, A. D.; Wieringa, B.: Genetic mapping of the beta-1- and gamma-subunits of the human skeletal muscle L-type voltage-dependent calcium channel on chromosome 17q and exclusion as candidate genes for malignant hyperthermia susceptibility. *Hum. Molec. Genet.* 2: 863–868, 1993.

[34266] 7818. Pragnell, M.; Sakamoto, J.; Jay, S. D.; Campbell, K. P.: Cloning and tissue-specific expression of the brain calcium channel beta-subunit. *FEBS Lett.* 291: 253–258, 1991.

[34267] 7819. Bulun, S. E.: Aromatase deficiency in women and men: would you have predicted the phenotypes? *J. Clin.*

Endocr. Metab. 81: 867–871,1996.

- [34268] 7820.Carani, C.; Qin, K.; Simoni, M.; Faustini-Fustini, M.; Serpente,S.; Boyd, J.; Korach, K. S.; Simpson, E. R.: Effect of testosteroneand estradiol in a man with aromatase deficiency. New Eng. J. Med. 337:91–95, 1997.
- [34269] 7821.Chen, S.; Besman, M. J.; Sparkes, R. S.; Zollman, S.; Klisak, I.;Mohandas, T.; Hall, P. F.; Shively, J. E.: Human aromatase: cDNAcloning, Southern blot analysis, and assignment of the gene to chromosome15. DNA 7: 27–38, 1988.
- [34270] 7822.Chen, S.; Shively, J. E.; Nakajin, S.; Shinoda, M.; Hall, P. F.: Amino terminal sequence analysis of human placenta aromatase. Biochem.Biophys. Res. Commun. 135: 713–719, 1986.
- [34271] 7823.Conte, F. A.; Grumbach, M. M.; Ito, Y.; Fisher, C. R.; Simpson,E. R.: A syndrome of female pseudohermaphrodism, hypergonadotropichypogonadism, and multicystic ovaries associated with missense mutationsin the gene encoding aromatase (P450arom). J. Clin. Endocr. Metab. 78:1287–1292, 1994.
- [34272] 7824.Corbin, C. J.; Graham-Lorence, S.; McPhaul, M.; Mason, J. I.; Mendelson,C. R.; Simpson, E. R.: Isolation of a full-length cDNA insert encodinghuman aromatase system

cytochrome P-450 and its expression in nonsteroidogenic cells. *Proc. Nat. Acad. Sci.* 85: 8948–8952, 1988.

- [34273] 7825. Deladoey, J.; Fluck, C.; Bex, M.; Yoshimura, N.; Harada, N.; Mullis, P. E.: Aromatase deficiency caused by a novel P450(arom) gene mutation: impact of absent estrogen production on serum gonadotropin concentration in a boy. *J. Clin. Endocr. Metab.* 84: 4050–4054, 1999.
- [34274] 7826. Ellis, J. A.; Stebbing, M.; Harrap, S. B.: Significant population variation in adult male height associated with the Y chromosome and the aromatase gene. *J. Clin. Endocr. Metab.* 86: 4147–4150, 2001.
- [34275] 7827. Evans, C. T.; Ledesma, D. B.; Schulz, T. Z.; Simpson, E. R.; Mendelson, C. R.: Isolation and characterization of a complementary DNA specific for human aromatase–system cytochrome P-450 mRNA. *Proc. Nat. Acad. Sci.* 83: 6387–6391, 1986.
- [34276] 7828. Fisher, C. R.; Graves, K. H.; Parlow, A. F.; Simpson, E. R.: Characterization of mice deficient in aromatase (ArKO) because of targeted disruption of the *cyp19* gene. *Proc. Nat. Acad. Sci.* 95: 6965–6970, 1998.
- [34277] 7829. George, F. W.; Matsumine, H.; McPhaul, M. J.; Somes, R. G., Jr.; Wilson, J. D.: Inheritance of the henry feathering trait in the Golden Campine chicken: evidence for allelism

with the gene that causes henneyfeathering in the Sebright Bantam. *J. Hered.* 81: 107–110, 1990.

[34278] 7830. George, F. W.; Wilson, J. D.: Pathogenesis of the henney feathering trait in the Sebright Bantam chicken. *J. Clin. Invest.* 66: 57–65, 1980.

[34279] 7831. Harada, N.: Cloning of a complete cDNA encoding human aromatase: immunochemical identification and sequence analysis. *Biochem. Biophys. Res. Commun.* 156: 725–732, 1988.

[34280] 7832. Harada, N.; Ogawa, H.; Shozu, M.; Yamada, K.: Genetic studies to characterize the origin of the mutation in placental aromatase deficiency. *Am. J. Hum. Genet.* 51: 666–672, 1992.

[34281] 7833. Harada, N.; Ogawa, H.; Shozu, M.; Yamada, K.; Suhara, K.; Nishida, E.; Takagi, Y.: Biochemical and molecular genetic analyses on placental aromatase (P-450-AROM) deficiency. *J. Biol. Chem.* 267: 4781–4785, 1992.

[34282] 7834. Hemsell, D. L.; Edman, C. D.; Marks, J. F.; Siiteri, P. K.; MacDonald, P. C.: Massive extraglandular aromatization of plasma androstenedione resulting in feminization of a prepubertal boy. *J. Clin. Invest.* 60: 455–464, 1977.

[34283] 7835. Ito, Y.; Fisher, C. R.; Conte, F. A.; Grumbach, M. M.;

Simpson, E. R.: Molecular basis of aromatase deficiency in an adult female with sexual infantilism and polycystic ovaries. *Proc. Nat. Acad. Sci.* 90: 11673–11677, 1993.

[34284] 7836. Leiberman, E.; Zachmann, M.: Familial adrenal feminization probably due to increased steroid aromatization. *Hormone Res.* 37: 96–102, 1992.

[34285] 7837. Leshin, M.; Baron, J.; George, F. W.; Wilson, J. D.: Increased estrogen formation and aromatase activity in fibroblasts cultured from the skin of chickens with the Henny feathering trait. *J. Biol. Chem.* 256: 4341–4344, 1981.

[34286] 7838. Leshin, M.; George, F. W.; Wilson, J. D.: Increased estrogen synthesis in the Sebright bantam is due to a mutation that causes increased aromatase activity. *Trans. Assoc. Am. Phys.* 94: 97–105, 1981.

[34287] 7839. Mango, D.; Montemurro, A.; Scirpa, P.; Bompiani, A.; Menini, E.: Four cases of pregnancy with low estrogen production due to placental enzymatic deficiency. *Europ. J. Obstet. Gynec. Reprod. Biol.* 8: 65–71, 1978.

[34288] 7840. McTernan, P. G.; Anderson, L. A.; Anwar, A. J.; Eggo, M. C.; Crocker, J.; Barnett, A. H.; Stewart, P. M.; Kumar, S.: Glucocorticoid regulation of P450 aromatase activity in human adipose tissue: gender and site differences. *J. Clin.*

Endocr. Metab. 87: 1327–1336, 2002.

- [34289] 7841. Morishima, A.; Grumbach, M. M.; Simpson, E. R.; Fisher, C.; Qin, K.: Aromatase deficiency in male and female siblings caused by a novel mutation and the physiological role of estrogens. *J. Clin. Endocr. Metab.* 80: 3689–3698, 1995.
- [34290] 7842. Mullis, P. E.; Yoshimura, N.; Kuhlmann, B.; Lippuner, K.; Jaeger, P.; Harada, H.: Aromatase deficiency in a female who is compound heterozygote for two new point mutations in the P450(arom) gene: impact of estrogens on hypergonadotropic hypogonadism, multicystic ovaries, and bone densitometry in childhood. *J. Clin. Endocr. Metab.* 82: 1739–1745, 1997.
- [34291] 7843. Phornphutkul, C.; Okubo, T.; Wu, K.; Harel, Z.; Tracy, T. F., Jr.; Pinar, H.; Chen, S.; Gruppuso, P. A.; Goodwin, G.: Aromatase P450 expression in a feminizing adrenal adenoma presenting as isosexual precocious puberty. *J. Clin. Endocr. Metab.* 86: 649–652, 2001.
- [34292] 7844. Robertson, K. M.; O'Donnell, L.; Jones, M. E. E.; Meachem, S. J.; Boon, W. C.; Fisher, C. R.; Graves, K. H.; McLachlan, R. I.; Simpson, E. R.: Impairment of spermatogenesis in mice lacking a functional aromatase (cyp 19) gene. *Proc. Nat. Acad. Sci.* 96: 7986–7991, 1999.

- [34293] 7845. Sebastian, S.; Bulun, S. E.: A highly complex organization of the regulatory region of the human CYP19 (aromatase) gene revealed by the Human Genome Project. *J. Clin. Endocr. Metab.* 86: 4600–4602, 2001.
- [34294] 7846. Mohr, J.; Eiberg, H.: Colton blood groups: indication of linkage with the Kidd (Jk) system as support for assignment to chromosome 7. *Clin. Genet.* 11: 372–374, 1977.
- [34295] 7847. Everman, D. B.; Bartels, C. F.; Yang, Y.; Yanamandra, N.; Goodman, F. R.; Mendoza-Londono, J. R.; Savarirayan, R.; White, S. M.; Graham, J. M., Jr.; Gale, R. P.; Svarch, E.; Newman, W. G.; Kleckers, A. R.; Francomano, C. A.; Govindaiah, V.; Singh, L.; Morrison, S.; Thomas, J. T.; Warman, M. L.: The mutational spectrum of brachydactyly type C. *Am. J. Med. Genet.* 112: 291–296, 2002.
- [34296] 7848. Galjaard, R. J. H.; van der Ham, L. I.; Posch, N. A. S.; Dijkstra, P. F.; Oostra, B. A.; Hovius, S. E. R.; Timmenga, E. J. F.; Sonneveld, G. J.; Hoogeboom, A. J. M.; Heutink, P.: Differences in complexity of isolated brachydactyly type C cannot be attributed to locus heterogeneity alone. *Am. J. Med. Genet.* 98: 256–262, 2001.
- [34297] 7849. Haws, D. V.: Inherited brachydactyly and hypoplasia of the bones of the extremities. *Ann. Hum. Genet.* 26: 201–212, 1963.

- [34298] 7850. Polinkovsky, A.; Robin, N. H.; Thomas, J. T.; Irons, M.; Lynn, A.; Goodman, F. R.; Reardon, W.; Kant, S. G.; Brunner, H. G.; vander Burgt, I.; Chitayat, D.; McGaughan, J.; Donnai, D.; Luyten, F. P.; Warman, M. L.: Mutations in CDMP1 cause autosomal dominant brachydactyly type C. (Letter) *Nature Genet.* 17: 18–19, 1997.
- [34299] 7851. Polymeropoulos, M. H.; Ide, S. E.; Magyari, T.; Francomano, C. A.: Brachydactyly type C gene maps to human chromosome 12q24. *Genomics* 38:45–50, 1996.
- [34300] 7852. Robin, N. H.; Gunay-Aygun, M.; Polinkovsky, A.; Warman, M. L.; Morrison, S.: Clinical and locus heterogeneity in brachydactyly type C. *Am. J. Med. Genet.* 68: 369–377, 1997.
- [34301] 7853. Storm, E. E.; Huynh, T. V.; Copeland, N. G.; Jenkins, N. A.; Kingsley, D. M.; Lee, S. J.: Limb alterations in brachypodism mice due to mutations in a new member of the TGF- β superfamily. *Nature* 368: 639–643, 1994.
- [34302] 7854. Dayhoff, M. O.: *Atlas of Protein Sequence and Structure. Hormones, active peptides and toxins.* Washington: National Biomedical Research Foundation (pub.) 5: 1972. Pp. D205 only.
- [34303] 7855. Edbrooke, M. R.; Parker, D.; McVey, J. H.; Riley, J. H.; Sorenson, G. D.; Pettengill, O. S.; Craig, R. K.: Expression

of the human calcitonin/CGRP gene in lung and thyroid carcinoma. EMBO J. 4: 715–724, 1985.

- [34304] 7856. Girgis, S. I.; Macdonald, D. W. R.; Stevenson, J. C.; Bevis, P. J. R.; Lynch, C.; Wimalawansa, S. J.; Self, C. H.; Morris, H. R.; MacIntyre, I.: Calcitonin gene-related peptide: potent vasodilator and major product of calcitonin gene. Lancet II: 14–16, 1985.
- [34305] 7857. Goltzman, D.; Mitchell, J.: Interaction of calcitonin and calcitonin gene-related peptide at receptor sites in target tissues. Science 227:1343–1345, 1985.
- [34306] 7858. Henry, I.; Grandjouan, S.; Barichard, F.; Huerre-Jeanpierre, C.; Junien, C.: Mitotic deletions of 11p15.5 in two different tumors indicate that the CALCA locus is distal to the PTH locus. Cytogenet. Cell Genet. 50: 155–157, 1989.
- [34307] 7859. Hillyard, C. J.; Myers, C.; Abeyasekera, G.; Stevenson, J. C.; Craig, R. K.; MacIntyre, I.: Katalcalcin: a new plasma calcium-lowering hormone. Lancet I: 846–848, 1983.
- [34308] 7860. Hoovers, J. M. N.; Redeker, E.; Speleman, F.; Hoppenier, J. W. M.; Bhola, S.; Blik, J.; van Roy, N.; Leschot, N. J.; Westerveld, A.; Mannens, M.: High-resolution chromosomal localization of the human calcitonin/CGRP/IAPP

gene family members. *Genomics* 15: 525–529,1993.

[34309] 7861.Hoppener, J. W. M.; Steenbergh, P. H.; Zandberg, J.; Adema, G.J.; Geurts van Kessel, A. H. M.; Lips, C. J. M.; Jansz, H. S.: A third human CALC (pseudo)gene on chromosome 11. *FEBS Lett.* 233:57–63, 1988.

[34310] 7862.Hoppener, J. W. M.; Steenbergh, P. H.; Zandberg, J.; Bakker, E.; Pearson, P. L.; Geurts van Kessel, A. H. M.; Jansz, H. S.; Lips, C.J. M.: Localization of the polymorphic human calcitonin gene on chromosome 11. *Hum. Genet.* 66: 309–312, 1984.

[34311] 7863.Jacobs, J. W.; Goodman, R. H.; Chin, W. W.; Dee, P. C.; Habener, J. F.; Bell, N. H.; Potts, J. T., Jr.: Calcitonin messenger RNA encodes multiple polypeptides in a single precursor. *Science* 213: 457–459,1981.

[34312] 7864.Jonas, V.; Lin, C. R.; Kawashima, E.; Semon, D.; Swanson, L. W.; Mermoud, J.-J.; Evans, R. M.; Rosenfeld, M. G.: Alternative RNA processing events in human calcitonin/calcitonin gene-related peptide gene expression. *Proc.Nat. Acad. Sci.* 82: 1994–1998, 1985.

[34313] 7865.Kittur, S. D.; Hoppener, J. W. M.; Antonarakis, S. E.; Daniels, J. D. J.; Meyers, D. A.; Maestri, N. E.; Jansen, M.; Korneluk, R.G.; Nelkin, B. D.; Kazazian, H. H., Jr.: Linkage map of the short arm of human chromosome 11: location

of the genes for catalase, calcitonin, and insulin-like growth factor II. *Proc. Nat. Acad. Sci.* 82: 5064–5067, 1985.

- [34314] 7866. MacIntyre, I.; Hillyard, C. J.; Murphy, P. K.; Reynolds, J. J.; Gaines-Das, R. E.; Craig, R. K.: A second plasma calcium-lowering peptide from the human calcitonin precursor. *Nature* 300: 460–462, 1982.
- [34315] 7867. Mathe, A. A.; Agren, H.; Lindstrom, L.; Theodorsson, E.: Increased concentration of calcitonin gene-related peptide in cerebrospinal fluid of depressed patients: a possible trait marker of major depressive disorder. *Neurosci. Lett.* 182: 138–142, 1994.
- [34316] 7868. Neher, R.; Riniker, B.; Rittel, W.; Zuber, H.: Thyrocalcitonin. II. Struktur von alpha-Thyrocalcitonin. *Helv. Chim. Acta* 51: 917–924, 1968.
- [34317] 7869. New, H. V.; Mudge, A. W.: Calcitonin gene-related peptide regulates muscle acetylcholine receptor synthesis. *Nature* 323: 809–811, 1986.
- [34318] 7870. Przepiorka, D.; Baylin, S. B.; McBride, D. W.; Testa, J. R.; deBustros, A.; Nelkin, B. D.: The human calcitonin gene is located on the short arm of chromosome 11. *Biochem. Biophys. Res. Commun.* 120: 493–499, 1984.
- [34319] 7871. Rosenfeld, M. G.; Lin, C. R.; Amara, S. G.; Stolarsky,

L.; Roos, B. A.; Ong, E. S.; Evans, R. M.: Calcitonin mRNA polymorphism: peptideswitching associated with alternative RNA splicing events. *Proc. Nat. Acad. Sci.* 79: 1717–1721, 1982.

[34320] 7872. Vora, S.; Seaman, C.; Durham, S.; Piomelli, S.: Isozymes of human phosphofructokinase: identification and subunit structural characterization of a new system. *Proc. Nat. Acad. Sci.* 77: 62–66, 1980.

[34321] 7873. Pizon, V.; Chardin, P.; Leroisey, I.; Olofsson, B.; Tavian, A.: Human cDNAs RAP1 and RAP2 homologous to the *Drosophila* gene *Dras3* encode proteins closely related to ras in the 'effector' region. *Oncogene* 3:201–204, 1988.

[34322] 7874. Mimmack, M. L.; Ryan, M.; Baba, H.; Navarro-Ruiz, J.; Iritani, S.; Faull, R. L. M.; McKenna, P. J.; Jones, P. B.; Arai, H.; Starkey, M.; Emson, P. C.; Bahn, S.: Gene expression analysis in schizophrenia: reproducible up-regulation of several members of the apolipoprotein L family located in a high-susceptibility locus for schizophrenia on chromosome 22. *Proc. Nat. Acad. Sci.* 99: 4680–4685, 2002.

[34323] 7875. Schmauss, C.; McAllister, G.; Ohosone, Y.; Hardin, J. A.; Lerner, M. R.: A comparison of snRNP-associated Sm-autoantigens: human N, rat N and human B/B-prime. *Nucleic Acids Res.* 17: 1733–1743, 1989.

- [34324] 7876.Kovacs, G.; Kung, H.: Nonhomologous chromatid exchange in hereditary and sporadic renal cell carcinomas. Proc. Nat. Acad. Sci. 88: 194–198, 1991.
- [34325] 7877.Montini, E.; Rugarli, E. I.; Van de Vosse, E.; Andolfi, G.; Mariani, M.; Puca, A. A.; Consalez, G. G.; den Dunnen, J. T.; Ballabio, A.; Franco, B.: A novel human serine–threonine phosphatase related to the *Drosophila* retinal degeneration C (rdgC) gene is selectively expressed in sensory neurons of neural crest origin. Hum. Molec. Genet. 6:1137–1145, 1997.
- [34326] 7878.Johnson, M. D.; Tho, S. P. T.; Behzadian, A.; McDonough, P. G.: Molecular scanning of Yq11 (interval 6) in men with Sertoli–cell–only syndrome. Am. J. Obstet. Gynec. 161: 1732–1737, 1989.
- [34327] 7879.Bolger, G.; Michaeli, T.; Martins, T.; St. John, T.; Steiner, B.; Rodgers, L.; Riggs, M.; Wigler, M.; Ferguson, K.: A family of human phosphodiesterases homologous to the *Drosophila* dunce learning and memory gene product of *Drosophila melanogaster* are potential targets for antidepressant–drugs. Molec. Cell. Biol. 13: 6558–6571, 1993.
- [34328] 7880.Argyrokastitis, A.; Kamakari, S.; Kapsetaki, M.; Kritis, A.; Talianidis, I.; Moschonas, N. K.: Human hepatocyte nuclear factor–4 (hHNF–4) gene maps to 20q12–q13.1 be–

tween PLCG1 and D20S17. Hum. Genet. 99:233–236, 1997.

[34329] 7881.Coovert, D. D.; Le, T. T.; Morris, G. E.; Man, N. T.; Kralewski, M.; Sendtner, M.; Burghes, A. H. M.: Does the survival motor neuron protein (SMN) interact with Bcl-2? (Letter) J. Med. Genet. 37: 536–539, 2000.

[34330] 7882.Nizet, V.; Ohtake, T.; Lauth, X.; Trowbridge, J.; Rudisill, J.; Dorschner, R. A.; Pestonjamas, V.; Piraino, J.; Huttner, K.; Gallo, R. L.: Innate antimicrobial peptide protects the skin from invasive bacterial infection. Nature 414: 454–457, 2001.

[34331] 7883.Weaver, D. R.; Rivkees, S. A.; Carlson, L. L.; Reppert, S. M.: Localization of melatonin receptors in mammalian brain. In: Klein, D. C.; Moore, R. Y.; Reppert, S. M.: Suprachiasmatic Nucleus: The Mind's Clock. New York: Oxford Press (pub.) 1991.

[34332] 7884.Bohm, H.; Gross, B.; Gaestel, M.; Bommer, U.-A.; Ryffel, G.; Bielka, H.: The 5-prime-untranslated region of p23 mRNA from the Ehrlich ascites tumor is involved in translation control of the growth related protein p23. Biomed. Biochim. Acta 50: 1193–1203, 1991.

[34333] 7885.Wei, Y.-F.; Robins, P.; Carter, K.; Caldecott, K.; Pappin, D. J. C.; Yu, G.-L.; Wang, R.-P.; Shell, B. K.; Nash, R. A.;

Schar, P.; Barnes, D. E.; Haseltine, W. A.; Lindahl, T.: Molecular cloning and expression of human cDNAs encoding a novel DNA ligase IV and DNA ligase III, an enzyme active in DNA repair and recombination. *Molec. Cell. Biol.* 15: 3206–3216, 1995.

[34334] 7886. Pickard, R. T.; Striffler, B. A.; Kramer, R. M.; Sharp, J. D.: Molecular cloning of two new human paralogs of 85-kDa cytosolic phospholipase A₂. *J. Biol. Chem.* 274: 8823–8831, 1999.

[34335] 7887. Hegele, R. A.; Cao, H.; Harris, S. B.; Zinman, B.; Hanley, A. J. G.; Anderson, C. M.: Peroxisome proliferator-activated receptor- γ P12A and type 2 diabetes in Canadian Oji-Cree. *J. Clin. Endocr. Metab.* 85: 2014–2019, 2000.

[34336] 7888. Chua, A. O.; Chizzonite, R.; Desai, B. B.; Truitt, T. P.; Nunes, P.; Minetti, L. J.; Warriar, R. R.; Presky, D. H.; Levine, J. F.; Gately, M. K.; Gubler, U.: Expression cloning of a human IL-12 receptor component: a new member of the cytokine receptor superfamily with strong homology to gp130. *J. Immun.* 153: 128–136, 1994.

[34337] 7889. Wedemeyer, N.; Peoples, R.; Himmelbauer, H.; Lehrach, H.; Francke, U.; Wanker, E. E.: Localization of the human HIP1 gene close to the elastin (ELN) locus on

7q11.23. Genomics 46: 313–315, 1997.

- [34338] 7890.Zetterstrom, R. H.; Solomin, L.; Jansson, L.; Hoffer, B. J.; Olson,L.; Perlmann, T.: Dopamine neuron agenesis in Nurr1-deficient mice. Science 276:248–250, 1997.
- [34339] 7891.Gong, D.–W.; Monemdjou, S.; Gavrilova, O.; Leon, L. R.; Marcus–Samuels,B.; Chou, C. J.; Everett, C.; Kozak, L. P.; Li, C.; Deng, C.; Harper,M.–E.; Reitman, M. L.: Lack of obesity and normal response to fastingand thyroid hormone in mice lacking uncoupling protein–3. J. Biol.Chem. 275: 16251–16257, 2000.
- [34340] 7892.Palmiter, R. D.; Cole, T. B.; Findley, S. D.: ZnT–2, a mammalianprotein that confers resistance to zinc by facilitating vesicularsequestration. EMBO J. 15: 1784–1791, 1996.
- [34341] 7893.Bryan, J.; Kane, R. E.: Actin gelation in sea urchin egg extracts. MethodsCell Biol. 25: 175–199, 1982.
- [34342] 7894.Dosanjh, M. K.; Collins, D. W.; Fan, W.; Lennon, G. G.; Albala,J. S.; Shen, Z.; Schild, D.: Isolation and characterization of RAD51C,a new human member of the RAD51 family of related genes. NucleicAcids Res. 26: 1179–1184, 1998.
- [34343] 7895.Carpino, N.; Wisniewski, D.; Strife, A.; Marshak, D.; Kobayashi,R.; Stillman, B.; Clarkson, B.: p62(dok): a con–

stitutively tyrosine-phosphorylated,GAP-associated protein in chronic myelogenous leukemia progenitorcells. Cell 88: 197-204, 1997.

- [34344] 7896.Bittner, R. E.; Anderson, L. V. B.; Burkhardt, E.; Bashir, R.;Vafiadaki, E.; Ivanova, S.; Raffelsberger, T.; Maerk, I.; Hoyer, H.;Jung, M.; Karbasiyan, M.; Storch, M.; Lassmann, H.; Moss, J. A.; Davison,K.; Harrison, R.; Bushby, K. M. D.; Reis, A.: Dysferlin deletionin SJL mice (SJL-Dysf) defines a natural model for limb girdle muscular dystrophy 2B. (Letter) Nature Genet. 23: 141-142, 1999.
- [34345] 7897.Koi, M.; Johnson, L. A.; Kalikin, L. M.; Little, P. F. R.; Nakamura,Y.; Feinberg, A. P.: Tumor cell growth arrest caused by subchromosomaltransferable DNA fragments from chromosome 11. Science 260: 361-364,1993.
- [34346] 7898.Makoff, A.; Pilling, C.; Harrington, K.; Emson, P.: Human metabotropicglutamate receptor type 7: molecular cloning and mRNA distributionin the CNS. Molec. Brain Res. 40: 165-170, 1996.
- [34347] 7899.Esterbauer, H.; Oberkofler, H.; Krempler, F.; Patsch, W.: Humanperoxisome proliferator activated receptor gamma coactivator 1 (PPARGC1)gene: cDNA sequence, genomic organization, chromosomal localization,and tis-

sue expression. *Genomics* 62: 98–102, 1999.

- [34348] 7900. Lietzke, S. E.; Bose, S.; Cronin, T.; Klarlund, J.; Chawla, A.; Czech, M. P.; Lambright, D. G.: Structural basis of 3-phosphoinositide recognition by pleckstrin homology domains. *Molec. Cell* 6: 385–394, 2000.
- [34349] 7901. Xu, F.; Xia, W.; Luo, R. Z.; Peng, H.; Zhao, S.; Dai, J.; Long, Y.; Zou, L.; Le, W.; Liu, J.; Parlow, A. F.; Hung, M.-C.; Bast, R.C., Jr.; Yu, Y.: The human ARHI tumor suppressor gene inhibits lactation and growth in transgenic mice. *Cancer Res.* 60: 4913–4920, 2000.
- [34350] 7902. Tsuchida, K.; Arai, K. Y.; Kuramoto, Y.; Yamakawa, N.; Hasegawa, Y.; Sugino, H.: Identification and characterization of a novel follistatin-like protein as a binding protein for the TGF- β family. *J. Biol. Chem.* 275: 40788–40796, 2000.
- [34351] 7903. Takatsu, H.; Yoshino, K.; Nakayama, K.: Adaptor gamma-ear homology domain conserved in gamma-adaptin and GGA proteins that interact with gamma-synergin. *Biochem. Biophys. Res. Commun.* 271: 719–725, 2000.
- [34352] 7904. Burbelo, P. D.; Snow, D. M.; Bahou, W.; Spiegel, S.: MSE55, a Cdc42 effector protein, induces long cellular extensions in fibroblasts. *Proc. Nat. Acad. Sci.* 96:

9083–9088, 1999.

- [34353] 7905.Tsavalier, L.; Shaper, M. H.; Morkowski, S.; Laus, R.: Trp-p8, a novel prostate-specific gene, is up-regulated in prostate cancer and other malignancies and shares high homology with transient receptor potential calcium channel proteins. *Cancer Res.* 61: 3760–3769, 2001.
- [34354] 7906.Rafi, M. A.; Luzi, P.; Chen, Y. Q.; Wenger, D. A.: A large deletion together with a point mutation in the GALC gene is a common mutant allele in patients with infantile Krabbe disease. *Hum. Molec. Genet.* 4:1285–1289, 1995.
- [34355] 7907.Ohta, T.; Michel, J. J.; Schottelius, A. J.; Xiong, Y.: ROC1, a homolog of APC11, represents a family of cullin partners with an associated ubiquitin ligase activity. *Molec. Cell* 3: 535–541, 1999.
- [34356] 7908.Andreev, J.; Simon, J.-P.; Sabatini, D. D.; Kam, J.; Plowman, G.; Randazzo, P. A.; Schlessinger, J.: Identification of a new Pyk2 target protein with Arf-GAP activity. *Molec. Cell. Biol.* 19: 2338–2350, 1999.
- [34357] 7909.Burden, S.; Yarden, Y.: Neuregulins and their receptors: a versatile signaling module in organogenesis and oncogenesis. *Neuron* 18: 847–855, 1997.
- [34358] 7910.Busfield, S. J.; Michnick, D. A.; Chickering, T. W.; Revett, T.L.; Ma, J.; Woolf, E. A.; Comrack, C. A.; Dussault,

B. J.; Woolf, J.; Goodearl, A. D. J.; Gearing, D. P.: Characterization of a neuregulin-related gene, Don-1, that is highly expressed in restricted regions of the cerebellum and hippocampus. *Molec. Cell. Biol.* 17: 4007–4014, 1997.

[34359] 7911. Carraway, K. L., III; Weber, J. L.; Unger, M. J.; Ledesma, J.; Yu, N.; Gassmann, M.; Lai, C.: Neuregulin-2, a new ligand of ErbB3/ErbB4-receptor tyrosine kinases. *Nature* 387: 512–516, 1997.

[34360] 7912. Chang, H.; Riese, D. J., II; Gilbert, W.; Stern, D. F.; McMahon, U. J.: Ligands for ErbB-family receptors encoded by a neuregulin-like gene. *Nature* 387: 509–512, 1997.

[34361] 7913. Ring, H. Z.; Chang, H.; Guilbot, A.; Brice, A.; LeGuern, E.; Francke, U.: The human neuregulin-2 (NRG2) gene: cloning, mapping and evaluation as a candidate for the autosomal recessive form of Charcot-Marie-Tooth disease linked to 5q. *Hum. Genet.* 104: 326–332, 1999.

[34362] 7914. Yamada, K.; Ichino, N.; Nishii, K.; Sawada, H.; Higashiyama, S.; Ishiguro, H.; Nagatsu, T.: Characterization of the human NTAK gene structure and distribution of the isoforms for rat NTAK mRNA. *Gene* 255:15–24, 2000.

[34363] 7915. Hu, R.-J.; Lee, M. P.; Connors, T. D.; Johnson, L. A.; Burn, T. C.; Su, K.; Landes, G. M.; Feinberg, A. P.: A 2.5-Mb

transcript map of a tumor-suppressing subchromosomal transferable fragment from 11p15.5, and isolation and sequence analysis of three novel genes. *Genomics* 46:9–17, 1997.

- [34364] 7916. Stephan, D.; Bon, C.; Holzwarth, J. A.; Galvan, M.; Pruss, R.M.: Human metabotropic glutamate receptor 1: mRNA distribution, chromosomal localization and functional expression of two splice variants. *Neuropharmacology* 35:1649–1660, 1996.
- [34365] 7917. Chen, M. S.; Huber, A. B.; van der Haar, M. E.; Frank, M.; Schnell, L.; Spillmann, A. A.; Christ, F.; Schwab, M. E.: Nogo-A is a myelin-associated neurite outgrowth inhibitor and an antigen for monoclonal antibody IN-1. *Nature* 403: 434–439, 2000.
- [34366] 7918. Fournier, A. E.; GrandPre, T.; Strittmatter, S. M.: Identification of a receptor mediating Nogo-66 inhibition of axonal regeneration. *Nature* 409:341–346, 2001.
- [34367] 7919. GrandPre, T.; Li, S.; Strittmatter, S. M.: Nogo-66 receptor antagonist peptide promotes axonal regeneration. *Nature* 417: 547–551, 2002.
- [34368] 7920. GrandPre, T.; Nakamura, F.; Vartanian, T.; Strittmatter, S. M.: Identification of the Nogo inhibitor of axon regeneration as a reticulon protein. *Nature* 403: 439–444,

2000.

- [34369] 7921. Prinjha, R.; Moore, S. E.; Vinson, M.; Blake, S.; Morrow, R.; Christie, G.; Michalovich, D.; Simmons, D. L.; Walsh, F. S.: Inhibitor of neurite outgrowth in humans. (Letter) *Nature* 403: 383–384, 2000.
- [34370] 7922. Spillmann, A. A.; Bandtlow, C. E.; Lottspeich, F.; Keller, F.; Schwab, M. E.: Identification and characterization of a bovine neurite growth inhibitor (bNI-220). *J. Biol. Chem.* 273: 19283–19293, 1998.
- [34371] 7923. Yang, J.; Yu, L.; Bi, A. D.; Zhao, S.-Y.: Assignment of the human reticulon 4 gene (RTN4) to chromosome 2p14–2p13 by radiation hybrid mapping. *Cytogenet. Cell Genet.* 88: 101–102, 2000.
- [34372] 7924. Korner, C. G.; Wahle, E.: Poly(A) tail shortening by a mammalian poly(A)-specific 3-prime-exoribonuclease. *J. Biol. Chem.* 272: 10448–10456, 1997.
- [34373] 7925. Korner, C. G.; Wormington, M.; Muckenthaler, M.; Schneider, S.; Dehlin, E.; Wahle, E.: The deadenylating nuclease (DAN) is involved in poly(A) tail removal during the meiotic maturation of *Xenopus* oocytes. *EMBO J.* 17: 5427–5437, 1998.
- [34374] 7926. Lofberg, H.; Stromblad, L.-G.; Grubb, A. O.; Olsson, S.-O.: Demonstration of gamma-trace in normal and neo-

plastic endocrine A-cells of the pancreatic islets: an immunohistochemical study in monkey, rat and man.

Biomed.Res. 2: 527–535, 1981.

[34375] 7927.Merz, G. S.; Benedikz, E.; Schwenk, V.; Johansen, T. E.; Vogel, L. K.; Rushbrook, J. I.; Wisniewski, H. M.: Human cystatin C forms an inactive dimer during intracellular trafficking in transfected CHO cells. J. Cell. Physiol. 173: 423–432, 1997.

[34376] 7928.Schnittger, S.; Gopal Rao, V. V. N.; Abrahamson, M.; Hansmann, I.: Cystatin C (CST3), the candidate gene for hereditary cystatin C amyloid angiopathy (HCCAA), and other members of the cystatin gene family are clustered on chromosome 20p11.2. Genomics 16: 50–55, 1993.

[34377] 7929.Shi, G.-P.; Sukhova, G. K.; Grubb, A.; Ducharme, A.; Rhode, L.H.; Lee, R. T.; Ridker, P. M.; Libby, P.; Chapman, H. A.: Cystatin C deficiency in human atherosclerosis and aortic aneurysms. J. Clin. Invest. 104: 1191–1197, 1999.

[34378] 7930.Ai, Y.; Jenkins, N. A.; Copeland, N. G.; Gilbert, D. J.; Bergsma, D. J.; Stambolian, D.: Mouse galactokinase: isolation, characterization, and location on chromosome 11. Genome Res. 5: 53–59, 1995.

[34379] 7931.Ai, Y.; Zheng, Z.; O'Brien-Jenkins, A.; Bernard, D. J.; Wynshaw-Boris, T.; Ning, C.; Reynolds, R.; Segal, S.;

Huang, K.; Stambolian, D.: A mouse model of galactose-induced cataracts. *Hum. Molec. Genet.* 9:1821–1827, 2000.

- [34380] 7932. Bergsma, D. J.; Ai, Y.; Skach, W. R.; Nesburn, K.; Anoaia, E.; VanHorn, S.; Stambolian, D.: Fine structure of the human galactokinase GALK1 gene. *Genome Res.* 6: 980–985, 1996.
- [34381] 7933. Croce, C. M.; Huebner, K.; Koprowski, H.: Chromosome assignment of the T-antigen gene of simian virus 40 in African green monkey cells transformed by adeno 7-SV 40 hybrid. *Proc. Nat. Acad. Sci.* 71: 4116–4119, 1974.
- [34382] 7934. de Jonge, A. J. R.; de Smit, S.; Kroos, M. A.; Reuser, A. J. J.: Cotransfer of syntenic human genes into mouse cells using isolated metaphase chromosomes or cellular DNA. *Hum. Genet.* 69: 32–38, 1985.
- [34383] 7935. Elsevier, S. M.; Kucherlapati, R. S.; Nichols, E. A.; Willecke, K.; Creagan, R. P.; Giles, R. E.; McDougall, J. K.; Ruddle, F. H.: Assignment and regional localization of a gene coding for galactokinase to human chromosome 17q21–22. *Birth Defects Orig. Art. Ser.* 11(3):117–119, 1975. Note: *Alternate: Cytogenet. Cell Genet.* 14: 287–289, 1975.
- [34384] 7936. Klobutcher, L. A.; Ruddle, F. H.: Phenotype stabilization and integration of transferred material in chromo-

some-mediated gene transfer. *Nature* 280: 657–660, 1979.

[34385] 7937.Kolosha, V.; Anoaia, E.; de Cespedes, C.; Gitzelmann, R.; Shih,L.; Casco, T.; Saborio, M.; Trejos, R.; Buist, N.; Tedesco, T.; Skach,W.; Mitelmann, O.; Ledee, D.; Huang, K.; Stambolian, D.: Novel mutationsin 13 probands with galactokinase deficiency. *Hum. Mutat.* 15: 447–453,2000.

[34386] 7938.Okajima, K.; Yazaki, M.; Wada, Y.: Thymidine kinase activityin individuals with galactokinase deficiency. (Letter) *Am. J. Hum.Genet.* 41: 503–504, 1987.

[34387] 7939.Okano, Y.; Asada, M.; Fujimoto, A.; Ohtake, A.; Murayama, K.;Hsiao, K.-J.; Choeh, K.; Yang, Y.; Cao, Q.; Reichardt, J. K. V.; Niihira,S.; Imamura, T.; Yamano, T.: A genetic factor for age-related cataract:identification and characterization of a novel galactokinase variant,'Osaka,' in Asians. *Am. J. Hum. Genet.* 68: 1036–1042, 2001.

[34388] 7940.Ruddle, F. H.: Personal Communication. New Haven, Conn. 5/4/1982.

[34389] 7941.Schoen, R. C.; Cox, S. H.; Wagner, R. P.: Thymidine-kinase activityof cultured cells from individuals with inherited galactokinase deficiency. *Am.J. Hum. Genet.* 36: 815–822, 1984.

[34390] 7942.Scott, A. F.: Personal Communication. Baltimore, Md.

2/20/2001.

- [34391] 7943.Schmidt, A.; Wolde, M.; Thiele, C.; Fest, W.; Kratzin, H.; Podtelejnikov,A. V.; Witke, W.; Huttner, W. B.; Soling, H.-D.: Endophilin I mediates synaptic vesicle formation by transfer of arachidonate to lysophosphatidic acid. *Nature* 401: 133–141, 1999.
- [34392] 7944.Aiba, A.; Chen, C.; Herrup, K.; Rosenmund, C.; Stevens, C. F.; Tonegawa, S.: Reduced hippocampal long-term potentiation and context-specific deficit in associative learning in mGluR1 mutant mice. *Cell* 79:365–375, 1994.
- [34393] 7945.Aiba, A.; Kano, M.; Chen, C.; Stanton, M. E.; Fox, G. D.; Herrup,K.; Zwingman, T. A.; Tonegawa, S.: Deficient cerebellar long-term depression and impaired motor learning in mGluR1 mutant mice. *Cell* 79:377–388, 1994.
- [34394] 7946.Conquet, F.; Bashir, Z. I.; Davies, C. H.; Daniel, H.; Ferraguti,F.; Bordi, F.; Franz–Bacon, K.; Reggiani, A.; Matarese, V.; Conde,F.; Collingridge, G. L.; Crepel, F.: Motor deficit and impairment of synaptic plasticity in mice lacking mGluR1. *Nature* 372: 237–243, 1994.
- [34395] 7947.Ganesh, S.; Amano, K.; Yamakawa, K.: Assignment of the gene GRM1 coding for metabotropic glutamate receptor 1 to human chromosome band 6q24 by in situ hy-

bridization. *Cytogenet. Cell Genet.* 88: 314–315, 2000.

[34396] 7948. Ichise, T.; Kano, M.; Hashimoto, K.; Yanagihara, D.; Nakao, K.; Shigemoto, R.; Katsuki, M.; Alba, A.: mGluR1 in cerebellar Purkinje cells essential for long-term depression, synapse elimination, and motor coordination. *Science* 288: 1832–1835, 2000.

[34397] 7949. Kunishima, N.; Shimada, Y.; Tsuji, Y.; Sato, T.; Yamamoto, M.; Kumasaka, T.; Nakanishi, S.; Jingami, H.; Morikawa, K.: Structural basis of glutamate recognition by a dimeric metabotropic glutamate receptor. *Nature* 407: 971–977, 2000.

[34398] 7950. Okamoto, T.; Sekiyama, N.; Otsu, M.; Shimada, Y.; Sato, A.; Nakanishi, S.; Jingami, H.: Expression and purification of the extracellular ligand binding region of metabotropic glutamate receptor subtype 1. *J. Biol. Chem.* 273: 13089–13096, 1998.

[34399] 7951. Smitt, P. S.; Kinoshita, A.; De Leeuw, B.; Moll, W.; Coesmans, M.; Jaarsma, D.; Henzen-Logmans, S.; Vecht, C.; De Zeeuw, C.; Sekiyama, N.; Nakanishi, S.; Shigemoto, R.: Paraneoplastic cerebellar ataxia due to autoantibodies against a glutamate receptor. *New Eng. J. Med.* 342: 21–27, 2000.

[34400] 7952. Nagase, T.; Ishikawa, K.; Suyama, M.; Kikuno, R.; Hi-

rosawa, M.;Miyajima, N.; Tanaka, A.; Kotani, H.; Nomura, N.; Ohara, O.: Predictionof the coding sequences of unidentified human genes. XII. The complete sequences of 100 new cDNA clones from brain which code for large proteinsin vitro. DNA Res. 5: 355–364, 1998.

[34401] 7953.Chadwick, B. P.; Kidd, T.; Sgouros, J.; Ish-Horowicz, D.; Frischauf,A.–M.: Cloning, mapping and expression of UBL3, a novel ubiquitin–likegene. Gene 233: 189–195, 1999.

[34402] 7954.Hodges, M.; Tissot, C.; Freemont, P. S. Protein regulation:tag wrestling with relatives of ubiquitin. Curr. Biol. 8: R749–R752,1998.

[34403] 7955.Bora, R. S.; Kanamori, A.; Hirabayashi, Y.: Assignment of a putativeacetyl–CoA transporter gene (Acatn) to mouse chromosome band 3E1–E3by in situ hybridization. Cytogenet. Cell Genet. 83: 78–79, 1998.

[34404] 7956.Andree, B.; Hillemann, T.; Kessler–leckson, G.; Schmitt–John, T.;Jockusch, H.; Arnold, H.–H.; Brand, T.: Isolation and characterizationof the novel Popeye gene family expressed in skeletal muscle and heart. Dev.Biol. 223: 371–382, 2000.

[34405] 7957.Reese, D. E.; Bader, D. M.: Cloning and expression of hbves, anovel and highly conserved mRNA expressed in

the developing and adult heart and skeletal muscle in the human. *Mammalian Genome* 10: 913–915, 1999.

[34406] 7958. Reese, D. E.; Zavaljevski, M.; Streiff, N. L.; Bader, D.: Bves: a novel gene expressed during coronary blood vessel development. *Dev. Biol.* 209: 159–171, 1999.

[34407] 7959. Kowal, R. C.; Jolsin, J. M.; Olson, E. N.; Schultz, R. A.: Assignment of fibulin-5 (FBLN5) to human chromosome 14q31 by in situ hybridization and radiation hybrid mapping. *Cytogenet. Cell Genet.* 87: 2–3, 1999.

[34408] 7960. Nakamura, T.; Lozano, P. R.; Ikeda, Y.; Iwanaga, Y.; Hinek, A.; Minamisawa, S.; Cheng, C.-F.; Kobuke, K.; Dalton, N.; Takada, Y.; Tashiro, K.; Ross, J., Jr.; Honjo, T.; Chien, K. R.: Fibulin-5/DANCE is essential for elastogenesis in vivo. *Nature* 415: 171–175, 2002.

[34409] 7961. Nakamura, T.; Ruiz-Lozano, P.; Lindner, V.; Yabe, D.; Taniwaki, M.; Furukawa, Y.; Kobuke, K.; Tashiro, K.; Lu, Z.; Andon, N. L.; Schaub, R.; Matsumori, A.; Sasayama, S.; Chien, K. R.; Honjo, T.: DANCE, a novel secreted RGD protein expressed in developing, atherosclerotic, and balloon-injured arteries. *J. Biol. Chem.* 274: 22476–22483, 1999.

[34410] 7962. Yanagisawa, H.; Davis, E. C.; Starcher, B. C.; Ouchi, T.; Yanagisawa, M.; Richardson, J. A.; Olson, E. N.: Fibulin-5 is an elastin-binding protein essential for elastic fibre

development in vivo. *Nature* 415:168–171, 2002.

- [34411] 7963.Dong, F.; Feldmesser, M.; Casadevall, A.; Rubin, C. S.: Molecular characterization of a cDNA that encodes six isoforms of a novel murine A kinase anchor protein. *J. Biol. Chem.* 273: 6533–6541, 1998.
- [34412] 7964.Dent, A. L.; Yewdell, J.; Puvion-Dutilleul, F.; Koken, M. H.; deThe, H.; Staudt, L. M.: LYSP100 associated nuclear domains (LANDs): description of a new class of sub-nuclear structures and their relationship to PML nuclear bodies. *Blood* 88: 1423–1426, 1996.
- [34413] 7965.Seeler, J. S.; Marchio, A.; Sitterlin, D.; Transy, C.; Dejean, A.: Interaction of SP100 with HP1 proteins: a link between the promyelocytic leukemia-associated nuclear bodies and the chromatin compartment. *Proc. Nat. Acad. Sci.* 95: 7316–7321, 1998.
- [34414] 7966.Scanlan, M. J.; Chen, Y.-T.; Williamson, B.; Gure, A. O.; Stockert, E.; Gordan, J. D.; Tureci, O.; Sahin, U.; Pfrendschuh, M.; Old, L.J.: Characterization of human colon cancer antigens recognized by autologous antibodies. *Int. J. Cancer* 76: 652–658, 1998.
- [34415] 7967.Halford, S.; Dulai, K. S.; Daw, S. C.; Fitzgibbon, J.; Hunt, D.M.: Isolation and chromosomal localization of two human CDP-diacylglycerol synthase (CDS) genes. *Ge-*

nomics 54: 140–144, 1998.

- [34416] 7968.Heacock, A. M.; Uhler, M. D.; Agranoff, B. W.: Cloning of CDP–diacylglycerolsynthase from a human neuronal cell line. *J. Neurochem.* 67: 2200–2203,1996.
- [34417] 7969.Weeks, R.; Dowhan, W.; Shen, H.; Balantac, N.; Meengs, B.; Nudelman,E.; Leung, D. W.: Isolation and expression of an isoform of humanCDP–diacylglycerol synthase cDNA. *DNA Cell Biol.* 16: 281–289, 1997.
- [34418] 7970.Volta, M.; Bulfone, A.; Gattuso, C.; Rossi, E.; Mariani, M.; Consalez,G. G.; Zuffardi, O.; Ballabio, A.; Banfi, S.; Franco, B.: Identificationand characterization of CDS2, a mammalian homolog of the *Drosophila*CDP–diacylglycerol synthase gene. *Genomics* 55: 68–77, 1999.
- [34419] 7971.Borsani, G.; DeGrandi, A.; Ballabio, A.; Bulfone, A.; Bernard,L.; Banfi, S.; Gattuso, C.; Mariani, M.; Dixon, M.; Donnai, D.; Metcalfe,K.; Winter, R.; Robertson, M.; Axton, R.; Brown, A.; van Heyningen,V.; Hanson, I.: EYA4, a novel vertebrate gene related to *Drosophila*eyes absent. *Hum. Molec. Genet.* 8: 11–23, 1999.
- [34420] 7972.Lepperdinger, G.; Strobl, B.; Kreil, G.: HYAL2, a human gene expressedin many cells, encodes a lysosomal hyaluronidase with a novel typeof specificity. *J. Biol. Chem.* 273: 22466–22470, 1998.

- [34421] 7973.Kanamori, A.; Nakayama, J.; Fukuda, M. N.; Stallcup, W. B.; Sasaki,K.; Fukuda, M.; Hirabayashi, Y.: Expression cloning and characterization of a cDNA encoding a novel membrane protein required for the formation of O-acetylated ganglioside: a putative acetyl-CoA transporter. *Proc.Nat. Acad. Sci.* 94: 2897–2902, 1997.
- [34422] 7974.Holmes, M.; Turner, J.; Fox, A.; Chisholm, O.; Crossley, M.; Chong,B.: hFOG-2, a novel zinc finger protein, binds the co-repressor mCtBP2 and modulates GATA-mediated activation. *J. Biol. Chem.* 274: 23491–23498, 1999.
- [34423] 7975.Svensson, E. C.; Huggins, G. S.; Lin, H.; Clendenin, C.; Jiang,F.; Tufts, R.; Dardik, F. B.; Leiden, J. M.: A syndrome of tricuspid atresia in mice with a targeted mutation of the gene encoding Fog-2. *Nature Genet.* 25: 353–356, 2000.
- [34424] 7976.Svensson, E. C.; Tufts, R. L.; Polk, C. E.; Leiden, J. M.: Molecular cloning of FOG-2: a modulator of transcription factor GATA-4 in cardiomyocytes. *Proc.Nat. Acad. Sci.* 96: 956–961, 1999.
- [34425] 7977.Tevosian, S. G.; Deconinck, A. E.; Tanaka, M.; Schinke, M.; Litovsky,S. H.; Izumo, S.; Fujiwara, Y.; Orkin, S. H.: FOG-2, a cofactor for GATA transcription factors, is

essential for heart morphogenesis and development of coronary vessels from epicardium. Cell 101: 729–739, 2000.

[34426] 7978. Behrends, S.; Kazmierczak, B.; Steenpass, A.; Knauf, B.; Bullerdiek, J.; Scholz, H.; Eiberg, H.: Assignment of GUCY1B2, the gene coding for the beta-2 subunit of human guanylyl cyclase to chromosomal band 13q14.3 between markers D13S168 and D13S155. Genomics 55: 126–127, 1999.

[34427] 7979. Malterer, A.; Gupta, G.; Danziger, R. S.: Assignment of GUCY1B2, the human homologue of a candidate gene for hypertension, to chromosome bands 13q14.2–q14.3 by in situ hybridization. Cytogenet. Cell Genet. 85: 256–257, 1999.

[34428] 7980. Yuen, P. S. T.; Potter, L. R.; Garbers, D. L.: A new form of guanylyl cyclase is preferentially expressed in rat kidney. Biochemistry 29: 10872–10878, 1990.

[34429] 7981. Paavola, P.; Horelli-Kuitunen, N.; Palotie, A.; Peltonen, L.: Characterization of a novel gene, PNUTL2, on human chromosome 17q22–q23 and its exclusion as the Meckel syndrome gene. Genomics 55: 122–125, 1999.

[34430] 7982. Tedesco, F.; Roncelli, L.; Petersen, B. H.; Agnello, V.; Sodetz, J. M.: Two distinct abnormalities in patients with

C8-alpha-gammadeficiency: low level of C8-beta chain and presence of dysfunctional C8-alpha-gamma subunit. J. Clin. Invest. 86: 884-888, 1990.

- [34431] 7983.Savov, A.; Angelicheva, D.; Balassopoulou, A.; Jordanova, A.;Noussia-Arvanitakis, S.; Kalaydjieva, L.: Double mutant alleles:are they rare? Hum. Molec. Genet. 4: 1169-1171, 1995.
- [34432] 7984.Theriault, A.; Boyd, E.; Whaley, K.; Sodetz, J. M.; Connor, J.M.: Regional chromosomal assignment of genes encoding the alpha andbeta subunits of human complement protein c8 to 1p32. (Abstract) Cytogenet.Cell Genet. 58: 1864 only, 1991.
- [34433] 7985.Theriault, A.; Boyd, E.; Whaley, K.; Sodetz, J. M.; Connor, J.M.: Regional chromosomal assignment of genes encoding the alpha andbeta subunits of human complement protein C8 to 1p32. Hum. Genet. 88:703-704, 1992.
- [34434] 7986.Zhang, L.; Rittner, C.; Sodetz, J. M.; Schneider, P. M.; Kaufmann,T.: The eighth component of human complement: molecular basis ofC8A (C81) polymorphism. Hum. Genet. 96: 281-284, 1995.
- [34435] 7987.Bahary, N.; Zorich, G.; Pachter, J. E.; Leibel, R. L.; Friedman,J. M.: Molecular genetic linkage maps of mouse chromosomes 4 and6. Genomics 11: 33-47, 1991.

- [34436] 7988.Arnaout, M. A.; Gupta, S. K.; Pierce, M. W.; Tenen, D. G.: Aminoacid sequence of the alpha subunit of human leukocyte adhesion receptor Mo1 (complement receptor type 3). J. Cell Biol. 106: 2153–2158, 1988.
- [34437] 7989.Arnaout, M. A.; Remold–O'Donnell, E.; Pierce, M. W.; Harris, P.; Tenen, D. G.: Molecular cloning of the alpha–subunit of human and guinea pig leukocyte adhesion glycoprotein Mo1: chromosomal localization and homology to the alpha–subunits of integrins. Proc. Nat. Acad. Sci. 85: 2776–2780, 1988.
- [34438] 7990.Callen, D. F.; Chen, L. Z.; Nancarrow, J.; Whitmore, S. A.; Apostolou, S.; Thompson, A. D.; Lane, S. A.; Stallings, R. L.; Hildebrand, C. E.; Harris, P. G.; Sutherland, G. R.: Current state of the physical map of human chromosome 16. (Abstract) Cytogenet. Cell Genet. 58:1998 only, 1991.
- [34439] 7991.Corbi, A. L.; Kishimoto, T. K.; Miller, L. J.; Springer, T. A.: The human leukocyte adhesion glycoprotein Mac–1 (complement receptor type 3, CD11b) alpha subunit: cloning, primary structure, and relation to the integrins, von Willebrand factor and factor B. J. Biol. Chem. 263:12403–12411, 1988.
- [34440] 7992.Corbi, A. L.; Larson, R. S.; Kishimoto, T. K.; Springer, T. A.; Morton, C. C.: Chromosomal location of the genes

encoding the leukocyte adhesion receptors LFA-1, Mac-1 and p150,95: identification of a gene cluster involved in cell adhesion. *J. Exp. Med.* 167: 1597–1607, 1988.

- [34441] 7993. Pierce, M. W.; Remold-O'Donnell, E.; Todd, R. F., III; Arnaout, M. A.: N-terminal sequence of human leukocyte glycoprotein Mo1: conservation across species and homology to platelet IIb/IIIa. *Biochim. Biophys. Acta* 874: 368–371, 1986.
- [34442] 7994. Simon, D. I.; Chen, Z.; Seifert, P.; Edelman, E. R.; Balantyne, C. M.; Rogers, C.: Decreased neointimal formation in Mac-1 $-/-$ mice reveals a role for inflammation in vascular repair after angioplasty. *J. Clin. Invest.* 105: 293–300, 2000.
- [34443] 7995. Springer, T. A.; Teplow, D. B.; Dreyer, W. J.: Sequence homology of the LFA-1 and Mac-1 leukocyte adhesion glycoproteins and unexpected relation to leukocyte interferon. *Nature* 314: 540–542, 1985.
- [34444] 7996. Cartier, M.; Breitman, M. L.; Tsui, L.-C.: A frameshift mutation in the gamma-E-crystallin gene of the Elo mouse. *Nature Genet.* 2:42–45, 1992.
- [34445] 7997. Petersen, M. B.; Slaugenhaupt, S. A.; Lewis, J. G.; Warren, A. C.; Chakravarti, A.; Antonarakis, S. E.: A genetic linkage map of 27 markers on human chromosome 21.

Genomics 9: 407–419, 1991.

- [34446] 7998.Amagai, M.; Klaus-Kovtun, V.; Stanley, J. R.: Autoantibodies against a novel epithelial cadherin in pemphigus vulgaris, a disease of cell adhesion. *Cell* 67: 869–877, 1991.
- [34447] 7999.Koch, P. J.; Mahoney, M. G.; Ishikawa, H.; Pulkkinen, L.; Uitto, J.; Schultz, L.; Murphy, G. F.; Whitaker-Menezes, D.; Stanley, J.R.: Targeted disruption of the pemphigus vulgaris antigen (desmoglein3) gene in mice causes loss of keratinocyte cell adhesion with a phenotype similar to pemphigus vulgaris. *J. Cell Biol.* 137: 1091–1102, 1997.
- [34448] 8000.Wang, Y.; Amagai, M.; Minoshima, S.; Sakai, K.; Green, K. J.; Nishikawa, T.; Shimizu, N.: The human genes for desmogleins (DSG1 and DSG3) are located in a small region on chromosome 18q12. *Genomics* 20:492–495, 1994.
- [34449] 8001.Arneemann, J.; Spurr, N. K.; Magee, A. I.; Buxton, R. S.: The human gene (DSG2) coding for HDGC, a second member of the desmoglein subfamily of the desmosomal cadherins, is, like DSG1 coding for desmoglein DGI, assigned to chromosome 18. *Genomics* 13: 484–486, 1992.
- [34450] 8002.Koch, P. J.; Goldschmidt, M. D.; Walsh, M. J.; Zimbel-

mann, R.; Franke, W. W.: Complete amino acid sequence of the epidermal desmoglein precursor polypeptide and identification of a second type of desmoglein gene. *Europ. J. Cell Biol.* 55: 200–208, 1991.

[34451] 8003. Inskip, A.; Elexperu–Camiruaga, J.; Buxton, N.; Dias, P. S.; MacIntosh, J.; Campbell, D.; Jones, P. W.; Yengi, L.; Talbot, J. A.; Strange, R. C.; Fryer, A. A.: Identification of polymorphism at the glutathione S–transferase, GSTM3 locus: evidence for linkage with GSTM1* A. *Biochem. J.* 312: 713–716, 1995.

[34452] 8004. Van Cong, N.; Laisney, V.; Gross, M. S.; Frezal, J.: Glutathione–S–transferases—tissue distribution, number of loci, polymorphism, chromosome localization. (Abstract) *Cytogenet. Cell Genet.* 37: 554, 1984.

[34453] 8005. Benham, F. J.; Povey, S.: Members of the human glyceraldehyde–3–phosphatedehydrogenase–related gene family map to dispersed chromosomal locations. *Genomics* 5: 209–214, 1989.

[34454] 8006. Bruns, G. A. P.; Gerald, P. S.: Human glyceraldehyde–3–phosphatedehydrogenase in man–rodent somatic cell hybrids. *Science* 192: 54–56, 1976.

[34455] 8007. Edwards, Y. H.; Clark, P.; Harris, H.: Isozymes of glyceraldehyde–3–phosphatedehydrogenase in man and

other mammals. Ann. Hum. Genet. 40: 67–77,1976.

- [34456] 8008.Goodfellow, P. N.; Davies, K. E.; Ropers, H.–H.: Report of the committee on the genetic constitution of the X and Y chromosomes. Cytogenet.Cell Genet. 40: 296–352, 1985.
- [34457] 8009.Law, M. L.; Kao, F.–T.: Induced segregation of human syntenic genes by 5–bromodeoxyuridine plus near-visible light. Somat. Cell Genet. 4: 465–476, 1978.
- [34458] 8010.Lebherz, H. G.; Rutter, W. J.: Glyceraldehyde–3–phosphate dehydrogenase variants in phylogenically diverse organisms. Science 157: 1198–1199,1967.
- [34459] 8011.Piechaczyk, M.; Blanchard, J. M.; Riaad–el Sabouty, S.; Dani,C.; Marty, L.; Jeanteur, P.: Unusual abundance of glyceraldehyde3–phosphate dehydrogenase pseudogenes in vertebrate genomes. Nature 312:469–471, 1984.
- [34460] 8012.Rethore, M.–O.; Junien, C.; Malpuech, G.; Baccichetti, C.; Tenconi,R.; Kaplan, J.–C.; de Romeuf, J.; Lejeune, J.: Localisation du gene de la glyceraldehyde–3–phosphate dehydrogenase (G3PD) sur le segment distal du bras court de chromosome 12. Ann. Genet. 19: 140–142,1976.
- [34461] 8013.Rivas, F.; Vaca, G.; Zuniga, G.; Gonzalez, R. M.; Ruiz, C.; Rivera,H.; Moller, M.; Cantu, J. M.:
46,XX,–12,+der(12),rcp(3;12)(p25.1;p13.31)patkaryotype

in a girl: probably subregional assignment of glyceraldehyde-3-phosphatedehydrogenase locus to 12p13.1-p13.31 by exclusion. *Ann. Genet.* 28:189-192, 1985.

- [34462] 8014.Serville, F.; Junien, C.; Kaplan, J. C.; Gachet, M.; Cadoux, J.;Broustet, A.: Gene dosage effect for human triosephosphate isomeraseand glyceraldehyde-3-phosphate dehydrogenase in partial trisomy 12p13and trisomy 18p. *Hum. Genet.* 45: 63-69, 1978.
- [34463] 8015.Tso, J. Y.; Sun, X.-H.; Kao, T.; Reece, K. S.; Wu, R.: Isolationand characterization of rat and human glyceraldehyde-3-phosphate dehydrogenasecDNAs: genomic complexity and molecular evolution of the gene. *NucleicAcids Res.* 13: 2485-2502, 1985.
- [34464] 8016.Hashimoto, S.; Chiorazzi, N.; Gregersen, P. K.: The complete sequenceof the human CD79b (Ig-beta/B29) gene: identification of a conservedexon/intron organization, immunoglobulin-like regulatory regions,and allelic polymorphism. *Immunogenetics* 40: 145-149, 1994.
- [34465] 8017.Hashimoto, S.; Gregersen, P. K.; Chiorazzi, N.: The human Ig-betaDNA sequence, a homologue of murine B29, is identical in B cell andplasma cell lines producing all the human Ig isotypes. *J. Immun.* 150:491-498, 1993.

- [34466] 8018.Wood, W. J., Jr.; Thompson, A. A.; Korenberg, J.; Chen, X.; May,W.; Wall, R.; Denny, C. T.: Isolation and chromosomal mapping ofthe human immunoglobulin-associated B29 gene (IGB). *Genomics* 16:187–192, 1993.
- [34467] 8019.Okabe, I.; Nussbaum, R. L.: Identification and chromosomal mappingof the mouse inositol polyphosphate 1–phosphatase gene. *Genomics* 30:358–360, 1995.
- [34468] 8020.Woodcock, E. A.; Wang, B. H.; Arthur, J. F.; Lennard, A.; Matkovich,S. J.; Du, X.–J.; Brown, J. H.; Hannan, R. D.: Inositol polyphosphate1–phosphatase is a novel antihypertrophic factor. *J. Biol. Chem.* 277:22734–22742, 2002.
- [34469] 8021.York, J. D.; Veile, R. A.; Donis–Keller, H.; Majerus, P. W.: Cloning,heterologous expression, and chromosomal localization of human inositolpolyphosphate 1–phosphatase. *Proc. Nat. Acad. Sci.* 90: 5833–5837,1993.
- [34470] 8022.Janne, P. A.; Dutra, A. S.; Dracopoli, N. C.; Charnas, L. R.; Puck,J. M.; Nussbaum, R. L.: Localization of the 75–kDa inositol polyphosphate–5–phosphatase(INPP5B) to human chromosome band 1p34. *Cytogenet. Cell Genet.* 66:164–166, 1994.
- [34471] 8023.Janne, P. A.; Rochelle, J. M.; Martin–DeLeon, P. A.; Stambolian,D.; Seldin, M. F.; Nussbaum, R. L.: Mapping of

the 75-kDa inositolpolyphosphate-5-phosphatase (Inpp5b) to distal mouse chromosome 4 and its exclusion as a candidate gene for dysgenetic lens. *Genomics* 28:280–285, 1995.

[34472] 8024. Ross, T. S.; Jefferson, A. B.; Mitchell, C. A.; Majerus, P. W.: Cloning and expression of human 75-kDa inositol polyphosphate-5-phosphatase. *J. Biol. Chem.* 266: 20283–20289, 1991.

[34473] 8025. Matsumoto, M.; Nakagawa, T.; Inoue, T.; Nagata, E.; Tanaka, K.; Takano, H.; Minowa, O.; Kuno, J.; Sakakibara, S.; Yamada, M.; Yoneshima, H.; Miyawaki, A.; Fukuichi, T.; Furuichi, T.; Okano, H.; Mikoshiba, K.; Noda, T.: Ataxia and epileptic seizures in mice lacking type 1 inositol 1,4,5-triphosphate receptor. *Nature* 379: 168–171, 1996.

[34474] 8026. Nucifora, F. C., Jr.; Li, S.-H.; Danoff, S.; Ullrich, A.; Ross, C. A.: Molecular cloning of a cDNA for the human inositol 1,4,5-trisphosphate receptor type 1, and the identification of a third alternatively spliced variant. *Molec. Brain Res.* 32: 291–296, 1995.

[34475] 8027. Bradham, D. M.; Igarashi, A.; Potter, R. L.; Groten-dorst, G. R.: Connective tissue growth factor: a cysteine-rich mitogen secreted by human vascular endothelial cells is related to the SRC-induced immediate early gene prod-

uct CEF-10. J. Cell Biol. 114: 1285-1294,1991.

[34476] 8028.Aguilar-Salinas, C. A.; Reyes-Rodriguez, E.; Ordonez-Sanchez, M.L.; Torres, M. A.; Ramirez-Jimenez, S.; Dominguez-Lopez, A.; Martinez-Francois, J. R.; Velasco-Perez, M. L.; Alpizar, M.; Garcia-Garcia, E.; Gomez-Perez, F.; Rull, J.; Tusie-Luna, M. T.: Early-onset type 2 diabetes: metabolic and genetic characterization in the Mexican population. J. Clin. Endocr. Metab. 86: 220-226, 2001.

[34477] 8029.DerKinderen, D. J.; Kotev, J. W.; Tan, K. E. W. P.; Beemer, F.A.; Van Romunde, L. K. J.; Den Otter, W.: Parental age in sporadic hereditary retinoblastoma. Am. J. Ophthalmol. 110: 605-609, 1990.

[34478] 8030.Orphanides, G.; Wu, W.-H.; Lane, W. S.; Hampsey, M.; Reinberg, D.: The chromatin-specific transcription elongation factor FACT comprises human SPT16 and SSRP1 proteins. Nature 400: 284-288, 1999.

[34479] 8031.Li, H.; Chen, J.; Huang, A.; Stinson, J.; Heldens, S.; Foster, J.; Dowd, P.; Gurney, A. L.; Wood, W. I.: Cloning and characterization of IL-17B and IL-17C, two new members of the IL-17 cytokine family. Proc. Nat. Acad. Sci. 97: 773-778, 2000.

[34480] 8032.Shi, Y.; Ullrich, S. J.; Zhang, J.; Connolly, K.; Grze-

gorzewski, K. J.; Barber, M. C.; Wang, W.; Wathen, K.; Hodge, V.; Fisher, C. L.; Olsen, H.; Ruben, S. M.; Knyazev, I.; Cho, Y. H.; Kao, V.; Wilkinson, K. A.; Carrell, J. A.; Ebner, R.: A novel cytokine receptor–ligand pair: identification, molecular characterization, and in vivo immunomodulatory activity. *J. Biol. Chem.* 275: 19167–19176, 2000.

[34481] 8033. Friedman, L. S.; Ostermeyer, E. A.; Lynch, E. D.; Welcsh, P.; Szabo, C. I.; Meza, J. E.; Anderson, L. A.; Dowd, P.; Lee, M. K.; Rowell, S. E.; Ellison, J.; Boyd, J.; King, M.–C.: 22 genes from chromosome 17q21: cloning, sequencing, and characterization of mutations in breast cancer families and tumors. *Genomics* 25: 256–163, 1995.

[34482] 8034. Linial, M.; Miller, K.; Scheller, R. H.: VAT–1: an abundant membrane protein from *Torpedo* cholinergic synaptic vesicles. *Neuron* 2: 1265–1273, 1989.

[34483] 8035. Smith, T. M.; Lee, M. K.; Szabo, C. I.; Jerome, N.; McEuen, M.; Taylor, M.; Hood, L.; King, M.–C.: Complete genomic sequence and analysis of 117 kb of human DNA containing the gene BRCA1. *Genome Res.* 6: 1029–1049, 1996.

[34484] 8036. Johnstone, R. W.; Tommerup, N.; Hansen, C.; Vissing, H.; Shi, Y.: Structural organization, tissue expression, and chromosomal localization of Ciao 1, a functional mod–

ulator of the Wilms' tumor suppressor, WT1. Immunogenetics 49: 900–905, 1999.

[34485] 8037. Johnstone, R. W.; Wang, J.; Tommerup, N.; Vissing, H.; Roberts, T.; Shi, Y.: Ciao 1 is a novel WD40 protein that interacts with the tumor suppressor protein WT1. J. Biol. Chem. 273: 10880–10887, 1998.

[34486] 8038. Huebner, K.; Cannizzaro, L. A.; Nakamura, T.; Hillova, J.; Mariage-Samson, R.; Hecht, F.; Hill, M.; Croce, C. M.: A rearranged transforming gene, *trc*, is made up of human sequences derived from chromosome regions 5q, 17q and 18q. Oncogene 3: 449–455, 1988.

[34487] 8039. Nakamura, T.; Hillova, J.; Mariage-Samson, R.; Hill, M.: Molecular cloning of a novel oncogene generated by DNA recombination during transfection. Oncogene Res. 2: 357–370, 1988.

[34488] 8040. Nakamura, T.; Hillova, J.; Mariage-Samson, R.; Onno, M.; Huebner, K.; Cannizzaro, L. A.; Boghosian-Sell, L.; Croce, C. M.; Hill, M.: A novel transcriptional unit of the *trc* oncogene widely expressed in human cancer cells. Oncogene 7: 733–741, 1992.

[34489] 8041. Chen, H.; Rossier, C.; Morris, M. A.; Scott, H. S.; Gos, A.; Bairoch, A.; Antonarakis, S. E.: A testis-specific gene, TPTE, encodes a putative transmembrane tyrosine phos-

phatase and maps to the pericentromeric region of human chromosomes 21 and 13, and to chromosomes 15, 22, and Y. Hum. Genet. 105: 399–409, 1999.

[34490] 8042. Guipponi, M.; Tapparel, C.; Jousson, O.; Scamuffa, N.; Mas, C.; Rossier, C.; Hutter, P.; Meda, P.; Lyle, R.; Raymond, A.; Antonarakis, S. E.: The murine orthologue of the Golgi-localized TPTE protein provides clues to the evolutionary history of the human TPTE gene family. Hum. Genet. 109: 569–575, 2001.

[34491] 8043. Guipponi, M.; Yaspo, M.-L.; Riesselman, L.; Chen, H.; De Sario, A.; Roizes, G.; Antonarakis, S. E.: Genomic structure of a copy of the human TPTE gene which encompasses 87 kb on the short arm of chromosome 21. Hum. Genet. 107: 127–131, 2000.

[34492] 8044. Gruber, A. D.; Pauli, B. U.: Molecular cloning and biochemical characterization of a truncated, secreted member of the human family of Ca^{2+} -activated Cl^- channels. Biochim. Biophys. Acta 1444: 418–423, 1999.

[34493] 8045. Bause, E.; Bieberich, E.; Rolfs, A.; Volker, C.; Schmidt, B.: Molecular cloning and primary structure of Man(9)-mannosidase from human kidney. Eur. J. Biochem. 217: 535–540, 1993.

[34494] 8046. Tremblay, L. O.; Campbell Dyke, N.; Herscovics, A.:

Molecular cloning, chromosomal mapping and tissue-specific expression of a novel human α -1,2-mannosidase gene involved in N-glycan maturation. *Glycobiology* 8:585–595, 1998.

- [34495] 8047. Ueno, M.; Kimura, N.; Nakashima, K.; Saito-Ohara, F.; Inazawa, J.; Taga, T.: Genomic organization, sequence and chromosomal localization of the mouse *Tbr2* gene and a comparative study with *Tbr1*. *Gene* 254:29–35, 2000.
- [34496] 8048. Baker, E.; Crawford, J.; Sutherland, G. R.; Freeman, C.; Parish, C. R.; Hulett, M. D.: Human HPA endoglycosidase heparanase. *Chromosome Res.* 7: 319 only, 1999.
- [34497] 8049. Hulett, M. D.; Freeman, C.; Hamdorf, B. J.; Baker, R. T.; Harris, M. J.; Parish, C. R.: Cloning of mammalian heparanase, an important enzyme in tumor invasion and metastasis. *Nature Med.* 5: 803–809, 1999.
- [34498] 8050. Kussie, P. H.; Hulmes, J. D.; Ludwig, D. L.; Patel, S.; Navarro, E. C.; Seddon, A. P.; Giorgio, N. A.; Bohlen, P.: Cloning and functional expression of a human heparanase gene. *Biochem. Biophys. Res. Commun.* 261:183–187, 1999.
- [34499] 8051. Toyoshima, M.; Nakajima, M.: Human heparanase: purification, characterization, cloning, and expression. *J. Biol. Chem.* 274: 24153–24160, 1999.

- [34500] 8052.Vlodavsky, I.; Friedmann, Y.; Elkin, M.; Aingorn, H.; Atzmon, R.;Ishai-Michaeli, R.; Bitan, M.; Pappo, O.; Peretz, T.; Michal, I.;Spector, L.; Pecker, I.: Mammalian heparanase: gene cloning, expressionand function in tumor progression and metastasis. *Nature Med.* 5:793–802, 1999.
- [34501] 8053.Ikeda, A.; Ikeda, S.; Gridley, T.; Nishina, P. M.; Nagert, J.K.: Neural tube defects and neuroepithelial cell death in Tulp3 knockoutmice. *Hum. Molec. Genet.* 10: 1325–1334, 2001.
- [34502] 8054.Nishina, P. M.; North, M. A.; Ikeda, A.; Yan, Y.; Nagert, J. K.: Molecular characterization of a novel tubby gene family member,TULP3, in mouse and humans. *Genomics* 54: 215–220, 1998.
- [34503] 8055.Wu, X.; Kekuda, R.; Huang, W.; Fei, Y.–J.; Leibach, F. H.; Chen,J.; Conway, S. J.; Ganapathy, V.: Identity of the organic cationtransporter OCT3 as the extraneuronal monoamine transporter (uptake–2)and evidence for the expression of the transporter in the brain. *J.Biol. Chem.* 273: 32776–32786, 1998.
- [34504] 8056.Seki, N.; Ohira, M.; Nagase, T.; Ishikawa, K.; Miyajima, N.; Nakajima,D.; Nomura, N.; Ohara, O.: Characterization of cDNA clones in size–fractionatedcDNA libraries

from human brain. DNA Res. 4: 345–349, 1997.

[34505] 8057. Biassoni, R.; Cantoni, C.; Falco, M.; Verdiani, S.; Bottino, C.; Vitale, M.; Conte, R.; Poggi, A.; Moretta, A.; Moretta, L.: The human leukocyte antigen (HLA)–C–specific 'activatory' or 'inhibitory' natural killer cell receptors display highly homologous extracellular domains but differ in their transmembrane and intracytoplasmic portions. J. Exp. Med. 183: 645–650, 1996.

[34506] 8058. Bottino, C.; Sivori, S.; Vitale, M.; Cantoni, C.; Falco, M.; Pende, D.; Morelli, L.; Augugliaro, R.; Semenzato, G.; Biassoni, R.; Moretta, L.; Moretta, A.: A novel surface molecule homologous to the p58/p50 family of receptors is selectively expressed on a subset of human natural killer cells and induces both triggering of cell functions and proliferation. Europ. J. Immun. 26: 1816–1824, 1996.

[34507] 8059. Sumoy, L.; Pluvinet, R.; Andreu, N.; Estivill, X.; Escarceller, M.: PACSIN 3 is a novel SH3 domain cytoplasmic adapter protein of the pacsin–syndapin–FAP52 gene family. Gene 262: 199–205, 2001.

[34508] 8060. Hubener, C.; Mincheva, A.; Lichter, P.; Schraven, B.; Bruyns, E.: Genomic organization and chromosomal localization of the human gene encoding the T-cell receptor–interacting molecule (TRIM). Immunogenetics

51:154–158, 2000.

- [34509] 8061. Hubener, C.; Mincheva, A.; Lichter, P.; Schraven, B.; Bruyns, E.: Complete sequence, genomic organization, and chromosomal localization of the human gene encoding the SHP2-interacting transmembrane adaptor protein (SIT). *Immunogenetics* 53: 337–341, 2001.
- [34510] 8062. Bruyns, E.; Marie-Cardine, A.; Kirchgessner, H.; Sagolla, K.; Shevchenko, A.; Mann, M.; Autschbach, F.; Bensussan, A.; Meuer, S.; Schraven, B.: T cell receptor (TCR) interacting molecule (TRIM), a novel disulfide-linked dimer associated with the TCR-CD3-zeta complex, recruits intracellular signaling proteins to the plasma membrane. *J. Exp. Med.* 188: 561–575, 1998.
- [34511] 8063. Zhao, R.; Qi, Y.; Chen, J.; Zhao, Z. J.: FYVE-DSP2, a FYVE domain-containing dual specificity protein phosphatase that dephosphorylates phosphatidylinositol(3)-phosphate. *Exp. Cell Res.* 265: 329–338, 2001.
- [34512] 8064. Cui, X.; De Vivo, I.; Slany, R.; Miyamoto, A.; Firestein, R.; Cleary, M. L.: Association of SET domain and myotubularin-related proteins modulates growth control. *Nature Genet.* 18: 331–337, 1998.
- [34513] 8065. Kahyo, T.; Nishida, T.; Yasuda, H.: Involvement of PIAS1 in the sumoylation of tumor suppressor p53. *Molec.*

Cell 8: 713–718, 2001.

- [34514] 8066.Liu, B.; Liao, J.; Rao, X.; Kushner, S. A.; Chung, C. D.; Chang,D. D.; Shuai, K.: Inhibition of Stat1–mediated gene activation byPIAS1. *Proc. Nat. Acad. Sci.* 95: 10626–10631, 1998.
- [34515] 8067.Valdez, B. C.; Henning, D.; Perlaky, L.; Busch, R. K.; Busch, H.: Cloning and characterization of Gu/RH-II binding protein. *Biochem.Biophys. Res. Commun.* 234: 335–340, 1997.
- [34516] 8068.Mansour, S. J.; Herbrick, J.–A.; Scherer, S. W.; Melancon, P.:Human GBF1 is a ubiquitously expressed gene of the Sec7 domain familymapping to 10q24. *Genomics* 54: 323–327, 1998.
- [34517] 8069.Heisenberg, C.–P.; Tada, M.; Rauch, G.–J.; Saude, L.; Concha, M.L.; Geisler, R.; Stemple, D. L.; Smith, J. C.; Wilson, S. W.: Silberblick/Wnt11mediates convergent extension movements during zebrafish gastrulation. *Nature* 405:76–81, 2000.
- [34518] 8070.Lako, M.; Strachan, T.; Bullen, P.; Wilson, D. I.; Robson, S. C.;Lindsay, S.: Isolation, characterisation and embryonic expressionof WNT11, a gene which maps to 11q13.5 and has possible roles in thedevelopment of skeleton, kidney and lung. *Gene* 219: 101–110, 1998.

- [34519] 8071.Pandur, P.; Lasche, M.; Eisenberg, L. M.; Kuhl, M.: Wnt-11 activation of a non-canonical Wnt signalling pathway is required for cardiogenesis. *Nature* 418:636-641, 2002.
- [34520] 8072.Frye, R. A.: Characterization of five human cDNAs with homology to the yeast SIR2 gene: Sir2-like proteins (sirtuins) metabolize NAD and may have protein ADP-ribosyltransferase activity. *Biochem. Biophys. Res. Commun.* 260: 273-279, 1999.
- [34521] 8073.Imai, S.; Armstrong, C. M.; Kaeberlein, M.; Guarente, L.: Transcriptional silencing and longevity protein Sir2 is an NAD-dependent histone deacetylase. *Nature* 403:795-800, 2000.
- [34522] 8074.Kimura, A.; Umehara, T.; Horikoshi, M.: Chromosomal gradient of histone acetylation established by Sas2p and Sir2p functions as a shield against gene silencing. *Nature Genet.* 15 Oct.: , 2002. Note: Advance Electronic Publication.
- [34523] 8075.Aula, N.; Salomaki, P.; Timonen, R.; Verheijen, F.; Mancini, G.; Mansson, J.-E.; Aula, P.; Peltonen, L.: The spectrum of SLC17A5-gene mutations resulting in free sialic acid-storage diseases indicates some genotype-phenotype correlation. *Am. J. Hum. Genet.* 67:

832–840,2000.

- [34524] 8076.Biancheri, R.; Verbeek, E.; Rossi, A.; Gaggero, R.; Roccatagliata,L.; Gatti, R.; van Diggelen, O.; Verheijen, F. W.; Mancini, G. M.S.: An Italian severe Salla disease variant associated with a SLC17A5mutation earlier described in infantile sialic acid storage disease. Clin.Genet. 61: 443–447, 2002.
- [34525] 8077.Belinsky, M. G.; Bain, L. J.; Balsara, B. B.; Testa, J. R.; Kruh,G. D.: Characterization of MOAT–C and MOAT–D, new members of theMRP/cMOAT subfamily of transporter proteins. J. Nat. Cancer Inst. 90:1735–1741, 1998.
- [34526] 8078.Fromm, M. F.; Leake, B.; Roden, D. M.; Wilkinson, G. R.; Kim, R.B.: Human MRP3 transporter: identification of the 5–prime flankingregion, genomic organization and alternative splice variants. Biochim.Biophys. Acta 1415: 369–374, 1999.
- [34527] 8079.Kiuchi, Y.; Suzuki, H.; Hirohashi, T.; Tyson, C. A.; Sugiyama,Y.: cDNA cloning and inducible expression of human multidrug resistanceassociated protein 3 (MRP3). FEBS Lett. 433: 149–152, 1998.
- [34528] 8080.Konig, J.; Rost, D.; Cui, Y.; Keppler, D.: Characterization ofthe human multidrug resistance protein isoform MRP3 localized to thebasolateral hepatocyte membrane.

Hepatology 29: 1156–1163, 1999.

- [34529] 8081.Kool, M.; van der Linden, M.; de Haas, M.; Scheffer, G. L.; deVree, J. M. L.; Smith, A. J.; Jansen, G.; Peters, G. J.; Ponne, N.;Scheper, R. J.; Oude Elferink, R. P. J.; Baas, F.; Borst, P.: MRP3,an organic anion transporter able to transport anti–cancer drugs. Proc.Nat. Acad. Sci. 96: 6914–6919, 1999.
- [34530] 8082.Ortiz, D. F.; Li, S.; Iyer, R.; Zhang, X.; Novikoff, P.; Arias,I. M.: MRP3, a new ATP–binding cassette protein localized to thecanalicular domain of the hepatocyte. Am. J. Physiol. 276: G1493–G1500,1999.
- [34531] 8083.Hjalt, T. A.; Murray, J. C.: The human BARX2 gene: genomic structure,chromosomal localization, and single nucleotide polymorphisms. Genomics 62:456–459, 1999.
- [34532] 8084.Jones, F. S.; Kioussi, C.; Copertino, D. W.; Kallunki, P.; Holst,B. D.; Edelman, G. M.: Barx2, a new homeobox gene of the Bar class,is expressed in neural and craniofacial structures during development. Proc.Nat. Acad. Sci. 94: 2632–2637, 1997.
- [34533] 8085.Arking, D. E.; Krebsova, A.; Macek, M., Sr.; Macek, M., Jr.; Arking,A.; Mian, I. S.; Fried, L.; Hamosh, A.; Dey, S.; McIntosh, I.; Dietz,H. C.: Association of human aging with a functional variant of klotho. Proc.Nat. Acad. Sci. 99:

856–861, 2002.

- [34534] 8086.Koh, N.; Fujimori, T.; Nishiguchi, S.; Tamori, A.; Sh-iomi, S.;Nakatani, T.; Sugimura, K.; Kishimoto, T.; Ki-noshita, S.; Kuroki,T.; Nabeshima, Y.: Severely reduced production of Klotho in humanchronic renal failure kidney. Biochem. Biophys. Res. Commun. 280:1015–1020, 2001.
- [34535] 8087.Kuro-o, M.; Matsumura, Y.; Aizawa, H.; Kawaguchi, H.; Suga, T.;Utsugi, T.; Ohyama, Y.; Kurabayashi, M.; Kaname, T.; Kume, E.; Iwasaki,H.; Iida, A.; Shiraki-Iida, T.; Nishikawa, S.; Nagai, R.; Nabeshima,Y.: Mutation of the mouse klotho gene leads to a syndrome resemblingage-ing. Nature 390: 45–51, 1997.
- [34536] 8088.Matsumura, Y.; Aizawa, H.; Shiraki-Iida, T.; Nagai, R.; Kuro-o,M.; Nabeshima, Y.: Identification of the human klotho gene and itstwo transcripts encoding membrane and secreted klotho protein. Biochem.Biophys. Res. Com-mun. 242: 626–630, 1998.
- [34537] 8089.Mori, K.; Yahata, K.; Mukoyama, M.; Suganami, T.; Makino, H.; Nagae,T.; Masuzaki, H.; Ogawa, Y.; Sugawara, A.; Nabeshima, Y.; Nakao, K.: Disruption of klotho gene causes an abnormal energy homeostasisin mice. Biochem. Biophys. Res. Commun. 278: 665–670, 2000.
- [34538] 8090.Saito, Y.; Nakamura, T.; Ohyama, Y; Suzuki, T.; Iida,

A.; Shiraki-Iida, T.; Kuro-o, M.; Nabeshima, Y.; Kurabayashi, M.; Nagai, R.: In vivo clothe gene delivery protects against endothelial dysfunction in multiple risk factor syndrome. *Biochem. Biophys. Res. Commun.* 276: 767–772, 2000.

[34539] 8091. Little, N. A.; Hastie, N. D.; Davies, R. C.: Identification of WTAP, a novel Wilms' tumour 1-associated protein. *Hum. Molec. Genet.* 9:2231–2239, 2000.

[34540] 8092. Vazquez, F.; Hastings, G.; Ortega, M.-A.; Lane, T. F.; Oikemus, S.; Lombardo, M.; Iruela-Arispe, M. L.: METH-1, a human ortholog of ADAMTS-1, and METH-2 are members of a new family of proteins with angiogenic activity. *J. Biol. Chem.* 274: 23349–23357, 1999.

[34541] 8093. Georgiadis, K. E.; Hirohata, S.; Seldin, M. F.; Apte, S. S.: ADAM-TS8, a novel metalloprotease of the ADAM-TS family located on mouse chromosome 9 and human chromosome 11. *Genomics* 62: 312–315, 1999.

[34542] 8094. Sugawara, M.; Nakanishi, T.; Fei, Y.-J.; Huang, W.; Ganapathy, M. E.; Leibach, F. H.; Ganapathy, V.: Cloning of an amino acid transporter with functional characteristics and tissue expression pattern identical to that of system A. *J. Biol. Chem.* 275: 16473–16477, 2000.

[34543] 8095. Fedi, P.; Bafico, A.; Soria, A. N.; Burgess, W. H.; Miki,

T.; Bottaro, D. P.; Kraus, M. H.; Aaronson, S. A.: Isolation and biochemical characterization of the human Dkk-1 homologue, a novel inhibitor of mammalian Wnt signaling. *J. Biol. Chem.* 274: 19465–19472, 1999.

- [34544] 8096. Krupnik, V. E.; Sharp, J. D.; Jiang, C.; Robison, K.; Chickering, T. W.; Amaravadi, L.; Brown, D. E.; Guyot, D.; Mays, G.; Leiby, K.; Chang, B.; Duong, T.; Goodearl, A. D. J.; Gearing, D. P.; Sokol, S. Y.; McCarthy, S. A.: Functional and structural diversity of the human Dickkopf gene family. *Gene* 238: 301–313, 1999.
- [34545] 8097. Roessler, E.; Du, Y.; Glinka, A.; Dutra, A.; Niehrs, C.; Muenke, M.: The genomic structure, chromosome location, and analysis of the human DKK1 head inducer gene as a candidate for holoprosencephaly. *Cytogenet. Cell Genet.* 89: 220–224, 2000.
- [34546] 8098. Gatignol, A.; Buckler-White, A.; Berkhout, B.; Jeang, K. T.: Characterization of a human TAR RNA-binding protein that activates the HIV-1 LTR. *Science* 251: 1597–1600, 1991.
- [34547] 8099. Gatignol, A.; Duarte, M.; Daviet, L.; Chang, Y.-N.; Jeang, K.-T.: Sequential steps in Tat trans-activation of HIV-1 mediated through cellular DNA, RNA, and protein binding factors. *Gene Expr.* 5: 217–228, 1996.

- [34548] 8100.Kozak, C. A.; Gatignol, A.; Graham, K.; Jeang, K. T.; McBride, O. W.: Genetic mapping in human and mouse of the locus encoding TRBP, a protein that binds the TAR region of the human immunodeficiencyvirus (HIV-1). *Genomics* 25: 66-72, 1995.
- [34549] 8101.Bauer, M. F.; Gempel, K.; Reichert, A. S.; Rappold, G. A.; Lichtner, P.; Gerbitz, K. D.; Neupert, W.; Brunner, M.; Hofmann, S.: Genetic and structural characterization of the human mitochondrial inner membrane translocase. *J. Molec. Biol.* 289: 69-82, 1999.
- [34550] 8102.Bomer, U.; Rassow, J.; Zufall, N.; Pfanner, N.; Meijer, M.; Maarse, A. C.: The preprotein translocase of the inner mitochondrial membrane: evolutionary conservation of targeting and assembly of Tim17. *J. Molec. Biol.* 262: 389-395, 1996.
- [34551] 8103.Phornphutkul, C.; Anikster, Y.; Huizing, M.; Braun, P.; Brodie, C.; Chou, J. Y.; Gahl, W. A.: The promoter of a lysosomal membrane transporter gene, CTNS, binds Sp-1, shares sequences with the promoter of an adjacent gene, CARKL, and causes cystinosis if mutated in a critical region. *Am. J. Hum. Genet.* 69: 712-721, 2001.
- [34552] 8104.Lebre, A.-S.; Jamot, L.; Takahashi, J.; Spasskey, N.; Leprince, C.; Ravise, N.; Zander, Fujigasaki, H.; Kussel-

Andermann, P.; Duyckaerts, C.; Camonis, J. H.; Brice, A.: Ataxin-7 interacts with a Cbl-associated protein that it recruits into neuronal intranuclear inclusions. *Hum. Molec. Genet.* 10: 10:1201-1213, 2001.

[34553] 8105. Lin, W.-H.; Chiu, K. C.; Chang, H.-M.; Lee, K.-C.; Tai, T.-Y.; Chuang, L.-M.: Molecular scanning of the human sorbin and SH3-domain-containing-1 (SORBS1) gene: positive association of the T228A polymorphism with obesity and type 2 diabetes. *Hum. Molec. Genet.* 10: 1753-1760, 2001.

[34554] 8106. Scott, A. F.: Personal Communication. Baltimore, Md. 9/13/2000.

[34555] 8107. Nishi, M.; Mizushima, A.; Nakagawara, K.; Takeshima, H.: Characterization of human junctophilin subtype genes. *Biochem. Biophys. Res. Commun.* 273:920-927, 2000.

[34556] 8108. Takeshima, H.; Komazaki, S.; Nishi, M.; Iino, M.; Kangawa, K.: Junctophilins: a novel family of junctional membrane complex proteins. *Molec. Cell* 6: 11-22, 2000.

[34557] 8109. Holmes, S. E.; O'Hearn, E.; Rosenblatt, A.; Callahan, C.; Hwang, H. S.; Ingersoll-Ashworth, R. G.; Fleisher, A.; Stevanin, G.; Brice, A.; Potter, N. T.; Ross, C. A.; Margolis, R. L.: A repeat expansion in the gene encoding junc-

tophilin-3 is associated with Huntingtondisease-like 2. Nature Genet. 29: 377-378, 2001. Note: Erratum:Nature Genet. 30: 123 only, 2002.

[34558] 8110.Chavez, R. A.; Gray, A. T.; Zhao, B. B.; Kindler, C. H.; Mazurek,M. J.; Mehta, Y.; Forsayeth, J. R.; Yost, C. S.: TWIK-2, a new weakinward rectifying member of the tandem pore domain potassium channelfamily. J. Biol. Chem. 274: 7887-7892, 1999.

[34559] 8111.Gray, A. T.; Kindler, C. H.; Sampson, E. R.; Yost, C. S.: Assignmentof KCNK6 encoding the human weak inward rectifier potassium channelTWIK-2 to chromosome band 19q13.1 by radiation hybrid mapping. Cyto-genet.Cell Genet. 84: 190-191, 1999.

[34560] 8112.Pountney, D. J.; Gulkarov, I.; Vega-Saenz de Miera, E.; Holmes,D.; Saganich, M.; Rudy, B.; Artman, M.; Coetzee, W. A.: Identificationand cloning of TWIK-originated similarity sequence (TOSS): a novelhuman 2-pore K(+) channel principal subunit. FEBS Lett. 450: 191-196,1999.

[34561] 8113.Salinas, M.; Reyes, R.; Lesage, F.; Fosset, M.; Heurteaux, C.;Romey, G.; Lazdunski, M.: Cloning of a new mouse two-P domain channelsubunit and a human homologue with a unique pore structure. J. Biol.Chem. 274: 11751-11760, 1999.

- [34562] 8114. Barbetti, F.; Rocchi, M.; Bossolasco, M.; Cordera, R.; Sbraccia, P.; Finelli, P.; Consalez, G. G.: The human skeletal muscle glycogenin gene: cDNA, tissue expression, and chromosomal localization. *Biochem. Biophys. Res. Commun.* 220: 72–77, 1996.
- [34563] 8115. Machesky, L. M.; Reeves, E.; Wientjes, F.; Mattheyse, F. J.; Grogan, A.; Totty, N. F.; Burlingame, A. L.; Hsuan, J. J.; Segal, A. W.: Mammalian actin-related protein 2/3 complex localizes to regions of lamellipodial protrusion and is composed of evolutionarily conserved proteins. *Biochem. J.* 328: 105–112, 1997.
- [34564] 8116. Luo, J.; Nikolaev, A. Y.; Imai, S.; Chen, D.; Su, F.; Shiloh, A.; Guarente, L.; Gu, W.: Negative control of p53 by Sir2- α promotes cell survival under stress. *Cell* 107: 137–148, 2001.
- [34565] 8117. Bulfone, A.; Smiga, S. M.; Shimamura, K.; Peterson, A.; Puellas, L.; Rubenstein, J. L. R.: T-brain-1: a homolog of Brachyury whose expression defines molecularly distinct domains within the cerebral cortex. *Neuron* 15: 63–78, 1995.
- [34566] 8118. Makoff, A.; Lelchuk, R.; Oxer, M.; Harrington, K.; Emson, P.: Molecular characterization and localization of human metabotropic glutamate receptor type 4. *Molec.*

Brain Res. 37: 239–248, 1996.

- [34567] 8119. Pekhletski, R.; Gerlai, R.; Overstreet, L. S.; Huang, X. P.; Agopyan, N.; Slater, N. T.; Abramow-Newerly, W.; Roder, J. C.; Hampson, D. R.: Impaired cerebellar synaptic plasticity and motor performance in mice lacking the mGluR4 subtype of metabotropic glutamate receptor. J. Neurosci. 16: 6364–6373, 1996.
- [34568] 8120. Barbon, A.; Ferraboli, S.; Barlati, S.: Assignment of the human metabotropic glutamate receptor gene GRM7 to chromosome 3p26.1–p25.2 by radiation hybrid mapping. Cytogenet. Cell Genet. 88: 288 only, 2000.
- [34569] 8121. Shore, D.; Squire, M.; Nasmyth, K. A.: Characterization of two genes required for the position-effect control of yeast mating-type genes. EMBO J. 3: 2817–2823, 1984.
- [34570] 8122. Suka, N.; Luo, K.; Grunstein, M.: Sir2p and Sas2p opposingly regulate acetylation of yeast histone H4 lysine 16 and spreading of heterochromatin. Nature Genet. 15 Oct.: , 2002. Note: Advance Electronic Publication.
- [34571] 8123. Tanny, J. C.; Dowd, G. J.; Huang, J.; Hilz, H.; Moazed, D.: A enzymatic activity in the yeast Sir2 protein that is essential for gene silencing. Cell 99: 735–745, 1999.
- [34572] 8124. Vaziri, H.; Dessain, S. K.; Eaton, E. N.; Imai, S.-I.; Frye, R. A.; Pandita, T. K.; Guarente, L.; Weinberg, R. A.:

hSIR2–SIRT1 functions as an NAD–dependent p53 deacetylase. *Cell* 107: 149–159, 2001.

[34573] 8125. Verhaagh, S.; Schweifer, N.; Barlow, D. P.; Zwart, R.: Cloning of the mouse and human solute carrier 22a3 (Slc22a3/SLC22A3) identifies a conserved cluster of three organic cation transporters on mouse chromosome 17 and human 6q26–q27. *Genomics* 55: 209–218, 1999.

[34574] 8126. Raich, N.; Mattei, M. G.; Romeo, P. H.; Beaupain, D.: PHTF, a novel atypical homeobox gene on chromosome 1p13, is evolutionarily conserved. *Genomics* 59: 108–109, 1999.

[34575] 8127. Liu, J.; Shworak, N. W.; Sinay, P.; Schwartz, J. J.; Zhang, L.; Fritze, L. M.; Rosenberg, R. D.: Expression of heparan sulfate D–glucosaminyl 3–O–sulfotransferase isoforms reveals novel substrate specificities. *J. Biol. Chem.* 274: 5185–5192, 1999.

[34576] 8128. Shworak, N. W.; Liu, J.; Petros, L. M.; Zhang, L.; Kobayashi, M.; Copeland, N. G.; Jenkins, N. A.; Rosenberg, R. D.: Multiple isoforms of heparan sulfate D–glucosaminyl 3–O–sulfotransferase: isolation, characterization, and expression of human cDNAs and identification of distinct genomic loci. *J. Biol. Chem.* 274: 5170–5184, 1999.

[34577] 8129. McIlhatton, M. A.; Burrows, J. F.; Donaghy, P. G.;

Chanduloy, S.; Johnston, P. G.; Russell, S. E. H.: Genomic organization, complex splicing pattern and expression of a human septin gene on chromosome 17q25.3. *Oncogene* 20: 5930–5939, 2001.

[34578] 8130. Osaka, M.; Rowley, J. D.; Zeleznik-Le, N. J.: MSF (MLL septin-like fusion), a fusion partner gene of MLL, in a therapy-related acute myeloid leukemia with a t(11;17)(q23;q25). *Proc. Nat. Acad. Sci.* 96:6428–6433, 1999.

[34579] 8131. Russell, S. E. H.; McIlhatton, M. A.; Burrows, J. F.; Donaghy, P. G.; Chanduloy, S.; Petty, E. M.; Kalikin, L. M.; Church, S. W.; McIlroy, S.; Harkin, D. P.; Keilty, G. W.; Cranston, A. N.; Weissenbach, J.; Hickey, I.; Johnston, P. G.: Isolation and mapping of a human septin gene to a region on chromosome 17q, commonly deleted in sporadic epithelial ovarian tumors. *Cancer Res.* 60: 4729–4734, 2000.

[34580] 8132. Karpinski, B. A.; Morle, G. D.; Huggenvik, J.; Uhler, M. D.; Leiden, J. M.: Molecular cloning of human CREB-2: an ATF/CREB transcription factor that can negatively regulate transcription from the cAMP response element. *Proc. Nat. Acad. Sci.* 89: 4820–4824, 1992.

[34581] 8133. Kasukabe, T.; Kobayashi, H.; Kaneko, Y.; Okabe–

Kado, J.; Honma, Y.: Identity of human normal counterpart (MmTRA1b) of mouse leukemogenesis-associated gene (MmTRA1a) product as a plasma membrane phospholipid scramblase and chromosome mapping of the human Mm-TRA1b/phospholipid scramblase gene. *Biochem. Biophys. Res. Commun.* 249: 449–455, 1998.

[34582] 8134. Zhou, Q.; Sims, P. J.; Wiedmer, T.: Identity of a conserved motif in phospholipid scramblase that is required for Ca^{2+} -accelerated transbilayer movement of membrane phospholipids. *Biochemistry* 37: 2356–2360, 1998.

[34583] 8135. Zhou, Q.; Zhao, J.; Stout, J. G.; Luhm, R. A.; Wiedmer, T.; Sims, P. J.: Molecular cloning of human plasma membrane phospholipid scramblase: a protein mediating transbilayer movement of plasma membrane phospholipids. *J. Biol. Chem.* 272: 18240–18244, 1997.

[34584] 8136. Robinson, R. C.; Turbedsky, K.; Kaiser, D. A.; Marchand, J.-B.; Higgs, H. N.; Choe, S.; Pollard, T. D.: Crystal structure of Arp2/3 complex. *Science* 294: 1679–1684, 2001.

[34585] 8137. Volkman, N.; Amann, K. J.; Stoilova-McPhie, S.; Egile, C.; Winter, D. C.; Hazelwood, L.; Heuser, J. E.; Li, R.; Pollard, T. D.; Hanein, D.: Structure of Arp2/3 complex in its activated state and in actin filament branch junctions.

Science 293: 2456–2459, 2001.

[34586] 8138. Welch, M. D.; DePace, A. H.; Verma, S.; Iwamatsu, A.; Mitchison, T. J.: The human Arp2/3 complex is composed of evolutionarily conserved subunits and is localized to cellular regions of dynamic actin filament assembly. *J. Cell Biol.* 138: 375–384, 1997.

[34587] 8139. Michel, J. J.; Xiong, Y.: Human CUL-1, but not other cullin family members, selectively interacts with SKP1 to form a complex with SKP2 and cyclin A. *Cell Growth Differ.* 9: 435–449, 1998.

[34588] 8140. Du, M.; Sansores-Garcia, L.; Zu, Z.; Wu, K. K.: Cloning and expression analysis of a novel salicylate suppressible gene, Hs-CUL-3, a member of cullin/Cdc53 family. *J. Biol. Chem.* 273: 24289–24292, 1998.

[34589] 8141. Bao, S.; Shen, X.; Shen, K.; Liu, Y.; Wang, X.-F.: The mammalian Rad24 homologous to yeast *Saccharomyces cerevisiae* Rad24 and *Schizosaccharomyces pombe* Rad17 is involved in DNA damage checkpoint. *Cell Growth Differ.* 9: 961–967, 1998.

[34590] 8142. Bao, S.; Chang, M.-S.; Auclair, D.; Sun, Y.; Wang, Y.; Wong, W.-K.; Zhang, J.; Liu, Y.; Qian, X.; Sutherland, R.; Magi-Galluzzi, C.; Weisberg, E.; Cheng, E. Y. S.; Hao, L.; Sasaki, H.; Campbell, M. S.; Kraeft, S.-K.; Loda, M.; Lo,

K.-M.; Chen, L. B.: Hrad17, a human homologue of the *Schizosaccharomyces pombe* checkpoint gene rad17, is overexpressed in colon carcinoma. *Cancer Res.* 59: 2023–2028, 1999.

[34591] 8143. Bluysen, H. A. R.; Naus, N. C.; van Os, R. I.; Jaspers, I.; Hoeijmakers, J. H. J.; de Klein, A.: Human and mouse homologues of the *Schizosaccharomyces pombe* rad17+ cell cycle checkpoint control gene. *Genomics* 55: 219–228, 1999.

[34592] 8144. Loijens, J. C.; Anderson, R. A.: Type I phosphatidylinositol-4-phosphate 5-kinases are distinct members of this novel lipid kinase family. *J. Biol. Chem.* 271: 32937–32943, 1996.

[34593] 8145. Xie, Y.; Zhu, L.; Zhao, G.: Assignment of type I phosphatidylinositol-4-phosphate 5-kinase (PIP5K1A) to human chromosome bands 1q22–q24 by in situ hybridization. *Cytogenet. Cell Genet.* 88: 197–199, 2000.

[34594] 8146. Seelig, H. P.; Moosbrugger, I.; Ehrfeld, H.; Fink, T.; Renz, M.; Genth, E.: The major dermatomyositis-specific Mi-2 autoantigen is a presumed helicase involved in transcriptional activation. *Arthritis Rheum.* 38: 1389–1399, 1995.

[34595] 8147. Wade, P. A.; Geronzi, A.; Jones, P. L.; Ballestar, E.;

Aubry, F.;Wolffe, A. P.: Mi-2 complex couples DNA methylation to chromatinremodelling and histone deacetylation. Nature Genet. 23: 62–66,1999.

[34596] 8148.Zhang, Y.; LeRoy, G.; Seelig, H.–P.; Lane, W. S.; Reinberg, D.: The dermatomyositis–specific autoantigen Mi2 is a component of acomplex containing histone deacetylase and nucleosome remodeling activities. Cell 95:279–289, 1998.

[34597] 8149.Aihara, T.; Miyoshi, Y.; Koyama, K.; Suzuki, M.; Takahashi, E.;Monden, M.; Nakamura, Y.: Cloning and mapping of SMARCA5 encodinghSNF2H, a novel human homologue of Drosophila ISWI. Cytogenet. CellGenet. 81: 191–193, 1998.

[34598] 8150.Bochar, D. A.; Savard, J.; Wang, W.; Lafleur, D. W.; Moore, P.;Cote, J.; Shiekhattar, R.: A family of chromatin remodeling factorsrelated to Williams syndrome transcription factor. Proc. Nat. Acad.Sci. 97: 1038–1043, 2000.

[34599] 8151.Bozhenok, L.; Wade, P. A.; Varga–Weisz, P.: WSTF–ISWI chromatinremodeling complex targets heterochromatic replication foci. EMBOJ. 21: 2231–2241, 2002.

[34600] 8152.LeRoy, G.; Orphanides, G.; Lane, W. S.; Reinberg, D.: Requirementof RSF and FACT for transcription of chromatin templates in vitro. Science 282:1900–1904, 1998.

- [34601] 8153.Poot, R. A.; Dellaire, G.; Hulsman, B. B.; Grimaldi, M. A.; Corona,D. F. V.; Becker, P. B.; Bickmore, W. A.; Varga-Weisz, P. D.: HuCHRAC,a human ISWI chromatin re-modelling complex contains hACF1 and twonovel histone-fold proteins. EMBO J. 19: 3377–3387, 2000.
- [34602] 8154.Alberati-Giani, D.; Cesura, A. M.; Broger, C.; Warren, W. D.; Rover,S.; Malherbe, P.: Cloning and functional expression of human kynurenine3-monooxygenase. FEBS Lett. 410: 407–412, 1997.
- [34603] 8155.Bounous, G.; Kongshavn, P. A. L.; Taveroff, A.; Gold, P.: Evolutionarytraits in human milk proteins. Med. Hypotheses 27: 133–140, 1988.
- [34604] 8156.Davies, M. S.; West, L. F.; Davis, M. B.; Povey, S.; Craig, R.K.: The gene for human alpha-lactalbumin is assigned to chromosome12q13. Ann. Hum. Genet. 51: 183–188, 1987.
- [34605] 8157.Dayhoff, M. O.: Atlas of Protein Sequence and Structure. Lactalbuminand Lysozymes. Washington: National Biomedical Research Foundation(pub.) 5: 1972. Pp. D133–D140.
- [34606] 8158.Hakansson, A.; Zhivotovsky, B.; Orrenius, S.; Sabharwal, H.; Svanborg,C.: Apoptosis induced by a human milk protein. Proc. Nat. Acad.Sci. 92: 8064–8068, 1995.

- [34607] 8159.Hall, L.; Davies, M. S.; Craig, R. K.: The construction, identification and characterisation of plasmids containing human alpha-lactalbumin cDNA sequences. *Nucleic Acids Res.* 9: 65–84, 1981.
- [34608] 8160.Hall, L.; Emery, D. C.; Davies, M. S.; Parker, D.; Craig, R. K.: Organization and sequence of the human alpha-lactalbumin gene. *Biochem.J.* 242: 735–742, 1987.
- [34609] 8161.Stacey, A.; Schnieke, A.; Kerr, M.; Scott, A.; McKee, C.; Cottingham, I.; Binas, B.; Wilde, C.; Colman, A.: Lactation is disrupted by alpha-lactalbumin deficiency and can be restored by human alpha-lactalbumin gene replacement in mice. *Proc. Nat. Acad. Sci.* 92: 2835–2839, 1995.
- [34610] 8162.Boyer, S. H.; Fainer, D. C.; Watson-Williams, E. J.: Lactate dehydrogenase variant from human blood: evidence for molecular subunits. *Science* 141:642–643, 1963.
- [34611] 8163.Ben-Neriah, Y.; Bauskin, A. R.: Leukocytes express a novel gene encoding a putative transmembrane protein-kinase devoid of an extracellular domain. (Letter) *Nature* 333: 672–676, 1988.
- [34612] 8164.Krolewski, J. J.; Lee, R.; Eddy, R.; Shows, T. B.; Dalla-Favera, R.: Identification and chromosomal mapping of new human tyrosine kinase genes. *Oncogene* 5: 277–282,

1990.

- [34613] 8165.Liao, X.; Zhou, R.; Gilbert, D. J.; Copeland, N. G.; Jenkins, N.A.: Receptor tyrosine kinase gene Tyro3 maps to mouse chromosome2, closely linked to Ltk. *Mammalian Genome* 7: 395–396, 1996.
- [34614] 8166.Toyoshima, H.; Kozutsumi, H.; Maru, Y.; Hagiwara, K.; Furuya, A.;Mioh, H.; Hanai, N.; Takaku, F.; Yazaki, Y.; Hirai, H.: Differentlyspliced cDNAs of human leukocyte tyrosine kinase receptor tyrosinekinase predict receptor proteins with and without a tyrosine kinasedomain and a soluble receptor protein. *Proc. Nat. Acad. Sci.* 90:5404–5408, 1993.
- [34615] 8167.Knobeloch, K.–P.; Wright, M. D.; Ochsenbein, A. F.; Liesenfeld,O.; Lohler, J.; Zinkernagel, R. M.; Horak, I.; Orinska, Z.: Targetedinactivation of the tetraspanin CD37 impairs T–cell–dependent B–cellresponse under suboptimal costimulatory conditions. *Molec. Cell.Biol.* 20: 5363–5369, 2000.
- [34616] 8168.Virtaneva, K. I.; Angelisova, P.; Baumruker, T.; Horejsi, V.; Nevanlinna,H.; Schroder, J.: The genes for CD37, CD53, and R2, all members ofa novel gene family, are located on different chromosomes. *Immunogenetics* 37:461–465, 1993.

- [34617] 8169.Delmas, B.; Gelfi, J.; L'Haridon, R.; Vogel, L. K.; Sjostrom, H.;Noren, O.; Laude, H.: Aminopeptidase N is a major receptor for the enteropathogenic coronavirus TGEV. *Nature* 357: 417–420, 1992.
- [34618] 8170.Kruse, T. A.; Bolund, L.; Grzeschik, K.–H.; Ropers, H. H.; Olsen, J.; Sjostrom, H.; Noren, O.: Assignment of the human aminopeptidase N (peptidase E) gene to chromosome 15q13–qter. *FEBS Lett.* 239: 305–308, 1988.
- [34619] 8171.Look, A. T.; Ashmun, R. A.; Shapiro, L. H.; Peiper, S. C.: Human myeloid plasma membrane glycoprotein CD13 (gp150) is identical to aminopeptidase N. *J. Clin. Invest.* 83: 1299–1307, 1989.
- [34620] 8172.Look, A. T.; Peiper, S. C.; Rebentisch, M. B.; Ashmun, R. A.; Roussel, M. F.; Lemons, R. S.; Le Beau, M. M.; Rubin, C. M.; Sherr, C. J.: Molecular cloning, expression, and chromosomal localization of the gene encoding a human myeloid membrane antigen (gp150). *J. Clin. Invest.* 78: 914–921, 1986.
- [34621] 8173.Watt, V. M.; Willard, H. F.: The human aminopeptidase N gene: isolation, chromosome localization, and DNA polymorphism analysis. *Hum. Genet.* 85: 651–654, 1990.
- [34622] 8174.Yeager, C. L.; Ashmun, R. A.; Williams, R. K.; Cardelli, C. B.; Shapiro, L. H.; Look, A. T.; Holmes, K. V.: Hu–

man aminopeptidaseN is a receptor for human coronavirus 229E. *Nature* 357: 420–422, 1992.

[34623] 8175. Yoshimura, K.; Toibana, A.; Nakahama, K.: Human lysozyme: sequencing of a cDNA, and expression and secretion by *Saccharomyces cerevisiae*. *Biochem. Biophys. Res. Commun.* 150: 794–801, 1988.

[34624] 8176. Salmikangas, P.; Mykkanen, O.–M.; Gronholm, M.; Heiska, L.; Kere, J.; Carpen, O.: Myotilin, a novel sarcomeric protein with two Ig-like domains, is encoded by a candidate gene for limb-girdle muscular dystrophy. *Hum. Molec. Genet.* 8: 1329–1336, 1999.

[34625] 8177. Hauser, M. A.; Horrigan, S. K.; Salmikangas, P.; Torian, U. M.; Viles, K. D.; Dancel, R.; Tim, R. W.; Taivainen, A.; Bartoloni, L.; Gilchrist, J. M.; Stajich, J. M.; Gaskell, P. C.; Gilbert, J. R.; Vance, J. M.; Pericak-Vance, M. A.; Carpen, O.; Westbrook, C. A.; Speer, M. C.: Myotilin is mutated in limb girdle muscular dystrophy 1A. *Hum. Molec. Genet.* 9: 2141–2147, 2000.

[34626] 8178. Riegman, P. H. J.; Vlietstra, R. J.; van der Korput, J. A. G. M.; Romijn, J. C.; Trapman, J.: Characterization of the prostate-specific antigen gene: a novel human kallikrein-like gene. *Biochem. Biophys. Res. Commun.* 159: 95–102, 1989.

- [34627] 8179.Santoro, M.; Carlomagno, F.; Hay, I. D.; Herrmann, M. A.; Grieco, M.; Melillo, R.; Pierotti, M. A.; Bongarzone, I.; Della Porta, G.; Berger, N.; Peix, J. L.; Paulin, C.; Fabien, N.; Vecchio, G.; Jenkins, R. B.; Fusco, A.: Ret oncogene activation in human thyroid neoplasms restricted to the papillary cancer subtype. *J. Clin. Invest.* 89:1517–1522, 1992.
- [34628] 8180.Santoro, M.; Carlomagno, F.; Romano, A.; Bottaro, D. P.; Dathan, N. A.; Grieco, M.; Fusco, A.; Vecchio, G.; Matoskova, B.; Kraus, M.H.; Di Fiore, P. P.: Activation of RET as a dominant transforming gene by germline mutations of MEN2A and MEN2B. *Science* 267: 381–383, 1995.
- [34629] 8181.Schuchardt, A.; D'Agati, V.; Larsson-Blomberg, L.; Costantini, F.; Pachnis, V.: Defects in the kidney and enteric nervous system of mice lacking the tyrosine kinase receptor Ret. *Nature* 367: 380–383, 1994.
- [34630] 8182.Shirahama, S.; Ogura, K.; Takami, H.; Ito, K.; Tohsen, T.; Miyauchi, A.; Nakamura, Y.: Mutational analysis of the RET proto-oncogene in 71 Japanese patients with medullary thyroid carcinoma. *J. Hum. Genet.* 43:101–106, 1998.
- [34631] 8183.Seri, M.; Yin, L.; Barone, A.; Bolino, A.; Celli, I.; Bocciardi, R.; Pasini, B.; Ceccherini, I.; Lerone, M.; Kristoffersson, U.; Larsson, L. T.; Casasa, J. M.; Cass, D. T.;

Abramowicz, M. J.; Vanderwinden, J.-M.; Kravcenkiene, I.; Baric, I.; Silengo, M.; Martucciello, G.; Romeo, G.: Frequency of RET mutations in long- and short-segment Hirschsprung disease. *Hum. Mutat.* 9: 243–249, 1997.

[34632] 8184. Takahashi, M.; Buma, Y.; Hiai, H.: Isolation of ret proto-oncogene cDNA with an amino-terminal signal sequence. *Oncogene* 4: 805–806, 1989.

[34633] 8185. Takahashi, M.; Buma, Y.; Iwamoto, T.; Inaguma, Y.; Ikeda, H.; Hiai, H.: Cloning and expression of the ret proto-oncogene encoding a tyrosine kinase with two potential transmembrane domains. *Oncogene* 3: 571–578, 1988.

[34634] 8186. Takahashi, M.; Ritz, J.; Cooper, G. M.: Activation of a novel human transforming gene, ret, by DNA rearrangement. *Cell* 42: 581–588, 1985.

[34635] 8187. Tessitore, A.; Sinisi, A. A.; Pasquali, D.; Cardone, M.; Vitale, D.; Bellastella, A.; Colantuoni, V.: A novel case of multiple endocrine neoplasia type 2A associated with two de novo mutations of the RET proto-oncogene. *J. Clin. Endocr. Metab.* 84: 3522–3527, 1999.

[34636] 8188. van Heyningen, V.: One gene—four syndromes. *Nature* 367: 319–320, 1994.

- [34637] 8189.Xue, F.; Yu, H.; Maurer, L. H.; Memoli, V. A.; Nutile-McMenemey, N.; Schuster, M. K.; Bowden, D. W.; Mao, J.; Noll, W. W.: Germline RET mutations in MEN 2A and FMTC and their detection by simple DNA diagnostic tests. *Hum. Molec. Genet.* 3: 635–638, 1994.
- [34638] 8190.Yin, L.; Ceccherini, I.; Pasini, B.; Matera, I.; Bicocchi, M.P.; Barone, V.; Bocciardi, R.; Kaariainen, H.; Weber, D.; Devoto, M.; Romeo, G.: Close linkage with the RET proto-oncogene and boundaries of deletion mutations in autosomal dominant Hirschsprung disease. *Hum. Molec. Genet.* 2: 1803–1808, 1993.
- [34639] 8191.Feller, S. M.; Ren, R. B.; Hanafusa, H.; Baltimore, D.: SH2 and SH3 domains as molecular adhesives: the interactions of crk and abl. *Trends Biochem. Sci.* 19: 453–458, 1994.
- [34640] 8192.Fioretos, T.; Heisterkamp, N.; Groffen, J.; Benjes, S.; Morris, C.: CRK proto-oncogene maps to human chromosome band 17p13. *Oncogene* 8:2853–2855, 1993.
- [34641] 8193.Matsuda, M.; Tanaka, S.; Nagata, S.; Kojima, A.; Kurata, T.; Shibuya, M.: Two species of human CRK cDNA encode proteins with distinct biological activities. *Molec. Cell. Biol.* 12: 3482–3489, 1992.
- [34642] 8194.Reichman, C. T.; Mayer, B. J.; Keshav, S.; Hanafusa,

H.: The product of the cellular *crk* gene consists primarily of SH2 and SH3 regions. *Cell Growth Differ.* 3: 451–460, 1992.

[34643] 8195. Schuuring, E.; Verhoeven, E.; Mooi, W. J.; Michalides, R. J. A.M.: Identification and cloning of two overexpressed genes, U21B31/PRAD1 and EMS1, within the amplified chromosome 11q13 region in human carcinomas. *Oncogene* 7:355–361, 1992.

[34644] 8196. van Damme, H.; Brok, H.; Schuuring-Scholtes, E.; Schuuring, E.: The redistribution of cortactin into cell-matrix contact sites in human carcinoma cells with 11q13 amplification is associated with both overexpression and post-translational modification. *J. Biol. Chem.* 272: 7374–7380, 1997.

[34645] 8197. Benz-Lemoine, E.; Brizard, A.; Huret, J.-L.; Babin, P.; Guilhot, F.; Couet, D.; Tanzer, J.: Malignant histiocytosis: a specific t(2;5)(p23;q35) translocation? Review of the literature. *Blood* 72: 1045–1047, 1988.

[34646] 8198. Boulton, J.; Rack, K.; Kelly, S.; Madden, J.; Sakaguchi, A. Y.; Wang, L.-M.; Oscier, D. G.; Buckle, V. J.; Wainscoat, J. S.: Loss of both CSF1R (FMS) alleles in patients with myelodysplasia and chromosome 5 deletion. *Proc. Nat. Acad. Sci.* 88: 6176–6180, 1991.

- [34647] 8199.De Qi Xu; Guilhot, S.; Galibert, F.: Restriction fragment length polymorphism of the human c-fms gene. Proc. Nat. Acad. Sci. 82:2862–2865, 1985.
- [34648] 8200.Eccles, M. R.: Genes encoding the platelet-derived growth factor(PDGF) receptor and colony-stimulating factor 1 (CSF-1) receptor are physically associated in mice as in humans. Gene 108: 285–288, 1991.
- [34649] 8201.Gisselbrecht, S.; Fichelson, S.; Sola, B.; Bordereaux, D.; Hampe,A.; Andre, C.; Galibert, F.; Tambourin, P. E.: Frequent c-fms activation by proviral insertion in mouse myeloblastic leukaemias. Nature 329:259–261, 1987.
- [34650] 8202.Groffen, J.; Heisterkamp, N.; Spurr, N. K.; Dana, S. L.; Wasmuth,J. J.; Stephenson, J. R.: Regional assignment of the human c-fms oncogene to band q34 of chromosome 5. (Abstract) Cytogenet. Cell Genet. 37: 484 only, 1984.
- [34651] 8203.Hampe, A.; Gobet, M.; Sherr, C. J.; Galibert, F.: Nucleotide sequence of the feline retroviral oncogene v-fms shows unexpected homology with oncogenes encoding tyrosine-specific protein kinases. Proc.Nat. Acad. Sci. 81: 85–89, 1984.
- [34652] 8204.Hampe, A.; Shamoon, B.-M.; Gobet, M.; Sherr, C. J.; Galibert, F.: Nucleotide sequence and structural organization of the human FMS proto-oncogene. Oncogene Res. 4:

9–17, 1989.

[34653] 8205.How, G.–F.; Venkatesh, B.; Brenner, S.: Conserved linkage between the puffer fish (*Fugu rubripes*) and human genes for platelet–derived growth factor receptor and macrophage colony–stimulating factor receptor.

Genome Res. 6: 1185–1191, 1996.

[34654] 8206.Hibbs, M. L.; Tarlinton, D. M.; Armes, J.; Grail, D.; Hodgson, G.; Maglitto, R.; Stacker, S. A.; Dunn, A. R.: Multiple defects in the immune system of Lyn–deficient mice, culminating in autoimmune disease. Cell 83: 301–311, 1995.

[34655] 8207.Webster, M. A.; Cardiff, R. D.; Muller, W. J.: Induction of mammary epithelial hyperplasias and mammary tumors in transgenic mice expressing a murine mammary tumor virus/activated c–src fusion gene. Proc. Nat. Acad. Sci. 92: 7849–7853, 1995.

[34656] 8208.Yamanashi, Y.; Fukushige, S.–I.; Semba, K.; Sukegawa, J.; Miyajima, N.; Matsubara, K.–I.; Yamamoto, T.; Toyoshima, K.: The yes–related cellular gene lyn encodes a possible tyrosine kinase similar to p56(lck). Molec. Cell. Biol. 7: 237–243, 1987.

[34657] 8209.Berger, A.; Rosenthal, D.; Spiegel, S.: Sphingophosphocholine, a signaling molecule which accumu–

lates in Niemann–Pick disease typeA, stimulates DNA–binding activity of the transcription activatorprotein AP–1. Proc. Nat. Acad. Sci. 92: 5885–5889, 1995.

[34658] 8210.Bohmann, D.; Bos, T. J.; Admon, A.; Nishimura, T.; Vogt, P. K.;Tjian, R.: Human proto–oncogene c–jun encodes a DNA binding proteinwith structural and functional properties of transcription factorAP–1. Science 238: 1386–1392, 1987.

[34659] 8211.Bos, T. J.; Bohmann, D.; Tsuchie, H.; Tjian, R.; Vogt, P. K.:v–jun encodes a nuclear protein with enhancer binding properties ofAP–1. Cell 52: 705–712, 1988.

[34660] 8212.Haluska, F. G.; Huebner, K.; Isobe, M.; Nishimura, T.; Croce, C.M.; Vogt, P. K.: Localization of the human JUN protooncogene to chromosomeregion 1p31–32. Proc. Nat. Acad. Sci. 85: 2215–2218, 1988.

[34661] 8213.Hattori, K.; Angel, P.; Le Beau, M. M.; Karin, M.: Structure andchromosomal localization of the functional intronless human JUN protooncogene. Proc.Nat. Acad. Sci. 85: 9148–9152, 1988.

[34662] 8214.Lamph, W. W.; Wamsley, P.; Sassone–Corsi, P.; Verma, I. M.: Inductionof proto–oncogene JUN/AP–1 by serum and TPA. Nature 334: 629–631,1988.

[34663] 8215.Marx, J. L.: 'Jun' is bustin' out all over. (Research

News). Science 242:1377–1378, 1988.

[34664] 8216. Mattei, M. G.; Simon–Chazottes, D.; Hirai, S.–I.; Ryseck, R.–P.; Galcheva–Gargova, Z.; Guenet, J. L.; Mattei, J. F.; Bravo, R.; Yaniv, M.: Chromosomal localization of the three members of the jun proto–oncogene family in mouse and man. Oncogene 5: 151–156, 1990.

[34665] 8217. Shaulian, E.; Karin, M.: AP–1 as a regulator of cell life and death. Nature Cell Biol. 4: E131–E136, 2002.

[34666] 8218. Shaulian, E.; Schreiber, M.; Piu, F.; Beeche, M.; Wagner, E. F.; Karin, M.: The mammalian UV response: c–Jun induction is required for exit from p53–imposed growth arrest. Cell 103: 897–907, 2000.

[34667] 8219. Mattei, M. G.; Simon–Chazottes, D.; Hirai, S.; Ryseck, R. P.; Galcheva–Gargova, Z.; Guenet, J. L.; Mattei, J. F.; Bravo, R.; Yaniv, M.: Chromosomal localization of the three members of the jun proto–oncogene family in mouse and man. Oncogene 5: 151–156, 1990.

[34668] 8220. Passegue, E.; Jochum, W.; Behrens, A.; Ricci, R.; Wagner, E. F.: JunB can substitute for Jun in mouse development and cell proliferation. Nature Genet. 30: 158–166, 2002.

[34669] 8221. Passegue, E.; Jochum, W.; Schorpp–Kistner, M.; Mohle–Steinlein, U.; Wagner, E. F.: Chronic myeloid

leukemia with increased granulocyte progenitors in mice lacking JunB expression in the myeloid lineage. *Cell* 104:21–32, 2001.

- [34670] 8222. Phinney, D. G.; Tseng, S. W.; Ryder, K.: Complex genetic organization of junB: multiple blocks of flanking evolutionarily conserved sequence at the murine and human junB loci. *Genomics* 28: 228–234, 1995.
- [34671] 8223. Sullivan, M.; Olsen, A. S.; Houslay, M. D.: Genomic organisation of the human cyclic AMP-specific phosphodiesterase PDE4C gene and its chromosomal localisation to 19p13.1, between RAB3A and JUND. *Cell. Signal.* 11: 735–742, 1999.
- [34672] 8224. Weitzman, J. B.; Fiette, L.; Matsuo, K.; Yaniv, M.: JunD protects cells from p53-dependent senescence and apoptosis. *Molec. Cell* 6:1109–1119, 2000.

- [34673] 8225.DeKoter, R. P.; Lee, H.-J.; Singh, H.: PU.1 regulates expression of the interleukin-7 receptor in lymphoid progenitors. *Immunity* 16:297-309, 2002.
- [34674] 8226.DeKoter, R. P.; Singh, H.: Regulation of B lymphocyte and macrophage development by graded expression of PU.1. *Science* 288: 1439-1441, 2000.
- [34675] 8227.Li, S.-L.; Valente, A. J.; Zhao, S.-J.; Clark, R. A.: PU.1 is essential for p47(phox) promoter activity in myeloid cells. *J. Biol.Chem.* 272: 17802-17809, 1997.
- [34676] 8228.Moreau-Gachelin, F.; Tavitian, A.; Tambourin, P.: Spi-1 is a putative oncogene in virally induced murine erythroleukaemias. *Nature* 331:277-280, 1988.
- [34677] 8229.Ray, D.; Culine, S.; Tavitian, A.; Moreau-Gachelin, F.: The human homologue of the putative proto-oncogene Spi-1: characterization and expression in tumors. *Oncogene* 5: 663-667, 1990.
- [34678] 8230.Tondravi, M. M.; McKercher, S. R.; Anderson, K.; Erdmann, J. M.; Quiroz, M.; Maki, R.; Teitelbaum, S. L.: Osteopetrosis in mice lacking haematopoietic transcription factor PU.1 *Nature* 386: 81-84, 1997.
- [34679] 8231.Xie, C.-W.; Sayah, D.; Chen, Q.-S.; Wei, W.-Z.; Smith, D.; Liu, X.: Deficient long-term memory and long-lasting long-term potentiation in mice with a targeted deletion of

neurotrophin-4 gene. *Proc. Nat. Acad. Sci.* 97: 8116–8121, 2000.

[34680] 8232. Steiglitz, B. M.; Greenspan, D. S.: Assignment of the mouse *Pcolce2* gene, which encodes procollagen C-proteinase enhancer protein 2, to chromosome 9 and localization of *PCOLCE2* to human chromosome 3q23. *Cytogenet. Cell Genet.* 95: 244–245, 2001.

[34681] 8233. Xu, H.; Acott, T. S.; Wirtz, M. K.: Identification and expression of a novel type I procollagen C-proteinase enhancer protein gene from the glaucoma candidate region on 3q21–q24. *Genomics* 66: 264–273, 2000.

[34682] 8234. Ke, N.; Godzik, A.; Reed, J. C.: Bcl-B, a novel Bcl-2 family member that differentially binds and regulates Bax and Bak. *J. Biol. Chem.* 276:12481–12484, 2001.

[34683] 8235. Lee, R.; Chen, J.; Matthews, C. P.; McDougall, J. K.; Neiman, P. E.: Characterization of NR13-related human cell death regulator, Boo/Diva, in normal and cancer tissues. *Biochim. Biophys. Acta* 1520:187–194, 2001.

[34684] 8236. Naumann, U.; Weit, S.; Wischhusen, J.; Weller, M.: Diva/Boo is a negative regulator of cell death in human glioma cells. *FEBS Lett.* 505:23–26, 2001.

[34685] 8237. Zhang, H.; Holzgreve, W.; De Geyter, C.: Bcl2-L-10, a novel anti-apoptotic member of the Bcl-2 family, blocks

apoptosis in the mitochondria death pathway but not in the death receptor pathway. *Hum. Molec. Genet.* 10:2329–2339, 2001.

[34686] 8238.Brand, S. H.; Castle, J. D.: SCAMP–37, a new marker within the general cell surface recycling system. *EMBO J.* 12: 3753–3761, 1993.

[34687] 8239.Singleton, D. R.; Wu, T. T.; Castle, J. D.: Three mammalian SCAMPs (secretory carrier membrane proteins) are highly related products of distinct genes having similar subcellular distributions. *J. Cell Sci.* 110: 2099–2107, 1997.

[34688] 8240.Yamashita, A.; Ohnishi, T.; Kashima, I.; Taya, Y.; Ohno, S.: Human SMG–1, a novel phosphatidylinositol 3–kinase–related protein kinase, associates with components of the mRNA surveillance complex and is involved in the regulation of nonsense–mediated mRNA decay. *Genes Dev.* 15: 2215–2228, 2001.

[34689] 8241.Spataro, V.; Toda, T.; Craig, R.; Seeger, M.; Dubiel, W.; Harris, A. L.; Norbury, C.: Resistance to diverse drugs and ultraviolet light conferred by overexpression of a novel human 26 S proteasome subunit. *J. Biol. Chem.* 272: 30470–30475, 1997.

[34690] 8242.Yao, T.; Cohen, R. E.: A cryptic protease couples

deubiquitination and degradation by the proteasome. Nature 1 Sep, 2002. Note: Advance Electronic Publication.

- [34691] 8243. Kamberov, E.; Makarova, O.; Roh, M.; Liu, A.; Karnak, D.; Straight, S.; Margolis, B.: Molecular cloning and characterization of Pals, proteins associated with mLin-7. J. Biol. Chem. 275: 11425–11431, 2000.
- [34692] 8244. Tseng, T.-C.; Marfatia, S. M.; Bryant, P. J.; Pack, S.; Zhuang, A.; O'Brien, J. E.; Lin, L.; Hanada, T.; Chishti, A. H.: VAM-1: a new member of the MAGUK family binds to human Veli-1 through a conserved domain. Biochim. Biophys. Acta 1518: 249–259, 2001.
- [34693] 8245. Wei, X.; Malicki, J. M.: nagie oko, encoding a MAGUK-family protein, is essential for cellular patterning of the retina. Nature Genet. 31: 150–157, 2002. Note: Erratum: Nature Genet. 31: 439 only, 2002.
- [34694] 8246. Esser, V.; Russell, D. W.: Transport-deficient mutations in the low density lipoprotein receptor: alterations in the cysteine-rich and cysteine-poor regions of the protein block intracellular transport. J. Biol. Chem. 263: 13276–13281, 1988.
- [34695] 8247. Fumeron, F.; Grandchamp, B.; Fricker, J.; Krempf, M.; Wolf, L.-M.; Khayat, M.-C.; Boiffard, O.; Apfelbaum, M.: Presence of the French Canadian deletion in a French pa-

tient with familial hypercholesterolemia.(Letter) New Eng. J. Med. 326: 69 only, 1992.

- [34696] 8248.Gilbert, W.: Genes-in-pieces revisited. Science 228: 823–824,1985.
- [34697] 8249.Goldfarb, L. G.; Petersen, R. B.; Tabaton, M.; Brown, P.; LeBlanc,A. C.; Montagna, P.; Cortelli, P.; Julien, J.; Vital, C.; Pendelbury,W. W.; Haltia, M.; Wills, P. R.; and 9 others: Fatal familial insomniaand familial Creutzfeldt–Jakob disease: disease phenotype determinedby a DNA polymorphism. Science 258: 806–808, 1992.
- [34698] 8250.Graadt van Roggen, F.; van der Westhuyzen, D. R.; Marais, A. D.;Gevers, W.; Coetzee, G. A.: Low density lipoprotein receptor foundermutations in Afrikaner familial hypercholesterolaemic patients: acomparison of two geographical areas. Hum. Genet. 88: 204–208, 1991.
- [34699] 8251.Gudnason, V.; King–Underwood, L.; Seed, M.; Sun, X.–M.; Soutar,A. K.; Humphries, S. E.: Identification of recurrent and novel mutationsin exon 4 of the LDL receptor gene in patients with familial hypercholesterolemia in the United Kingdom. Arteriosclerosis Thromb. 13: 56–63, 1993.
- [34700] 8252.Gudnason, V.; Sigurdsson, G.; Nissen, H.; Humphries, S. E.: Commonfounder mutation in the LDL

receptor gene causing familial hypercholesterolemia in the Icelandic population. *Hum. Mutat.* 10: 36–44, 1997.

[34701] 8253. Henderson, H. E.; Berger, G. M. B.; Marais, A. D.: A new LDL receptor gene deletion mutation in the South African population. *Hum. Genet.* 80: 371–374, 1988.

[34702] 8254. Hobbs, H. H.: Personal Communication. Dallas, Tex. 12/1/1990.

[34703] 8255. Hobbs, H. H.; Brown, M. S.; Russell, D. W.; Davignon, J.; Goldstein, J. L.: Deletion in the gene for the low-density-lipoprotein receptor in a majority of French Canadians with familial hypercholesterolemia. *New Eng. J. Med.* 317: 734–737, 1987.

[34704] 8256. Hobbs, H. H.; Lehrman, M. A.; Yamamoto, T.; Russell, D. W.: Polymorphism and evolution of Alu sequences in the human low density lipoprotein receptor gene. *Proc. Nat. Acad. Sci.* 82: 7651–7655, 1985.

[34705] 8257. Hobbs, H. H.; Leitersdorf, E.; Goldstein, J. L.; Brown, M. S.; Russell, D. W.: Multiple CRM- mutations in familial hypercholesterolemia: evidence for 13 alleles, including four deletions. *J. Clin. Invest.* 81: 909–917, 1988.

[34706] 8258. Horsthemke, B.; Dunning, A.; Humphries, S.: Identification of deletions in the human low density lipoprotein receptor gene. *J. Med. Genet.* 24: 144–147, 1987.

- [34707] 8259.Horsthemke, B.; Kessling, A. M.; Seed, M.; Wynn, V.; Williamson,R.; Humphries, S. E.: Identification of a deletion in the low densitylipoprotein (LDL) receptor gene in a patient with familial hypercholesterolaemia. Hum.Genet. 71: 75–78, 1985.
- [34708] 8260.Jensen, H. K.; Jensen, T. G.; Faergeman, O.; Jensen, L. G.; Andresen,B. S.; Corydon, M. J.; Andreassen, P. H.; Hansen, P. S.; Heath, F.;Bolund, L.; Gregersen, N.: Two mutations in the same low–densitylipoprotein receptor allele act in synergy to reduce receptor functionin heterozygous familial hypercholesterolemia. Hum. Mutat. 9: 437–444,1997.
- [34709] 8261.Jensen, J. M.; Kruse, T. A.; Brorholt–Petersen, J. U.; Christiansen,T. M.; Jensen, H. K.; Kolvraa, S.; Faergeman, O.: Linking genotypeto aorto–coronary atherosclerosis: a model using familial hypercholesterolemiaand aorto–coronary calcification. Ann. Hum. Genet. 63: 511–520,1999.
- [34710] 8262.Kajinami, K.; Fujita, H.; Koizumi, J.; Mabuchi, H.; Takeda, R.;Ohta, M.: Genetically determined mild type of familial hypercholesterolemiaincluding normocholesterolemic patients: FH–Tonami–2. Circulation 80(suppl. 2): 278 only, 1989.

- [34711] 8263.Kajinami, K.; Mabuchi, H.; Itoh, H.; Michishita, I.; Takeda, M.;Wakasugi, T.; Koizumi, J.; Takeda, R.: New variant of low densitylipoprotein receptor gene FH-Tonami. *Arteriosclerosis* 8: 187–192,1988.
- [34712] 8264.Podos, S. D.; Reddy, P.; Ashkenas, J.; Krieger, M.: LDLC encodesa brefeldin A-sensitive, peripheral Golgi protein required for normalGolgi function. *J. Cell Biol.* 127: 679–691, 1994.
- [34713] 8265.Suvorova, E. S.; Kurten, R. C.; Lupashin, V. V.: Identificationof a human orthologue of Sec34p as a component of the cis-Golgi vesicletethering machinery. *J. Biol. Chem.* 276: 22810–22818, 2001.
- [34714] 8266.Whyte, J. R. C.; Munro, S.: The Sec34/35 Golgi transport complexis related to the exocyst, defining a family of complexes involvedin multiple steps of membrane traffic. *Dev. Cell* 1: 527–537, 2001.
- [34715] 8267.Haataja, L.; Groffen, J.; Heisterkamp, N.: Identification of anovel Rac3-interacting protein C1D. *Int. J. Molec. Med.* 1: 665–670,1998.
- [34716] 8268.Nehls, P.; Keck, T.; Greferath, R.; Spiess, E.; Glaser, T.; Rothbarth,K.; Stammer, H.; Werner, D.: cDNA cloning, recombinant expressionand characterization of polypeptides with exceptional DNA affinity. *NucleicAcids Res.* 26:

1160–1166, 1998.

- [34717] 8269. Rothbarth, K.; Hunziker, A.; Stammer, H.; Werner, D.: Promoter of the gene encoding the 16 kDa DNA-binding and apoptosis-inducing C1D protein. *Biochim. Biophys. Acta* 1518: 271–275, 2001.
- [34718] 8270. Zamir, I.; Dawson, J.; Lavinsky, R. M.; Glass, C. K.; Rosenfeld, M. G.; Lazar, M. A.: Cloning and characterization of a corepressor and potential component of the nuclear hormone receptor repression complex. *Proc. Nat. Acad. Sci.* 94: 14400–14405, 1997.
- [34719] 8271. Bera, T. K.; Lee, S.; Salvatore, G.; Lee, B.; Pastan, I.: MRP8, a new member of ABC transporter superfamily, identified by EST database mining and gene prediction program, is highly expressed in breast cancer. *Molec. Med.* 7: 509–516, 2001.
- [34720] 8272. Tammur, J.; Prades, C.; Arnould, I.; Rzhetsky, A.; Hutchinson, A.; Adachi, M.; Schuetz, J. D.; Swoboda, K. J.; Ptacek, L. J.; Rosier, M.; Dean, M.; Allikmets, R.: Two new genes from the human ATP-binding cassette transporter superfamily, ABCC11 and ABCC12, tandemly duplicated on chromosome 16q12. *Gene* 273: 89–96, 2001.
- [34721] 8273. Yabuuchi, H.; Shimizu, H.; Takayanagi, S.; Ishikawa, T.: Multiple splicing variants of two new human ATP–

binding cassette transporters, ABCC11 and ABCC12.

Biochem. Biophys. Res. Commun. 288: 933–939, 2001.

[34722] 8274. Hoefler, G.; Forstner, M.; McGuinness, M. C.; Hulla, W.; Hiden, M.; Krisper, P.; Kenner, L.; Ried, T.; Lengauer, C.; Zechner, R.; Moser, H. W.; Chen, G. L.: cDNA cloning of the human peroxisomal enoyl-CoA hydratase: 3-hydroxyacyl-CoA dehydrogenase bifunctional enzyme and localization to chromosome 3q26.3–3q28: a free left Alu arm is inserted in the 3-prime noncoding region. Genomics 19: 60–67, 1994.

[34723] 8275. Bera, T. K.; Iavarone, C.; Kumar, V.; Lee, S.; Lee, B.; Pastan, I.: MRP9, an unusual truncated member of the ABC transporter superfamily, is highly expressed in breast cancer. Proc. Nat. Acad. Sci. 99: 6997–7002, 2002.

[34724] 8276. Nagle, D. L.; McGrail, S. H.; Vitale, J.; Woolf, E. A.; Dussault, B. J., Jr.; DiRocco, L.; Holmgren, L.; Montagno, J.; Bork, P.; Huszar, D.; Fairchild-Huntress, V.; Ge, P.; Keilty, J.; Ebell, C.; Baldini, L.; Gilchrist, J.; Burr, P.; Carlson, G. A.; Moore, K. J.: The mahogany protein is a receptor involved in suppression of obesity. Nature 398: 148–151, 1999.

[34725] 8277. Zhang, Y.; Ng, H.-H.; Erdjument-Bromage, H.; Tempst, P.; Bird, A.; Reinberg, D.: Analysis of the NuRD

subunits reveals a histone deacetylase core complex and a connection with DNA methylation. *Genes Dev.* 13: 1924–1935, 1999.

- [34726] 8278. Strobl, B.; Wechselberger, C.; Beier, D. R.; Lepperdinger, G.: Structural organization and chromosomal localization of Hyal2, a gene encoding a lysosomal hyaluronidase. *Genomics* 53: 214–219, 1998.
- [34727] 8279. Pineda, M.; Fernandez, E.; Torrents, D.; Estevez, R.; Lopez, C.; Camps, M.; Lloberas, J.; Zorzano, A.; Palacin, M.: Identification of a membrane protein, LAT-2, that co-expresses with 4F2 heavy chain, an L-type amino acid transport activity with broad specificity for small and large zwitterionic amino acids. *J. Biol. Chem.* 274: 19738–19744, 1999.
- [34728] 8280. Alexander, W. S.; Rakar, S.; Robb, L.; Farley, A.; Willson, T. A.; Zhang, J.-G.; Hartley, L.; Kikuchi, Y.; Kojima, T.; Nomura, H.; Hasegawa, M.; Maeda, M.; Fabri, L.; Jachno, K.; Nash, A.; Metcalf, D.; Nicola, N. A.; Hilton, D. J.: Suckling defect in mice lacking the soluble haemopoietin receptor NR6. *Curr. Biol.* 9: 605–608, 1999.
- [34729] 8281. Elson, G. C. A.; Graber, P.; Losberger, C.; Herren, S.; Gretener, D.; Menoud, L. N.; Wells, T. N. C.; Kosco-Vilbois, M. H.; Gauchat, J.-F.: Cytokine-like factor-1, a novel solu-

ble protein, shares homology with members of the cytokine type I receptor family. *J. Immun.* 161:1371–1379, 1998.

- [34730] 8282. Hinderlich, S.; Stasche, R.; Zeitler, R.; Reutter, W.: A bifunctional enzyme catalyzes the first two steps in N-acetylneuraminic acid biosynthesis of rat liver: purification and characterization of UDP-N-acetylglucosamine 2-epimerase/N-acetylmannosamine kinase. *J. Biol. Chem.* 272: 24313–24318, 1997.
- [34731] 8283. Huizing, M.; Anikster, Y.: Personal Communication. Bethesda, Md. 1/10/2000.
- [34732] 8284. Kayashima, T.; Matsuo, H.; Satoh, A.; Ohta, T.; Yoshiura, K.; Matsumoto, N.; Nakane, Y.; Niikawa, N.; Kishino, T.: Nonaka myopathy is caused by mutations in the UDP-N-acetylglucosamine-2-epimerase/N-acetylmannosamine kinase gene (GNE). *J. Hum. Genet.* 47: 77–79, 2002.
- [34733] 8285. Keppler, O. T.; Hinderlich, S.; Langner, J.; Schwartz-Albiez, R.; Reutter, W.; Pawlita, M.: UDP-GlcNAc 2-epimerase: a regulator of cell surface sialylation. *Science* 284: 1372–1376, 1999.
- [34734] 8286. Nonaka, I.; Sunohara, N.; Ishiura, S.; Satoyoshi, E.:

Familial distal myopathy with rimmed vacuole and lamellar (myeloid) body formation. *J. Neurol. Sci.* 51: 141–155, 1981.

[34735] 8287. Schwarzkopf, M.; Knobloch, K.-P.; Rohde, E.; Hinderlich, S.; Wiechens, N.; Lucka, L.; Horak, I.; Reutter, W.; Horstkorte, R.: Sialylation is essential for early development in mice. *Proc. Nat. Acad. Sci.* 99: 5267–5270, 2002.

[34736] 8288. Stasche, R.; Hinderlich, S.; Weise, C.; Effertz, K.; Lucka, L.; Moormann, P.; Reutter, W.: A bifunctional enzyme catalyzes the first two steps in N-acetylneuraminic acid biosynthesis of rat liver: molecular cloning and functional expression of UDP-N-acetyl-glucosamine 2-epimerase/N-acetylmannosamine kinase. *J. Biol. Chem.* 272: 24319–24324, 1997.

[34737] 8289. Carter, M. G.; Johns, M. A.; Zeng, X.; Zhou, L.; Zink, M. C.; Mankowski, J. L.; Donovan, D. M.; Baylin, S. B.: Mice deficient in the candidate tumor suppressor gene *Hic1* exhibit developmental defects of structures affected in the Miller-Dieker syndrome. *Hum. Molec. Genet.* 9: 413–419, 2000.

[34738] 8290. Chong, S. S.; Tanigami, A.; Roschke, A. V.; Ledbetter, D. H.: 14-3-3 epsilon has no homology to LIS1 and lies telomeric to it on chromosome 17p13.3 outside the Miller-

Dieker syndrome chromosome region. *GenomeRes.* 6: 735–741, 1996.

- [34739] 8291. Grimm, C.; Sporle, R.; Schmid, T. E.; Adler, I.-D.; Adamski, J.; Schughart, K.; Graw, J.: Isolation and embryonic expression of the novel mouse gene *Hic1*, the homologue of *HIC1*, a candidate gene for the Miller–Dieker syndrome. *Hum. Molec. Genet.* 8: 697–710, 1999.
- [34740] 8292. Makos Wales, M.; Biel, M. A.; El Deiry, W.; Nelkin, B. D.; Issa, J.-P.; Cavenee, W. K.; Kuerbitz, S. J.; Baylin, S. B.: p53 activates expression of *HIC-1*, a new candidate tumour suppressor gene on 17p13.3. *Nature Med.* 1: 570–577, 1995.
- [34741] 8293. Smith, D. J.; Salmi, M.; Bono, P.; Hellman, J.; Leu, T.; Jalkanen, S.: Cloning of vascular adhesion protein 1 reveals a novel multifunctional adhesion molecule. *J. Exp. Med.* 188: 17–27, 1998.
- [34742] 8294. Zhang, X.; McIntire, W. S.: Cloning and sequencing of a copper-containing, topa quinone-containing monoamine oxidase from human placenta. *Gene* 179: 279–286, 1996.
- [34743] 8295. Lomako, J.; Mazuruk, K.; Lomako, W. M.; Alonso, M. D.; Whelan, W. J.; Rodriguez, I. R.: The human intron-containing gene for glycogenin maps to chromosome 3,

band q24. Genomics 33: 519–522, 1996.

- [34744] 8296. Tanaka, T.; Tsujimura, T.; Takeda, K.; Sugihara, A.; Maekawa, A.; Terada, N.; Yoshida, N.; Akira, S.: Targeted disruption of ATF4 discloses its essential role in the formation of eye lens fibres. Genes Cells 3:801–810, 1998.
- [34745] 8297. Maas, S.; Gerber, A. P.; Rich, A.: Identification and characterization of a human tRNA-specific adenosine deaminase related to the ADAR family of pre-mRNA editing enzymes. Proc. Nat. Acad. Sci. 96: 8895–8900, 1999.
- [34746] 8298. Viskupic, E.; Cao, Y.; Zhang, W.; Cheng, C.; DePaoli-Roach, A.A.; Roach, P. J.: Rabbit skeletal muscle glycogenin. Molecular cloning and production of fully functional protein in Escherichia coli. J. Biol. Chem. 267: 25759–25763, 1992.
- [34747] 8299. Hosokawa, Y.; Matsumoto, A.; Oka, J.; Itakura, H.; Yamaguchi, K.: Isolation and characterization of a cDNA for rat liver cysteine dioxygenase. Biochem. Biophys. Res. Commun. 168: 473–478, 1990.
- [34748] 8300. Jeremiah, S.; McCann, K. P.; Williams, A. C.; Ramsden, D. B.; Pilz, A. J.; Fox, M. F.; Povey, S.: Chromosomal localisation of genes coding for human and mouse liver cytosolic cysteine dioxygenase. Ann. Hum. Genet. 60: 29–33, 1996.

- [34749] 8301.McCann, K. P.; Akbari, M. T.; Williams, A. C.; Ramsden, D. B.:Human cysteine dioxygenase type I: primary structure derived frombase sequencing of cDNA. *Biochim. Biophys. Acta* 1209: 107–110, 1994.
- [34750] 8302.Ramsden, D. B.; Kapadi, A.; Fitch, N. J. S.; Farmer, M. J.; Bennett,P.; Williams, A. C.: Human cysteine dioxygenase type I (CDO-I; EC1.13.11.20): 5–prime flanking region and intron–exon structure ofthe gene. *Molec. Path.* 50: 269–271, 1997.
- [34751] 8303.Fox, M. F.; Lahbib, F.; Pratt, W.; Attwood, J.; Gum, J.; Kim, Y.;Swallow, D. M.: Regional localization of the intestinal mucin geneMUC3 to chromosome 7q22. *Ann. Hum. Genet.* 56: 281–287, 1992.
- [34752] 8304.Kyo, K.; Muto, T.; Nagawa, H.; Lathrop, G. M.; Nakamura, Y.: Associationsof distinct variants of the intestinal mucin gene MUC3A with ulcerativecolitis and Crohn's disease. *J. Hum. Genet.* 46: 5–20, 2001.
- [34753] 8305.Hill, A. V. S.; Gentile, B.; Bonnardot, J. M.; Roux, J.; Weatherall,D. J.; Clegg, J. B.: Polynesian origins and affinities: globin genevariants in eastern Polynesia. *Am. J. Hum. Genet.* 40: 453–463, 1987.
- [34754] 8306.Zou, Y.–R.; Kottmann, A. H.; Kuroda, M.; Taniuchi, I.; Littman,D. R.: Function of the chemokine receptor CXCR4

in haematopoiesis and in cerebellar development. *Nature* 393: 595–599, 1998.

[34755] 8307. Laurent, P.; Clerc, P.; Mattei, M.-G.; Forgez, P.; Dumont, X.; Ferrara, P.; Caput, D.; Rostene, W.: Chromosomal localization of mouse and human neurotensin receptor genes. *Mammalian Genome* 5:303–306, 1994.

[34756] 8308. Le, F.; Groshan, K.; Zeng, X. P.; Richelson, E.: Characterization of the genomic structure, promoter region, and a tetranucleotide repeat polymorphism of the human neurotensin receptor gene. *J. Biol. Chem.* 272:1315–1322, 1997.

[34757] 8309. Vincent, J.-P.: Neurotensin receptors: binding properties, transduction pathways, and structure. *Cell. Molec. Neurobiol.* 15: 501–512, 1995.

[34758] 8310. Vita, N.; Laurent, P.; Lefort, S.; Chalon, P.; Dumont, X.; Kaghad, M.; Gully, D.; Le Fur, G.; Ferrara, P.; Caput, D.: Cloning and expression of a complementary DNA encoding a high affinity human neurotensin receptor. *FEBS Lett.* 317: 139–142, 1993.

[34759] 8311. Jones, K. R.; Reichardt, L. F.: Molecular cloning of a human gene that is a member of the nerve growth factor family. *Proc. Nat. Acad. Sci.* 87: 8060–8064, 1990.

[34760] 8312. Berkemeier, L. R.; Ozcelik, T.; Francke, U.; Rosen-

thal, A.: Human chromosome 19 contains the neurotrophin-5 gene locus and three related genes that may encode novel acidic neurotrophins. *Somat. Cell Molec. Genet.* 18: 233–245, 1992.

[34761] 8313. Ibanez, C. F.: Neurotrophin-4: the odd one out in the neurotrophin family. *Neurochem. Res.* 21: 787–793, 1996.

[34762] 8314. Ip, N. Y.; Ibanez, C. F.; Nye, S. H.; McClain, J.; Jones, P. F.; Gies, D. R.; Belluscio, L.; Le Beau, M. M.; Espinosa, R., III; Squinto, S. P.; Persson, H.; Yancopoulos, G. D.: Mammalian neurotrophin-4: structure, chromosomal localization, tissue distribution, and receptor specificity. *Proc. Nat. Acad. Sci.* 89: 3060–3064, 1992.

[34763] 8315. Hill, A. V. S.; Nicholls, R. D.; Thein, S. L.; Higgs, D. R.: Recombination within the human embryonic zeta-globin locus: a common zeta-zeta chromosome produced by gene conversion of the psi-zeta gene. *Cell* 42: 809–819, 1985.

[34764] 8316. Housman, D.: Personal Communication. Boston, Mass. 1979.

[34765] 8317. Kamuzora, H.; Lehmann, H.: Human embryonic haemoglobins including a comparison by homology of the human zeta and alpha chains. *Nature* 256: 511–513, 1975.

- [34766] 8318.Melderis, H.; Steinheider, G.; Ostertag, W.: Evidence for a unique kind of alpha-type globin chain in early mammalian embryos. *Nature* 250:774–776, 1974.
- [34767] 8319.Pressley, L.; Higgs, D. R.; Clegg, J. B.; Weatherall, D. J.: Gene deletions in alpha-thalassemia prove that the 5-prime zeta locus is functional. *Proc. Nat. Acad. Sci.* 77: 3586–3589, 1980.
- [34768] 8320.Proudfoot, N. J.; Shander, M. H. M.; Manley, J. L.; Gefer, M.L.; Maniatis, T.: Structure and in vitro transcription of human globin genes. *Science* 209: 1329–1336, 1980.
- [34769] 8321.Randhawa, Z. I.; Jones, R. T.; Lie-Injo, L. E.: Separation of the tryptic peptides and cyanogen bromide fragments of the human embryonic zeta chains of hemoglobin Portland I and II by reverse phase high performance liquid chromatography. *Hemoglobin* 8: 463–482, 1984.
- [34770] 8322.Whitney, J. B., III; Russell, E. S.: Linkage of genes for adult alpha-globin and embryonic alpha-like globin chains. *Proc. Nat. Acad. Sci.* 77: 1087–1090, 1980.
- [34771] 8323.Bluteau, O.; Jeannot, E.; Bioulac-Sage, P.; Marques, J. M.; Blanc, J.-F.; Bui, H.; Beaudoin, J.-C.; Franco, D.; Balabaud, C.; Laurent-Puig, P.; Zucman-Rossi, J.: Bi-allelic inactivation of TCF1 in hepatic adenomas. *Nature Genet.* 32:

312–315, 2002.

- [34772] 8324.Foster, J. H.; Donohue, T. A.; Berman, M. M.: Familial liver–celladenomas and diabetes mellitus. *New Eng. J. Med.* 299: 239–241, 1978.
- [34773] 8325.Quintrell, N.; Lebo, R.; Varmus, H.; Bishop, J. M.; Pettenati,M. J.; Le Beau, M. M.; Diaz, M. O.; Rowley, J. D.: Identificationof a human gene (HCK) that encodes a protein–tyrosine kinase and isexpressed in hemopoietic cells. *Molec. Cell. Biol.* 7: 2267–2275,1987.
- [34774] 8326.Ziegler, S. F.; Marth, J. D.; Lewis, D. B.; Perlmutter, R. M.:Novel protein–tyrosine kinase gene (hck) preferentially expressedin cells of hematopoietic origin. *Molec. Cell. Biol.* 7: 2276–2285,1987.
- [34775] 8327.Honda, H.; Inaba, T.; Suzuki, T.; Oda, H.; Ebihara, Y.; Tsuiji,K.; Nakahata, T.; Ishikawa, T.; Yazaki, Y.; Hirai, H.: Expressionof E2A–HLF chimeric protein induced T–cell apoptosis, B–cell maturationarrest, and development of acute lymphoblastic leukemia. *Blood* 93:2780–2790, 1999.
- [34776] 8328.Hunger, S. P.: Chromosomal translocations involving the E2A genein acute lymphoblastic leukemia: clinical features and molecular pathogenesis. *Blood* 87:1211–1224, 1996.

- [34777] 8329.Inaba, T.; Roberts, W. M.; Shapiro, L. H.; Jolly, K. W.; Raimondi, S. C.; Smith, S. D.; Look, A. T.: Fusion of the leucine zipper gene HLF to the E2A gene in human acute B-lineage leukemia. *Science* 257:531–534, 1992.
- [34778] 8330.Smith, K. S.; Rhee, J. W.; Naumovski, L.; Cleary, M. L.: Disrupted differentiation and oncogenic transformation of lymphoid progenitors in E2A–HLF transgenic mice. *Molec. Cell. Biol.* 19: 4443–4451, 1999.
- [34779] 8331.Acampora, D.; D'Esposito, M.; Faiella, A.; Pannese, M.; Migliaccio, E.; Morelli, F.; Stornaiuolo, A.; Nigro, V.; Simeone, A.; Boncinelli, E.: The human HOX gene family. *Nucleic Acids Res.* 17: 10385–10402, 1989.
- [34780] 8332.Balling, R.; Mutter, G.; Gruss, P.; Kessel, M.: Cranio-facial abnormalities induced by ectopic expression of the homeobox gene Hox–1.1 in transgenic mice. *Cell* 58: 337–347, 1989.
- [34781] 8333.Bucan, M.; Yang–Feng, T.; Colberg–Poley, A. M.; Wolgemuth, D. J.; Guenet, J.–L.; Francke, U.; Lehrach, H.: Genetic and cytogenetic localisation of the homeo box containing genes on mouse chromosome 6 and human chromosome 7. *EMBO J.* 5: 2899–2905, 1986.
- [34782] 8334.Ferguson–Smith, A. C.; Fienberg, A.; Ruddle, F. H.: Isolation, chromosomal localization, and nucleotide se–

quence of the human HOX1.4 homeobox. *Genomics* 5: 250–258, 1989.

[34783] 8335. Gaunt, S. J.; Singh, P. B.: Homeogene expression patterns and chromosomal imprinting. *Trends Genet.* 6: 208–212, 1990.

[34784] 8336. Gehring, W. J.: The homeo box: a key to the understanding of development? *Cell* 40:3–5, 1985.

[34785] 8337. Joyner, A. L.; Lebo, R. V.; Kan, Y. W.; Tjian, R.; Cox, D. R.; Martin, G. R.: Comparative chromosome mapping of a conserved homeobox region in mouse and human. *Nature* 314: 173–175, 1985.

[34786] 8338. McGinnis, W.; Garber, R. L.; Wirz, J.; Kuroiwa, A.; Gehring, W. J.: A homologous protein-coding sequence in *Drosophila* homeotic genes and its conservation in other metazoans. *Cell* 37: 403–408, 1984.

[34787] 8339. McGinnis, W.; Levine, M. S.; Hafen, E.; Kuroiwa, A.; Gehring, W. J.: A conserved DNA sequence in homeotic genes of the *Drosophila* Antennapedia and bithorax complexes. *Nature* 308: 428–433, 1984.

[34788] 8340. Ohno, S.: *Evolution by Gene Duplication*. Heidelberg: Springer(pub.) 1970.

[34789] 8341. Rabin, M.; Ferguson-Smith, A.; Hart, C. P.; Ruddle, F. H.: Cognate homeo-box loci mapped on homologous hu-

man and mouse chromosomes. Proc.Nat. Acad. Sci. 83: 9104–9108, 1986.

[34790] 8342.Rabin, M.; Hart, C. P.; Ferguson–Smith, A.; McGinnis, W.; Levine,M.; Ruddle, F. H.: Two homoeo box loci mapped in evolutionarily related mouse and human chromosomes. Nature 314: 175–178, 1985.

[34791] 8343.Rieger, R.; Michaelis, A.; Green, M. M.: Glossary of Genetics and Cytogenetics. New York: Springer–Verlag (pub.) 1976. Pp.281 only.

[34792] 8344.Schughart, K.; Kappen, C.; Ruddle, F. H.: Duplication of large genomic regions during the evolution of vertebrate homeobox genes. Proc.Nat. Acad. Sci. 86: 7067–7071, 1989.

[34793] 8345.Scott, M. P.: Vertebrate homeobox gene nomenclature. (Letter) Cell 71:551–553, 1992.

[34794] 8346.Greer, J. M.; Puetz, J.; Thomas, K. R.; Capecchi, M. R.: Maintenance of functional equivalence during paralogous Hox gene evolution. Nature 403:661–665, 2000.

[34795] 8347.Apiou, F.; Flagiello, D.; Cillo, C.; Malfoy, B.; Poupon, M.–F.; Dutrillaux, B.: Fine mapping of human HOX gene clusters. Cytogenet.Cell Genet. 73: 114–115, 1996.

[34796] 8348.Chan, H. Y. E.; Warrick, J. M.; Gray–Board, G. L.; Paulson, H.L.; Bonini, N. M.: Mechanisms of chaperone

suppression of polyglutaminedisease: selectivity, synergy and modulation of protein solubilityin *Drosophila*. *Hum. Molec. Genet.* 9: 2811–2820, 2000.

- [34797] 8349.Dunah, A. W.; Jeong, H.; Griffin, A.; Kim, Y.–M.; Standaert, D.G.; Hersch, S. M.; Mouradian, M. M.; Young, A. B.; Tanese, N.; Krainc,D.: Sp1 and TAFII130 transcriptional activity disrupted in earlyHuntington's disease. *Science* 296: 2238–2243, 2002.
- [34798] 8350.Man in't Veld, A. J.; Boomsma, F.; Moleman, P.; Schalekamp, M.A. D. H.: Congenital dopamine–beta–hydroxylase deficiency: a novelorthostatic syndrome. *Lancet* I: 183–188, 1987.
- [34799] 8351.Robertson, D.; Goldberg, M. R.; Onrot, J.; Hollister, A. S.; Wiley,R.; Thompson, J. G., Jr.; Robertson, R. M.: Isolated failure of autonomicnoradrenergic neurotransmission: evidence for impaired beta–hydroxylationof dopamine. *New Eng. J. Med.* 314: 1494–1497, 1986.
- [34800] 8352.Ogawa, O.; McNoe, L. A.; Eccles, M. R.; Morison, I. M.; Reeve,A. E.: Human insulin–like growth factor type I and type II receptorsare not imprinted. *Hum. Molec. Genet.* 2: 2163–2165, 1993.
- [34801] 8353.Oshima, A.; Nolan, C. M.; Kyle, J. W.; Grubb, J. H.; Sly, W. S.: The human cation–independent mannose

6-phosphate receptor: cloning and sequence of the full-length cDNA and expression of functional receptor in COS cells. *J. Biol. Chem.* 263: 2553–2562, 1988.

[34802] 8354. Puertollano, R.; Aguilar, R. C.; Gorshkova, I.; Crouch, R. J.; Bonifacino, J. S.: Sorting of mannose 6-phosphate receptors mediated by the GGAs. *Science* 292: 1712–1716, 2001.

[34803] 8355. Rao, P. H.; Murty, V. V. V. S.; Gaidano, G.; Hauptschein, R.; Dalla-Favera, R.; Chaganti, R. S. K.: Sub-regional mapping of 8 single copy loci to chromosome 6 by fluorescence in situ hybridization. *Cytogenet. Cell Genet.* 66: 272–273, 1994.

[34804] 8356. Roth, R. A.: Structure of the receptor for insulin-like growth factor II: the puzzle amplified. *Science* 239: 1269–1271, 1988.

[34805] 8357. Sleutels, F.; Zwart, R.; Barlow, D. P.: The non-coding Air RNAs required for silencing autosomal imprinted genes. *Nature* 415: 810–813, 2002.

[34806] 8358. Souza, R. F.; Appel, R.; Yin, J.; Wang, S.; Smolinski, K. N.; Abraham, J. M.; Zou, T.-T.; Shi, Y.-Q.; Lei, J.; Cottrell, J.; Cymes, K.; Biden, K.; Simms, L.; Leggett, B.; Lynch, P. M.; Frazier, M.; Powell, S. M.; Harpaz, N.; Sugimura, H.; Young, J.; Meltzer, S. J.: Microsatellite instability in the insulin-like

growth factor II receptor gene in gastrointestinal tumours.
(Letter) Nature Genet. 14:255–257, 1996.

- [34807] 8359.Szebeny, G.; Rotwein, P.: The mouse insulin-like growth factorII/cation-independent mannose 6-phosphate (IGF-II/MPR) receptor gene:molecular cloning and genomic organization. Genomics 19: 120–129,1994.
- [34808] 8360.Tong, P. Y.; Tollefsen, S. E.; Kornfeld, S.: The cation-independentmannose 6-phosphate receptor binds insulin-like growth factor II. J.Biol. Chem. 263: 2585–2588, 1988.
- [34809] 8361.Waheed, A.; Braulke, T.; Junghans, U.; von Figura, K.: Mannose6-phosphate/insulin like growth factor II receptor: the two typesof ligands bind simultaneously to one receptor at different sites. Biochem.Biophys. Res. Commun. 152: 1248–1254, 1988.
- [34810] 8362.Xu, Y.; Goodyer, C. G.; Deal, C.; Polychronakos, C.: Functionalpolymorphism in the parental imprinting of the human IGF2R gene. Biochem.Biophys. Res. Commun. 197: 747–754, 1993.
- [34811] 8363.Young, L. E.; Fernandes, K.; McEvoy, T. G.; Butterwith, S. C.;Gutierrez, C. G.; Carolan, C.; Broadbent, P. J.; Robinson, J. J.;Wilmut, I.; Sinclair, K. D.: Epigenetic change

in IGF2R is associated with fetal overgrowth after sheep embryo culture. *Nature Genet.* 27:153–154, 2001.

[34812] 8364. Zhu, Y.; Doray, B.; Poussu, A.; Lehto, V.-P.; Kornfeld, S.: Binding of GGA2 to the lysosomal enzyme sorting motif of the mannose 6-phosphate receptor. *Science* 292: 1716–1718, 2001.

[34813] 8365. Brown, C. W.; Houston-Hawkins, D. E.; Woodruff, T. K.; Matzuk, M. M.: Insertion of *Inhbb* into the *Inhba* locus rescues the *Inhba*-null phenotype and reveals new activin functions. *Nature Genet.* 25: 453–457, 2000.

[34814] 8366. Ferguson, C. A.; Tucker, A. S.; Christensen, L.; Lau, A. L.; Matzuk, M. M.; Sharpe, P. T.: Activin is an essential early mesenchymal signal in tooth development that is required for patterning of the murine dentition. *Genes Dev.* 12: 2636–2649, 1998.

[34815] 8367. Burger, H. G.; Igarashi, M.; Baird, D.; Mason, T.; Bardin, W.; McLachlan, R.; Chappel, S.; Miyamoto, K.; de Jong, F.; Moudgal, A.; Demoulin, A.; Nieschlag, E.; de Kretser, D.; Robertson, D.; Findlay, J.; Sasamoto, S.; Forage, R.; Schwartz, N.; Fukuda, M.; Steinberger, A.; Hasegawa, Y.; Tanabe, K.; Ling, N.; Ying, S.-Y.: Inhibin: definition and nomenclature, including related substances. (Letter) *J. Clin. Endocr. Metab.* 66: 885–886, 1988.

- [34816] 8368.Lumpkin, M. D.; Moltz, J. H.; Yu, W. H.; Samson, W. K.; McCann,S. M.: Purification of FSH-releasing factor: its dissimilarity fromLHRH of mammalian, avian, and piscian origin. Brain Res. Bull. 18:175–178, 1987.
- [34817] 8369.Matzuk, M. M.; Kumar, T. R.; Vassalli, A.; Bickenbach, J. R.; Roop,D. R.; Jaenisch, R.; Bradley, A.: Functional analysis of activinsduring mammalian development. Nature 374: 354–356, 1995.
- [34818] 8370.Mellor, S. L.; Cranfield, M.; Ries, R.; Pedersen, J.; Cancilla,B.; de Kretser, D.; Groome, N. P.; Mason, A. J.; Risbridger, G. P.: Localization of activin beta(A)–, beta(B)–, and beta(C)–subunitsin human prostate and evidence for formation of new activin heterodimersof beta(C)–subunit. J. Clin. Endocr. Metab. 85: 4851–4858, 2000.
- [34819] 8371.Murata, M.; Eto, Y.; Shibai, H.; Sakai, M.; Muramatsu, M.: Erythroiddifferentiation factor is encoded by the same mRNA as that of theinhibin beta–A chain. Proc. Nat. Acad. Sci. 85: 2434–2438, 1988.
- [34820] 8372.You, L.; Kruse, F. E.: Differential effect of activin A and BMP–7on myofibroblast differentiation and the role of the Smad signalingpathway. Invest. Ophthal. Vis. Sci. 43: 72–81, 2002.
- [34821] 8373.Lacy, D. A.; Wang, Z.–E.; Symula, D. J.; McArthur, C.

J.; Rubin, E. M.; Frazer, K. A.; Locksley, R. M.: Faithful expression of the human 5q31 cytokine cluster in transgenic mice. *J. Immun.* 164: 4569–4574, 2000.

[34822] 8374. Taniyama, Y.; Kuroki, R.; Omura, F.; Seko, C.; Kikuchi, M.: Evidence for intramolecular disulfide bond shuffling in the folding of mutant human lysozyme. *J. Biol. Chem.* 266: 6456–6461, 1991.

[34823] 8375. Loots, G. G.; Locksley, R. M.; Blankespoor, C. M.; Wang, Z. E.; Miller, W.; Rubin, E. M.; Frazer, K. A.: Identification of a coordinate regulator of interleukins 4, 13, and 5 by cross-species sequence comparisons. *Science* 288:136–140, 2000.

[34824] 8376. McKenzie, A. N. J.; Culpepper, J. A.; de Waal Malefyt, R.; Briere, F.; Punnonen, J.; Aversa, G.; Sato, A.; Dang, W.; Cocks, B. G.; Menon, S.; de Vries, J. E.; Banchereau, J.; Zurawski, G.: Interleukin 13, a T-cell-derived cytokine that regulates human monocyte and B-cell function. *Proc. Nat. Acad. Sci.* 90: 3735–3739, 1993.

[34825] 8377. Minty, A.; Chalon, P.; Derocq, J.-M.; Dumont, X.; Guillemot, J.-C.; Kaghad, M.; Labit, C.; Leplatois, P.; Liauzun, P.; Miloux, B.; Minty, C.; Casellas, P.; Loison, G.; Lupker, J.; Shire, D.; Ferrara, P.; Caput, D.: Interleukin-13 is a new human lymphokine regulating inflammatory and

immune responses. *Nature* 362: 248–250, 1993.

[34826] 8378.Morgan, J. G.; Dolganov, G. M.; Robbins, S. E.; Hinton, L. M.; Lovett, M.: The selective isolation of novel cDNAs encoded by the regions surrounding the human interleukin 4 and 5 genes. *Nucleic Acids Res.* 20: 5173–5179, 1992.

[34827] 8379.Paul, W. E.: Personal Communication. Bethesda, Md. 3/6/1993.

[34828] 8380.Punnonen, J.; Aversa, G.; Cocks, B. G.; McKenzie, A. N. J.; Menon, S.; Zurawski, G.; de Waal Malefyt, R.; de Vries, J. E.: Interleukin 13 induces interleukin 4-independent IgG4 and IgE synthesis and CD23 expression by human B cells. *Proc. Nat. Acad. Sci.* 90: 3730–3734, 1993.

[34829] 8381.Smirnov, D. V.; Smirnova, M. G.; Korobko, V. G.; Frolova, E. I.: Tandem arrangement of human genes for interleukin-4 and interleukin-13: resemblance in their organization. *Gene* 155: 277–281, 1995.

[34830] 8382.Wills-Karp, M.; Luyimbazi, J.; Xu, X.; Schofield, B.; Neben, T.Y.; Karp, C. L.; Donaldson, D. D.: Interleukin-13: central mediator of allergic asthma. *Science* 282: 2258–2261, 1998.

[34831] 8383.Zhu, Z.; Homer, R. J.; Wang, Z.; Chen, Q.; Geba, G. P.; Wang, J.; Zhang, Y.; Elias, J. A.: Pulmonary expression of

interleukin-13 causes inflammation, mucus hypersecretion, subepithelial fibrosis, physiologic abnormalities, and eotaxin production. *J. Clin. Invest.* 103:779–788, 1999.

[34832] 8384. Zurawski, G.; de Vries, J. E.: Interleukin 13 elicits a subset of the activities of its close relative interleukin 4. *Stem Cells* 12:169–174, 1994.

[34833] 8385. Li, C.; Lai, C.; Sigman, D. S.; Gaynor, R. B.: Cloning of a cellular factor, interleukin binding factor, that binds to NFAT-like motifs in the human immunodeficiency virus long terminal repeat. *Proc. Nat. Acad. Sci.* 88: 7739–7743, 1991.

[34834] 8386. Li, C.; Lysis, A. J.; Sparkes, R.; Nirula, A.; Gaynor, R.: Characterization and chromosomal mapping of the gene encoding the cellular DNA binding protein ILF. *Genomics* 13: 665–671, 1992.

[34835] 8387. Boone, C.; Chen, T.-R.; Ruddle, F. H.: Assignment of three human genes to chromosomes (LDH-A to 11, TK to 17 and IDH to 20) and evidence for translocation between human and mouse chromosomes in somatic cell hybrids. *Proc. Nat. Acad. Sci.* 68: 510–514, 1972.

[34836] 8388. Chen, S.-H.; Fossum, B. L. G.; Giblett, E. R.: Genetic variation of the soluble form of NADP-dependent isocitric dehydrogenase in man. *Am. J. Hum. Genet.* 24: 325–329,

1972.

- [34837] 8389.Creagan, R. P.; Carritt, B.; Chen, S.-H.; Kucherlapati, R. S.;McMorris, F. A.; Ricciuti, F.; Tan, Y. H.; Tischfield, J. A.; Ruddle,F. H.: Chromosome assignments of genes in man using mouse-human somaticcell hybrids: cytoplasmic isocitrate dehydrogenase (IDH 1) and malatedehydrogenase (MDH 1) to chromosome 2. *Am. J. Hum. Genet.* 26: 604-613,1974.
- [34838] 8390.Glass, I. A.; Swindlehurst, C. A.; Aitken, D. A.; McCrea, W.; Boyd,E.: Interstitial deletion of the long arm of chromosome 2 with normallevels of isocitrate dehydrogenase. *J. Med. Genet.* 26: 127-130,1989.
- [34839] 8391.Henderson, N. S.: Isozymes of isocitrate dehydrogenase: subunitstructure and intracellular location. *J. Exp. Zool.* 158: 263-273,1965.
- [34840] 8392.Henderson, N. S.: Intracellular location and genetic control ofisozymes of NADP-dependent isocitrate dehydrogenase and malate dehydrogenase. *Ann.N.Y. Acad. Sci.* 151: 429-440, 1968.
- [34841] 8393.Narahara, K.; Kimura, S.; Kikkawa, K.; Takahashi, Y.; Wakita, Y.;Kasai, R.; Nagai, S.; Nishibayashi, Y.; Kimoto, H.: Probable assignmentof soluble isocitrate dehydrogenase (IDH-1) to 2q33.3. *Hum. Genet.* 71:37-40, 1985.

- [34842] 8394.Ruddle, F. H.: Linkage analysis in man by somatic cell genetics. *Nature* 242:165–169, 1973.
- [34843] 8395.Shows, T. B.: . (Abstract) 4th Int. Cong. Hum. Genet., Paris 165,1971.
- [34844] 8396.Shows, T. B.: Genetics of human–mouse somatic cell hybrids: linkageof human genes for isocitrate dehydrogenase and malate dehydrogenase. *Biochem.Genet.* 7: 193–204, 1972.
- [34845] 8397.Turner, B. M.; Fisher, R. A.; Garthwaite, E.; Whale, R. J.; Harris,H.: An account of two new ICD–S variants not detectable in red bloodcells. *Ann. Hum. Genet.* 37: 469–476, 1974.
- [34846] 8398.Van Cong, N.: Personal Communication. Paris, France 1976.
- [34847] 8399.Weil, D.; Van Cong, N.; Finaz, C.; Rebourcet, R.; Cochet, C.;de Grouchy, J.; Frezal, J.: Localisation regionale des genes humainsIDH–S, MDH–S, PGK, alpha–GAL, G6PD par l'hybridation cellulaire interspecifique. *Hum.Genet.* 36: 205–211, 1977.
- [34848] 8400.Auron, P. E.; Webb, A. C.; Rosenwasser, L. J.; Mucci, S. F.; Rich,A.; Wolff, S. M.; Dinarello, C. A.: Nucleotide sequence of humanmonocyte interleukin 1 precursor cDNA. *Proc. Nat. Acad. Sci.* 81:7907–7911, 1984.

- [34849] 8401. Winter, H.; Labreze, C.; Chapalain, V.; Surleve-Bazeille, J. E.; Mercier, M.; Rogers, M. A.; Taieb, A.; Schweizer, J.: A variable monilethrix phenotype associated with a novel mutation, glu402lys, in the helix termination motif of the type II hair keratin hHb1. *J. Invest. Derm.* 111: 169–172, 1998.
- [34850] 8402. Francke, U.; Foellmer, B. E.; Haynes, B. F.: Chromosome mapping of human cell surface molecules: monoclonal anti-human lymphocyte antibodies 4F2, A3D8, and A1G3 define antigens controlled by different regions of chromosome 11. *Somat. Cell Genet.* 9: 333–344, 1983.
- [34851] 8403. Gottesdiener, K. M.; Karpinski, B. A.; Lindsten, T.; Strominger, J. L.; Jones, N. H.; Thompson, C. B.; Leiden, J. M.: Isolation and structural characterization of the human 4F2 heavy-chain gene, an inducible gene involved in T-lymphocyte activation. *Molec. Cell. Biol.* 8: 3809–3819, 1988.
- [34852] 8404. Haynes, B. F.; Hemler, M. E.; Mann, D. L.; Eisenbarth, G. S.; Shelhamer, J.; Mostowski, H. S.; Thomas, C. A.; Strominger, J. L.; Fauci, A. S.: Characterization of a monoclonal antibody (4F2) that binds to human monocytes and to a subset of activated lymphocytes. *J. Immun.* 126:1409–1414, 1981.

- [34853] 8405.Hemler, M. E.; Strominger, J. L.: Characterization of antigenrecognized by the monoclonal antibody (4F2): different molecular formson human T and B lymphoblastoid cell lines. *J. Immun.* 129: 623–628,1982.
- [34854] 8406.Lindsten, T.; June, C. H.; Thompson, C. B.; Leiden, J. M.: Regulationof 4F2 heavy-chain gene expression during normal human T-cell activationcan be mediated by multiple distinct molecular mechanisms. *Molec.Cell. Biol.* 8: 3820–3826, 1988.
- [34855] 8407.Lumadue, J. A.; Glick, A. B.; Ruddle, F. H.: Cloning, sequenceanalysis, and expression of the large subunit of the human lymphocyteactivation antigen 4F2. *Proc. Nat. Acad. Sci.* 84: 9204–9208, 1987.
- [34856] 8408.Mastroberardino, L.; Spindler, B.; Pfeiffer, R.; Skelly, P. J.;Loffing, J.; Shoemaker, C. B.; Verrey, F.: Amino-acid transport byheterodimers of 4F2hc/CD98 and members of a permease family. *Nature* 395:288–291, 1998.
- [34857] 8409.Copeland, N. G.; Hutchison, K. W.; Jenkins, N. A.: Excision ofthe DBA ecotropic provirus in dilute coat-color revertants of miceoccurs by homologous recombination involving the viral LTRs. *Cell* 33:379–387, 1983.
- [34858] 8410.El-Husseini, A. E.; Vincent, S. R.: Cloning and characterizationof a novel RING finger protein that interacts

with class V myosins. J.Biol. Chem. 274: 19771–19777, 1999.

[34859] 8411.Engle, L. J.; Kennett, R. H.: Cloning, analysis, and chromosomal localization of myoxin (MYH12): the human homologue to the mouse dilute gene. Genomics 19: 407–416, 1994.

[34860] 8412.Espresifco, E. M.; Coling, D. E.; Tsakraklides, V.; Krogh, K.; Wolenski, J. S.; Kalinec, G.; Kachar, B.: Localization of myosin-V in the centrosome. Proc. Nat. Acad. Sci. 95: 8636–8641, 1998.

[34861] 8413.Jenkins, N. A.; Copeland, N. G.; Taylor, B. A.; Lee, B. K.: Dilute(d) coat colour mutation of DNA/2J mice is associated with the site of integration of an ecotropic MuLV genome. Nature 293: 370–374, 1981.

[34862] 8414.Jones, J. M.; Huang, J.-D.; Mermall, V.; Hamilton, B. A.; Mooseker, M. S.; Escayg, A.; Copeland, N. G.; Jenkins, N. A.; Meisler, M. H.: The mouse neurological mutant flailar expresses a novel hybrid gene derived by exon shuffling between Gnb5 and Myo5a. Hum. Molec. Genet. 9:821–828, 2000.

[34863] 8415.Karcher, R. L.; Roland, J. T.; Zappacosta, F.; Huddleston, M. J.; Annan, R. S.; Carr, S. A.; Gelfand, V. I.: Cell cycle regulation of myosin-V by calcium/calmod-

ulin-dependent protein kinase II. *Science* 293:1317–1320, 2001.

[34864] 8416. Dooley, T. P.; Huang, Z.: Genomic organization and DNA sequences of two human phenol sulfotransferase genes (STP1 and STP2) on the short arm of chromosome 16. *Biochem. Biophys. Res. Commun.* 228:134–140, 1996.

[34865] 8417. Her, C.; Raftogianis, R.; Weinshilboum, R. M.: Human phenol sulfotransferase STP2 gene: molecular cloning, structural characterization, and chromosomal localization. *Genomics* 33: 409–420, 1996.

[34866] 8418. Jones, A. L.; Hagen, M.; Coughtrie, M. W. H.; Roberts, R. C.; Glatt, H.: Human platelet phenol sulfotransferases: cDNA cloning, stable expression in V79 cells and identification of a novel allelic variant of the phenol-sulfating form. *Biochem. Biophys. Res. Commun.* 208:855–862, 1995.

[34867] 8419. Barbosa, M. D. F. S.; Nguyen, Q. A.; Tchernev, V. T.; Ashley, J. A.; Detter, J. C.; Blaydes, S. M.; Brandt, S. J.; Chotai, D.; Hodgman, C.; Solari, R. C. E.; Lovett, M.; Kingsmore, S. F.: Identification of the homologous beige and Chediak-Higashi syndrome genes. *Nature* 382:262–265, 1996.

[34868] 8420. Fukai, K.; Oh, J.; Karim, M. A.; Moore, K. J.; Kandil, H.

H.; Ito, H.; Burger, J.; Spritz, R. A.: Homozygosity mapping of the gene for Chediak–Higashi syndrome to chromosome 1q42–q44 in a segment of conserved synteny that includes the mouse beige locus (bg). *Am. J. Hum. Genet.* 59:620–624, 1996.

[34869] 8421. Jackson, I. J.: Homologous pigmentation mutations in human, mouse and other model organisms. *Hum. Molec. Genet.* 6: 1613–1624, 1997.

[34870] 8422. Karim, M. A.; Nagle, D. L.; Kandil, H. H.; Burger, J.; Moore, K. J.; Spritz, R. A.: Mutations in the Chediak–Higashi syndrome gene (CHS1) indicate requirement for the complete 3801 amino acid CHS protein. *Hum. Molec. Genet.* 6: 1087–1089, 1997.

[34871] 8423. Nagle, D. L.; Karim, M. A.; Woolf, E. A.; Holmgren, L.; Bork, P.; Misumi, D. J.; McGrail, S. H.; Dussault, B. J., Jr.; Perou, C. M.; Boissy, R. E.; Duyk, G. M.; Spritz, R. A.; Moore, K. J.: Identification and mutation analysis of the complete gene for Chediak–Higashi syndrome. *Nature–Genet.* 14: 307–311, 1996.

[34872] 8424. Perou, C. M.; Moore, K. J.; Nagle, D. L.; Misumi, D. J.; Woolf, E. A.; McGrail, S. H.; Holmgren, L.; Brody, T. H.; Dussault, B. J., Jr.; Monroe, C. A.; Duyk, G. M.; Pryor, R. J.; Li, L.; Justice, M. J.; Kaplan, J.: Identification of the murine

beige gene by YAC complementation and positional cloning. *Nature Genet.* 13: 303–308, 1996.

- [34873] 8425. Baysal, B. E.; Potkin, S. G.; Farr, J. E.; Higgins, M. J.; Korcz, J.; Gollin, S. M.; James, M. R.; Evans, G. A.; Richard, C. W., III: Bipolar affective disorder partially cosegregates with a balanced $t(9;11)(p24;q23.1)$ chromosomal translocation in a small pedigree. *Am. J. Med. Genet.* 81: 81–91, 1998.
- [34874] 8426. Baysal, B. E.; Willett-Brozick, J. E.; Badner, J. A.; Corona, W.; Ferrell, R. E.; Nimgaonkar, V. L.; Detera-Wadleigh, S. D.: A mannosyltransferase gene at 11q23 is disrupted by a translocation breakpoint that cosegregates with bipolar affective disorder in a small family. *Neurogenetics* 4:43–53, 2002.
- [34875] 8427. Borg, J.-P.; Marchetto, S.; Le Bivic, A.; Ollendorff, V.; Jaulin-Bastard, F.; Saito, H.; Fournier, E.; Adelaide, J.; Margolis, B.; Birnbaum, D.: ERBIN: a basolateral PDZ protein that interacts with the mammalian ERBB2/HER2 receptor. *Nature Cell Biol.* 2: 407–414, 2000.
- [34876] 8428. Dustin, M. L.; Olszowy, M. W.; Holdorf, A. D.; Li, J.; Bromley, S.; Desai, N.; Widder, P.; Rosenberger, F.; van der Merwe, P. A.; Allen, P. M.; Shaw, A. S.: A novel adaptor protein orchestrates receptor patterning and cytoskeletal po-

larity in T-cell contacts. *Cell* 94:667–677, 1998.

[34877] 8429.Kirsch, K. H.; Georgescu, M.–M.; Ishimaru, S.; Hanafusa, H.: CMS:an adapter molecule involved in cytoskeletal rearrangements. *Proc.Nat. Acad. Sci.* 96: 6211–6216, 1999.

[34878] 8430.Shih, N.–Y.; Li, J.; Karpitskii, V.; Nguyen, A.; Dustin, M. L.;Kanagawa, O.; Miner, J. H.; Shaw, A. S.: Congenital nephrotic syndromein mice lacking CD2-associated protein. *Science* 286: 312–315, 1999.

[34879] 8431.Loughney, K.; Hill, T. R.; Florio, V. A.; Uher, L.; Rosman, G.J.; Wolda, S. L.; Jones, B. A.; Howard, M. L.; McAllister–Lucas, L.M.; Sonnenburg, W. K.; Francis, S. H.; Corbin, J. D.; Beavo, J. A.;Ferguson, K.: Isolation and characterization of cDNAs encoding PDE5A,a human cGMP-binding, cGMP-specific 3–prime,5–prime–cyclic nucleotidephosphodiesterase. *Gene* 216: 139–147, 1998.

[34880] 8432.Kim, J. M.; Sato, N.; Yamada, M.; Arai, K.; Masai, H.: Growthregulation of the expression of mouse cDNA and gene encoding a serine/threoninekinase related to *Saccharomyces cerevisiae* CDC7 essential for G(1)/Stransition: structure, chromosomal localization, and expression of–mouse gene for *S. cerevisiae* CDC7–related kinase. *J. Biol. Chem.* 273:23248–23257, 1998.

- [34881] 8433.Wei, M.-H.; Latif, F.; Bader, S.; Kashuba, V.; Chen, J.-Y.; Duh,F.-M.; Sekido, Y.; Lee, C.-C.; Geil, L.; Kuzmin, I.; Zabarovsky, E.;Klein, G.; Zbar, B.; Minna, J. D.; Lerman, M. I.: Construction of a 600-kilobase cosmid clone contig and generation of a transcriptional map surrounding the lung cancer tumor suppressor gene (TSG) locus on human chromosome 3p21.3: progress toward the isolation of a lung-cancer TSG. *Cancer Res.* 56: 1487-1492, 1996.
- [34882] 8434.Dolphin, C. T.; Beckett, D. J.; Janmohamed, A.; Cullingford, T.E.; Smith, R. L.; Shephard, E. A.; Phillips, I. R.: The flavin-containing monooxygenase 2 gene (FMO2) of humans, but not of other primates, encodes a truncated, nonfunctional protein. *J. Biol. Chem.* 273:30599-30607, 1998.
- [34883] 8435.Williams, D. E.; Kelly, J.; Dutchuk, M.: Flavin-containing monooxygenase in pulmonary microsomes of primates. In: Ingelman-Sundberg, M.; Gustafsson, J.-A.; Orrenius, S. (eds.): *Drug Metabolizing Enzymes: Genetics, Regulation and Toxicology: Proceedings of the 8th International Symposium on Microsomes and Drug Oxidations*. Stockholm: Karolinska Institutet 1990. P. 173.
- [34884] 8436.Gelb, B. D.; Zhang, J.; Cotter, P. D.; Gershin, I. F.; Desnick, R. J.: Physical mapping of the human connexin 40

(GJA5), flavin-containing monooxygenase 5, and natriuretic peptide receptor A genes on 1q21. *Genomics* 39:409–411, 1997.

[34885] 8437. Overby, L. H.; Buckpitt, A. R.; Lawton, M. P.; Atta-Asafo-Adjei, E.; Schulze, J.; Philpot, R. M.: Characterization of flavin-containing monooxygenase 5 (FMO5) cloned from human and guinea pig: evidence that the unique catalytic properties of FMO5 are not confined to the rabbit ortholog. *Arch. Biochem. Biophys.* 317: 275–284, 1995.

[34886] 8438. Poindexter, K.; Nelson, N.; DuBose, R. F.; Black, R. A.; Cerretti, D. P.: The identification of seven metalloproteinase-disintegrin (ADAM) genes from genomic libraries. *Gene* 237: 61–70, 1999.

[34887] 8439. Li-Hawkins, J.; Lund, E. G.; Turley, S. D.; Russell, D. W.: Disruption of the oxysterol 7- α -hydroxylase gene in mice. *J. Biol. Chem.* 275:16536–16542, 2000.

[34888] 8440. Setchell, K. D. R.; Schwarz, M.; O'Connell, N. C.; Lund, E. G.; Davis, D. L.; Lathe, R.; Thompson, H. R.; Tyson, R. W.; Sokol, R. J.; Russell, D. W.: Identification of a new inborn error in bile acid synthesis: mutation of the oxysterol 7- α -hydroxylase gene causes severe neonatal liver disease. *J. Clin. Invest.* 102: 1690–1703, 1998.

[34889] 8441. Granadino, B.; Gallardo, M. E.; Lopez-Rios, J.; Sanz,

R.; Ramos,C.; Ayuso, C.; Bovolenta, P.; Rodriguez de Cordoba, S.: Genomic cloning,structure, expression pattern, and chromosomal location of the humanSIX3 gene. Genomics 55: 100–105, 1999.

[34890] 8442.Loosli, F.; Winkler, S.; Wittbrodt, J.: Six3 overexpression initiatesthe formation of ectopic retina. Genes Dev. 13: 649–654, 1999.

[34891] 8443.Oliver, G.; Mailhos, A.; Wehr, R.; Copeland, N. G.; Jenkins, N.A.; Gruss, P.: Six3, a murine homologue of the sine oculis gene,demarcates the most anterior border of the developing neural plateand is expressed during eye development. Development 121: 4045–4055,1995.

[34892] 8444.Pasquier, L.; Dubourg, C.; Blayau, M.; Lazaro, L.; Le Marec, B.;David, V.; Odent, S.: A new mutation in the six-domain of SIX3 genecauses holoprosencephaly. Europ. J. Hum. Genet. 8: 797–800, 2000.

[34893] 8445.Roessler, E.; Muenke, M.: Holoprosencephaly: a paradigm for thecomplex genetics of brain development. J. Inherit. Metab. Dis. 21:481–497, 1998.

[34894] 8446.Gunther, T.; Chen, Z.-F.; Kim, J.; Priemel, M.; Rueger, J. M.;Amling, M.; Moseley, J. M.; Martin, T. J.; Anderson, D. J.; Karsenty,G.: Genetic ablation of parathyroid glands reveals another sourceof parathyroid hormone.

Nature 406: 199–203, 2000.

- [34895] 8447.Kanemura, Y.; Hiraga, S.; Arita, N.; Ohnishi, T.; Izumoto, S.;Mori, K.; Matsumura, H.; Yamasaki, M.; Fushiki, S.; Yoshimine, T.: Isolation and expression analysis of a novel human homologue ofthe *Drosophila* glial cells missing (gcm) gene. FEBS Lett. 442: 151–156,1999.
- [34896] 8448.Schell, U.; Wienberg, J.; Kohler, A.; Bray–Ward, P.; Ward, D. E.;Wilson, W. G.; Allen, W. P.; Lebel, R. R.; Sawyer, J. R.; Campbell,P. L.; Aughton, D. J.; Punnett, H. H.; Lammer, E. J.; Kao, F.–T.;Ward, D. C.; Muenke, M.: Molecular characterization of breakpointsin patients with holoprosencephaly and definition of the HPE2 criticalregion 2p21. Hum. Molec. Genet. 5: 223–229, 1996.
- [34897] 8449.Bouillet, P.; Metcalf, D.; Huang, D. C. S.; Tarlinton, D. M.; Kay,T. W. H.; Kontgen, F.; Adams, J. M.; Strasser, A.: Proapoptotic Bcl–2relative Bim required for certain apoptotic responses, leukocyte homeostasis,and to preclude autoimmunity. Science 286: 1735–1738, 1999.
- [34898] 8450.Bouillet, P.; Purton, J. F.; Godfrey, D. I.; Zhang, L.–C.; Coultas,L.; Puthalakath, H.; Pellegrini, M.; Cory, S.; Adams, J. M.; Strasser,A.: BH3–only Bcl–2 family member Bim is required for apoptosis ofautoreactive thymocytes. Nature 415: 922–926, 2002.

- [34899] 8451. Bouillet, P.; Zhang, L. C.; Huang, D. C. S.; Webb, G. C.; Bottema, C. D. K.; Shore, P.; Eyre, H. J.; Sutherland, G. R.; Adams, J. M.: Gene structure, alternative splicing, and chromosomal localization of pro-apoptotic Bcl-2 relative Bim. *Mammalian Genome* 12: 163–168, 2001.
- [34900] 8452. Murray, S.; Halford, S.; Ebenezer, N. D.; Gregory-Evans, C. Y.; Bhattacharya, S. S.: Assignment of BCL2L11 to human chromosome band 2p13 with somatic cell and radiation hybrids. *Cytogenet. Cell Genet.* 92:353, 2001.
- [34901] 8453. O'Connor, L.; Strasser, A.; O'Reilly, L. A.; Hausmann, G.; Adams, J. M.; Cory, S.; Huang, D. C.: Bim: a novel member of the Bcl-2 family that promotes apoptosis. *EMBO J.* 17: 384–395, 1998.
- [34902] 8454. Puthalakath, H.; Huang, D. C. S.; O'Reilly, L. A.; King, S. M.; Strasser, A.: The proapoptotic activity of the Bcl-2 family member Bim is regulated by interaction with the dynein motor complex. *Molec. Cell* 3: 287–296, 1999.
- [34903] 8455. Custer, M.; Spindler, B.; Verrey, F.; Murer, H.; Biber, J.: Identification of a new gene product (diphospho-1) regulated by dietary phosphate. *Am. J. Physiol.* 273: F801–F806, 1997.
- [34904] 8456. Kocher, O.; Comella, N.; Gilchrist, A.; Pal, R.; Tognazzi, K.; Brown, L. F.; Knoll, J. H. M.: PDZK1, a novel PDZ

domain-containing protein up-regulated in carcinomas and mapped to chromosome 1q21, interacts with cMOAT (MRP2), the multidrug resistance-associated protein.

Lab. Invest. 79: 1161–1170, 1999.

[34905] 8457. Kocher, O.; Comella, N.; Tognazzi, K.; Brown, L. F.: Identification and partial characterization of PDZK1: a novel protein containing PDZ interaction domains. Lab. Invest. 78: 117–125, 1998.

[34906] 8458. Wang, S.; Yue, H.; Derin, R. B.; Guggino, W. B.; Li, M.: Accessory protein facilitated CFTR–CFTR interaction, a molecular mechanism to potentiate the chloride channel activity. Cell 103: 169–179, 2000.

[34907] 8459. White, K. E.; Biber, J.; Murer, H.; Econs, M. J.: A PDZ domain-containing protein with homology to Diphor-1 maps to human chromosome 1q21. Ann. Hum. Genet. 62: 287–290, 1998.

[34908] 8460. Pastorek, J.; Pastorekova, S.; Callebaut, I.; Mornon, J. P.; Zelnik, V.; Opavsky, R.; Zat'ovicova, M.; Liao, S.; Portetelle, D.; Stanbridge, E. J.; Zavada, J.; Burny, A.; Kettmann, R.: Cloning and characterization of MN, a human tumor-associated protein with a domain homologous to carbonic anhydrase and a putative helix–loop–helix DNA binding segment. Oncogene 9: 2877–2888, 1994.

- [34909] 8461. Matsumoto-Taniura, N.; Pirollet, F.; Monroe, R.; Gerace, L.; Westendorf, J. M.: Identification of novel M phase phosphoproteins by expression cloning. *Molec. Biol. Cell* 7: 1455–1469, 1996.
- [34910] 8462. Doi, T.; Minami, T.; Itoh, M.; Aburatani, H.; Kawabe, Y.; Kodama, T.; Kondo, N.; Satoh, Y.; Asayama, T.; Imanishi, T.: An alternative form of nucleolysin binds to a T-cluster DNA in the silencer element of platelet factor 4 gene. *Biochem. Biophys. Res. Commun.* 235: 625–630, 1997.
- [34911] 8463. Kawakami, A.; Tian, Q.; Duan, X.; Streuli, M.; Schlossman, S. F.; Anderson, P.: Identification and functional characterization of a TIA-1-related nucleolysin. *Proc. Nat. Acad. Sci.* 89: 8681–8685, 1992.
- [34912] 8464. Roebroek, A. J. M.; Contreras, B.; Pauli, I. G. L.; Van de Ven, W. J. M.: cDNA cloning, genomic organization, and expression of the human RTN2 gene, a member of a gene family encoding reticulons. *Genomics* 51:98–106, 1998.
- [34913] 8465. Kiriakidou, M.; Driscoll, D. A.; Lopez-Guisa, J. M.; Strauss, J. F., III: Cloning and expression of primate Daxx cDNAs and mapping of the human gene to chromosome 6p21.3 in the MHC region. *DNA Cell Biol.* 16: 1289–1298, 1997.

- [34914] 8466. Yang, X.; Khosravi-Far, R.; Chang, H. Y.; Baltimore, D.: Daxx, a novel Fas-binding protein that activates JNK and apoptosis. *Cell* 89:1067–1076, 1997.
- [34915] 8467. Parlati, F.; McNew, J. A.; Fukuda, R.; Miller, R.; Sollner, T.H.; Rothman, J. E.: Topological restriction of SNARE-dependent membranefusion. *Nature* 407: 194–198, 2000.
- [34916] 8468. Beckstead, R.; Ortiz, J. A.; Sanchez, C.; Prokopenko, S. N.; Chambon, P.; Losson, R.; Bellen, H. J.: Bonus, a *Drosophila* homolog of TIF1 proteins, interacts with nuclear receptors and can inhibit beta-FTZ-F1-dependent transcription. *Molec. Cell* 7: 753–765, 2001.
- [34917] 8469. Le Douarin, B.; Zechel, C.; Garnier, J.-M.; Lutz, Y.; Tora, L.; Pierrat, B.; Heery, D.; Gronemeyer, H.; Chambon, P.; Losson, R.: The N-terminal part of TIF1, a putative mediator of the ligand-dependent activation function (AF-2) of nuclear receptors, is fused to B-raf in the oncogenic protein T18. *EMBO J.* 14: 2020–2033, 1995.
- [34918] 8470. Thenot, S.; Henriquet, C.; Rochefort, H.; Cavailles, V.: Differential interaction of nuclear receptors with the putative human transcriptional coactivator hTIF1. *J. Biol. Chem.* 272: 12062–12068, 1997.
- [34919] 8471. Feral, C.; Mattei, M. G.; Pawlak, A.; Guellaen, G.:

Chromosomal localization of three human poly(A)-binding protein genes and four related pseudogenes. *Hum. Genet.* 105: 347-353, 1999.

- [34920] 8472. Houg, A. K.; Maggini, L.; Clement, C. Y.; Reed, G. L.: Identification and structure of activated-platelet protein-1, a protein with RNA-binding domain motifs that is expressed by activated platelets. *Europ. J. Biochem.* 243: 209-218, 1997.
- [34921] 8473. Yang, H.; Duckett, C. S.; Lindsten, T.: iPABP, an inducible poly(A)-binding protein detected in activated human T cells. *Molec. Cell. Biol.* 15: 6770-6776, 1995.
- [34922] 8474. Liu, T.; DeCostanzo, A. J.; Liu, X.; Wang, H.; Hallagan, S.; Moon, R. T.; Malbon, C. C.: G protein signaling from activated rat frizzled-1 to the beta-catenin-Lef-Tcf pathway. *Science* 292: 1718-1722, 2001.
- [34923] 8475. Sagara, N.; Toda, G.; Hirai, M.; Terada, M.; Katoh, M.: Molecular cloning, differential expression, and chromosomal localization of human frizzled-1, frizzled-2, and frizzled-7. *Biochem. Biophys. Res. Commun.* 252: 117-122, 1998.
- [34924] 8476. Tanaka, S.; Akiyoshi, T.; Mori, M.; Wands, J. R.; Sugimachi, K.: A novel frizzled gene identified in human esophageal carcinoma mediates APC/beta-catenin signals.

Proc. Nat. Acad. Sci. 95: 10164–10169,1998.

- [34925] 8477.Winklbauer, R.; Medina, A.; Swain, R. K.; Steinbeisser, H.: Frizzled–7signalling controls tissue separation during *Xenopus* gastrulation. *Nature* 413:856–860, 2001.
- [34926] 8478.Messina, A.; Oliva, M.; Rosato, C.; Huizing, M.; Ruitenbeek, W.;van den Heuvel, L. P.; Forte, M.; Rocchi, M.; De Pinto, V.: Mappingof the human voltage–dependent action channel isoforms 1 and 2 reconsidered. *Biochem. Biophys. Res. Commun.* 255: 707–710, 1999.
- [34927] 8479.Parnet, P.; Garka, K. E.; Bonnert, T. P.; Dower, S. K.; Sims, J.E.: IL–1Rrp is a novel receptor–like molecule similar to the type1 interleukin–1 receptor and its homologues T1/ST2 and IL–1R AcP. *J.Biol. Chem.* 271: 3967–3970, 1996.
- [34928] 8480.Torigoe, K.; Ushio, S.; Okura, T.; Kobayashi, S.; Taniai, M.; Kunikata,T.; Murakami, T.; Sanou, O.; Kojima, H.; Fujii, M.; Ohta, T.; Ikeda,M.; Ikegami, H.; Kurimoto, M.: Purification and characterizationof the human interleukin–18 receptor. *J. Biol. Chem.* 272: 25737–25742,1997.
- [34929] 8481.Akiba, H.; Atsuta, M.; Yagita, H.; Okumura, K.: Identificationof rat OX40 ligand by molecular cloning. *Biochem. Biophys. Res. Commun.* 251:131–136, 1998.
- [34930] 8482.Baum, P. R.; Gayle, R. B., III; Ramsdell, F.; Srinivasan,

S.; Sorensen, R. A.; Watson, M. L.; Seldin, M. F.; Baker, E.; Sutherland, G. R.; Clifford, K. N.; Alderson, M. R.; Goodwin, R. G.; Fanslow, W. C.: Molecular characterization of murine and human OX40/OX40 ligand systems: identification of a human OX40 ligand as the HTLV-1-regulated protein gp34. *EMBO J.* 13: 3992–4001, 1994.

[34931] 8483. Godfrey, W. R.; Fagnoni, F. F.; Harara, M. A.; Buck, D.; Engleman, E. G.: Identification of a human OX-40 ligand, a costimulator of CD4⁺ T cells with homology to tumor necrosis factor. *J. Exp. Med.* 180: 757–762, 1994.

[34932] 8484. Malmstrom, V.; Shipton, D.; Singh, B.; Al-Shamkhani, A.; Puklavec, M. J.; Barclay, A. N.; Powrie, F.: CD134L expression on dendritic cells in the mesenteric lymph nodes drives colitis in T cell-restored SCID mice. *J. Immunol.* 166: 6972–6981, 2001.

[34933] 8485. Miura, S.; Ohtani, K.; Numata, N.; Niki, M.; Ohbo, K.; Ina, Y.; Gojobori, T.; Tanaka, Y.; Tozawa, H.; Nakamura, M.; Sugamura, K.: Molecular cloning and characterization of a novel glycoprotein, gp34, that is specifically induced by the human T-cell leukemia virus type 1 transactivator p40-tax. *Mol. Cell. Biol.* 11: 1313–1325, 1991.

[34934] 8486. Cretney, E.; Takeda, K.; Yagita, H.; Glaccum, M.; Peschon, J. J.; Smyth, M. J.: Increased susceptibility to tu-

mor initiation and metastasis in TNF-related apoptosis-inducing ligand-deficient mice. *J. Immun.* 168:1356–1361, 2002.

[34935] 8487. Degli-Esposti, M. A.; Dougall, W. C.; Smolak, P. J.; Waugh, J. Y.; Smith, C. A.; Goodwin, R. G.: The novel receptor TRAIL-R4 induces NF- κ B and protects against TRAIL-mediated apoptosis, yet retains an incomplete death domain. *Immunity* 7: 813–820, 1997.

[34936] 8488. Nagashima, M.; Shiseki, M.; Miura, K.; Hagiwara, K.; Linke, S. P.; Pedoux, R.; Wang, X. W.; Yokota, J.; Riabowol, K.; Harris, C. C.: DNA damage-inducible gene p33^{ING2} negatively regulates cell proliferation through acetylation of p53. *Proc. Nat. Acad. Sci.* 98: 9671–9676, 2001.

[34937] 8489. Shimada, Y.; Saito, A.; Suzuki, M.; Takahashi, E.; Horie, M.: Cloning of a novel gene (ING1L) homologous to ING1, a candidate tumor suppressor. *Cytogenet. Cell Genet.* 83: 232–235, 1998.

[34938] 8490. Bardoni, B.; Giglio, S.; Schenck, A.; Rocchi, M.; Mandel, J. L.: Assignment of NUFIP1 (nuclear FMRP interacting protein 1) gene to chromosome 13q14 and assignment of a pseudogene to chromosome 6q12. *Cytogenet. Cell Genet.* 89: 11–13, 2000.

[34939] 8491. Bardoni, B.; Schenck, A.; Mandel, J. L.: A novel RNA-

binding nuclearprotein that interacts with the fragile X
mental retardation (FMR1)protein. Hum. Molec. Genet. 8:
2557–2566, 1999.

- [34940] 8492. Blagitko, N.; Schulz, U.; Schinzel, A. A.; Ropers, H.-H.; Kalscheuer, V. M.: Gamma-2-COP, a novel imprinted gene on chromosome 7q32, defines a new imprinting cluster in the human genome. Hum. Molec. Genet. 8:2387–2396, 1999.
- [34941] 8493. Maw, M. A.; Corbeil, D.; Koch, J.; Hellwig, A.; Wilson-Wheeler, J. C.; Bridges, R. J.; Kumaramanickavel, G.; John, S.; Nancarrow, D.; Roper, K.; Weigmann, A.; Huttner, W. B.; Denton, M. J.: A frameshift mutation in prominin (mouse)-like 1 causes human retinal degeneration. Hum. Molec. Genet. 9: 27–34, 2000.
- [34942] 8494. Miraglia, S.; Godfrey, W.; Yin, A. H.; Atkins, K.; Warnke, R.; Holden, J. T.; Bray, R. A.; Waller, E. K.; Buck, D. W.: A novel five-transmembrane hematopoietic stem cell antigen: isolation, characterization, and molecular cloning. Blood 90: 5013–5021, 1997.
- [34943] 8495. Yin, A. H.; Miraglia, S.; Zanjani, E. D.; Almeida-Porada, G.; Ogawa, M.; Leary, A. G.; Olweus, J.; Kearney, J.; Buck, D. W.: AC133, a novel marker for human hematopoietic stem and progenitor cells. Blood 90:5002–5012,

1997.

- [34944] 8496. Taylor, S. S.; Ha, E.; McKeon, F.: The human homologue of Bub3 is required for kinetochore localization of Bub1 and a Mad3/Bub1-related protein kinase. *J. Cell Biol.* 142: 1–11, 1998.
- [34945] 8497. Bonne, S.; van Hengel, J; van Roy, F.: Assignment of the plakophilin-2 gene (PKP2) and a plakophilin-2 pseudogene (PKP2P1) to human chromosome bands 12p11 and 12p13, respectively, by in situ hybridization. *Cytogenet. Cell Genet.* 88: 286–287, 2000.
- [34946] 8498. Mertens, C.; Kuhn, C.; Franke, W. W.: Plakophilins 2a and 2b: constitutive proteins of dual location in the karyoplasm and the desmosomal plaque. *J. Cell Biol.* 135: 1009–1025, 1996.
- [34947] 8499. Schmidt, A; Langbein, L.; Pratzel, S.; Rode, M.; Rackwitz, H.-R.; Franke, W. W.: Plakophilin 3—a novel cell-type-specific desmosomal plaque protein. *Differentiation* 64: 291–306, 1999.
- [34948] 8500. Bergstein, I.; Eisenberg, L. M.; Bhalerao, J.; Jenkins, N. A.; Copeland, N. G.; Osborne, M. P.; Bowcock, A. M.; Brown, A. M. C.: Isolation of two novel WNT genes, WNT14 and WNT15, one of which (WNT15) is closely linked to WNT3 on human chromosome 17q21. *Genomics*

46:450–458, 1997.

- [34949] 8501.Hartmann, C.; Tabin, C. J.: Wnt-14 plays a pivotal role in inducing synovial joint formation in the developing appendicular skeleton. *Cell* 104:341–351, 2001.
- [34950] 8502.Saitoh, T.; Hirai, M.; Katoh, M.: Molecular cloning and characterization of WNT3A and WNT14 clustered in human chromosome 1q42 region. *Biochem.Biophys. Res. Commun.* 284: 1168–1175, 2001.
- [34951] 8503.Habuchi, T.; Luscombe, M.; Elder, P. A.; Knowles, M. A.: Structure and methylation-based silencing of a gene (DBCCR1) within a candidate bladder cancer tumor suppressor region at 9q32–q33. *Genomics* 48:277–288, 1998.
- [34952] 8504.Habuchi, T.; Yoshida, O.; Knowles, M. A.: A novel candidate tumour suppressor locus at 9q32–33 in bladder cancer: localization of the candidate region within a single 840 kb YAC. *Hum. Molec. Genet.* 6:913–919, 1997.
- [34953] 8505.Nishiyama, H.; Takahashi, T.; Kakehi, Y.; Habuchi, T.; Knowles, M. A.: Homozygous deletion at the 9q32–33 candidate tumor suppressor locus in primary human bladder cancer. *Genes Chromosomes Cancer* 26:171–175, 1999.
- [34954] 8506.Murphy, M.; Pykett, M. J.; Harnish, P.; Zang, K. D.;

George, D.L.: Identification and characterization of genes differentially expressed in meningiomas. *Cell Growth Differ.* 4: 715–722, 1993.

[34955] 8507. Oh, Y.; Nagalla, S. R.; Yamanaka, Y.; Kim, H.-S.; Wilson, E.; Rosenfeld, R. G.: Synthesis and characterization of insulin-like growth factor-binding protein (IGFBP)-7. *J. Biol. Chem.* 271: 30322–30325, 1996.

[34956] 8508. Swisshelm, K.; Ryan, K.; Tsuchiya, K.; Sager, R.: Enhanced expression of an insulin growth factor-like binding protein (mac25) in senescent human mammary epithelial cells and induced expression with retinoic acid. *Proc. Nat. Acad. Sci.* 92: 4472–4476, 1995.

[34957] 8509. Yamauchi, T.; Umeda, F.; Masakado, M.; Isaji, M.; Mizushima, S.; Nawata, H.: Purification and molecular cloning of prostacyclin-stimulating factor from serum-free conditioned medium of human diploid fibroblast cells. *Biochem. J.* 303: 591–598, 1994.

[34958] 8510. Ayala-Madrigal, M. L.; Doerr, S.; Ramirez-Duenas, M. L.; Hansmann, I.: Assignment of KPNA4 and KPNB1 encoding karyopherin alpha 4 and beta 1 to human chromosome bands 11q22 and 17q21 respectively, by in situ hybridization. *Cytogenet. Cell Genet.* 89: 258–259, 2000.

[34959] 8511. Bayliss, R.; Littlewood, T.; Stewart, M.: Structural ba-

sis for the interaction between FxFG nucleoporin repeats and importin- β nuclear trafficking. *Cell* 102: 99–108, 2000.

[34960] 8512. Chi, N. C.; Adam, E. J. H.; Adam, S. A.: Sequence and characterization of cytoplasmic nuclear protein import factor p97. *J. Cell Biol.* 130:265–274, 1995.

[34961] 8513. Gorlich, D.; Kostka, S.; Kraft, R.; Dingwall, C.; Laskey, R. A.; Hartmann, E.; Prehn, S.: Two different subunits of importin cooperate to recognize nuclear localization signals and bind them to the nuclear envelope. *Curr. Biol.* 5: 383–392, 1995.

[34962] 8514. Kutay, U.; Izaurralde, E.; Bischoff, F. R.; Mattaj, J. W.; Gorlich, D.: Dominant-negative mutants of importin- β block multiple pathways of import and export through the nuclear pore complex. *EMBO J.* 16:1153–1163, 1997.

[34963] 8515. Matsuda, Y.; Hamatani, K.; Itoh, M.; Takahashi, E.; Araki, R.; Abe, M.: Localization of the importin- β gene to mouse chromosome 11D and rat chromosome 10q32.1. *Genomics* 36: 213–215, 1996.

[34964] 8516. Thornton, C.; Snowden, M. A.; Carling, D.: Identification of a novel AMP-activated protein kinase β subunit isoform that is highly expressed in skeletal muscle. *J. Biol. Chem.* 273: 12443–12450, 1998.

- [34965] 8517.Woods, A.; Cheung, P. C. F.; Smith, F. C.; Davison, M. D.; Scott,J.; Beri, R. K.; Carling, D.: Characterization of AMP-activated proteinkinase beta and gamma subunits: assembly of the heterotrimeric complexin vitro. J. Biol. Chem. 271: 10282-10290, 1996.
- [34966] 8518.Gao, G.; Fernandez, C. S.; Stapleton, D.; Auster, A. S.; Widmer,J.; Dyck, J. R. B.; Kemp, B. E.; Witters, L. A.: Non-catalytic beta-and gamma-subunit isoforms of the 5-prime-AMP-activated protein kinase. J.Biol. Chem. 271: 8675-8681, 1996.
- [34967] 8519.Cheung, P. C. F.; Salt, I. P.; Davies, S. P.; Hardie, D. G.; Carling,D.: Characterization of AMP-activated protein kinase gamma-subunitisoforms and their role in AMP binding. Biochem. J. 346: 659-669,2000.
- [34968] 8520.Gollob, M. H.; Green, M. S.; Tang, A. S.-L.; Gollob, T.; Karibe,A.; Al Sayegh, A. H.; Ahmad, F.; Lozado, R.; Shah, G.; Fananapazir,L.; Bachinski, L. L.; Roberts, R.: Identification of a gene responsiblefor familial Wolff-Parkinson-White syndrome. New Eng. J. Med. 344:1823-1831, 2001. Note: Erratum: New Eng. J. Med. 345: 552 only, 2001.
- [34969] 8521.Lang, T.; Yu, L.; Qiang, T.; Jiang, J.; Chen, Z.; Xin, Y.; Liu,G.; Zhao, S.: Molecular cloning, genomic organization,

and mapping of PRKAG2, a heart abundant gamma-2 subunit of 5-prime-AMP-activated protein kinase, to human chromosome 7q36. *Genomics* 70: 258–263, 2000.

- [34970] 8522. Sinha, S. C.; Nair, M.; Gambhir, D. S.; Mohan, J. C.; Kaul, U.A.; Arora, R.: Genetically transmitted ventricular pre-excitation in a family with hypertrophic cardiomyopathy. *Indian Heart J.* 52:76–78, 2000.
- [34971] 8523. Lee, S.-J.: Expression of growth/differentiation factor 1 in the nervous system: conservation of a bicistronic structure. *Proc. Nat. Acad. Sci.* 88: 4250–4254, 1991.
- [34972] 8524. Yamagoe, S.; Akasaka, T.; Uchida, T.; Hachiya, T.; Okabe, T.; Yamakawa, Y.; Arai, T.; Mizuno, S.; Suzuki, K.: Expression of a neutrophil chemotactic protein LECT2 in human hepatocytes revealed by immunochemical studies using polyclonal and monoclonal antibodies to a recombinant LECT2. *Biochem. Biophys. Res. Commun.* 237: 116–120, 1997.
- [34973] 8525. Yamagoe, S.; Kameoka, Y.; Hashimoto, K.; Mizuno, S.; Suzuki, K.: Molecular cloning, structural characterization, and chromosomal mapping of the human LECT2 gene. *Genomics* 48: 324–329, 1998.
- [34974] 8526. Yamagoe, S.; Mizuno, S.; Suzuki, K.: Molecular cloning of human and bovine LECT2 having a neutrophil

chemotactic activity and its specific expression in the liver.
Biochim. Biophys. Acta 1396: 105–113, 1998.

[34975] 8527. Yamagoe, S.; Watanabe, T.; Mizuno, S.; Suzuki, K.:
The mouse Lect2 gene: cloning of cDNA and genomic
DNA, structural characterization and chromosomal localization.
Gene 216: 171–178, 1998.

[34976] 8528. Yamagoe, S.; Yamakawa, Y.; Matsuo, Y.; Minowada,
J.; Mizuno, S.; Suzuki, K.: Purification and primary amino
acid sequence of a novel neutrophil chemotactic factor
LECT2. Immun. Lett. 52: 9–13, 1996.

[34977] 8529. Kullak-Ublick, G.-A.; Beuers, U.; Meier, P. J.;
Domdey, H.; Paumgartner, G.: Assignment of the human
organic anion transporting polypeptide (OATP) gene to
chromosome 12p12 by fluorescence in situ hybridization.
J. Hepatol. 25: 985–987, 1996.

[34978] 8530. Kullak-Ublick, G. A.; Hagenbuch, B.; Stieger, B.;
Schteingart, C. D.; Hofmann, A. F.; Wolkoff, A. W.; Meier, P.
J.: Molecular and functional characterization of an organic
anion transporting polypeptide cloned from human liver.
Gastroenterology 109: 1274–1282, 1995.

[34979] 8531. Ohya, C.; Smith, P. L.; Angata, K.; Fukuda, M. N.;
Lowe, J. B.; Fukuda, M.: Molecular cloning and expression
of GDP-D-mannose-4,6-dehydratase, a key enzyme for

fucose metabolism defective in Lec13 cells. *J. Biol.Chem.* 273: 14582–14587, 1998.

[34980] 8532.El-Husseini, A. E.-D.; Schnell, E.; Chetkovich, D. M.; Nicoll,R. A.; Brecht, D. S.: PSD-95 involvement in maturation of excitatorysynapses. *Science* 290: 1364–1368, 2000.

[34981] 8533.El-Husseini, A. E.-D.; Schnell, E.; Dakoji, S.; Sweeney, N.; Zhou,Q.; Prange, O.; Gauthier-Campbell, C.; Aguilera-Moreno, A.; Nicoll,R. A.; Brecht, D. S.: Synaptic strength regulated by palmitate cyclinon PSD-95. *Cell* 108: 849–863, 2002.

[34982] 8534.Kim, E.; Cho, K.-O.; Rothschild, A.; Sheng, M.: Heteromultimerizationand NMDA receptor-clustering activity of Chapsyn-110, a member ofthe PSD-95 family of proteins. *Neuron* 17: 103–113, 1996.

[34983] 8535.Kim, E.; Niethammer, M.; Rothschild, A.; Jan, Y. N.; Sheng, M.: Clustering of Shaker-type K⁺ channels by interaction with a familyof membrane-associated guanylate kinases. *Nature* 378: 85–88, 1995.

[34984] 8536.Kistner, U.; Wenzel, B. M.; Veh, R. W.; Cases-Langhoff, C.; Garner,A. M.; Appeltauer, U.; Voss, B.; Gundelfinger, E. D.; Garner, C. C.: SAP90, a rat presynaptic protein related to the product of the *Drosophila* tumor

suppressor gene, dLg-A. J. Biol. Chem. 268: 4580–4583, 1993.

- [34985] 8537. Migaud, M.; Charlesworth, P.; Dempster, M.; Webster, L. C.; Watabe, A. M.; Makhinson, M.; He, Y.; Ramsay, M. F.; Morris, R. G. M.; Morrison, J. H.; O'Dell, T. J.; Grant, S. G. N.: Enhanced long-term potentiation and impaired learning in mice with mutant postsynaptic density-95 protein. *Nature* 396: 433–439, 1998.
- [34986] 8538. Sattler, R.; Xiong, Z.; Lu, W.-Y.; Hafner, M.; MacDonald, J. F.; Tymianski, M.: Specific coupling of NMDA receptor activation to nitric oxide neurotoxicity by PSD-95 protein. *Science* 284: 1845–1848, 1999.
- [34987] 8539. Stathakis, D. G.; Hoover, K. B.; You, Z.; Bryant, P. J.: Human postsynaptic density-95 (PSD95): location of the gene (DLG4) and possible function in nonneural as well as in neural tissues. *Genomics* 44: 71–82, 1997.
- [34988] 8540. Strippoli, P.; Petrini, M.; Lenzi, L.; Carinci, P.; Zannotti, M.: The murine DSCR1-like (Down syndrome candidate region 1) gene family: conserved synteny with the human orthologous genes. *Gene* 257: 223–232, 2000.
- [34989] 8541. Yang, J.; Rothermel, B.; Vega, R. B.; Frey, N.; McKinsey, T. A.; Olson, E. N.; Bassel-Duby, R.; Williams, R. S.: Independent signals control expression of the calcineurin in-

hibitory proteins MCIP1 and MCIP2 in striated muscles.

Circ. Res. 87: 61e–68e, 2000.

- [34990] 8542. Zheng, B.; Larkin, D. W.; Albrecht, U.; Sun, Z. S.; Sage, M.; Eichele, G.; Lee, C. C.; Bradley, A.: The mPer2 gene encodes a functional component of the mammalian circadian clock. *Nature* 400: 169–173, 1999.
- [34991] 8543. Nakamura, H.; Sudo, T.; Tsuiki, H.; Miyake, H.; Morisaki, T.; Sasaki, J.; Masuko, N.; Kochi, M.; Ushio, Y.; Saya, H.: Identification of a novel human homolog of the *Drosophila* dlg, P-dlg, specifically expressed in the gland tissues and interacting with p55. *FEBS Lett.* 433: 63–67, 1998.
- [34992] 8544. Abrieu, A.; Magnaghi-Jaulin, L.; Kahana, J. A.; Peter, M.; Castro, A.; Vigneron, S.; Lorca, T.; Cleveland, D. W.; Labbe, J.-C.: Mps1 is a kinetochore-associated kinase essential for the vertebrate mitotic checkpoint. *Cell* 106: 83–93, 2001.
- [34993] 8545. Fisk, H. A.; Winey, M.: The mouse Mps1p-like kinase regulates centrosome duplication. *Cell* 106: 95–104, 2001.
- [34994] 8546. Lauze, E.; Stoelcker, B.; Luca, F. C.; Weiss, E.; Schutz, A. R.; Winey, M.: Yeast spindle pole body duplication gene MPS1 encodes an essential dual specificity protein kinase. *EMBO J.* 14: 1655–1663, 1995.

- [34995] 8547.Lindberg, R. A.; Fischer, W. H.; Hunter, T.: Characterization of a human protein threonine kinase isolated by screening an expression library with antibodies to phosphotyrosine. *Oncogene* 8: 351–359,1993.
- [34996] 8548.Funderburgh, J. L.; Perchellet, A. L.; Swiergiel, J.; Conrad, G.W.; Justice, M. J.: Keratocan (Kera), a corneal keratan sulfate proteoglycan, maps to the distal end of mouse chromosome 10. *Genomics* 52: 110–111,1998.
- [34997] 8549.Liu, C.-Y.; Shiraishi, A.; Kao, C. W.-C.; Converse, R. L.; Funderburgh, J. L.; Corpuz, L. M.; Conrad, G. W.; Kao, W. W.-Y.: The cloning of mouse keratocan cDNA and genomic DNA and the characterization of its expression during eye development. *J. Biol. Chem.* 273: 22584–22588,1998.
- [34998] 8550.Tasheva, E. S.; Funderburgh, J. L.; Funderburgh, M. L.; Corpuz, L. M.; Conrad, G. W.: Structure and sequence of the gene encoding human keratocan. *DNA Seq.* 10: 67–74, 1999.
- [34999] 8551.Tasheva, E. S.; Pettenati, M.; Von Kap-Her, C.; Conrad, G. W.: Assignment of keratocan gene (KERA) to human chromosome band 12q22 by in situ hybridization. *Cytogenet. Cell Genet.* 88: 244–245, 2000.
- [35000] 8552.Bruick, R. K.: Expression of the gene encoding the

proapoptotic Nip3 protein is induced by hypoxia. *Proc. Nat. Acad. Sci.* 97: 9082–9087, 2000.

[35001] 8553. Vaughan, K. T.; Mikami, A.; Paschal, B. M.; Holzbaur, E. L. F.; Hughes, S. M.; Echeverri, C. J.; Moore, K. J.; Gilbert, D. J.; Copeland, N. G.; Jenkins, N. A.; Vallee, R. B.: Multiple mouse chromosomal loci for dynein-based motility. *Genomics* 36: 29–38, 1996.

[35002] 8554. Soyombo, A. A.; Hofmann, S. L.: Molecular cloning and expression of palmitoyl-protein thioesterase 2 (PPT2), a homolog of lysosomal palmitoyl-protein thioesterase with a distinct substrate specificity. *J. Biol. Chem.* 272: 27456–27463, 1997.

[35003] 8555. Richter-Cook, N. J.; Dever, T. E.; Hensold, J. O.; Merrick, W. C.: Purification and characterization of a new eukaryotic protein translation factor: eukaryotic initiation factor 4H. *J. Biol. Chem.* 273: 7579–7587, 1998.

[35004] 8556. Hoogenraad, C. C.; Eussen, B. H. J.; Langeveld, A.; van Haperen, R.; Winterberg, S.; Wouters, C. H.; Grosveld, F.; De Zeeuw, C. I.; Galjart, N.: The murine CYLN2 gene: genomic organization, chromosomal localization, and comparison to the human gene that is located within the 7q11.23 Williams syndrome critical region. *Genomics* 53: 348–358, 1998.

- [35005] 8557.Araujo, H.; Danziger, N.; Cordier, J.; Glowinski, J.; Chneiweiss, H.: Characterization of PEA-15, a major substrate for protein kinase C in astrocytes. *J. Biol. Chem.* 268: 5911–5920, 1993.
- [35006] 8558.Bera, T. K.; Guzman, R. C.; Miyamoto, S.; Panda, D. K.; Sasaki, M.; Hanyu, K.; Enami, J.; Nandi, S.: Identification of a mammary transforming gene (MAT1) associated with mouse mammary carcinogenesis. *Proc. Nat. Acad. Sci.* 91: 9789–9793, 1994.
- [35007] 8559.Condorelli, G.; Vigliotta, G.; Iavarone, C.; Caruso, M.; Tocchetti, C. G.; Andreozzi, F.; Cafieri, A.; Tecce, M. F.; Formisano, P.; Beguinot, L.; Beguinot, F.: PED/PEA-15 gene controls glucose transport and is overexpressed in type 2 diabetes mellitus. *EMBO J.* 17: 3858–3866, 1998.
- [35008] 8560.Danziger, N.; Yokoyama, M.; Jay, T.; Cordier, J.; Glowinski, J.; Chneiweiss, H.: Cellular expression, developmental regulation, and phylogenetic conservation of PEA-15, the astrocytic major phosphoprotein and protein kinase C substrate. *J. Neurochem.* 64: 1016–1025, 1995.
- [35009] 8561.Estelles, A.; Yokoyama, M.; Nothias, F.; Vincent, J.-D.; Glowinski, J.; Vernier, P.; Chneiweiss, H.: The major astrocytic phosphoprotein PEA-15 is encoded by two mRNAs conserved on their full length in mouse and human. *J.*

Biol. Chem. 271: 14800–14806, 1996.

- [35010] 8562.Hwang, S.; Kuo, W.-L.; Cochran, J. F.; Guzman, R. C.; Tsukamoto,T.; Bandyopadhyay, G.; Myambo, K.; Collins, C. C.: Assignment ofHMAT1, the human homolog of the murine mammary transforming gene (MAT1)associated with tumorigenesis, to 1q21.1, a region frequently gainedin human breast cancers. Genomics 42: 540–542, 1997.
- [35011] 8563.Wolford, J. K.; Bogardus, C.; Ossowski, V.; Proc-hazka, M.: Molecularcharacterization of the human PEA15 gene on 1q21–q22 and associationwith type 2 diabetes mellitus in Pima Indians. Gene 241: 143–148,2000.
- [35012] 8564.Aurrand–Lions, M.; Galland, F.; Bazin, H.; Zakharyev, V. M.; Imhof,B. A.; Naquet, P.: Vanin–1, a novel GPI–linked perivascular moleculeinvolved in thymus homing. Immunity 5: 391–405, 1996.
- [35013] 8565.Galland, F.; Malergue, F.; Bazin, H.; Mattei, M. G.; Aurrand–Lions,M.; Theillet, C.; Naquet, P.: Two human genes related to murine vanin–1are located on the long arm of human chromosome 6. Genomics 53:203–213, 1998.
- [35014] 8566.Martin, F.; Malergue, F.; Pitari, G.; Philippe, J. M.; Philips,S.; Chabret, C.; Granjeaud, S.; Mattei, M. G.;

Mungall, A. J.; Naquet, P.; Galland, F.: Vanin genes are clustered (human 6q22–24 and mouse 10A2B1) and encode isoforms of pantetheinase ectoenzymes. *Immunogenetics* 53:296–306, 2001.

[35015] 8567. Pitari, G.; Malergue, F.; Martin, F.; Philippe, J. M.; Massucci, M. T.; Chabret, C.; Maras, B.; Dupre, S.; Naquet, P.; Galland, F.: Pantetheinase activity of membrane-bound vanin-1: lack of free cysteamine in tissues of vanin-1 deficient mice. *FEBS Lett.* 483: 149–154, 2000.

[35016] 8568. Hendrich, B.; Bird, A.: Identification and characterization of a family of mammalian methyl-CpG binding proteins. *Molec. Cell. Biol.* 18:6538–6547, 1998.

[35017] 8569. Rasooly, R. S.: Personal Communication. Baltimore, Md. 2/23/1999.

[35018] 8570. Zhang, Y.; Ng, H.-H.; Erdjument-Bromage, H.; Tempst, P.; Bird, A.; Reinberg, D.: Analysis of the NuRD subunits reveals a histone deacetylase core complex and a connection with DNA methylation. *Genes Dev.* 13:1924–1935, 1999.

[35019] 8571. Bellacosa, A.; Cicchillitti, L.; Schepis, F.; Riccio, A.; Yeung, A. T.; Matsumoto, Y.; Golemis, E. A.; Genuardi, M.; Neri, G.: MED1, a novel human methyl-CpG-binding endonuclease, interacts with DNA mismatch repair protein

MLH1. Proc. Nat. Acad. Sci. 96: 3969–3974,1999.

[35020] 8572.Boland, C. R.; Thibodeau, S. N.; Hamilton, S. R.; Sidransky, D.;Eshleman, J. R.; Burt, R. W.; Meltzer, S. J.; Rodriguez–Bigas, M.A.; Fodde, R.; Ranzani, G. N.; Srivastava, S.: A National CancerInstitute workshop on microsatellite instability for cancer detectionand familial predisposition: development of international criteriafor the determination of microsatellite instability in colorectal cancer. Cancer Res. 58: 5248–5257, 1998.

[35021] 8573.Hendrich, B.; Hardeland, U.; Ng, H.–H.; Jiricny, J.; Bird, A.:The thymine glycosylase MBD4 can bind to the product of deaminationat methylated CpG sites. Nature 401: 301–304, 1999.

[35022] 8574.Millar, C. B.; Guy, J.; Sansom, O. J.; Selfridge, J.; MacDougall,E.; Hendrich, B.; Keightley, P. D.; Bishop, S. M.; Clarke, A. R.;Bird, A.: Enhanced CpG mutability and tumorigenesis in MBD4–deficientmice. Science 297: 403–405, 2002.

[35023] 8575.Riccio, A.; Aaltonen, L. A.; Godwin, A. K.; Loukola, A.; Percesepe,A.; Salovaara, R.; Masciullo, V.; Genuardi M.; Paravatou–Petsotas,M.; Bassi, D. E.; Ruggeri, B. A.; Klein–Szanto, A. J. P.; Testa, J.R.; Neri, G.; Bellacosa, A.: The DNA repair gene MBD4 (MED1) is mutatedin human carcinomas

with microsatellite instability. (Letter) *NatureGenet.* 23: 266–268, 1999.

- [35024] 8576. Duncan, L. M.; Deeds, J.; Hunter, J.; Shao, J.; Holmgren, L. M.; Woolf, E. A.; Tepper, R. I.; Shyjan, A. W.: Down-regulation of the novel gene melastatin correlates with potential for melanoma metastasis. *CancerRes.* 58: 1515–1520, 1998.
- [35025] 8577. Fang, D.; Setaluri, V.: Expression and up-regulation of alternatively spliced transcripts of melastatin, a melanoma metastasis-related gene, in human melanoma cells. *Biochem. Biophys. Res. Commun.* 279: 53–61, 2000.
- [35026] 8578. Hunter, J. J.; Shao, J.; Smutko, J. S.; Dussault, B. J.; Nagle, D. L.; Woolf, E. A.; Holmgren, L. M.; Moore, K. J.; Shyjan, A. W.: Chromosomal localization and genomic characterization of the mouse melastatin gene (*Mlsn1*). *Genomics* 54: 116–123, 1998.
- [35027] 8579. Xu, X. Z.; Moebius, F.; Gill, D. L.; Montell, C.: Regulation of melastatin, a TRP-related protein, through interaction with a cytoplasmic isoform. *Proc. Nat. Acad. Sci.* 98: 10692–10697, 2001.
- [35028] 8580. Kalitsis, P.; Earle, E.; Fowler, K. J.; Choo, K. H. A.: *Bub3* gene disruption in mice reveals essential mitotic spindle checkpoint function during early embryogenesis.

Genes Dev. 14: 2277–2282, 2000.

- [35029] 8581. Ardley, H. C.; Moynihan, T. P.; Thompson, J.; Leek, J. P.; Markham, A. F.; Robinson, P. A.: Rapid isolation of genomic clones for individual members of human multigene families: identification and localisation of UBE2L4, a novel member of a ubiquitin conjugating enzyme dispersed gene family. *Cytogenet. Cell Genet.* 79: 188–192, 1997.
- [35030] 8582. Moynihan, T. P.; Cole, C. G.; Dunham, I.; O'Neil, L.; Markham, A. F.; Robinson, P. A.: Fine-mapping, genomic organization, and transcript analysis of the human ubiquitin-conjugating enzyme gene UBE2L3. *Genomics* 51:124–127, 1998.
- [35031] 8583. Hu, M. C.-T.; Qiu, W. R.; Wang, Y.-P.; Hill, D.; Ring, B. D.; Scully, S.; Bolon, B.; DeRose, M.; Luethy, R.; Simonet, W. S.; Arakawa, T.; Danilenko, D. M.: FGF-18, a novel member of the fibroblast growth factor family, stimulates hepatic and intestinal proliferation. *Molec. Cell. Biol.* 18: 6063–6074, 1998.
- [35032] 8584. Baens, M.; Chaffanet, M.; Cassiman, J.-J.; van den Berghe, H.; Marynen, P.: Construction and evaluation of a hncDNA library of human 12 transcribed sequences derived from a somatic cell hybrid. *Genomics* 16:214–218, 1993.

- [35033] 8585.Fearnley, I. M.; Finel, M.; Skehel, J. M.; Walker, J. E.: NADH:ubiquinoneoxidoreductase from bovine heart mitochondria. *Biochem. J.* 278:821–829, 1991.
- [35034] 8586.Baens, M.; Chaffanet, M.; Cassiman, J.-J.; van den Berghe, H.;Marynen, P.: Construction and evaluation of a hncDNA library of human12p transcribed sequences derived from a somatic cell hybrid. *Genomics* 16:214–218, 1993.
- [35035] 8587.Kawasaki, H.; Springett, G. M.; Toki, S.; Canales, J. J.; Harlan,P.; Blumenstiel, J. P.; Chen, E. J.; Bany, I. A.; Mochizuki, N.; Ashbacher,A.; Matsuda, M.; Housman, D. E.; Graybiel, A. M.: A Rap guanine nucleotideexchange factor enriched highly in the basal ganglia. *Proc. Nat.Acad. Sci.* 95: 13278–13283, 1998.
- [35036] 8588.Erle, D. J.; Sheppard, D.; Breuss, J.; Ruegg, C.; Pytela, R.:Novel integrin alpha and beta subunit cDNAs identified in airway epithelialcells and lung leukocytes using the polymerase chain reaction. *Am.J. Resp. Cell Molec. Biol.* 5: 170–177, 1991.
- [35037] 8589.Hibi, K.; Yamakawa, K.; Ueda, R.; Horio, Y.; Murata, Y.; Tamari,M.; Uchida, K.; Takahashi, T.; Nakamura, Y.; Takahashi, T.: Aberrantupregulation of a novel integrin alpha subunit gene at 3p21.3 in smallcell lung cancer.

Oncogene 9: 611–619, 1994.

- [35038] 8590. Palmer, E. L.; Ruegg, C.; Ferrando, R.; Pytela, R.; Sheppard, D.: Sequence and tissue distribution of the integrin alpha-9 subunit, a novel partner of beta-1 that is widely distributed in epithelia and muscle. *J. Cell Biol.* 123: 1289–1297, 1993.
- [35039] 8591. Yamakawa, K.; Takahashi, T.; Horio, Y.; Murata, Y.; Takahashi, E.; Hibi, K.; Yokoyama, S.; Ueda, R.; Takahashi, T.; Nakamura, Y.: Frequent homozygous deletions in lung cancer cell lines detected by a DNA marker located at 3p21.3–p22. *Oncogene* 8: 327–330, 1993.
- [35040] 8592. La Starza, R.; Wlodarska, I.; Aventin, A.; Falzetti, D.; Crescenzi, B.; Martelli, M. F.; Van den Berghe, H.; Mecucci, C.: Molecular delineation of 13q deletion boundaries in 20 patients with myeloid malignancies. *Blood* 91: 231–237, 1998.
- [35041] 8593. Favre, B.; Fontao, L.; Koster, J.; Shafaatian, R.; Jaunin, F.; Saurat, J.-H.; Sonnenberg, A.; Borradori, L.: The hemidesmosomal protein bullous pemphigoid antigen 1 and the integrin beta-4 subunit bind to ERBIN: molecular cloning of multiple alternative splice variants of ERBIN and analysis of their tissue expression. *J. Biol. Chem.* 276: 32427–32436, 2001.

- [35042] 8594. Agnello, V.; Abel, G.; Elfahal, M.; Knight, G. B.; Zhang, Q.-X.: Hepatitis C virus and other flaviviridae viruses enter cells via low density lipoprotein receptor. *Proc. Nat. Acad. Sci.* 96: 12766–12771, 1999.
- [35043] 8595. Allen, J. M.; Thompson, G. R.; Myant, N. B.; Steiner, R.; Oakley, C. M.: Cardiovascular complications of homozygous familial hypercholesterolaemia. *Brit. Heart J.* 44: 361–368, 1980.
- [35044] 8596. Benlian, P.; Amselem, S.; Loux, N.; Pastier, D.; Giraud, G.; deGennes, J. L.; Turpin, G.; Monnier, L.; Rieu, D.; Douste-Blazy, P.; Dastugue, B.; Goossens, M.; Junien, C.: A LDL receptor gene homozygous mutation: PCR amplification, direct genomic sequencing, associated haplotype, rapid screening for frequency. *Ann. Genet.* 33: 65–69, 1990.
- [35045] 8597. Bertolini, S.; Lelli, N.; Coviello, D. A.; Ghisellini, M.; Masturzo, P.; Tiozzo, R.; Elicio, N.; Gaddi, A.; Calandra, S.: A large deletion in the LDL receptor gene—the cause of familial hypercholesterolemia in three Italian families: a study that dates back to the 17th century (FH-Pavia). *Am. J. Hum. Genet.* 51: 123–134, 1992.
- [35046] 8598. Betard, C.; Kessling, A. M.; Roy, M.; Chamberland, A.; Lussier-Cacan, S.; Davignon, J.: Molecular genetic evi-

dence for a founder effect in familial hypercholesterolemia among French Canadians. Hum. Genet. 88:529–536, 1992.

[35047] 8599.Boehnke, M.; Arnheim, N.; Li, H.; Collins, F. S.: Fine-structure genetic mapping of human chromosomes using the polymerase chain reaction on single sperm: experimental design considerations. Am. J. Hum. Genet. 45: 21–32, 1989.

[35048] 8600.Brown, M. S.; Goldstein, J. L.: Receptor-mediated endocytosis: insights from the lipoprotein receptor system. Proc. Nat. Acad. Sci. 76:3330–3337, 1979.

[35049] 8601.Defesche, J. C.; van Diermen, D. E.; Lansberg, P. J.; Lamping, R. J.; Reymer, P. W. A.; Hayden, M. R.; Kastelein, J. J. P.: South African founder mutations in the low-density lipoprotein receptor gene causing familial hypercholesterolemia in the Dutch population. Hum. Genet. 92: 567–570, 1993.

[35050] 8602.Defesche, J. C.; van de Ree, M. A.; Kastelein, J. J. P.; van Diermen, D. E.; Janssens, N. W. E.; van Doormaal, J. J.; Hayden, M. R.: Detection of the pro664-to-leu mutation in the low-density lipoprotein receptor and its relation to lipoprotein(a) levels in patients with familial hypercholesterolemia of Dutch ancestry from The Netherlands and

Canada. Clin.Genet. 42: 273–280, 1992.

- [35051] 8603.Chen, H. I.; Einbond, A.; Kwak, S.–J.; Linn, H.; Koepf, E.; Peterson,S.; Kelly, J. W.; Sudol, M.: Characterization of the WW domain ofhuman Yes–associated protein and its polyproline containing ligands. J.Biol. Chem. 272: 17070–17077, 1997.
- [35052] 8604.Chen, H. I.; Sudol, M.: The WW domain of Yes–associated proteinbinds a proline–rich ligand that differs from the consensus establishedfor Src homology 3–binding modules. Proc. Nat. Acad. Sci. 92: 7819–7823,1995.
- [35053] 8605.Hoggard, N.; Brintell, B.; Howell, A.; Weissenbach, J.; Varley,J.: Allelic imbalance on chromosome 1 in human breast cancer. II.Microsatellite repeat analysis. Genes Chromosomes Cancer 12: 24–31,1995.
- [35054] 8606.White, G. R. M.; Varley, J. M.; Heighway, J.: Isolation and characterizationof a human homologue of the latrophilin gene from a region of 1p31.1implicated in breast cancer. Oncogene 17: 3513–3519, 1998.
- [35055] 8607.Tommasi, S.; Dammann, R.; Jin, S.–G.; Zhang, X.; Avruch, J.; Pfeifer,G. P.: RASSF3 and NORE1: identification and cloning of two humanhomologues of the putative tumor suppressor gene RASSF1. Oncogene 21:2713–2720,

2002.

- [35056] 8608.Yao, R.; Wang, Y.; You, M.: Chromosome mapping and sequence variation of the murine Ras effector gene Nore1. *Cytogenet. Cell Genet.* 95:126–128, 2001.
- [35057] 8609.Heyer, B. S.; Warsowe, J.; Solter, D.; Knowles, B. B.; Ackerman, S. L.: New member of the Snf1/AMPK kinase family, Melk, is expressed in the mouse egg and preimplantation embryo. *Molec. Reprod. Dev.* 47:148–156, 1997.
- [35058] 8610.Merrill, R. A.; Plum, L. A.; Kaiser, M. E.; Clagett-Dame, M.: A mammalian homolog of unc-53 is regulated by all-trans retinoic acid in neuroblastoma cells and embryos. *Proc. Nat. Acad. Sci.* 99: 3422–3427, 2002.
- [35059] 8611.van Hille, B.; Richener, H.; Evans, D. B.; Green, J. R.; Bilbe, G.: Identification of two subunit A isoforms of the vacuolar H(+)-ATPase in human osteoclastoma. *J. Biol. Chem.* 268: 7075–7080, 1993.
- [35060] 8612.van Hille, B.; Richener, H.; Green, J. R.; Bilbe, G.: The ubiquitous VA68 isoform of subunit A of the vacuolar H(+)-ATPase is highly expressed in human osteoclasts. *Biochem. Biophys. Res. Commun.* 214: 1108–1113, 1995.
- [35061] 8613.Boyhan, A.; Casimir, C. M.; French, J. K.; Teahan, C. G.; Segal, A. W.: Molecular cloning and characterization of

grancalcin, a novel EF-hand calcium-binding protein abundant in neutrophils and monocytes. *J. Biol. Chem.* 267: 2928–2933, 1992.

[35062] 8614. Teahan, C. G.; Totty, N. F.; Segal, A. W.: Isolation and characterization of grancalcin, a novel 28 kDa EF-hand calcium-binding protein from human neutrophils.

Biochem. J. 286: 549–554, 1992.

[35063] 8615. Denning, G.; Jamieson, L.; Maquat, L. E.; Thompson, E. A.; Fields, A. P.: Cloning of a novel phosphatidylinositol kinase-related kinase: characterization of the human SMG-1 RNA surveillance protein. *J. Biol. Chem.* 276:

22709–22714, 2001.

[35064] 8616. Diaz-Meco, M. T.; Municio, M. M.; Sanchez, P.; Lozano, J.; Moscat, J.: Lambda-interacting protein, a novel protein that specifically interacts with the zinc finger domain of the atypical protein kinase C isotype lambda/iota and stimulates its kinase activity in vitro and in vivo.

Molec. Cell. Biol. 16: 105–114, 1996.

[35065] 8617. Kondo, M.; Ji, L.; Kamibayashi, C.; Tomizawa, Y.; Randle, D.; Sekido, Y.; Yokota, J.; Kashuba, V.; Zabarovsky, E.; Kuzmin, I.; Lerman, M.; Roth, J.; Minna, J. D.: Overexpression of candidate tumor suppressor gene FUS1 isolated from the 3p21.3 homozygous deletion region lead–

sto G1 arrest and growth inhibition of lung cancer cells.
Oncogene 20:6258–6262, 2001.

- [35066] 8618.Deshpande, K. L.; Seubert, P. H.; Tillman, D. M.; Farkas, W. R.; Katze, J. R.: Cloning and characterization of cDNA encoding the rabbit tRNA–guanine transglycosylase 60–kilodalton subunit. *Arch. Biochem. Biophys.* 326: 1–7, 1996.
- [35067] 8619.Wilson, S. M.; Bhattacharyya, B.; Rachel, R. A.; Coppola, V.; Tessarollo, L.; Householder, D. B.; Fletcher, C. F.; Miller, R. J.; Copeland, N. G.; Jenkins, N. A.: Synaptic defects in ataxia mice result from a mutation in *Usp14*, encoding a ubiquitin–specific protease. *Nature Genet.* 7 Oct, 2002. Note: Advance Electronic Publication.
- [35068] 8620.Cunningham, S. A.; Arrate, M. P.; Rodriguez, J. M.; Bjercke, R. J.; Vanderslice, P.; Morris, A. P.; Brock, T. A.: A novel protein with homology to the junctional adhesion molecule: characterization of leukocyte interactions. *J. Biol. Chem.* 275: 34750–34756, 2000.
- [35069] 8621.Liang, T. W.; Chiu, H. H.; Gurney, A.; Sidle, A.; Tumas, D. B.; Schow, P.; Foster, J.; Klassen, T.; Dennis, K.; DeMarco, R. A.; Pham, T.; Frantz, G.; Fong, S.: Vascular endothelial–junctional adhesion molecule (VE–JAM)/JAM 2 interacts with T, NK, and dendritic cells through JAM 3. *J.*

Immun. 168: 1618–1626, 2002.

- [35070] 8622. Palmeri, D.; van Zante, A.; Huang, C.-C.; Hemmerich, S.; Rosen, S. D.: Vascular endothelial junction-associated molecule, a novel member of the immunoglobulin superfamily, is localized to intercellular boundaries of endothelial cells. *J. Biol. Chem.* 275: 19139–19145, 2000.
- [35071] 8623. Arrate, M. P.; Rodriguez, J. M.; Tran, T. T.; Brock, T. A.; Cunningham, S. A.: Cloning of human junctional adhesion molecule 3 (JAM3) and its identification as the JAM2 counter-receptor. *J. Biol. Chem.* 276: 45826–45832, 2001.
- [35072] 8624. Heng, Y. M.; Fox, M.; Sablitzky, F.: Assignment of the murine def-3 gene (Rbm6) to chromosome 9F1–F2 and its pseudogenes Rbm6-ps1 and Rbm6-ps2 to chromosome 1 by in situ hybridisation. *Cytogenet. Cell. Genet.* 89: 238–239, 2000.
- [35073] 8625. Garrett, R. M.; Bellissimo, D. B.; Rajagopalan, K. V.: Molecular cloning of human liver sulfite oxidase. *Biochim. Biophys. Acta* 1262: 147–149, 1995.
- [35074] 8626. Johnson, J. L.; Coyne, K. E.; Garrett, R. M.; Zabet, M.-T.; Dorche, C.; Kisker, C.; Rajagopalan, K. V.: Isolated sulfite oxidase deficiency: identification of 12 novel SUOX mutations in 10 patients. (Abstract) *Hum. Mutat.* 20: 74 only, 2002.

- [35075] 8627.Chen, Y. Q.; Rafi, M. A.; de Gala, G.; Wenger, D. A.: Cloning and expression of cDNA encoding human galactocerebrosidase, the enzyme deficient in globoid cell leukodystrophy. *Hum. Molec. Genet.* 2:1841–1845, 1993.
- [35076] 8628.Chen, Y. Q.; Wenger, D. A.: Galactocerebrosidase from human urine: purification and partial characterization. *Biochim. Biophys. Acta* 1170:53–61, 1993.
- [35077] 8629.Fiumara, A.; Pavone, L.; Siciliano, L.; Tine, A.; Parano, E.; Innico, G.: Late-onset globoid cell leukodystrophy: report on 7 new patients. *Child's Nerv. Syst.* 6: 194–197, 1990.
- [35078] 8630.Furuya, H.; Kukita, Y.; Nagano, S.; Sakai, Y.; Yamashita, Y.; Fukuyama, H.; Inatomi, Y.; Saito, Y.; Koike, R.; Tsuji, S.; Fukumaki, Y.; Hayashi, K.; Kobayashi, T.: Adult onset globoid cell leukodystrophy (Krabbe disease): analysis of galactosylceramidase cDNA from four Japanese patients. *Hum. Genet.* 100: 450–456, 1997.
- [35079] 8631.Kodama, S.; Igisu, H.; Siegel, D. A.; Suzuki, K.: Glycosylceramide synthesis in the developing spinal cord and kidney of the twitcher mouse, an enzymatically authentic model of human Krabbe disease. *J. Neurochem.* 39: 1314–1318, 1982.
- [35080] 8632.Luzi, P.; Rafi, M. A.; Victoria, T.; Baskin, G. B.;

Wenger, D.A.: Characterization of the rhesus monkey galactocerebrosidase (GALC)cDNA and gene and identification of the mutation causing globoid cellleukodystrophy (Krabbe disease) in this primate. *Genomics* 42: 319–324,1997.

[35081] 8633.Luzi, P.; Rafi, M. A.; Wenger, D. A.: Structure and organizationof the human galactocerebrosidase (GALC) gene. *Genomics* 26: 407–409,1995.

[35082] 8634.Wang, K.; Zhou, B.; Kuo, Y.–M.; Zemansky, J.; Gitschier, J.: Anovel member of a zinc transporter family is defective in acrodermatitisenteropathica. *Am. J. Hum. Genet.* 71: 66–73, 2002.

[35083] 8635.Freeman, B. C.; Yamamoto, K. R.: Disassembly of transcriptionalregulatory complexes by molecular chaperones. *Science* 296: 2232–2235,2002.

[35084] 8636.Johnson, J. L.; Beito, T. G.; Krco, C. J.; Toft, D. O.: Characterizationof a novel 23–kilodalton protein of unactive progesterone receptorcomplexes. *Molec. Cell. Biol.* 14: 1956–1963, 1994.

[35085] 8637.Smith, D. F.; Faber, L. E.; Toft, D. O.: Purification of unactivatedprogesterone receptor and identification of novel receptor–associatedproteins. *J. Biol. Chem.* 265: 3996–4003, 1990.

- [35086] 8638. Patterson, C. E.; Gao, J.; Rooney, A. P.; Davis, E. C.: Genomic organization of mouse and human 65 kDa FK506-binding protein genes and evolution of the FKBP multigene family. *Genomics* 79: 881–889, 2002.
- [35087] 8639. Patterson, C. E.; Schaub, T.; Coleman, E. J.; Davis, E. C.: Developmental regulation of FKBP65: an ER localized extracellular matrix-binding protein. *Molec. Biol. Cell* 11: 3925–3935, 2000.
- [35088] 8640. Cai, H.; Wang, Y.; McCarthy, D.; Wen, H.; Borchelt, D. R.; Price, D. L.; Wong, P. C.: BACE1 is the major beta-secretase for generation of A-beta peptides by neurons. *Nature Neurosci.* 4: 233–234, 2001.
- [35089] 8641. Meyaard, L.; Adema, G. J.; Chang, C.; Woollatt, E.; Sutherland, G. R.; Lanier, L. L.; Phillips, J. H.: LAIR-1, a novel inhibitory receptor expressed on human mononuclear leukocytes. *Immunity* 7: 283–290, 1997.
- [35090] 8642. Sathish, J. G.; Johnson, K. G.; Fuller, K. J.; LeRoy, F. G.; Meyaard, L.; Sims, M. J.; Matthews, R. J.: Constitutive association of SHP-1 with leukocyte-associated Ig-like receptor-1 in human T cells. *J. Immun.* 166: 1763–1770, 2001.
- [35091] 8643. Xu, M.; Zhao, R.; Zhao, Z. J.: Identification and characterization of leukocyte-associated Ig-like receptor-1 as

a major anchor protein of tyrosine phosphatase SHP-1 in hematopoietic cells. *J. Biol. Chem.* 275:17440–17446, 2000.

- [35092] 8644. Hofmann, R. M.; Pickart, C. M.: Noncanonical MMS2-encoded ubiquitin-conjugating enzyme functions in assembly of novel polyubiquitin chains for DNA repair. *Cell* 96: 645–653, 1999.
- [35093] 8645. Rothfisky, M. L.; Lin, S. L.: CROC-1 encodes a protein which mediates transcriptional activation of the human FOS promoter. *Gene* 195:141–149, 1997.
- [35094] 8646. Sancho, E.; Vila, M. R.; Sanchez-Pulido, L.; Lozano, J. J.; Paciucci, R.; Nadal, M.; Fox, M.; Harvey, C.; Bercovich, B.; Loukili, N.; Ciechanover, A.; Lin, S. L.; Sanz, F.; Estivill, X.; Valencia, A.; Thomson, T. M.: Role of UEV-1, an inactive variant of the E2 ubiquitin-conjugating enzymes, in in vitro differentiation and cell cycle behavior of HT-29-M6 intestinal mucosecretory cells. *Molec. Cell. Biol.* 18: 576–589, 1998.
- [35095] 8647. Charles, C. H.; Yoon, J. K.; Simske, J. S.; Lau, L. F.: Genomic structure, cDNA sequence, and expression of gly96, a growth factor-inducible immediate-early gene encoding a short-lived glycosylated protein. *Oncogene* 8:797–801, 1993.

- [35096] 8648.Kondratyev, A. D.; Chung, K.-N.; Jung, M. O.: Identification and characterization of a radiation-inducible glycosylated human early-response gene. *Cancer Res.* 56: 1498-1502, 1996.
- [35097] 8649.Pietzsch, A.; Buchler, C.; Aslanidis, C.; Schmitz, G.: Identification and characterization of a novel monocyte/macrophage differentiation-dependent gene that is responsive to lipopolysaccharide, ceramide, and lysophosphatidylcholine. *Biochem.Biophys. Res. Commun.* 235: 4-9, 1997.
- [35098] 8650.Pietzsch, A.; Buchler, C.; Schmitz, G.: Genomic organization, promoter cloning, and chromosomal localization of the Dif-2 gene. *Biochem.Biophys. Res. Commun.* 245: 651-657, 1998.
- [35099] 8651.Schafer, H.; Trauzold, A.; Siegel, E. G.; Folsch, U. R.; Schmidt, W. E.: PRG1: a novel early-response gene transcriptionally induced by pituitary adenylate cyclase activating polypeptide in a pancreatic carcinoma cell line. *Cancer Res.* 56: 2641, 1996.
- [35100] 8652.Wu, M. X.; Ao, Z.; Prasad, K. V. S.; Wu, R.; Schlossman, S. F.: IEX-1L, an apoptosis inhibitor involved in NF-kappa-B-mediated cell survival. *Science* 281: 998-1001, 1998.

- [35101] 8653.De Strooper, B.; Konig, G.: A firm base for drug development. *Nature* 402:471–472, 1999.
- [35102] 8654.Fan, W.; Bennett, B. D.; Babu–Khan, S.; Luo, Y.; Louis, J.–C.;McCaleb, M.; Citron, M.; Vassar, R.; Richards, W. G.: Response toSaunders et al. (1999). *Science* 286: 1255a, 1999. Note: ElectronicPublication.
- [35103] 8655.Haniu, M.; Denis, P.; Young, Y.; Mendiaz, E. A.; Fuller, J.; Hui,J. O.; Bennett, B. D.; Kahn, S.; Ross, S.; Burgess, T.; Katta, V.;Rogers, G.; Vassar, R.; Citron, M.: Characterization of Alzheimer'sbeta–secretase protein BACE: a pepsin family member with unusual properties. *J.Biol. Chem.* 275: 21099–21106, 2000.
- [35104] 8656.Hong, L.; Koelsch, G.; Lin, X.; Wu, S.; Terzyan, S.; Ghosh, A.K.; Zhang, X. C.; Tang, J.: Structure of the protease domain of memapsin2 (beta–secretase) complexed with inhibitor. *Science* 290: 150–153,2000.
- [35105] 8657.Hussain, I.; Powell, D.; Howlett, D. R.; Tew, D. G.; Meek, T. D.;Chapman, C.; Gloger, I. S.; Murphy, K. E.; Southan, C. D.; Ryan, D.M.; Smith, T. S.; Simmons, D. L.; Walsh, F. S.; Dingwall, C.; Christie,G.: Identification of a novel aspartic protease (asp2) as beta–secretase *Molec.Cell Neurosci.* 14: 419–427, 1999.
- [35106] 8658.Parker, A. E.; Van de Weyer, I.; Laus, M. C.; Verhas–

selt, P.; Luyten, W. H. M. L.: Identification of a human homologue of the *Schizosaccharomyces pombe* rad17+ checkpoint gene. *J. Biol. Chem.* 273: 18340–18346, 1998. Note: Erratum: *J. Biol. Chem.* 274: 24438–24439, 1999.

[35107] 8659. von Deimling, F.; Scharf, J. M.; Liehr, T.; Rothe, M.; Kelter, A.-R.; Albers, P.; Dietrich, W. F.; Kunkel, L. M.; Wernert, N.; Wirth, B.: Human and mouse RAD17 genes: identification, localization, genomic structure and histological expression pattern in normal testis and seminoma. *Hum. Genet.* 105: 17–27, 1999.

[35108] 8660. Serra-Pages, C.; Medley, Q. G.; Tang, M.; Hart, A.; Streuli, M.: Liprins, a family of LAR transmembrane protein-tyrosine phosphatase-interacting proteins. *J. Biol. Chem.* 273: 15611–15620, 1998.

[35109] 8661. Thomas, M. K.; Yao, K.-M.; Tenser, M. S.; Wong, G. G.; Habener, J. F.: Bridge-1, a novel PDZ-domain coactivator of E2A-mediated regulation of insulin gene transcription. *Molec. Cell. Biol.* 19: 8492–8504, 1999.

[35110] 8662. Watanabe, T. K.; Saito, A.; Suzuki, M.; Fujiwara, T.; Takahashi, E.; Slaughter, C. A.; DeMartino, G. N.; Hendil, K. B.; Chung, C. H.; Tanahashi, N.; Tanaka, K.: cDNA cloning and characterization of a human proteasomal modulator

subunit, p27 (PSMD9). Genomics 50: 241–250,1998.

- [35111] 8663.Kennedy, J.; Rossi, D. L.; Zurawski, S. M.; Vega, F., Jr.; Kastelein,R. A.; Wagner, J. L.; Hannum, C. H.; Zlotnick, A.: Mouse IL–17: acytokine preferentially expressed by alpha beta TCR + CD4–CD8–T cells. J.Interferon Cytokine Res. 16: 611–617, 1996.
- [35112] 8664.Kotake, S.; Udagawa, N.; Takahashi, N.; Matsuzaki, K.; Itoh, K.;Ishiyama, S.; Saito, S.; Inoue, K.; Kamatani, N.; Gillespie, M. T.;Martin, T. J.; Suda, T.: IL–17 in synovial fluids from patients withrheumatoid arthritis is a potent stimulator of osteoclastogenesis. J.Clin. Invest. 103: 1345–1352, 1999.
- [35113] 8665.Rouvier, E.; Luciani, M.–F.; Mattei, M.–G.; Denizot, F.; Golstein,P.: CTLA–8, cloned from an activated T cell, bearing AU–rich messengerRNA instability sequences, and homologous to a Herpesvirus saimirigene. J. Immun. 150: 5445–5456, 1993.
- [35114] 8666.Yao, Z.; Painter, S. L.; Fanslow, W. C.; Ulrich, D.; Macduff, B.M.; Spriggs, M. K.; Armitage, R. J.: Human IL–17: a novel cytokinederived from T cells. J. Immun. 155: 5483–5486, 1995.
- [35115] 8667.Higuti, T.; Tsurumi, C.; Kawamura, Y.; Tsujita, H.; Osaka, F.;Yoshihara, Y.; Tani, I.; Tanaka, K.; Ichihara, A.:

Molecular cloning of cDNA for the import precursor of human coupling factor 6 of H(+)-ATP synthase in mitochondria. *Biochem. Biophys. Res. Commun.* 178: 793–799, 1991.

- [35116] 8668. Javed, A. A.; Ogata, K.; Sanadi, D. R.: Human mitochondrial ATP synthase: cloning cDNA for the nuclear-encoded precursor of coupling factor 6. *Gene* 97: 307–310, 1991.
- [35117] 8669. Webster, K. A.; Oliver, N. A.; Wallace, D. C.: Assignment of an oligomycin-resistance locus to human chromosome 10. *Somat. Cell Genet.* 8: 223–244, 1982.
- [35118] 8670. Parker, A. E.; Van de Weyer, I.; Laus, M. C.; Oostveen, I.; Yon, J.; Verhasselt, P.; Luyten, W. H. M. L.: A human homologue of *Schizosaccharomyces pombe* rad1+ checkpoint gene encodes an exonuclease. *J. Biol. Chem.* 273: 18332–18339, 1998.
- [35119] 8671. Barbon, A.; Ferraboli, S.; Barlati, S.: Assignment of the human metabotropic glutamate receptor gene GRM4 to chromosome 6 band p21.3 by radiation hybrid mapping. *Cytogenet. Cell Genet.* 88: 210 only, 2000.
- [35120] 8672. Morita, R.; Miyazaki, E.; Fong, C. G.; Chen, X.-N.; Korenberg, J. R.; Delgado-Escueta, A. V.; Yamakawa, K.: JH8, a gene highly homologous to the mouse jerky gene,

maps to the region for childhood absence epilepsy on 8q24. *Biochem. Biophys. Res. Commun.* 248: 307–314, 1998.

[35121] 8673. Toth, M.; Grimsby, J.; Buzsaki, G.; Donovan, G. P.: Epileptic seizures caused by inactivation of a novel gene, jerky, related to centromere binding protein-B in transgenic mice. *Nature Genet.* 11:71–75, 1995. Note: Erratum: *Nature Genet.* 12: 110 only, 1996.

[35122] 8674. Zeng, Z.; Kyaw, H.; Gakenheimer, K. R.; Augustus, M.; Fan, P.; Zhang, X.; Su, K.; Carter, K. C.; Li, Y.: Cloning, mapping, and tissue distribution of a human homologue of the mouse jerky gene product. *Biochem. Biophys. Res. Commun.* 236: 389–395, 1997.

[35123] 8675. Kimura, M.; Matsuda, Y.; Eki, T.; Yoshioka, T.; Okumura, K.; Hanaoka, F.; Okano, Y.: Assignment of STK6 to human chromosome 20q13.2–q13.3 and a pseudogene STK6P to 1q41–q42. *Cytogenet. Cell Genet.* 79: 201–203, 1997.

[35124] 8676. Bryan, J.; Edwards, R.; Matsudaira, P.; Otto, J.; Wulfeuhle, J.: Fascin, an echinoid actin-bundling protein, is a homolog of the *Drosophila* singed gene product. *Proc. Nat. Acad. Sci.* 90: 9115–9119, 1993.

[35125] 8677. Dressel, U.; Thormeyer, D.; Altincicek, B.; Paululat,

A.; Eggert, M.; Schneider, S.; Tenbaum, S. P.; Renkawitz, R.; Baniahmad, A.: Alien, a highly conserved protein with characteristics of a corepressor for members of the nuclear hormone receptor superfamily. *Molec. Cell Biol.* 19: 3383–3394, 1999.

[35126] 8678. Schaefer, L.; Beermann, M. L.; Miller, J. B.: Coding sequence, genomic organization, chromosomal localization, and expression pattern of the signalosome component Cops2: the mouse homologue of *Drosophila* alien. *Genomics* 56: 310–316, 1999.

[35127] 8679. Scott, A. F.: Personal Communication. Baltimore, Md. 11/8/2000.

[35128] 8680. Seeger, M.; Kraft, R.; Ferrell, K.; Dawadshargal, B.-O.; Dumdey, R.; Schade, R.; Gordon, C.; Naumann, M.; Dubiel, W.: A novel protein complex involved in signal transduction possessing similarities to 26S proteasome subunits. *FASEB J.* 12: 469–478, 1998.

[35129] 8681. Barbosa, M. D.; Johnson, S. A.; Achey, K.; Gutierrez, M. J.; Wakeland, E. K.; Zerial, M.; Kingsmore, S. F.: The Rab protein family: genetic mapping of six Rab genes in the mouse. *Genomics* 30: 439–444, 1995.

[35130] 8682. Vitelli, R.; Chiariello, M.; Lattero, D.; Bruni, C. B.; Bucci, C.: Molecular cloning and expression analysis of the

human Rab7 GTP-ase complementary deoxyribonucleic acid. *Biochem. Biophys. Res. Commun.* 229:887–890, 1996.

[35131] 8683. Wang, L.; Mizzen, C.; Ying, C.; Candau, R.; Barlev, N.; Brownell, J.; Allis, C. D.; Berger, S. L.: Histone acetyltransferase activity is conserved between yeast and human GCN5 and is required for complementation of growth and transcriptional activation. *Molec. Cell. Biol.* 17:519–527, 1997.

[35132] 8684. Xu, W.; Edmondson, D. G.; Evrard, Y. A.; Wakamiya, M.; Behringer, R. R.; Roth, S. Y.: Loss of Gcn5l2 leads to increased apoptosis and mesodermal defects during mouse development. *Nature Genet.* 26: 229–232, 2000.

[35133] 8685. Xu, W.; Edmondson, D. G.; Roth, S. Y.: Mammalian GCN5 and P/CAF acetyltransferases have homologous amino-terminal domains important for recognition of nucleosomal substrates. *Molec. Cell. Biol.* 18:5659–5669, 1998.

[35134] 8686. Yang, X.-J.; Ogryzko, V. V.; Nishikawa, J.; Howard, B. H.; Nakatani, Y.: A p300/CBP-associated factor that competes with the adenoviral oncoprotein E1A. *Nature* 382: 319–324, 1996.

[35135] 8687. Miao, D.; He, B.; Karaplis, A. C.; Goltzman, D.:

Parathyroidhormone is essential for normal fetal bone formation. *J. Clin. Invest.* 109:1173–1182, 2002.

- [35136] 8688. Betticher, D. C.; Thatcher, N.; Altermatt, H. J.; Hoban, P.; Ryder, W. D. J.; Heighway, J.: Alternate splicing produces a novel cyclinD1 transcript. *Oncogene* 11: 1005–1011, 1995.
- [35137] 8689. Chesi, M.; Bergsagel, P. L.; Brents, L. A.; Smith, C. M.; Gerhard, D. S.; Kuehl, W. M.: Dysregulation of cyclin D1 by translocation into an IgH gamma switch region in two multiple myeloma cell lines. *Blood* 88:674–681, 1996.
- [35138] 8690. Gailani, M. R.; Petty, E. M.; Horsthemke, B.; Arnold, A.; Marx, S. J.; Bale, A. E.: Physical mapping of chromosome 11q12–13 by pulsed field gel electrophoresis (PFGE). (Abstract) *Cytogenet. Cell Genet.* 58:1959, 1991.
- [35139] 8691. Geng, Y.; Yu, Q.; Sicinska, E.; Das, M.; Bronson, R. T.; Sicinski, P.: Deletion of the p27(Kip1) gene restores normal development in cyclin D1-deficient mice. *Proc. Nat. Acad. Sci.* 98: 194–199, 2001.
- [35140] 8692. Hayette, S.; Gadoux, M.; Martel, S.; Bertrand, S.; Tigaud, I.; Magaud, J.-P.; Rimokh, R.: FLRG (follistatin-related gene), a new target of chromosomal rearrangement in malignant blood disorders. *Oncogene* 16:2949–2954, 1998.

- [35141] 8693. Tajima, T.; Kitagawa, H.; Yokoya, S.; Tachibana, K.; Adachi, M.; Nakae, J.; Suwa, S.; Katoh, S.; Fujieda, K.: A novel missense mutation of mineralocorticoid receptor gene in one Japanese family with a renal form of pseudo-hypoaldosteronism type 1. *J. Clin. Endocr. Metab.* 85:4690–4694, 2000.
- [35142] 8694. Viemann, M.; Peter, M.; Lopez-Siguero, J. P.; Simic-Schleicher, G.; Sippell, W. G.: Evidence for genetic heterogeneity of pseudohypoaldosteronism type 1: identification of a novel mutation in the human mineralocorticoid receptor in one sporadic case and no mutations in two autosomal dominant kindreds. *J. Clin. Endocr. Metab.* 86: 2056–2059, 2001.
- [35143] 8695. Lee, Y. L.; Helman, L.; Hoffman, T.; Laborda, J.: dlk, pG2 and Pref-1 mRNAs encode similar proteins belonging to the EGF-like superfamily. Identification of polymorphic variants of this RNA. *Biochim. Biophys. Acta* 1261: 223–232, 1995.
- [35144] 8696. Smas, C. M.; Sul, H. S.: Pref-1, a protein containing EGF-like repeats, inhibits adipocyte differentiation. *Cell* 73: 725–734, 1993.
- [35145] 8697. Takada, S.; Paulsen, M.; Tevendale, M.; Tsai, C.-E.; Kelsey, G.; Cattanach, B. M.; Ferguson-Smith, A. C.: Epige-

netic analysis of the Dlk1–Gtl2 imprinted domain on mouse chromosome 12: implications for imprinting control from comparison with Igf2–H19. *Hum. Molec. Genet.* 11:77–86, 2002.

- [35146] 8698. Wylie, A. A.; Murphy, S. K.; Orton, T. C.; Jirtle, R. L.: Novel imprinted DLK1/GTL2 domain on human chromosome 14 contains motifs that mimic those implicated in IGF2/H19 regulation. *Genome Res.* 10:1711–1718, 2000.
- [35147] 8699. Okumura, K.; Nogami, M.; Taguchi, H.; Hisamatsu, H.; Tanaka, K.: The genes for the alpha-type HC3 (PMSA2) and beta-type HC5 (PMSB1) subunits of human proteasomes map to chromosomes 6q27 and 7p12–p13 by fluorescence in situ hybridization. *Genomics* 27: 377–379, 1995.
- [35148] 8700. Tamura, T.; Osaka, F.; Kawamura, Y.; Higuchi, T.; Ishida, N.; Nothwang, H. G.; Tsurumi, C.; Tanaka, K.; Ichihara, A.: *J. Molec. Biol.* 244:1117–1124, 1994.
- [35149] 8701. Coux, O.; Tanaka, K.; Goldberg, A. L.: Structure and functions of the 20S and 26S proteasomes. *Ann. Rev. Biochem.* 65: 801–847, 1996.
- [35150] 8702. DeMartino, G. N.; Orth, K.; McCullough, M. L.; Lee, L. W.; Munn, T. Z.; Moomaw, C. R.; Dawson, P. A.; Slaughter, C. A.: The primary structures of four subunits of the hu-

man, high molecular weight proteinase,macropain (proteasome), are distinct but homologous. Biochim. Biophys.Acta 1079: 29–38, 1991.

- [35151] 8703.Cruz, M.; Elenich, L. A.; Smolarek, T. A.; Menon, A. G.; Monaco,J. J.: DNA sequence, chromosomal localization, and tissue expressionof the mouse proteasome subunit Lmp10 (Psm10) gene. Genomics 45:618–622, 1997.
- [35152] 8704.Goldberg, A. L.; Rock, K. L.: Proteolysis, proteasomes and antigenpresentation. Nature 357: 375–379, 1992.
- [35153] 8705.Larsen, F.; Gundersen, G.; Lopez, R.; Prydz, H.: CpG islands asgene markers in the human genome. Genomics 13: 1095–1107, 1992.
- [35154] 8706.Larsen, F.; Solheim, J.; Kristensen, T.; Kolsto, A.–B.; Prydz,H.: A tight cluster of five unrelated human genes on chromosome 16q22.1. Hum.Molec. Genet. 2: 1589–1595, 1993.
- [35155] 8707.DeVry, C. G.; Clarke, S.: Assignment of the protein L–isoaspartate(D–aspartate) O–methyltransferase gene (PCMT1) to human chromosomebands 6q24–q25 with radiation hybrid mapping. Cytogenet. Cell Genet. 84:130–131, 1999.
- [35156] 8708.Farrar, C.; Clarke, S.: Altered levels of S–

adenosylmethionine and S-adenosylhomocysteine in the brains of L-isoaspartyl

(D-aspartyl)O-methyltransferase-deficient mice. J. Biol. Chem. 277: 27856–27863, 2002.

[35157] 8709. Ingrosso, D.; Kagan, R. M.; Clarke, S.: Distinct C-terminal sequences of isozymes I and II of the human erythrocyte L-isoaspartyl/D-aspartyl protein methyltransferase. Biochem. Biophys. Res. Commun. 175: 351–358, 1991.

[35158] 8710. Kim, E.; Lowenson, J. D.; Clarke, S.; Young, S. G.: Phenotypic analysis of seizure-prone mice lacking L-isoaspartate (D-aspartate)O-methyltransferase. J. Biol. Chem. 274: 20671–20678, 1999.

[35159] 8711. Lowenson, J. D.; Kim, E.; Young, S. G.; Clarke, S.: Limited accumulation of damaged proteins in L-isoaspartyl (D-aspartyl) O-methyltransferase-deficient mice. J. Biol. Chem. 276: 20695–20702, 2001.

[35160] 8712. MacLaren, D. C.; Kagan, R. M.; Clarke, S.: Alternative splicing of the human isoaspartyl protein carboxyl methyltransferase leads to the generation of a C-terminal -RDEL sequence in isozyme II. Biochem. Biophys. Res. Commun. 185: 277–283, 1992.

[35161] 8713. MacLaren, D. C.; O'Connor, C. M.; Xia, Y.-R.; Mehra-

bian, M.; Klisak, I.; Sparkes, R. S.; Clarke, S.; Lusk, A. J.:
The L-isoaspartyl/D-aspartylprotein methyltransferase
gene (PCMT1) maps to human chromosome
6q22.3–6q24 and the syntenic region of mouse chromo-
some 10. *Genomics* 14: 852–856, 1992.

[35162] 8714. Ota, I. M.; Gilbert, J. M.; Clarke, S.: Two major
isozymes of the protein D-aspartyl/L-isoaspartyl methyl-
transferase from human erythrocytes. *Biochem. Biophys.
Res. Commun.* 151: 1136–1143, 1988.

[35163] 8715. Hunter, A. G. W.; Clifford, B.; Cox, D. M.: The char-
acteristic physiognomy and tissue specific karyotype dis-
tribution in the Pallister-Killian syndrome. *Clin. Genet.* 28:
47–53, 1985.

[35164] 8716. Astrom, A.-K.; Voz, M. L.; Kas, K.; Roijer, E.; Wedell,
B.; Mandahl, N.; Van de Ven, W.; Mark, J.; Stenman, G.:
Conserved mechanism of PLAG1 activation in salivary
gland tumors with and without chromosome 8q12 abnor-
malities: identification of SII as a new fusion partner gene.
Cancer Res. 59: 918–923, 1999.

[35165] 8717. Bullerdiek, J.; Bartnitzke, S.; Weinberg, M.; Chilla, R.;
Haubrich, J.; Schloot, W.: Rearrangements of chromosome
region 12q13–q15 in pleomorphic adenomas of the human
salivary gland (PSA). *Cytogenet. Cell Genet.* 45: 187–190,

1987.

- [35166] 8718. Bullerdiek, J.; Raabe, G.; Bartnitzke, S.; Boschen, C.; Schloot, W.: Structural rearrangements of chromosome #8 involving 8q12--a primary event in pleomorphic adenomas of the parotid gland. *Genetica* 72:85-92, 1987.
- [35167] 8719. Mark, J.; Dahlenfors, R.: Cytogenetical observations in 100 human benign pleomorphic adenomas: specificity of the chromosomal aberrations and their relationship to sites of localized oncogenes. *Anticancer Res.* 6: 299-308, 1986.
- [35168] 8720. Stenman, G.; Sandros, J.; Mark, J.; Nordkvist, A.: High p21(RAS) expression levels correlate with chromosome 8 rearrangements in benign human mixed salivary gland tumors. *Genes Chromosomes Cancer* 1: 59-66, 1989.
- [35169] 8721. Hinds, P. W.; Dowdy, S. F.; Eaton, E. N.; Arnold, A.; Weinberg, R. A.: Function of a human cyclin gene as an oncogene. *Proc. Nat. Acad. Sci.* 91: 709-713, 1994.
- [35170] 8722. Komatsu, H.; Iida, S.; Yamamoto, K.; Mikuni, C.; Nitta, M.; Takahashi, T.; Ueda, R.; Seto, M.: A variant chromosome translocation at 11q13 identifying PRAD1/cyclin D1 as the BCL-1 gene. *Blood* 84: 1226-1231, 1994.
- [35171] 8723. Kong, S.; Amos, C. I.; Luthra, R.; Lynch, P. M.; Levin,

B.; Frazier, M. L.: Effects of cyclin D1 polymorphism on age of onset of hereditary nonpolyposis colorectal cancer. *Cancer Res.* 60: 249–252, 2000.

- [35172] 8724. Kong, S.; Wei, Q.; Amos, C. I.; Lynch, P. M.; Zong, J.; Frazier, M. L.: Cyclin D1 polymorphism and increased risk of colorectal cancer at young age. *J. Nat. Cancer Inst.* 93: 1106–1108, 2001.
- [35173] 8725. Ma, C.; Papermaster, D.; Cepko, C. L.: A unique pattern of photoreceptor degeneration in cyclin D1 mutant mice. *Proc. Nat. Acad. Sci.* 95: 9938–9943, 1998.
- [35174] 8726. Motokura, T.; Bloom, T.; Kim, H. G.; Juppner, H.; Ruderhans, J. V.; Kronenberg, H. M.; Arnold, A.: A novel cyclin encoded by a *bcl1*-linked candidate oncogene. *Nature* 350: 512–515, 1991.
- [35175] 8727. Muller, H.; Lukas, J.; Schneider, A.; Warthoe, P.; Bartek, J.; Eilers, M.; Strauss, M.: Cyclin D1 expression is regulated by the retinoblastoma protein. *Proc. Nat. Acad. Sci.* 91: 2945–2949, 1994.
- [35176] 8728. Richard, C. W., III; Withers, D. A.; Meeker, T. C.; Maurer, S.; Evans, G. A.; Myers, R. M.; Cox, D. R.: A radiation hybrid map of the proximal long arm of human chromosome 11 containing the multiple endocrine neoplasia type I (MEN-1) and *bcl-1* disease loci. *Am. J. Hum. Genet.*

49: 1189–1196, 1991.

- [35177] 8729.Rimokh, R.; Berger, F.; Bastard, C.; Klein, B.; French, M.; Archimbaud, E.; Rouault, J. P.; Santa Lucia, B.; Duret, L.; Vuillaume, M.; et al.: Rearrangement of CCND1 (BCL1/PRAD1) 3-prime untranslated region in mantle-cell lymphomas and t(11q13)-associated leukemias. *Blood* 83:3689–3696, 1994.
- [35178] 8730.Rimokh, R.; Berger, F.; Delsol, G.; Charrin, C.; Bertheas, M.F.; French, M.; Garoscio, M.; Felman, P.; Coiffier, C.; Bryon, P.A.: Rearrangement and overexpression of the BCL-1/PRAD-1 gene in intermediate lymphocytic lymphomas and in t(11q13)-bearing leukemias. *Blood* 81:3063–3067, 1993.
- [35179] 8731.Rosenberg, C. L.; Kim, H. G.; Shows, T. B.; Kronenberg, H. M.; Arnold, A.: Rearrangement and overexpression of D11S287E, a candidate oncogene on chromosome 11q13 in benign parathyroid tumors. *Oncogene* 6:449–453, 1991.
- [35180] 8732.Sicinski, P.; Donaher, J. L.; Parker, S. B.; Li, T.; Fazeli, A.; Gardner, H.; Haslam, S. Z.; Bronson, R. T.; Elledge, S. J.; Weinberg, R. A.: Cyclin D1 provides a link between development and oncogenesis in the retina and breast. *Cell* 82: 621–630, 1995.

- [35181] 8733.Szepietowski, P.; Perucca-Lostanlen, D.; Gaudray, P.: Mappinggenes according to their amplification status in tumor cells: contributionto the map of 11q13. Genomics 16: 745–750, 1993.
- [35182] 8734.Clark, T. G.; Conway, S. J.; Scott, I. C.; Labosky, P. A.; Winnier,G.; Bundy, J.; Hogan, B. L. M.; Greenspan, D. S.: The mammalian Tolloid–like1 gene, Tll1, is necessary for normal septation and positioning ofthe heart. Development 126: 126:–2631–2642, 1999.
- [35183] 8735.Scott, I. C.; Clark, T. G.; Takahara, K.; Hoffman, G. G.; Eddy,R. L.; Haley, L. L.; Shows, T. B.; Greenspan, D. S.: Assignment ofTLL1 and TLL2, which encode human BMP–1/tolloid–related metalloproteases,to chromosomes 4q32–q33 and 10q23–q24 and assignment of murine Tll2to chromosome 19. Cytogenet. Cell Genet. 86: 64–65, 1999.
- [35184] 8736.Takahara, K.; Brevard, R.; Hoffman, G. G.; Suzuki, N.; Greenspan,D. S.: Characterization of a novel gene product (mammalian tolloid–like)with high sequence similarity to mammalian tolloid/bone morphogeneticprotein–1. Genomics 34: 157–165, 1996.
- [35185] 8737.Bauer, W. O.; Nanda, I.; Beck, G.; Schmid, M.; Jakob, F.: Humanpuromycin–sensitive aminopeptidase: cloning of

3-prime UTR, evidence for a polymorphism at aa 140 and refined chromosomal localization to 17q21. *Cytogenet. Cell Genet.* 92: 221–224, 2001.

- [35186] 8738. Huber, G.; Thompson, A.; Gruninger, F.; Mechler, H.; Hochstrasser, R.; Hauri, H.-P.; Malherbe, P.: cDNA cloning and molecular characterization of human brain metalloprotease MP100: a beta-secretase candidate? *J. Neurochem.* 72: 1215–1223, 1999.
- [35187] 8739. Osada, T.; Sakaki, Y.; Takeuchi, T.: Puromycin-sensitive aminopeptidase gene (Psa) maps to mouse chromosome 11. *Genomics* 56: 361–362, 1999.
- [35188] 8740. Schonlein, C.; Loffler, J.; Huber, G.: Purification and characterization of a novel metalloprotease from human brain with the ability to cleave substrates derived from the N-terminus of beta-amyloid protein. *Biochem. Biophys. Res. Commun.* 201: 45–53, 1994.
- [35189] 8741. Thompson, M. W.; Tobler, A.; Fontana, A.; Hersh, L. B.: Cloning and analysis of the gene for the human puromycin-sensitive aminopeptidase. *Biochem. Biophys. Res. Commun.* 258: 234–240, 1999.
- [35190] 8742. Tobler, A. R.; Constam, D. B.; Schmitt-Graff, A.; Malipiero, U.; Schlapbach, R.; Fontana, A.: Cloning of the human puromycin-sensitive aminopeptidase and evidence

for expression in neurons. *J. Neurochem.* 68:889–897, 1997.

- [35191] 8743. Zhang, Y.; Cai, X.; Schlegelberger, B.; Zheng, S.: Assignment of human putative tumor suppressor genes ST13 (alias SNC6) and ST14 (alias SNC19) to human chromosome bands 22q13 and 11q24–q25 by *in situ* hybridization. *Cytogenet. Cell Genet.* 83: 56–57, 1998.
- [35192] 8744. Friedrich, R.; Fuentes–Prior, P.; Ong, E.; Coombs, G.; Hunter, M.; Oehler, R.; Pierson, D.; Gonzalez, R.; Huber, R.; Bode, W.; Madison, E. L.: Catalytic domain structures of MT–SP1/matriptase, a matrix–degrading transmembrane serine proteinase. *J. Biol. Chem.* 277: 2160–2168, 2002.
- [35193] 8745. Lin, C.–Y.; Wang, J.–K.; Torri, J.; Dou, L.; Sang, Q. A.; Dickson, R. B.: Characterization of a novel, membrane–bound, 80–kDa matrix–degrading protease from human breast cancer cells: monoclonal antibody production, isolation, and localization. *J. Biol. Chem.* 272: 9147–9152, 1997.
- [35194] 8746. Gacy, A. M.; Goellner, G. M.; Spiro, C.; Chen, X.; Gupta, G.; Bradbury, E. M.; Dyer, R. B.; Mikesell, M. J.; Yao, J. Z.; Johnson, A. J.; Richter, A.; Melancon, S. B.; McMurray, C. T.: GAA instability in Friedreich's ataxia shares a common, DNA–directed and intraallelic mechanism with other

trinucleotide diseases. *Molec. Cell* 1: 583–593,1998.

- [35195] 8747.Gordon, D. M.; Kogan, M.; Knight, S. A. B.; Dancis, A.; Pain,D.: Distinct roles for two N-terminal cleaved domains in mitochondrialimport of the yeast frataxin homolog, Yfh1p. *Hum. Molec. Genet.* 10:259–269, 2001.
- [35196] 8748.Koenig, M.; Mandel, J.–L.: Deciphering the cause of Friedreichataxia. *Curr. Opin. Neurobiol.* 7: 689–694, 1997.
- [35197] 8749.Montermini, L.; Rodius, F.; Pianese, L.; Molto, M. D.; Cossee,M.; Campuzano, V.; Cavalcanti, F.; Monticelli, A.; Palau, F.; Gyapay,G.; Wenhert, M.; Zara, F.; Patel, P. I.; Cocozza, S.; Koenig, M.;Pandolfo, M.: The Friedreich ataxia critical region spans a 150–kbinterval on chromosome 9q13. *Am. J. Hum. Genet.* 57: 1061–1067, 1995.
- [35198] 8750.Pandolfo, M.: Personal Communication. Montreal, Canada 4/28/1997.
- [35199] 8751.Ristow, M.; Pfister, M. F.; Yee, A. J.; Schubert, M.; Michael,L.; Zhang, C.–Y.; Ueki, K.; Michael, M. D., II; Lowell, B. B.; Kahn,C. R.: Frataxin activates mitochondrial energy conversion and oxidativephosphorylation. *Proc. Nat. Acad. Sci.* 97: 12239–12243, 2000.
- [35200] 8752.Sakamoto, N.; Chastain, P. D.; Parniewski, P.; Ohshima, K.; Pandolfo,M.; Griffith, J. D.; Wells, R. D.: Sticky

DNA: self-association properties of long GAA-TTC repeats in R-R-Y triplex structures from Friedrich's ataxia. *Molec. Cell* 3: 465-475, 1999.

[35201] 8753. Santos, M. M.; Ohshima, K.; Pandolfo, M.: Frataxin deficiency enhances apoptosis in cells differentiating into neuroectoderm. *Hum. Molec. Genet.* 10: 1935-1944, 2001.

[35202] 8754. Shoichet, S. A.; Baumer, A. T.; Stamenkovic, D.; Sauer, H.; Pfeiffer, A. F. H.; Kahn, C. R.; Muller-Wieland, D.; Richter, C.; Ristow, M.: Frataxin promotes antioxidant defense in a thiol-dependent manner resulting in diminished malignant transformation in vitro. *Hum. Molec. Genet.* 11: 815-821, 2002.

[35203] 8755. Wilson, R. B.; Roof, D. M.: Respiratory deficiency due to loss of mitochondrial DNA in yeast lacking the frataxin homologue. *Nature Genet.* 16: 352-357, 1997.

[35204] 8756. Zuhlke, C.; Laccone, F.; Cossee, M.; Kohlschutter, A.; Koenig, M.; Schwinger, E.: Mutation of the start codon in the FRDA1 gene: linkage analysis of three pedigrees with the ATG to ATT transversion points to a unique common ancestor. *Hum. Genet.* 103: 102-105, 1998.

[35205] 8757. Fernandez-Gonzalez, A.; La Spada, A. R.; Treadaway, J.; Higdon, J. C.; Harris, B. S.; Sidman, R. L.; Morgan,

J. I.; Zuo, J.: Purkinjecell degeneration (pcd) phenotypes caused by mutations in the axotomy-induced gene, Nna1. Science 295: 1904–1906, 2002.

[35206] 8758. Harris, A.; Morgan, J. I.; Pecot, M.; Soumare, A.; Osborne, A.; Soares, H. D.: Regenerating motor neurons express Nna1, a novel ATP/GTP-binding protein related to zinc carboxypeptidases. Molec. Cell. Neurosci. 16:578–596, 2000.

[35207] 8759. Yuan, C.-X.; Ito, M.; Fondell, J. D.; Fu, Z.-Y.; Roeder, R. G.: The TRAP220 component of a thyroid hormone receptor-associated protein (TRAP) coactivator complex interacts directly with nuclear receptors in a ligand-dependent fashion. Proc. Nat. Acad. Sci. 95: 7939–7944, 1998.

[35208] 8760. Zhang, J.; Fondell, J. D.: Identification of mouse TRAP100: a transcriptional coregulatory factor for thyroid hormone and vitamin D receptors. Molec. Endocr. 13: 1130–1140, 1999.

[35209] 8761. Csoka, T. B.; Frost, G. I.; Heng, H. H. Q.; Scherer, S. W.; Mohapatra, G.; Stern, R.: The hyaluronidase gene HYAL1 maps to chromosome 3p21.2–p21.3 in human and 9F1–F2 in mouse, a conserved candidate tumor suppressor locus. Genomics 48: 63–70, 1998.

- [35210] 8762.Frost, G. I.; Csoka, T. B.; Wong, T.; Stern, R.: Purification, cloning, and expression of human plasma hyaluronidase. *Biochem. Biophys. Res. Commun.* 236: 10–15, 1997.
- [35211] 8763.Laurent, T. C.; Fraser, J. R. E.: Hyaluronan. *FASEB J.* 6: 2397–2404, 1992.
- [35212] 8764.Zegerman, P.; Bannister, A. J.; Kouzarides, T.: The putative tumour suppressor Fus-2 is an N-acetyltransferase. *Oncogene* 19: 161–163, 2000.
- [35213] 8765.Brodbeck, J.; Davies, A.; Courtney, J.-M.; Meir, A.; Balaguero, N.; Canti, C.; Moss, F. J.; Page, K. M.; Pratt, W. S.; Hunt, S. P.; Barclay, J.; Rees, M.; Dolphin, A. C.: The ducky mutation in *Cacna2d2* results in altered Purkinje cell morphology and is associated with the expression of a truncated alpha-2/delta-2 protein with abnormal function. *J. Biol. Chem.* 277: 7684–7693, 2002.
- [35214] 8766.Gao, B.; Sekido, Y.; Maximov, A.; Saad, M.; Forgacs, E.; Latif, F.; Wei, M. H.; Lerman, M.; Lee, J.-H.; Perez-Reyes, E.; Bezprozvanny, I.; Minna, J. D.: Functional properties of a new voltage-dependent calcium channel alpha-2/delta auxiliary subunit gene (*CACNA2D2*). *J. Biol. Chem.* 275: 12237–12242, 2000.
- [35215] 8767.Angrand, P.-O.; Apiou, F.; Stewart, A. F.; Dutrillaux,

B.; Losson,R.; Chambon, P.: NSD3, a new SET domain-containing gene, maps to8p12 and is amplified in human breast cancer cell lines. *Genomics* 74:79–88, 2001.

- [35216] 8768.Stec, I.; van Ommen, G.-J. B.; den Dunnen, J. T.: WHSC1L1, onhuman chromosome 8p11.2, closely resembles WHSC1 and maps to a duplicatedregion shared with 4p16.3. *Genomics* 76: 5–8, 2001.
- [35217] 8769.Drabkin, H. A.; West, J. D.; Hotfilder, M.; Heng, Y. M.; Erickson,P.; Calvo, R.; Dalmau, J.; Gemmill, R. M.; Sablitzky, F.: DEF-3(g16/NY-LU-12),an RNA binding protein from the 3p21.3 homozygous deletion regionin SCLC. *Oncogene* 18: 2589–2597, 1999.
- [35218] 8770.Gure, A. O.; Altorki, N. K.; Stockert, E.; Scanlan, M. J.; Old,L. J.; Chen, Y.-T.: Human lung cancer antigens recognized by autologousantibodies: definition of a novel cDNA derived from the tumor suppressorgene locus on chromosome 3p21.3. *Cancer Res.* 58: 1034–1041, 1998.
- [35219] 8771.Timmer, T.; Terpstra, P.; van den Berg, A.; Veldhuis, P. M. J.F.; Elst, A. T.; Voutsinas, G.; Hulsbeek, M. M. F.; Draaijers, T.G.; Looman, M. W. G.; Kok, K.; Naylor, S. L.; Buys, C. H. C. M.:A comparison of genomic structures and expression patterns of twoclosely related flanking genes in a critical lung cancer region at3p21.3. *Europ. J. Hum.*

Genet. 7: 478–486, 1999.

[35220] 8772.Singh, S.; Chao, L. Y.; Mishra, R.; Davies, J.; Saunders, G. F.: Missense mutation at the C-terminus of PAX6 negatively modulates homeodomain function. Hum. Molec. Genet. 10: 911–918, 2001.

[35221] 8773.Singh, S.; Mishra, R.; Arango, N. A.; Deng, J. M.; Behringer, R. R.; Saunders, G. F.: Iris hypoplasia in mice that lack the alternatively spliced Pax6(5a) isoform. Proc. Nat. Acad. Sci. 99: 6812–6815, 2002.

[35222] 8774.Singh, S.; Tang, H. K.; Lee, J.-Y.; Saunders, G. F.: Truncation mutations in the transactivation region of PAX6 result in dominant-negative mutants. J. Biol. Chem. 273: 21531–21541, 1998.

[35223] 8775.Ton, C. C. T.; Miwa, H.; Saunders, G. F.: Small eye (Sey): cloning and characterization of the murine homolog of the human aniridia gene. Genomics 13:251–256, 1992.

[35224] 8776.Walther, C.; Gruss, P.: Pax-6, a murine paired box gene, is expressed in the developing CNS. Development 113: 1435–1449, 1991.

[35225] 8777.Wawersik, S.; Maas, R. L.: Vertebrate eye development as modeled in Drosophila. Hum. Molec. Genet. 9: 917–925, 2000.

[35226] 8778.Xu, W.; Kozak, C. A.; Desnick, R. J.: Uroporphyrino-

gen-III synthase:molecular cloning, nucleotide sequence, expression of a mouse full-lengthcDNA, and its localization on mouse chromosome 7. *Genomics* 26: 556–562,1995.

- [35227] 8779.Xu, W.; Warner, C. A.; Desnick, R. J.: Congenital erythropoieticporphyria: identification and expression of 10 mutations in the uroporphyrinogenIII synthase gene. *J. Clin. Invest.* 95: 905–912, 1995.
- [35228] 8780.Winfield, S. L.; Tayebi, N.; Martin, B. M.; Ginns, E. I.; Sidransky,E.: Identification of three additional genes contiguous to the glucocerebrosidaselocus on chromosome 1q21: implications for Gaucher disease. *GenomeRes.* 7: 1020–1026, 1997.
- [35229] 8781.Kawasawa, Y.; Kume, K.; Izumi, T.; Shimizu, T.: Mammalian PSP24s(alpha and beta isoforms) are not responsive to lysophosphatidic acidin mammalian expression systems. *Biochem. Biophys. Res. Commun.* 276:957–964, 2000.
- [35230] 8782.Kawasawa, Y.; Kume, K.; Nakade, S.; Haga, H.; Izumi, T.; Shimizu,T.: Brain-specific expression of novel G-protein-coupled receptors,with homologies to *Xenopus* PSP24 and human GPR45. *Biochem. Biophys.Res. Commun.* 276: 952–956, 2000.

- [35231] 8783.Lee, D. K.; George, S. R.; Cheng, R.; Nguyen, T.; Liu, Y.; Brown,M.; Lynch, K. R.; O'Dowd, B. F.: Identification of four novel humanG protein–coupled receptors expressed in the brain. *Molec. BrainRes.* 86: 13–22, 2001.
- [35232] 8784.Cikos, S.; Gregor, P.; Koppel, J.: Cloning of a novel biogenicamine receptor–like G protein–coupled receptor expressed in humanbrain. *Biochim. Biophys. Acta* 1521: 66–72, 2001.
- [35233] 8785.Bascom, R. A.; Srinivasan, S.; Nussbaum, R. L.: Identificationand characterization of golgin–84, a novel Golgi integral membraneprotein with a cytoplasmic coiled–coil domain. *J. Biol. Chem.* 274:2953–2962, 1999. Note: Erratum: *J. Biol. Chem* 274: 12950 only, 1999.
- [35234] 8786.Ishizaka, Y.; Ochiai, M.; Tahira, T.; Sugimura, T.; Nagao, M.:Activation of the ret–II oncogene without a sequence encoding a transmembranedomain and transforming activity of two ret–II oncogene products differingin carboxy–termini due to alternative splicing. *Oncogene* 4: 789–794,1989.
- [35235] 8787.Klugbauer, S.; Demidchik, E. P.; Lengfelder, E.; Rabes, H. M.:Detection of a novel type of RET rearrangement (PTC5) in thyroid carcinomasafter Chernobyl and analysis of the involved RET–fused gene RFG5. *CancerRes.*

58: 198–203, 1998.

- [35236] 8788.Jiang, J. C.; Kirchman, P. A.; Zagulski, M.; Hunt, J.; Jazwinski, S. M.: Homologs of the yeast longevity gene LAG1 in *Caenorhabditis elegans* and human. *Genome Res.* 8: 1259–1272, 1998.
- [35237] 8789.Koivisto, U.–M.; Hubbard, A. L.; Mellman, I.: A novel cellular phenotype for familial hypercholesterolemia due to a defect in polarized targeting of LDL receptor. *Cell* 105: 575–585, 2001.
- [35238] 8790.Koivisto, U.–M.; Turtola, H.; Aalto–Setälä, K.; Top, B.; Frants, R. R.; Kovanen, P. T.; Syvänen, A.–C.; Kontula, K.: The familial hypercholesterolemia (FH)–North Karelia mutation of the low density lipoprotein receptor gene deletes seven nucleotides of exon 6 and is a common cause of FH in Finland. *J. Clin. Invest.* 90: 219–228, 1992.
- [35239] 8791.Koivisto, U.–M.; Viikari, J. S.; Kontula, K.: Molecular characterization of minor gene rearrangements in Finnish patients with heterozygous familial hypercholesterolemia: identification of two common missense mutations (Gly823–to–Asp and Leu380–to–His) and eight rare mutations of the LDL receptor gene. *Am. J. Hum. Genet.* 57: 789–797, 1995.
- [35240] 8792.Kotze, M. J.; Langenhoven, E.; Warnich, L.; du

Plessis, L.; Retief, A. E.: The molecular basis and diagnosis of familial hypercholesterolaemia in South African Afrikaners. *Ann. Hum. Genet.* 55: 115–121, 1991.

[35241] 8793. Kotze, M. J.; Theart, L.; Peeters, A.; Langenhoven, E.: A denovo duplication in the low density lipoprotein receptor gene. *Hum. Mutat.* 6: 181–183, 1995.

[35242] 8794. Kotze, M. J.; Warnich, L.; Langenhoven, E.; du Plessis, L.; Retief, A. E.: An exon 4 mutation identified in the majority of South African familial hypercholesterolaemics. *J. Med. Genet.* 27: 298–302, 1990.

[35243] 8795. Landsberger, D.; Meiner, V.; Reshef, A.; Levy, Y.; van der Westhuyzen, D. R.; Coetzee, G. A.; Leitersdorf, E.: A nonsense mutation in the LDL receptor gene leads to familial hypercholesterolemia in the Druze sect. *Am. J. Hum. Genet.* 50: 427–433, 1992.

[35244] 8796. Langlois, S.: Personal Communication. Vancouver, British Columbia, Canada 1989.

[35245] 8797. Langlois, S.; Kastelein, J. J. P.; Hayden, M. R.: Characterization of six partial deletions in the low-density-lipoprotein (LDL) receptor gene causing familial hypercholesterolemia (FH). *Am. J. Hum. Genet.* 43:60–68, 1988.

[35246] 8798. Lee, W. K.; Haddad, L.; Macleod, M. J.; Dorrance, A.

M.; Wilson, D. J.; Gaffney, D.; Dominiczak, M. H.; Packard, C. J.; Day, I. N.; Humphries, S. E.; Dominiczak, A. F.: Identification of a common low density lipoprotein receptor mutation (C163Y) in the West of Scotland. *J. Med. Genet.* 35: 573–578, 1998.

- [35247] 8799. Lehrman, M. A.; Goldstein, J. L.; Russell, D. W.; Brown, M. S.: Duplication of seven exons in LDL receptor gene caused by Alu–Alu recombination in a subject with familial hypercholesterolemia. *Cell* 48:827–835, 1987.
- [35248] 8800. Lehrman, M. A.; Russell, D. W.; Goldstein, J. L.; Brown, M. S.: Alu–Alu recombination deletes splice acceptor sites and produces secreted low density lipoprotein receptor in a subject with familial hypercholesterolemia. *J. Biol. Chem.* 262: 3354–3361, 1987.
- [35249] 8801. Lehrman, M. A.; Russell, D. W.; Goldstein, J. L.; Brown, M. S.: Exon–Alu recombination deletes 5 kilobases from the low density lipoprotein receptor gene, producing a null phenotype in familial hypercholesterolemia. *Proc. Nat. Acad. Sci.* 83: 3679–3683, 1986.
- [35250] 8802. Lehrman, M. A.; Schneider, W. J.; Sudhof, T. C.; Brown, M. S.; Goldstein, J. L.; Russell, D. W.: Mutation in LDL receptor: Alu–Alu recombination deletes exons encoding transmembrane and cytoplasmic domains. *Science*

227: 140–146, 1985.

- [35251] 8803. Leidersdorf, E.; Hobbs, H. H.: Personal Communication. Dallas, Tex. 12/1990.
- [35252] 8804. Leidersdorf, E.; Hobbs, H. H.; Fourie, A. M.; Jacobs, M.; van der Westhuyzen, D. R.; Coetzee, G. A.: Deletion in the first cysteine-rich repeat of low-density lipoprotein receptor impairs its transport but not lipoprotein binding in fibroblasts from a subject with familial hypercholesterolemia. *Proc. Nat. Acad. Sci.* 85: 7912–7916, 1988.
- [35253] 8805. Leidersdorf, E.; Tobin, E. J.; Davignon, J.; Hobbs, H. H.: Common low-density lipoprotein receptor mutations in the French Canadian population. *J. Clin. Invest.* 85: 1014–1023, 1990.
- [35254] 8806. Leidersdorf, E.; van der Westhuyzen, D. R.; Coetzee, G. A.; Hobbs, H. H.: Two common low density lipoprotein receptor gene mutations cause familial hypercholesterolemia in Afrikaners. *J. Clin. Invest.* 84: 954–961, 1989.
- [35255] 8807. Lelli, N.; Ghisellini, M.; Calandra, S.; Gaddi, A.; Ciarrocchi, A.; Coviello, D. A.; Bertolini, S.: Duplication of exons 13, 14 and 15 of the LDL-receptor gene in a patient with heterozygous familial hypercholesterolemia. *Hum. Genet.* 86: 359–362, 1991.
- [35256] 8808. Leren, T. P.; Solberg, K.; Rodningen, O. K.; Tonstad,

S.; Ose, L.: Two founder mutations in the LDL receptor gene in Norwegian familial hypercholesterolemia subjects. *Atherosclerosis* 111: 175–182, 1994.

[35257] 8809. Li, H.; Gyllenstein, U. B.; Cui, X.; Saiki, R. K.; Erlich, H. A.; Arnheim, N.: Amplification and analysis of DNA sequences in single human sperm and diploid cells. *Nature* 335: 414–417, 1988.

[35258] 8810. Downes, G. B.; Gilbert, D. J.; Copeland, N. G.; Gautam, N.; Jenkins, N. A.: Chromosomal mapping of five mouse G protein gamma subunits. *Genomics* 57: 173–176, 1999.

[35259] 8811. Modarressi, M. H.; Taylor, K. E.; Wolfe, J.: Cloning, characterization, and mapping of the gene encoding the human G protein gamma-2 subunit. *Biochem. Biophys. Res. Commun.* 272: 610–615, 2000.

[35260] 8812. Yu, Y.; Zhang, C.; Zhou, G.; Wu, S.; Qu, X.; Wei, H.; Xing, G.; Dong, C.; Zhai, Y.; Wan, J.; Ouyang, S.; Li, L.; Zhang, S.; Zhou, K.; Zhang, Y.; Wu, C.; He, F.: Gene expression profiling in human fetal liver and identification of tissue- and developmental-stage-specific genes through compiled expression profiles and efficient cloning of full-length cDNAs. *Genome Res.* 11: 1392–1403, 2001.

[35261] 8813. Ericsson, J.; Greene, J. M.; Carter, K. C.; Shell, B. K.;

Duan,D. R.; Florence, C.; Edwards, P. A.: Human geranyl-geranyl diphosphatesynthase: isolation of the cDNA, chromosomal mapping and tissue expression. J.Lipid Res. 39: 1731–1739, 1998.

[35262] 8814.Kainou, T.; Kawamura, K.; Tanaka, K.; Matsuda, H.; Kawamukai, M.: Identification of the GGPS1 genes encoding geranylgeranyl diphosphatesynthases from mouse and human. Biochim. Biophys. Acta 1437: 333–340,1999.

[35263] 8815.Kuzuguchi, T.; Morita, Y.; Sagami, I.; Sagami, H.; Ogura, K.:Human geranylgeranyl diphosphate synthase: cDNA cloning and expression. J.Biol. Chem. 274: 5888–5894, 1999.

[35264] 8816.Vicent, D.; Maratos–Flier, E.; Kahn, C. R.: The branch point enzymeof the mevalonate pathway for protein prenylation is overexpressedin the ob/ob mouse and induced by adipogenesis. Molec. Cell. Biol. 20:2158–2166, 2000.

[35265] 8817.Chen, C.–K.; Zhang, K.; Church–Kopish, J.; Huang, W.; Zhang, H.;Chen, Y.–J.; Frederick, J. M.; Baehr, W.: Characterization of humanGRK7 as a potential cone opsin kinase. Molec. Vision 7: 305–313,2001.

[35266] 8818.Weiss, E. R.; Ducceschi, M. H.; Horner, T. J.; Li, A.; Craft, C.M.; Osawa, S.: Species–specific differences in ex–

pression of G-protein-coupled receptor kinase (GRK) 7 and GRK1 in mammalian cone photoreceptor cells: implications for cone cell phototransduction. *J. Neurosci.* 21: 9175–9184, 2001.

[35267] 8819. Borregaard, N.; Cowland, J. B.: Granules of the human neutrophilic polymorphonuclear leukocyte. *Blood* 89: 3503–3521, 1997.

[35268] 8820. Chang, K. S.; Schroeder, W.; Siciliano, M. J.; Thompson, L. H.; McCredie, K.; Beran, M.; Freireich, E. J.; Liang, J. C.; Trujillo, J. M.; Stass, S. A.: The localization of the human myeloperoxidase gene is in close proximity to the translocation breakpoint in acute promyelocytic leukemia. *Leukemia* 1: 458–462, 1987.

[35269] 8821. DeLeo, F. R.; Goedken, M.; McCormick, S. J.; Nauseef, W. M.: A novel form of hereditary myeloperoxidase deficiency linked to endoplasmic reticulum/proteasome degradation. *J. Clin. Invest.* 101: 2900–2909, 1998.

[35270] 8822. Eiserich, J. P.; Baldus, S.; Brennan, M.-L.; Ma, W.; Zhang, C.; Tousson, A.; Castro, L.; Lusis, A. J.; Nauseef, W. M.; White, C. R.; Freeman, B. A.: Myeloperoxidase, a leukocyte-derived vascular NO oxidase. *Science* 296: 2391–2394, 2002.

[35271] 8823. Inazawa, J.; Inoue, K.; Nishigaki, H.; Tsuda, S.; Tani-

waki, M.; Misawa, S.; Abe, T.: Assignment of the human myeloperoxidase gene(MPO) to bands q21.3–q23 of chromosome 17. *Cytogenet. Cell Genet.* 50:135–136, 1989.

[35272] 8824. Johnson, K.; Gemperlein, I.; Hudson, S.; Shane, S.; Rovera, G.: Complete nucleotide sequence of the human myeloperoxidase gene. *Nucleic Acids Res.* 17: 7985–7986, 1989.

[35273] 8825. Kizaki, M.; Miller, C. W.; Selsted, M. E.; Koeffler, H. P.: Myeloperoxidase(MPO) gene mutation in hereditary MPO deficiency. *Blood* 83: 1935–1940, 1994.

[35274] 8826. Klebanoff, S. J.: Myeloperoxidase. *Proc. Assoc. Am. Physicians* 111:383–389, 1999.

[35275] 8827. Kudoh, J.; Minoshima, S.; Hashinaka, K.; Nishio, C.; Yamada, M.; Shimizu, Y.; Shimizu, N.: Assignment of the myeloperoxidase gene MPO to human chromosome 17 using somatic cell hybrids and flow-sorted chromosomes. *Jpn. J. Hum. Genet.* 33: 315–324, 1988.

[35276] 8828. Kudoh, J.; Minoshima, S.; Hashinaka, K.; Nishio, C.; Yamada, M.; Shimizu, Y.; Shimizu, N.: Assignment of the myeloperoxidase (MPO) gene to human chromosome 17. (Abstract) *Cytogenet. Cell Genet.* 46:641–642, 1987.

[35277] 8829. Law, D. J.; Prasad, M. A.; King, S. E.; Spranger, K. D.; Lee, Y. H.; Fox, R. E.; Collins, E. E.; Gebuhr, T. C.; Miller, D.

E.; Petty, E. M.: Localization of the human estrogen-responsive finger protein(EFP) gene (ZNF147) within a YAC contig containing the myeloperoxidase(MPO) gene. *Genomics* 28: 361–363, 1995.

[35278] 8830. Williams, M.; Lyu, M.-S.; Yang, Y.-L.; Lin, E. P.; Dunbrack, R.; Birren, B.; Cunningham, J.; Hunter, K.: *Ier5*, a novel member of the slow-kinetics immediate-early genes. *Genomics* 55: 327–334, 1999.

[35279] 8831. Arce, I.; Roda-Navarro, P.; Montoya, M. C.; Hernanz-Falcon, P.; Puig-Kroger, A.; Fernandez-Ruiz, E.: Molecular and genomic characterization of human DLEC, a novel member of the C-type lectin receptor gene family preferentially expressed on monocyte-derived dendritic cells. *Europ. J. Immun.* 31: 2733–2740, 2001.

[35280] 8832. Dzionek, A.; Sohma, Y.; Nagafune, J.; Cella, M.; Colonna, M.; Facchetti, F.; Gunther, G.; Johnston, I.; Lanza-vecchia, A.; Nagasaka, T.; Okada, T.; Vermi, W.; Winkels, G.; Yamamoto, T.; Zysk, M.; Yamaguchi, Y.; Schmitz, J.: BDCA-2, a novel plasmacytoid dendritic cell-specific type II C-type lectin, mediates antigen capture and is a potent inhibitor of interferon alpha/beta induction. *J. Exp. Med.* 194: 1823–1834, 2001.

[35281] 8833. Antonarakis, S. E.: Personal Communication. Balti-

more, Md. 3/25/2002.

- [35282] 8834.McKemy, D. D.; Neuhausser, W. M.; Julius, D.: Identification of a cold receptor reveals a general role for TRP channels in thermosensation. *Nature* 416:52–58, 2002.
- [35283] 8835.Peier, A. M.; Moqrich, A.; Hergarden, A. C.; Reeve, A. J.; Andersson, D. A.; Story, G. M.; Earley, T. J.; Dragoni, I.; McIntyre, P.; Bevan, S.; Patapoutian, A.: A TRP channel that senses cold stimuli and menthol. *Cell* 108:705–715, 2002.
- [35284] 8836.Ashery-Padan, R.; Marquardt, T.; Zhou, X.; Gruss, P.: Pax6 activity in the lens primordium is required for lens formation and for correct placement of a single retina in the eye. *Genes Dev.* 14: 2701–2711, 2000.
- [35285] 8837.Azuma, N.; Yamaguchi, Y.; Handa, H.; Hayakawa, M.; Kanai, A.; Yamada, M.: Missense mutation in the alternative splice region of the PAX6 gene in eye anomalies. *Am. J. Hum. Genet.* 65: 656–663, 1999.
- [35286] 8838.Roginski, R. S.; Mohan Raj, B. K.; Finkernagel, S. W.; Sciorra, L. J.: Assignment of an ionotropic glutamate receptor-like gene (GRINL1A) to human chromosome 15q22.1 by in situ hybridization. *Cytogenet. Cell Genet.* 93: 143–144, 2001.
- [35287] 8839.Wydner, K. S.; Mohan Raj, B. K.; Sciorra, L. J.; Roginski, R. S.: The mouse orthologue of the human ionotropic

glutamate receptor-like gene (GRINL1A) maps to mouse chromosome 9. *Cytogenet. Cell Genet.* 95:240–241, 2001.

[35288] 8840. Akiyama, H.; Hiraki, Y.; Noda, M.; Shigeno, C.; Ito, H.; Nakamura, T.: Molecular cloning and biological activity of a novel Ha-Ras suppressor gene predominantly expressed in skeletal muscle, heart, brain, and bone marrow by differential display using clonal mouse EC cells, ATDC5. *J. Biol. Chem.* 274: 32192–32197, 1999.

[35289] 8841. Ito, H.; Akiyama, H.; Shigeno, C.; Nakamura, T.: Isolation, characterization, and chromosome mapping of a human A-C1 Ha-Ras suppressor gene (HRASLS). *Cytogenet. Cell Genet.* 93: 36–39, 2001.

[35290] 8842. Nakamura, T.; Yamazaki, Y.; Saiki, Y.; Moriyama, M.; Largaespada, D. A.; Jenkins, N. A.; Copeland, N. G.: Evi9 encodes a novel zinc finger protein that physically interacts with BCL6, a known human B-cell proto-oncogene product. *Molec. Cell Biol.* 20: 3178–3186, 2000.

[35291] 8843. Saiki, Y.; Yamazaki, Y.; Yoshida, M.; Katoh, O.; Nakamura, T.: Human EVI9, a homologue of the mouse myeloid leukemia gene, is expressed in the hematopoietic progenitors and down-regulated during myeloid differentiation of HL60 cells. *Genomics* 70: 387–391, 2000.

[35292] 8844. Satterwhite, E.; Sonoki, T.; Willis, T. G.; Harder, L.;

Nowak,R.; Arriola, E. L.; Liu, H.; Price, H. P.; Gesk, S.; Steinemann, D.;Schlegelberger, B.; Oscier, D. G.; Siebert, R.; Tucker, P. W.; Dyer,M. J. S.: The BCL11 gene family: involvement of BCL11A in lymphoid malignancies. *Blood* 98: 3413–3420, 2001.

[35293] 8845.Frohlich, O.; Po, C.; Young, L. G.: Organization of the human gene encoding the epididymis-specific EP2 protein variants and its relationship to defensin genes. *Biol. Reprod.* 64: 1072–1079, 2001.

[35294] 8846.Hamil, K. G.; Sivashanmugam, P.; Richardson, R. T.; Grossman, G.;Ruben, S. M.; Mohler, J. L.; Petrusz, P.; O'rand, M. G.; French, F.S.; Hall, S. H.: HE2–beta and HE2–gamma, new members of an epididymis-specific family of androgen-regulated proteins in the human. *Endocrinology* 141:1245–1253, 2000.

[35295] 8847.Jia, H. P.; Schutte, B. C.; Schudy, A.; Linzmeier, R.; Guthmiller,J. M.; Johnson, G. K.; Tack, B. F.; Mitros, J. P.; Rosenthal, A.;Ganz, T.; McCray, P. B., Jr.: Discovery of new human beta-defensins using a genomics-based approach. *Gene* 263: 211–218, 2001.

[35296] 8848.Li, P.; Chan, H. C.; He, B.; So, S. C.; Chung, Y. W.; Shang, Q.;Zhang, Y.–D.; Zhang, Y.–L.: An antimicrobial peptide gene found in the male reproductive system of

rats. Science 291: 1783–1785, 2001.

- [35297] 8849.Osterhoff, C.; Kirchhoff, C.; Krull, N.; Ivell, R.: Molecularcloning and characterization of a novel human sperm antigen (HE2)specifically expressed in the proximal epididymis. Biol. Reprod. 50:516–525, 1994.
- [35298] 8850.Carim–Todd, L.; Escarceller, M.; Estivill, X.; Sumoy, L.: Cloningof the novel gene TM6SF1 reveals conservation of clusters of paralogousgenes between human chromo–somes 15q24–q26 and 19p13.3–p12. Cytogenet.Cell Genet. 90: 255–260, 2000.
- [35299] 8851.Nagase, T.; Kikuno, R.; Ohara, O.: Prediction of the coding sequencesof unidentified human genes. XXI. The complete sequences of 60 newcDNA clones from brain which code for large proteins. DNA Res. 8:179–187, 2001.
- [35300] 8852.Gress, T. M.; Wallrapp, C.; Frohme, M.; Muller–Pillasch, F.; Lacher,U.; Friess, H.; Buchler, M.; Adler, G.; Ho–heisel, J. D.: Identificationof genes with specific expres–sion in pancreatic cancer by cDNA representationaldiffer–ence analysis. Genes Chromosomes Cancer 19: 97–103, 1997.
- [35301] 8853.Wallrapp, C.; Hahnel, S.; Muller–Pillasch, F.; Burghardt, B.; Iwamura,T.; Ruthenburger, M.; Lerch, M. M.; Adler, G.; Gress, T. M.: A noveltransmembrane serine pro–

tease (TMPRSS3) overexpressed in pancreatic cancer. *Cancer Res.* 60: 2602–2606, 2000.

[35302] 8854. Kiss, H.; Kedra, D.; Kiss, C.; Kost-Alimova, M.; Yang, Y.; Klein, G.; Imreh, S.; Dumanski, J. P.: The LZTFL1 gene is a part of a transcriptional map covering 250 kb within the common eliminated region 1 (C3CER1) in 3p21.3. *Genomics* 73: 10–19, 2001.

[35303] 8855. Quentmeier, H.; Drexler, H. G.; Fleckenstein, D.; Zaborski, M.; Armstrong, A.; Sims, J. E.; Lyman, S. D.: Cloning of human thymic stromal lymphopoietin (TSLP) and signaling mechanisms leading to proliferation. *Leukemia* 15: 1286–1292, 2001.

[35304] 8856. Soumelis, V.; Reche, P. A.; Kanzler, H.; Yuan, W.; Edward, G.; Homey, B.; Gilliet, M.; Ho, S.; Antonenko, S.; Lauerma, A.; Smith, K.; Gorman, D.; Zurawski, S.; Abrams, J.; Menon, S.; McClanahan, T.; de Waal-Malefyt, R.; Bazan, F.; Kastelein, R. A.; Liu, Y.-J.: Human epithelial cells trigger dendritic cell-mediated allergic inflammation by producing TSLP. *Nature Immun.* 3: 673–680, 2002.

[35305] 8857. Guo, D.; Hasham, S.; Kuang, S.-Q.; Vaughan, C. J.; Boerwinkle, E.; Chen, H.; Abuelo, D.; Dietz, H. C.; Basson, C. T.; Shete, S. S.; Milewicz, D. M.: Familial thoracic aortic aneurysms and dissections: genetic heterogeneity with a

major locus mapping to 5q13–14. *Circulation* 103:2461–2468, 2001.

- [35306] 8858. Vaughan, C. J.; Casey, M.; He, J.; Veuglers, M.; Henderson, K.; Guo, D.; Campagna, R.; Roman, M. J.; Milewicz, D. M.; Devereux, R.B.; Basson, C. T.: Identification of a chromosome 11q23.2–q24 locus for familial aortic aneurysm disease, a genetically heterogeneous disorder. *Circulation* 103: 2469–2475, 2001.
- [35307] 8859. Tapon, N.; Harvey, K. F.; Bell, D. W.; Wahrer, D. C. R.; Schiripo, T. A.; Haber, D. A.; Hariharan, I. K.: salvador promotes both cell cycle exit and apoptosis in *Drosophila* and is mutated in human cancer cell lines. *Cell* 110: 467–478, 2002.
- [35308] 8860. Valverde, P.: Cloning, expression, and mapping of hWW45, a novel human WW domain-containing gene. *Biochem. Biophys. Res. Commun.* 276:990–998, 2000.
- [35309] 8861. Wang, X.; McLachlan, J.; Zamore, P. D.; Hall, T. M. T.: Modular recognition of RNA by a human Pumilio-homology domain. *Cell* 110:501–512, 2002.
- [35310] 8862. Ballinger, C. A.; Connell, P.; Wu, Y.; Hu, Z.; Thompson, L. J.; Yin, L.-Y.; Patterson, C.: Identification of CHIP, a novel tetratricopeptide repeat-containing protein that interacts with heat shock proteins and negatively regulates

chaperone functions. *Molec. Cell. Biol.* 19:4535–4545, 1999.

[35311] 8863.Jiang, J.; Ballinger, C. A.; Wu, Y.; Dai, Q.; Cyr, D. M.; Hohfeld,J.; Patterson, C.: CHIP is a U-box-dependent E3 ubiquitin ligase:identification of Hsc70 as a target for ubiquitylation. *J. Biol.Chem.* 276: 42938–42944, 2001.

[35312] 8864.Gaide, O.; Martinon, F.; Micheau, O.; Bonnet, D.; Thome, M.; Tschopp,J.: Carma1, a CARD-containing binding partner of Bcl10, induces Bcl10phosphorylation and NF-kappa-B activation. *FEBS Lett.* 496: 121–127,2001.

[35313] 8865.McAllister-Lucas, L. M.; Inohara, N.; Lucas, P. C.; Ruland, J.;Benito, A.; Li, Q.; Chen, S.; Chen, F. F.; Yamaoka, S.; Verma, I.M.; Mak, T. W.; Nunez, G.: Bimp1, a MAGUK family member linking proteinkinase C activation to Bcl10-mediated NF-kappa-B induction. *J. Biol.Chem.* 276: 30589–30597, 2001.

[35314] 8866.Wang, L.; Guo, Y.; Huang, W.-J.; Ke, X.; Poyet, J.-L.; Manji, G.A.; Merriam, S.; Glucksmann, M. A.; DiStefano, P. S.; Alnemri, E.S.; Bertin, J.: CARD10 is a novel caspase recruitment domain/membrane-associatedguanylate kinase family member that interacts with BCL10 and activatesNF-kappa-B. *J. Biol. Chem.* 276: 21405–21409, 2001.

[35315] 8867.Wagner, D. S.; Gan, L.; Klein, W. H.: Identification of

a differentially expressed RNA helicase by gene trapping.

Biochem. Biophys. Res. Commun. 262:677–684, 1999.

[35316] 8868. Takeuchi, T.; Shuman, M. A.; Craik, C. S.: Reverse biochemistry: use of macromolecular protease inhibitors to dissect complex biological processes and identify a membrane-type serine protease in epithelial cancer and normal tissue. Proc. Nat. Acad. Sci. 96: 11054–11061, 1999.

[35317] 8869. Loux, N.; Benlian, P.; Pastier, D.; Boileau, C.; Cambou, J. P.; Monnier, L.; Percheron, C.; Junien, C.: Recurrent mutation at aa792 in the LDL receptor gene in a French patient. Hum. Genet. 87:373–375, 1991.

[35318] 8870. Le Beau, M. M.; Lemons, R. S.; Rosner, G. L.; Carrino, J. C.; Reid, M. S.; Chisholm, R. L.; Diaz, M. O.; Weil, S. C.: Chromosomal localization of the gene encoding myeloperoxidase. (Abstract) Cytogenet. Cell Genet. 46: 645, 1987.

[35319] 8871. De Deken, X.; Wang, D.; Many, M.-C.; Costagliola, S.; Libert, F.; Vassart, G.; Dumont, J. E.; Miot, F.: Cloning of two human thyroid cDNAs encoding new members of the NADPH oxidase family. J. Biol. Chem. 275: 23227–23233, 2000.

[35320] 8872. Lacroix, L.; Nocera, M.; Mian, C.; Caillou, B.; Virion, A.; Dupuy, C.; Filetti, S.; Bidart, J. M.; Schlumberger, M.: Expression of nicotinamide adenine dinucleotide phos-

phate oxidase flavoprotein DUOX genes and proteins in human papillary and follicular thyroid carcinomas. *Thyroid* 11:1017–1023, 2001.

- [35321] 8873. Dupuy, C.; Ohayon, R.; Valent, A.; Noel-Hudson, M.-S.; Deme, D.; Virion, A.: Purification of a novel flavoprotein involved in the thyroid NADPH oxidase: cloning of the porcine and human cDNAs. *J. Biol. Chem.* 274: 37265–37269, 1999.
- [35322] 8874. Moreno, J. C.; Bikker, H.; Kempers, M. J. E.; van Trotsenburg, A. S. P.; Baas, F.; de Vijlder, J. J. M.; Vulsma, T.; Ris-Stalpers, C.: Inactivating mutations in the gene for thyroid oxidase 2 (THOX2) and congenital hypothyroidism. *New Eng. J. Med.* 347: 95–102, 2002.
- [35323] 8875. Okajima, T.; Fukumoto, S.; Miyazaki, H.; Ishida, H.; Kiso, M.; Furukawa, K.; Urano, T.; Furukawa, K.: Molecular cloning of a novel α -2,3-sialyltransferase (ST3Gal VI) that sialylates type II lactosamine structures on glycoproteins and glycolipids. *J. Biol. Chem.* 274:11479–11486, 1999.
- [35324] 8876. Richardson, J.; Cvekl, A.; Wistow, G.: Pax-6 is essential for lens-specific expression of zeta-crystallin. *Proc. Nat. Acad. Sci.* 92:4676–4680, 1995.
- [35325] 8877. Sander, M.; Neubuser, A.; Kalamaras, J.; Ee, H. C.;

Martin, G.R.; German, M. S.: Genetic analysis reveals that PAX6 is required for normal transcription of pancreatic hormone genes and islet development. *GenesDev.* 11: 1662–1673, 1997.

[35326] 8878. Taniguchi, A.; Kaneta, R.; Morishita, K.; Matsumoto, K.: Gene structure and transcriptional regulation of human Gal beta–1,4(3)GlcNac alpha–2,3–sialyltransferase VI (hST3Gal VI) gene in prostate cancer cell line. *Biochem. Biophys. Res. Commun.* 287: 1148–1156, 2001.

[35327] 8879. Schwientek, T.; Nomoto, M.; Levery, S. B.; Merx, G.; van Kessel, A. G.; Bennett, E. P.; Hollingsworth, M. A.; Clausen, H.: Control of O–glycan branch formation: molecular cloning of human cDNA encoding a novel beta–1,6–N–acetylglucosaminyltransferase forming core 2 and–core 4. *J. Biol. Chem.* 274: 4504–4512, 1999.

[35328] 8880. Chano, T.; et al; et al: Isolation, characterization and mapping of the mouse and human RB1CC1 genes. *Gene* (in–press), 2002.

[35329] 8881. Chano, T.; Kontani, K.; Teramoto, K.; Okabe, H.; Ikegawa, S.: Truncating mutations of RB1CC1 in human breast cancers. *Nature Genet.* 31: 285–288, 2002.

[35330] 8882. Kaneko, M.; Kudo, T.; Iwasaki, H.; Ikehara, Y.; Nishihara, S.; Nakagawa, S.; Sasaki, K.; Shiina, T.; Inoko, H.;

Saitou, N.; Narimatsu, H.: Alpha-1,3-fucosyltransferase (sic) IX (Fuc-TIX) is very highly conserved between human and mouse; molecular cloning, characterization and tissue distribution of human Fuc-TIX. FEBS Lett. 452: 237-242, 1999.

[35331] 8883. Kaneko, M.; Kudo, T.; Iwasaki, H.; Shiina, T.; Inoko, H.; Kozaki, T.; Saitou, N.; Narimatsu, H.: Assignment of the human alpha-1,3-fucosyltransferase IX gene (FUT9) to chromosome band 6q16 by in situ hybridization. Cytogenet. Cell Genet. 86: 329-330, 1999.

[35332] 8884. Lane, J. D.; Lucocq, J.; Pryde, J.; Barr, F. A.; Woodman, P. G.; Allan, V. J.; Lowe, M.: Caspase-mediated cleavage of the stacking protein GRASP65 is required for Golgi fragmentation during apoptosis. J. Cell Biol. 156: 495-509, 2002.

[35333] 8885. Sutterlin, C.; Hsu, P.; Mallabiabarrena, A.; Malhotra, V.: Fragmentation and dispersal of the pericentriolar Golgi complex is required for entry into mitosis in mammalian cells. Cell 109: 359-369, 2002.

[35334] 8886. Davis, A.; Cowell, J. K.: Mutations in the PAX6 gene in patients with hereditary aniridia. Hum. Molec. Genet. 2: 2093-2097, 1993.

[35335] 8887. Gronskov, K.; Rosenberg, T.; Sand, A.; Brondum-

Nielsen, K.: Mutational analysis of PAX6: 16 novel mutations including 5 missense mutations with a mild aniridia phenotype. *Europ. J. Hum. Genet.* 7: 274–286, 1999.

[35336] 8888. Halder, G.; Callaerts, P.; Gehring, W. J.: Induction of ectopic eyes by targeted expression of the eyeless gene in *Drosophila*. *Science* 267:1788–1792, 1995.

[35337] 8889. Hanson, I.; Brown, A.; van Heyningen, V.: A new PAX6 mutation in familial aniridia. *J. Med. Genet.* 32: 488–489, 1995.

[35338] 8890. Hanson, I.; Van Heyningen, V.: Pax6: more than meets the eye. *Trends Genet.* 11: 268–272, 1995.

[35339] 8891. Hanson, I. M.; Fletcher, J. M.; Jordon, T.; Brown, A.; Taylor, D.; Adams, R. J.; Punnett, H. H.; van Heyningen, V.: Mutations at the PAX6 locus are found in heterogeneous anterior segment malformations including Peters' anomaly. *Nature Genet.* 6: 168–173, 1994.

[35340] 8892. Heins, N.; Malatesta, P.; Cecconi, F.; Nakafuku, M.; Tucker, K. L.; Hack, M. A.; Chapouton, P.; Barde, Y.-A.; Gotz, M.: Glial cells generate neurons: the role of the transcription factor Pax6. *Nature Neurosci.* 5: 308–315, 2002.

[35341] 8893. Holmstrom, G. E.; Reardon, W. P.; Baraitser, M.; Elston, J. S.; Taylor, D. S.: Heterogeneity in dominant anterior segment malformations. *Brit. J. Ophthalmol.* 75: 591–597,

1991.

- [35342] 8894.Kioussi, C.; O'Connell, S.; St-Onge, L.; Treier, M.; Gleiberman, A. S.; Gruss, P.; Rosenfeld, M. G.: Pax6 is essential for establishing ventral–dorsal cell boundaries in pituitary gland development. *Proc. Nat. Acad. Sci.* 96: 14378–14382, 1999.
- [35343] 8895.Kleinjan, D. A.; Seawright, A.; Schedl, A.; Quinlan, R. A.; Danes, S.; van Heyningen, V.: Aniridia–associated translocations, DNase hypersensitivity, sequence comparison and transgenic analysis redefine the functional domain of PAX6. *Hum. Molec. Genet.* 10: 2049–2059, 2001.
- [35344] 8896.Lauderdale, J. D.; Wilensky, J. S.; Oliver, E. R.; Walton, D. S.; Glaser, T.: 3–prime deletions cause aniridia by preventing PAX6 gene expression. *Proc. Nat. Acad. Sci.* 97: 13755–13759, 2000.
- [35345] 8897.Marquardt, T.; Ashery-Padan, R.; Andrejewski, N.; Scardigli, R.; Guillemot, F.; Gruss, P.: Pax6 is required for the multipotent state of retinal progenitor cells. *Cell* 105: 43–55, 2001.
- [35346] 8898.Croze, E.; Russell-Harde, D.; Wagner, T. C.; Pu, H.; Pfeffer, L. M.; Perez, H. D.: The human type I interferon receptor: identification of the interferon beta–specific receptor–associated phosphoprotein. *J. Biol. Chem.* 271:

33165–33168, 1996.

[35347] 8899.Croze, E.; Usacheva, A.; Asarnow, D.; Minshall, R.D.; Perez, H.D.; Colamonici, O.: Receptor for activated C-kinase (RACK-1), a WD motif-containing protein, specifically associates with the human type I IFN receptor. *J. Immun.* 165: 5127–5132, 2000.

[35348] 8900.Domanski, P.; Witte, M.; Kellum, M.; Rubinstein, M.; Hackett, R.; Pitha, P.; Colamonici, O. R.: Cloning and expression of a long form of the beta subunit of the interferon alpha/beta receptor that is required for signaling. *J. Biol. Chem.* 270: 21606–21611, 1995.

[35349] 8901.Lutfalla, G.; Holland, S. J.; Cinato, E.; Monneron, D.; Reboul, J.; Rogers, N. C.; Smith, J. M.; Stark, G. R.; Gardiner, K.; Mogensen, K. E.; Kerr, I. M.; Uze, G.: Mutant U5A cells are complemented by an interferon-alpha/beta receptor subunit generated by alternative processing of a new member of a cytokine receptor gene cluster. *EMBO J.* 14: 5100–5108, 1995.

[35350] 8902.Platanias, L. C.; Uddin, S.; Domanski, P.; Colamonici, O. R.: Differences in interferon alpha and beta signaling: interferon beta selectively induces the interaction of the alpha and beta(L) subunits of the type I interferon receptor. *J. Biol. Chem.* 271: 23630–23633, 1996.

- [35351] 8903.Raz, R.; Cheung, K.; Ling, L.; Levy, D. E.: Three distinct loci on human chromosome 21 contribute to interferon- α /beta responsiveness. *Somat.Cell Molec. Genet.* 21: 139–145, 1995.
- [35352] 8904.Klocke, R.; Augustin, A.; Ronsiek, M.; Stief, A.; van der Putten, H.; Jockusch, H.: Dynamin genes Dnm1 and Dnm2 are located on proximal mouse chromosomes 2 and 9, respectively. *Genomics* 41: 290–292, 1997.
- [35353] 8905.Marks, B.; Stowell, M. H. B.; Vallis, Y.; Mills, I. G.; Gibson, A.; Hopkins, C. R.; McMahon, H. T.: GTPase activity of dynamin and resulting conformation change are essential for endocytosis. *Nature* 410:231–235, 2001.
- [35354] 8906.Newman-Smith, E. D.; Shurland, D.-L.; van der Bliek, A. M.: Assignment of the dynamin-1 gene (DNM1) to human chromosome 9q34 by fluorescence in situ hybridization and somatic cell hybrid analysis. *Genomics* 41:286–289, 1997.
- [35355] 8907.Sontag, J.-M.; Fykse, E. M.; Ushkaryov, Y.; Liu, J.-P.; Robinson, P. J.; Sudhof, T. C.: Differential expression and regulation of multiple dynamins. *J. Biol. Chem.* 269: 4547–4554, 1994.
- [35356] 8908.Sweitzer, S. M.; Hinshaw, J. E.: Dynamin undergoes a GTP-dependent conformational change causing vesicula-

tion. Cell 93: 1021–1029,1998.

- [35357] 8909.van der Bliek, A. M.; Redelmeier, T. E.; Damke, H.; Tisdale, E.J.; Meyerowitz, E. M.; Schmid, S. L.: Mutations in human dynaminblock an intermediate stage in coated vesicle formation. J. CellBiol. 122: 553–563, 1993.
- [35358] 8910.Diatloff–Zito, C.; Gordon, A. J. E.; Duchaud, E.; Merlin, G.:Isolation of an ubiquitously expressed cDNA encoding human dynaminII, a member of the large GTP-binding protein family. Gene 163:301–306, 1995.
- [35359] 8911.Finch, J. L.; Webb, G. C.; Evdokiou, A; Cowled, P. A.: Chromosomallocalization of the human urothelial tetraspan gene, UPK1B, to 3q13.3–q21and detection of a Taq1 polymorphism. Genomics 40: 501–503, 1997.
- [35360] 8912.Yu, J.; Lin, J.–H.; Wu, X.–R.; Sun, T.–T.: Uroplakins Ia and Ib,two major differentiation products of bladder epithelium, belong toa family of four transmembrane domain (4TM) proteins. J. Cell Biol. 125:171–182, 1994.
- [35361] 8913.Svaren, J.; Sevetson, B. R.; Apel, E. D.; Zimonjic, D. B.; Popescu,N. C.; Milbrandt, J.: NAB2, a corepressor of NGFI–A (Egr–1) and Krox20,is induced by proliferative and differentiative stimuli. Molec. Cell.Biol. 16: 3545–3553, 1996.
- [35362] 8914.Colley, W. C.; Altshuller, Y. M.; Sue–Ling, C. K.;

Copeland, N.G.; Gilbert, D. J.; Jenkins, N. A.; Branch, K. D.;
Tsirka, S. E.;Bollag, R. J.; Bollag, W. B.; Frohman, M. A.:
Cloning and expression analysis of murine phospholipase
D1. *Biochem. J.* 326: 745–753, 1997.

[35363] 8915.Bentz, H.; Nathan, R. M.; Rosen, D. M.; Armstrong,
R. M.; Thompson,A. Y.; Segarini, P. R.; Mathews, M. C.;
Dasch, J. R.; Piez, K. A.;Seyedin, S. M.: Purification and
characterization of a unique osteoinductive factor from
bovine bone. *J. Biol. Chem.* 264: 20805–20810, 1989.

[35364] 8916.Madisen, L.; Neubauer, M.; Plowman, G.; Rosen, D.;
Segarini, P.;Dasch, J.; Thompson, A.; Ziman, J.; Bentz, H.;
Purchio, A. F.: Molecular cloning of a novel bone-forming
compound: osteoinductive factor. *DNACell Biol.* 9:
303–309, 1990.

[35365] 8917.Fischer, G.; Perez–Rodriguez, M.; Arguello, J. R.;
Cox, S. T.;McWhinnie, A.; Travers, P. J.; Madrigal, J. A.:
Three novel MICBalleles. *Tissue Antigens* 55: 166–170,
2000.

[35366] 8918.Groh, V.; Bahram, S.; Bauer, S.; Herman, A.;
Beauchamp, M.; Spies,T.: Cell stress-regulated human
major histocompatibility complex class I gene expressed in
gastrointestinal epithelium. *Proc. Nat.Acad. Sci.* 93:
12445–12450, 1996.

- [35367] 8919.Nakai, A.; Tanabe, M.; Kawazoe, Y.; Inazawa, J.; Morimoto, R. I.; Nagata, K.: HSF4, a new member of the human heat shock factor family which lacks properties of a transcriptional activator. *Molec. Cell.Biol.* 17: 469–481, 1997.
- [35368] 8920.Uchida, K.; Yoshimura, A.; Inazawa, J.; Yanagisawa, K.; Osada, H.; Masuda, A.; Saito, T.; Takahashi, T.; Miyajima, A.; Takahashi, T.: Molecular cloning of CISH, chromosome assignment to 3p21.3, and analysis of expression in fetal and adult tissues. *Cytogenet. Cell Genet.* 78: 209–212, 1997.
- [35369] 8921.Yoshimura, A.; Ohkubo, T.; Kiguchi, T.; Jenkins, N. A.; Gilbert, D. J.; Copeland, N. G.; Hara, T.; Miyajima, A.: A novel cytokine-inducible gene CIS, encodes an SH2-containing protein that binds to tyrosine-phosphorylated interleukin 3 and erythropoietin receptors. *EMBO J.* 14: 2816–2826, 1995.
- [35370] 8922.Chen, H.; Antonarakis, S. E.: The SH3D1A gene maps to human chromosome 21q22.1–q22.2. *Cytogenet. Cell Genet.* 78: 213–215, 1997.
- [35371] 8923.Pucharcos, C.; Estivill, X.; de la Luna, S.: Intersectin 2, a new multimodular protein involved in clathrin-mediated endocytosis. *FEBS Lett.* 478: 43–51, 2000.

- [35372] 8924.Szepietowski, P.; Simon, M.-P.; Grosgeorge, J.; Huebner, K.; Bastard,C.; Evans, G. A.; Tsujimoto, Y.; Birnbaum, D.; Theillet, C.; Gaudray,P.: Localization of 11q13 loci with respect to regional chromosomalbreakpoints. *Genomics* 12: 738–744, 1992.
- [35373] 8925.Wang, T. C.; Cardiff, R. D.; Zukerberg, L.; Lees, E.; Arnold,A.; Schmidt, E. V.: Mammary hyperplasia and carcinoma in MMTV–cyclinD1 transgenic mice. *Nature* 369: 669–671, 1994.
- [35374] 8926.Xiong, Y.; Connelly, T.; Futcher, B.; Beach, D.: Human D–typecyclin. *Cell* 65: 691–699, 1991.
- [35375] 8927.Yu, Q.; Geng, Y.; Sicinski, P.: Specific protection against breastcancers by cyclin D1 ablation. *Nature* 411: 1017–1021, 2001.
- [35376] 8928.Zatyka, M.; da Silva, N. F.; Clifford, S. C.; Morris, M. R.; Wiesener,M. S.; Eckardt, K.–U.; Houlston, R. S.; Richards, F. M.; Latif, F.;Maher, E. R.: Identification of cyclin D1 and other novel targetsfor the von Hippel–Lindau tumor suppressor gene by expression arrayanalysis and investigation of cyclin D1 genotype as a modifier invon Hippel–Lindau disease. *Cancer Res.* 62: 3803–3811, 2002.
- [35377] 8929.Skoldberg, F.; Grimelius, L.; Woodward, E. R.; Rorsman, F.; VanSchothorst, E. W.; Winqvist, O.; Karlsson, F.

A.; Akerstrom, G.; Kampe, O.; Husebye, E. S.: A family with hereditary extra-adrenal paragangliomas without evidence for mutations in the von Hippel-Lindau disease or ret genes. Clin. Endocr. 48: 11-16, 1998.

[35378] 8930. Gibbons, B.; Scott, D.; Hungerford, J. L.; Cheung, K. L.; Harrison, C.; Attard-Montalto, S.; Evans, M.; Birch, J. M.; Kingston, J. E.: Retinoblastoma in association with the chromosome breakage syndromes Fanconi's anaemia and Bloom's syndrome: clinical and cytogenetic findings. Clin. Genet. 47: 311-317, 1995.

[35379] 8931. Girardet, A.; McPeck, M. S.; Leeflang, E. P.; Munier, F.; Arnheim, N.; Claustres, M.; Pellestor, F.: Meiotic segregation analysis of RB1 alleles in retinoblastoma pedigrees by use of single-sperm typing. Am. J. Hum. Genet. 66: 167-175, 2000.

[35380] 8932. Godbout, R.; Dryja, T. P.; Squire, J.; Gallie, B. L.; Phillips, R. A.: Somatic inactivation of genes on chromosome 13 is a common event in retinoblastoma. Nature 304: 451-453, 1983.

[35381] 8933. Goodrich, D. W.; Wang, N. P.; Qian, Y.-W.; Lee, E. Y.-H. P.; Lee, W.-H.: The retinoblastoma gene product regulates progression through the G1 phase of the cell cycle. Cell 67: 293-302, 1991.

- [35382] 8934.Grace, E.; Drennan, J.; Colver, D.; Gordon, R. R.: The 13q deletionsyndrome. *J. Med. Genet.* 8: 351–357, 1971.
- [35383] 8935.Green, A. R.; Wyke, J. A.: Anti-oncogenes: a subset of regulatorygenes involved in carcinogenesis? *Lancet II*: 475–477, 1985.
- [35384] 8936.Greger, V.; Kerst, S.; Messmer, E.; Hopping, W.; Passarge, E.;Horsthemke, B.: Application of linkage analysis to genetic counsellingin families with hereditary retinoblastoma. *J. Med. Genet.* 25: 217–221,1988.
- [35385] 8937.Greger, V.; Passarge, E.; Horsthemke, B.: Somatic mosaicism ina patient with bilateral retinoblastoma. *Am. J. Hum. Genet.* 46:1187–1193, 1990.
- [35386] 8938.Hagstrom, S. A.; Dryja, T. P.: Mitotic recombination map of 13cen–13q14derived from an investigation of loss of heterozygosity in retinoblastomas. *Proc.Nat. Acad. Sci.* 96: 2952–2957, 1999.
- [35387] 8939.Hall, J. G.: Personal Communication. Vancouver, British Columbia,Canada 5/29/1993.
- [35388] 8940.Hanahan, D.; Weinberg, R. A.: The hallmarks of cancer. *Cell* 100:57–70, 2000.
- [35389] 8941.Harbour, J. W.: Molecular basis of low-penetrance retinoblastoma. *Arch.Ophthal.* 119: 1699–1704, 2001.
- [35390] 8942.Harbour, J. W.; Lai, S.–L.; Whang–Peng, J.; Gazdar, A.

F.; Minna, J. D.; Kaye, F. J.: Abnormalities in structure and expression of the human retinoblastoma gene in SCLC. Science 241: 353–357, 1988.

- [35391] 8943. Hensel, C.; Hsieh, C.-L.; Lee, W.-H.; Pam-Lee, E.; Gazdar, A.; Sakaguchi, A. Y.; Naylor, S. L.: Allele loss and lack of expression of the RB-1 locus in small cell lung cancer. (Abstract) Am. J. Hum. Genet. 43: A25, 1988.
- [35392] 8944. Henson, J. W.; Schnitker, B. L.; Correa, K. M.; von Diemling, A.; Fassbender, F.; Xu, H.-J.; Benedict, W. F.; Yandell, D. W.; Louis, D. N.: The retinoblastoma gene is involved in malignant progression of astrocytomas. Ann. Neurol. 36: 714–721, 1994.
- [35393] 8945. Higgins, M. J.; Hansen, M. F.; Cavenee, W. K.; Lalande, M.: Molecular detection of chromosomal translocations that disrupt the putative retinoblastoma susceptibility locus. Molec. Cell. Biol. 9: 1–5, 1989.
- [35394] 8946. Hoegerman, S. F.: Chromosome 13 long arm interstitial deletion may result from maternal inverted insertion. Science 205: 1035–1036, 1979.
- [35395] 8947. Hogg, A.; Bia, B.; Onadim, Z.; Cowell, J. K.: Molecular mechanisms of oncogenic mutations in tumours from patients with bilateral and unilateral retinoblastoma. Proc. Nat. Acad. Sci. 90: 7351–7355, 1993.

- [35396] 8948.Honavar, S. G.; Shields, C. L.; Shields, J. A.; Demirci, H.; Naduvilath,T. J.: Intraocular surgery after treatment of retinoblastoma. Arch.Ophthal. 119: 1613–1621, 2001.
- [35397] 8949.Honavar, S. G.; Singh, A. D.; Shields, C. L.; Meadows, A. T.;Demirci, H.; Cater, J.; Shields, J. A.: Postenucleation adjuvanttherapy in high–risk retinoblastoma. Arch. Ophthal. 120: 923–931,2002.
- [35398] 8950.Hong, F. D.; Huang, H.–J. S.; To, H.; Young, L.–J. S.; Oro, A.;Bookstein, R.; Lee, E. Y.–H. P.; Lee, W.–H.: Struc–ture of the humanretinoblastoma gene. Proc. Nat. Acad. Sci. 86: 5502–5506, 1989.100. Horowitz, J. M.; Park, S.–H.; Bogenmann, E.; Cheng, J.–C.; Yandell,D. W.; Kaye, F. J.; Minna, J. D.; Dryja, T. P.; Weinberg, R. A.:Frequent inac–tivation of the retinoblastoma anti–oncogene is restrict–edto a subset of human tumor cells. Proc. Nat. Acad. Sci. 87: 2775–2779,1990.101. Horowitz, J. M.; Park, S. H.; Yandell, D. W.; Weinberg, R. A.: Involvement of the retinoblastoma gene in the genesis of varioushuman tu–mors:.In: Kavenee, W.; Hastie, N.; Stanbridge, E.: Reces–siveOncogenes and Tumor Suppression: Current Commu–nications in MolecularBiology. Cold Spring Harbor, New York: Cold Spring Harbor LaboratoryPress (pub.) 1989. Pp. 101–108.102. Horsthemke, B.; Greger, V.; Barnert, H. J.;

Hopping, W.; Passarge, E.: Detection of submicroscopic deletions and a DNA polymorphism at the retinoblastoma locus. *Hum. Genet.* 76: 257–261, 1987.103. Horsthemke, B.; Greger, V.; Becher, R.; Passarge, E.: Mechanism of i(6p) formation in retinoblastoma tumor cells. *Cancer Genet. Cytogenet.* 37:95–102, 1989.104. Hsieh, J.-K.; Chan, F. S. G.; O'Connor, D. J.; Mittnacht, S.; Zhong, S.; Lu, X.: RB regulates the stability and the apoptotic function of p53 via MDM2. *Molec. Cell* 3: 181–193, 1999.105. Huang, H.-J. S.; Yee, J.-K.; Shew, J.-Y.; Chen, P.-L.; Bookstein, R.; Friedmann, T.; Lee, E. Y.-H. P.; Lee, W.-H.: Suppression of the neoplastic phenotype by replacement of the RB gene in human cancer cells. *Science* 242: 1563–1566, 1988.106. Janson, M.; Nordenskjold, M.: A constitutional mutation within the retinoblastoma gene detected by PFGE. *Clin. Genet.* 45: 5–10, 1994.107. Jensen, R. D.; Miller, R. W.: Retinoblastoma: epidemiologic characteristics. *New Eng. J. Med.* 285: 307–311, 1971.108. Kimchi, A.; Wang, X.-F.; Weinberg, R. A.; Cheifetz, S.; Massague, J.: Absence of TGF- β receptors and growth inhibitory responses in retinoblastoma cells. *Science* 240: 196–199, 1988.109. Kitchin, F. D.; Ellsworth, R. M.: Pleiotropic effects of the gene for retinoblastoma. *J. Med. Genet.* 11: 244–246,

1974.110. Kivela, T.: Trilateral retinoblastoma: a meta-analysis of hereditary retinoblastoma associated with primary ectopic intracranial retinoblastoma. *J.Clin. Oncol.* 17: 1829–1837, 1999.111. Kivela, T.; Asko-Seljavaara, S.; Pihkala, U.; Hovi, L.; Heikkonen, J.: Sebaceous carcinoma of the eyelid associated with retinoblastoma. *Ophthalmology* 108:1124–1128, 2001.112. Klutz, M.; Brockmann, D.; Lohmann, D. R.: A parent-of-origin effect in two families with retinoblastoma is associated with a distinct splice mutation in the RB1 gene. *Am. J. Hum. Genet.* 71: 174–179, 2002.113. Knight, L. A.; Gardner, H. A.; Gallie, B. L.: Familial retinoblastoma: segregation of chromosome 13 in four families. *Am. J. Hum. Genet.* 32:194–201, 1980.114. Knudson, A. G.: Hereditary cancer, oncogenes and anti-oncogenes. *Cancer Res.* 45: 1437–1443, 1985.115. Knudson, A. G., Jr.: Mutation and cancer: statistical study of retinoblastoma. *Proc. Nat. Acad. Sci.* 68: 820–823, 1971.116. Knudson, A. G., Jr.: Genetics of human cancer. *Annu. Rev. Genet.* 20:231–251, 1986.117. Knudson, A. G., Jr.; Hethcote, H. W.; Brown, B. W.: Mutation and childhood cancer: a probabilistic model for the incidence of retinoblastoma. *Proc. Nat. Acad. Sci.* 72: 5116–5120, 1975.118. Knudson, A. G., Jr.; Meadows, A.

T.; Nichols, W. W.; Hill, R.: Chromosomal deletion and retinoblastoma. *New Eng. J. Med.* 295:1120–1123, 1976.119. Laquis, S. J.; Rodriguez–Galindo, C.; Wilson, M. W.; Fleming, J. C.; Haik, B. G.: Retinoblastoma in a patient with an X;13 translocation and facial abnormalities consistent with 13q–syndrome. *Am. J. Ophthal.* 133:285–287, 2002.120. Lee, W.–H.; Bookstein, R.; Hong, F.; Young, L.–J.; Shew, J.–Y.; Lee, E. Y.–H. P.: Human retinoblastoma susceptibility gene: cloning, identification, and sequence. *Science* 235: 1394–1399, 1987.121. Lee, W.–H.; Shew, J.–Y.; Hong, F. D.; Sery, T. W.; Donoso, L.A.; Young, L.–J.; Bookstein, R.; Lee, E. Y.–H. P.: The retinoblastoma susceptibility gene encodes a nuclear phosphoprotein associated with DNA binding activity. *Nature* 329: 642–645, 1987.122. Lele, K. P.; Penrose, L. S.; Stallard, H. B.: Chromosome deletion in a case of retinoblastoma. *Ann. Hum. Genet.* 27: 171–174, 1963.123. Lemieux, N.; Messier, P. E.; Jacob, J. L.; Milot, J.; Richer, C. L.: Precise cytogenetic localization of the Rb locus at subband 13q14.11 by ultrastructural detection after immunochemical chromosome banding. (Abstract) *Am. J. Hum. Genet.* 45 (suppl.): A27, 1989.124. Liu, Z.; Song, Y.; Bia, B.; Cowell, J. K.: Germline mutations in the RB1 gene in patients with hereditary

retinoblastoma. *GenesChromosomes Cancer* 14: 277–284, 1995.125. Lohmann, D. R.; Brandt, B.; Hopping, W.; Passarge, E.; Horsthemke,B.: Spectrum of small length germline mutations in the RB1 gene. *Hum.Molec. Genet.* 3: 2187–2193, 1994.126. Lohmann, D. R.; Brandt, B.; Hopping, W.; Passarge, E.; Horsthemke,B.: The spectrum of RB1 germ–line mutations in hereditary retinoblastoma. *Am.J. Hum. Genet.* 58: 940–949, 1996.127. Lohmann, D. R.; Gerick, M.; Brandt, B.; Oelschlager, U.; Lorenz,B.; Passarge, E.; Horsthemke, B.: Constitutional RB1–gene mutationsin patients with isolated unilateral retinoblastoma. *Am. J. Hum.Genet.* 61: 282–294, 1997.128. Lomazzi, M.; Moroni, M. C.; Jensen, M. R.; Frittoli, E.; Helin,K.: Suppression of the p53– or pRB–mediated G1 checkpoint is requiredfor E2F–induced S–phase entry. *Nature Genet* 31: 190–194, 2002.129. Lueder, G. T.; Judisch, G. F.; Wen, B.–C.: Heritable retinoblastomaand pinealoma. *Arch. Ophthalmol.* 109: 1707–1709, 1991.130. Luo, R. X.; Postigo, A. A.; Dean, D. C.: Rb interacts with histonedacetylase to repress transcription. *Cell* 92: 463–473, 1998.131. Maat-Kievit, J. A.; Oepkes, D.; Hartwig, N. G.; Vermeij–Keers,C.; van Kamp, I. L.; van de Kamp, J. J. P.: A large retinoblastomadetected in a fetus at 21 weeks of gestation. *Prenatal*

Diag. 13:377–384, 1993.132. MacKay, C. J.; Abramson, D. H.; Ellsworth, R. M.: Metastatic patterns of retinoblastoma. Arch. Ophthalmol. 102: 391–396, 1984.133. Macklin, M. T.: A study of retinoblastoma in Ohio. Am. J. Hum. Genet. 12: 1–43, 1960.134. Macklin, M. T.: Inheritance of retinoblastoma in Ohio. Arch. Ophthalmol. 62: 842–851, 1959.135. Manchester, P. T., Jr.: Retinoblastoma among offspring of adult survivors. Arch. Ophthalmol. 65: 546–549, 1961.136. Mancini, D.; Singh, S.; Ainsworth, P.; Rodenhiser, D.: Constitutively methylated CpG dinucleotides as mutation hot spots in the retinoblastoma gene (RB1). Am. J. Hum. Genet. 61: 80–87, 1997.137. Mancini, M. A.; Shan, B.; Nickerson, J. A.; Penman, S.; Lee, W.-H.: The retinoblastoma gene product is a cell cycle-dependent, nuclear matrix-associated protein. Proc. Nat. Acad. Sci. 91: 418–422, 1994.138. Marino, S.; Vooijs, M.; van der Gulden, H.; Jonker, J.; Berns, A.: Induction of medulloblastomas in p53-null mutant mice by somatic inactivation of Rb in the external granular layer cells of the cerebellum. Genes Dev. 14: 994–1004, 2000.139. Matsunaga, E.: Recurrence risks to relatives of patients with retinoblastoma. Jpn. J. Ophthalmol. 22: 313–319, 1978.140. Matsunaga, E.: Almost synchronous appearance of bilateral retinoblas-

tomas.(Letter) Am. J. Med. Genet. 11: 485–487, 1982.141.

Matsunaga, E.: Retinoblastoma: mutational mosaicism or hostresistance? Am. J. Med. Genet. 8: 375–387, 1981.142.

Matsunaga, E.: Hereditary retinoblastoma: host resistance andsecond primary tumors. J. Nat. Cancer Inst. 65: 47–51, 1980.143.

Matsunaga, E.: Hereditary retinoblastoma: de-layed mutation orhost resistance? Am. J. Hum. Genet. 30: 406–425, 1978.144.

Matsunaga, E.; Minoda, K.; Sasaki, M. S.: Parental age and seasonalvariation in the births of chil-dren with sporadic retinoblastoma:a mutation–epi–demiologic study. Hum. Genet. 84: 155–158, 1990.145.

Michalova, K.; Kloucek, F.; Musilova, J.: Deletion of 13q intwo patients with retinoblastoma, one probably due to 13q– mosaicismin the mother. Hum. Genet. 61: 264–266, 1982.146.

Moll, A. C.; Imhof, S. M.; Schouten–Van Meeteren, A. Y. N.; Kuik,D. J.; Hofman, P.; Boers, M.: Sec-ond primary tumors in hereditaryretinoblastoma: a regis-ter–based study, 1945–1997. Is there an ageeffect on ra-diation–related risk? Ophthalmology 108: 1109–1114,2001.147.

Motegi, T.: Lymphocyte chromo-some survey in 42 patients withretinoblastoma: effort to detect 13q14 deletion mosaicism. Hum. Genet. 58:168–173, 1981.148.

Motegi, T.: High rate of detection

of 13q14 deletion mosaicism among retinoblastoma patients (using more extensive methods). Hum.Genet. 61: 95–97, 1982.149. Motegi, T.; Kaga, M.; Yanagawa, Y.; Kadowaki, H.; Watanabe, K.; Inoue, A.; Komatsu, M.; Minoda, K.: A recognizable pattern of the midface of retinoblastoma patients with interstitial deletion of 13q. Hum.Genet. 64: 160–162, 1983.150. Motegi, T.; Komatsu, M.; Minoda, K.: Is the interstitial deletion of 13q in retinoblastoma patients not transmissible? (Letter) Hum.Genet. 64: 205, 1983.151. Motegi, T.; Komatsu, M.; Nakazato, Y.; Ohuchi, M.; Minoda, K.: Retinoblastoma in a boy with a de novo mutation of a 13/18 translocation: the assumption that the retinoblastoma locus is at 13q141, particularly at the distal portion of it. Hum. Genet. 60: 193–195, 1982.152. Munier, F.; Spence, M. A.; Pescia, G.; Balmer, A.; Gailloud, C.; Thonney, F.; van Melle, G.; Rutz, H. P.: Paternal selection favoring mutant alleles of the retinoblastoma susceptibility gene. Hum. Genet. 89:508–512, 1992.153. Munier, F. L.; Wang, M. X.; Spence, M. A.; Thonney, F.; Balmer, A.; Pescia, G.; Donoso, L. A.; Murphree, A. L.: Pseudo low penetrance in retinoblastoma: fortuitous familial aggregation of sporadic cases caused by independently derived mutations in two

large pedigrees. Arch.Ophthal. 111: 1507–1511, 1993.154. Murphree, A. L.; Benedict, W. F.: Retinoblastoma: clues to human oncogenesis. Science 223: 1028–1033, 1984.155. Naumova, A.; Hansen, M.; Strong, L.; Jones, P. A.; Hadjistilianou, D.; Mastrangelo, D.; Griegel, S.; Rajewsky, M. F.; Shields, J.; Donoso, L.; Wang, M.; Sapienza, C.: Concordance between parental origin of chromosome 13q loss and chromosome 6p duplication in sporadic retinoblastoma. Am.J. Hum. Genet. 54: 274–281, 1994.156. Naumova, A.; Sapienza, C.: The genetics of retinoblastoma, revisited. Am.J. Hum. Genet. 54: 264–273, 1994.157. Nevins, J. R.: The Rb/E2F pathway and cancer. Hum. Molec. Genet. 10:699–703, 2001.158. Nichols, W. W.; Miller, R. C.; Sobel, M.; Hoffman, E.; Sparkes, R. S.; Mohandas, T.; Veomett, I.; Davis, J. R.: Further observations on a 13qXp translocation associated with retinoblastoma. Am. J. Ophthal. 89:621–627, 1980.159. Nielsen, S. J.; Schneider, R.; Bauer, U.-M.; Bannister, A. J.; Morrison, A.; O'Carroll, D.; Firestein, R.; Cleary, M.; Jenuwein, T.; Herrera, R. E.; Kouzarides, T.: Rb targets histone H3 methylation and HP1 to promoters. Nature 412: 561–565, 2001.160. Nirankari, M. S.; Gulati, G. C.; Chadah, M. R.: Retinoblastoma: genetics and report of a fam-

ily. Am. J. Ophthal. 53: 523–532, 1962.161. Noorani, H. Z.; Khan, H. N.; Gallie, B. L.; Detsky, A. S.: Costcomparison of molecular versus conventional screening of relatives at risk for retinoblastoma. Am. J. Hum. Genet. 59: 301–307, 1996.162. Nussbaum, R.; Puck, J.: Recurrence risks for retinoblastoma: a model for autosomal dominant disorders with complex inheritance. J. Pediatr. Ophthal. 13: 89–98, 1976.163. Onadim, Z.; Hogg, A.; Baird, P. N.; Cowell, J. K.: Oncogenic point mutations in exon 20 of the RB1 gene in families showing incomplete penetrance and mild expression of the retinoblastoma phenotype. Proc. Nat. Acad. Sci. 89: 6177–6181, 1992.164. Onadim, Z.; Woolford, A. J.; Kingston, J. E.; Hungerford, J. L.: The RB1 gene mutation in a child with ectopic intracranial retinoblastoma. Brit. J. Cancer 76: 1405–1409, 1997.165. Ono, T.; Yoshida, M. C.: Chromosomal assignment of retinoblastoma 1 gene (RB1) to mouse 14D3 and rat 15q12 by fluorescence in situ hybridization. Jpn. J. Genet. 68: 617–621, 1993.166. Orye, E.; Benoit, Y.; Coppieters, R.; Jeannin, P.; Ver-cruysse, C.; Delaey, J.; Delbeke, M.-J.: A case of retinoblastoma, associated with histiocytosis-X and mosaicism of a deleted D-group chromosome (13q14–q31). Clin. Genet. 22: 37–39, 1982.167. Orye, E.; Delbeke, M. J.; Vanden-

abeele, B.: Retinoblastoma and long arm deletion of chromosome 13. Attempts to define the deleted segment. Clin. Genet. 5: 457–464, 1974.168. Orye, E.; Delbeke, M. J.; Vandenabeele, B.: Retinoblastoma and D–chromosome deletions. (Letter) Lancet II: 1376, 1971.169. Otterson, G. A.; Modi, S.; Nguyen, K.; Coxon, A. B.; Kaye, F.J.: Temperature-sensitive RB mutations linked to incomplete penetrance of familial retinoblastoma in 12 families. Am. J. Hum. Genet. 65:1040–1046, 1999.170. Otterson, G. W.; Chen, W.; Coxon, A. B.; Khleif, S. N.; Kaye, F. J.: Incomplete penetrance of familial retinoblastoma linked to germ-line mutations that result in partial loss of RB function. Proc. Nat. Acad. Sci. 94: 12036–12040, 1997.171. Pendergrass, T. W.; Davis, S.: Incidence of retinoblastoma in the United States. Arch. Ophthalmol. 98: 1204–1210, 1980.172. Pennaneach, V.; Salles-Passador, I.; Munshi, A.; Brickner, H.; Regazzoni, K.; Dick, F.; Dyson, N.; Chen, T.–T.; Wang, J. Y. J.; Fotadar, R.; Fotadar, A.: The large subunit of replication factor C promotes cell survival after DNA damage in an LxCxE motif– and Rb–dependent manner. Molec. Cell 7: 715–727, 2001.173. Riccardi, V. M.; Hittner, H. M.; Francke, U.; Pippin, S.; Holmquist, G. P.; Kretzer, F. L.; Ferrell, R.: Partial triplication and deletion of 13q: study of a

family presenting with bilateral retinoblastomas.

Clin.Genet. 15: 332–345, 1979.174. Rivera, H.; Turleau, C.; de Grouchy, J.; Junien, C.; Despoisse, S.; Zucker, J.-M.: Retinoblastoma–del(13q14): report of two patients, one with a trisomic sib due to maternal insertion; gene–dosage effect for esterase D. Hum. Genet. 59: 211–214, 1981.175. Sakai, T.; Ohtani, N.; McGee, T. L.; Robbins, P. D.; Dryja, T.P.: Oncogenic germ–line mutations in Sp1 and ATF sites in the human retinoblastoma gene. Nature 353: 83–86, 1991.176. Sakai, T.; Ohtani, N.; McGee, T. L.; Robbins, P. D.; Dryja, T.P.: Oncogenic germ–line mutations in Sp1 and ATF sites in the human retinoblastoma gene. Nature 353: 83–86, 1991.177. Sakai, T.; Toguchida, J.; Ohtani, N.; Yandell, D. W.; Rapaport, J. M.; Dryja, T. P.: Allele–specific hypermethylation of the retinoblastoma tumor–suppressor gene. Am. J. Hum. Genet. 48: 880–888, 1991.178. Schappert–Kimmijser, J.; Hemmes, G. D.; Nijland, R.: The heredity of retinoblastoma. Ophthalmologica 151: 197–213, 1966.179. Scheffer, H.; te Meerman, G. J.; Kruize, Y. C. M.; van den Berg, A. H. M.; Penninga, D. P.; Tan, K. E. W. P.; der Kinderen, D. J.; Buys, C. H. C. M.: Linkage analysis of families with hereditary retinoblastoma: nonpenetrance of mutation, revealed by combined

use of markers within and flanking the RB1 gene. *Am. J. Hum. Genet.* 45: 252–260, 1989.180. Schimke, R. N.; Lowman, J.; Cowan, G.: Retinoblastoma and osteogenic-sarcoma in sibs. *Cancer* 34: 2077–2079, 1974.181. Schubert, E. L.; Strong, L. C.; Hansen, M. F.: A splicing mutation in RB1 in low penetrance retinoblastoma. *Hum. Genet.* 100: 557–563, 1997.182. Shields, C. L.; Honavar, S.; Shields, J. A.; Demirci, H.; Meadows, A. T.: Vitrectomy in eyes with unsuspected retinoblastoma. *Ophthalmology* 107:2250–2255, 2000.183. Shio, Y.; Yamamoto, T.; Yamaguchi, N.: Negative regulation of Rb expression by the p53 gene product. *Proc. Nat. Acad. Sci.* 89:5206–5210, 1992.184. Shroeder, W. T.; Chao, L.-Y.; Dao, D. D.; Strong, L. C.; Pathak, S.; Riccardi, V.; Lewis, W. H.; Saunders, G. F.: Nonrandom loss of maternal chromosome 11 alleles in Wilms tumors. *Am. J. Hum. Genet.* 40:413–420, 1987.185. Sippel, K. C.; Fraieli, R. E.; Smith, G. D.; Schalkoff, M. E.; Sutherland, J.; Gallie, B. L.; Dryja, T. P.: Frequency of somatic and germ-line mosaicism in retinoblastoma: implications for genetic counseling. *Am. J. Hum. Genet.* 62: 610–619, 1998.186. Smith, S. M.; Sorsby, A.: Retinoblastoma: some genetic aspects. *Ann. Hum. Genet.* 23: 50–58, 1958.187. Sparkes, R. S.:

The genetics of retinoblastoma. *Biochim. Biophys. Acta* 780: 95–118, 1985.188. Sparkes, R. S.; Muller, H.; Klisak, I.; Abram, J. A.: Retinoblastoma with 13q; chromosomal deletion associated with maternal paracentric inversion of 13q. *Science* 203: 1027–1029, 1979.189. Sparkes, R. S.; Murphree, A. L.; Lingua, R. W.; Sparkes, M. C.; Field, L. L.; Funderburk, S. J.; Benedict, W. F.: Gene for hereditary retinoblastoma assigned to human chromosome 13 by linkage to esterase D. *Science* 219: 971–973, 1983.190. Sparkes, R. S.; Sparkes, M. C.; Wilson, M. G.; Towner, J. W.; Benedict, W.; Murphree, A. L.; Yunis, J. J.: Regional assignment of genes for human esterase D and retinoblastoma to chromosome band 13q14. *Science* 208: 1042–1044, 1980.191. Sparkes, R. S.; Sparkes, M. C.; Wilson, M. G.; Towner, J. W.; Benedict, W.; Murphree, A. L.; Yunis, J. J.: Regional assignment of genes for human esterase D and retinoblastoma to chromosome band 13q14. (Abstract) *Cytogenet. Cell Genet.* 25: 209, 1979.192. Squire, J.; Gallie, B. L.; Phillips, R. A.: A detailed analysis of chromosomal changes in heritable and non-heritable retinoblastoma. *Hum. Genet.* 70: 291–301, 1985.193. Squire, J.; Phillips, R. A.; Boyce, S.; Godbout, R.; Rogers, B.; Gallie, B. L.: Isochromosome 6p, a unique chromosomal

abnormality in retinoblastoma: verification by standard staining techniques, new densitometric methods, and somatic cell hybridization. *Hum. Genet.* 66:46–53, 1984.194. Stallard, H. B.: The conservation treatment of retinoblastoma. *Trans. Ophthalm. Soc.* 82: 473, 1962.195. Stone, J. C.; Crosby, J. L.; Kozak, C. A.; Schievella, A. R.; Bernards, R.; Nadeau, J. H.: The murine retinoblastoma homolog maps to chromosome 14 near Es-10. *Genomics* 5: 70–75, 1989.196. Strong, L. C.; Riccardi, V. M.; Ferrell, R. E.; Sparkes, R. S.: Familial retinoblastoma and chromosome 13 deletion transmitted via an insertional translocation. *Science* 213: 1501–1503, 1981.197. Taylor, A. I.: Dq-, Dr and retinoblastoma. *Humangenetik* 10:209–217, 1970.198. Thomas, D. M.; Carty, S. A.; Piscopo, D. M.; Lee, J.-S.; Wang, W.-F.; Forrester, W. C.; Hinds, P. W.: The retinoblastoma protein acts as a transcriptional coactivator required for osteogenic differentiation. *Molec. Cell* 8: 303–316, 2001.199. Toguchida, J.; Ishizaki, K.; Sasaki, M. S.; Nakamura, Y.; Ikenaga, M.; Kato, M.; Sugimoto, M.; Koutoura, Y.; Yamamoto, T.: Preferential mutation of paternally derived RB gene as the initial event in sporadic osteosarcoma. *Nature* 338: 156–158, 1989.200. Toguchida, J.; McGee, T. L.; Paterson, J. C.; Eagle, J. R.; Tucker, S.; Yan-

dell, D. W.; Dryja, T. P.: Complete genomic sequence of the human retinoblastoma susceptibility gene. *Genomics* 17: 535–543, 1993. 201. Turleau, C.; de Grouchy, J.; Chavin-Colin, F.; Despoisses, S.; Leblanc, A.: Two cases of del(13q)–retinoblastoma and two cases of partial trisomy due to a familial insertion. *Ann. Genet.* 26: 158–160, 1983. 202. Turleau, C.; de Grouchy, J.; Chavin-Colin, F.; Junien, C.; Seger, J.; Schlienger, P.; Leblanc, A.; Haye, C.: Cytogenetic forms of retinoblastoma: their incidence in a survey of 66 patients. *Cancer Genet. Cytogenet.* 16: 321–334, 1985. 203. Verma, R. S.; Ramesh, K. H.; Samonte, R. V.; Conte, R. A.: Mapping the homolog of the human Rb1 gene to chromosome 14 of higher primates. *Mammalian Genome* 7: 591–592, 1996. 204. Vogel, F.: Genetics of retinoblastoma. *Modern Trends in Ophthalmology*. (pub.) 1968. 205. Vogel, F.: Genetics of retinoblastoma. In: *Genetic Counseling*. Heidelberg University, Science Library. Trans. by Sabine Kurth. New York: Springer Verlag (pub.) 1969. 206. Vogel, F.: The genetics of retinoblastoma. *Hum. Genet.* 52: 1–54, 1979. 207. Warburg, M.: Retinoblastoma. In: *Goldberg, M. F.: Genetic and Metabolic Eye Disease*. Boston: Little, Brown and Co. (pub.) 1974. Pp. 447–461. 208. Weichselbaum, R. R.; Beckett, M.;

Diamond, A.: Some retinoblastomas, osteosarcomas, and soft tissue sarcomas may share a common etiology.

Proc. Nat. Acad. Sci. 85: 2106–2109, 1988.209.

Weichselbaum, R. R.; Nove, J.; Little, J. B.: Fibroblasts from a D-deletion type retinoblastoma patient are abnormally x-ray sensitive. Nature 266:726–727, 1977.210.

Weinberg, R. A.: The retinoblastoma protein and cell cycle control. Cell 81:323–330, 1995.211.

Whyte, P.; Buchkovich, K. J.; Horowitz, J. M.; Friend, S. H.; Raybuck, M.; Weinberg, R. A.; Harlow, E.: Association between an oncogene and an anti-oncogene: the adenovirus E1A proteins bind to the retinoblastoma gene product. Nature 334: 124–129, 1988.212.

Wiggs, J.; Nordenskjold, M.; Yandell, D.; Rapaport, J.; Grondin, V.; Janson, M.; Werelius, B.; Petersen, R.; Craft, A.; Riedel, K.; Liberfarb, R.; Walton, D.; Wilson, W.;

Dryja, T. P.: Prediction of the risk of hereditary retinoblastoma, using DNA polymorphisms within the retinoblastoma gene. New Eng. J. Med. 318: 151–157, 1988.213.

Wilson, M. G.; Ebbin, A. J.; Towner, J. W.; Spencer, W.

H.: Chromosomal anomalies in patients with retinoblastoma. Clin. Genet. 12:1–8, 1977.214.

Wilson, M. G.; Melnyk, J.; Towner, J. W. J.: Retinoblastoma and deletion D(14) syndrome. J. Med. Genet. 6: 322–327, 1969.215.

Wilson,

M. G.; Towner, J. W.; Fujimoto, A.: Retinoblastoma and D-chromosome deletions. *Am. J. Hum. Genet.* 25: 57–61, 1973.216. Windle, J. J.; Albert, D. M.; O'Brien, J. M.; Marcus, D. M.; Distèche, C. M.; Bernards, R.; Mellon, P. L.: Retinoblastoma in transgenic mice. *Nature* 343: 665–669, 1990.217. Yandell, D. W.; Campbell, T. A.; Dayton, S. H.; Petersen, R.; Walton, D.; Little, J. B.; McConkie-Rosell, A.; Buckley, E.; Dryja, T.: Oncogenic point mutations in the human retinoblastoma gene: their application to genetic counseling. *New Eng. J. Med.* 321: 1689–1695, 1989.218. Yokota, J.; Akiyama, T.; Fung, Y.-K. T.; Benedict, W. F.; Namba, Y.; Hanaoka, M.; Wada, M.; Terasaki, T.; Shimamoto, Y.; Sugimura, T.; Terada, M.: Altered expression of the retinoblastoma (RB) gene in small-cell carcinoma of the lung. *Oncogene* 3: 471–475, 1988.219. Zeschnigk, M.; Lohmann, D.; Horsthemke, B.: A PCR test for the detection of hypermethylated alleles at the retinoblastoma locus. *J. Med. Genet.* 36: 793–794, 1999.220. Zhang, H. S.; Postigo, A. A.; Dean, D. C.: Active transcriptional repression by the Rb-E2F complex mediates G1 arrest triggered by p16(INK4a), TGF- β , and contact inhibition. *Cell* 97: 53–61, 1999.221. Zhu, X.; Dunn, J. M.; Phillips, R. A.; Goddard, A. D.; Paton, K. E.; Becker, A.; Gallie, B. L.: Pref-

erential germline mutation of the paternal allele in retinoblastoma. *Nature* 340: 312–313, 1989.

- [35399] 8951. Baens, M.; Aerssens, J.; Van Zand, K.; Van den Berghe, H.; Marynen, P.: Isolation and regional assignment of human chromosome 12p cDNAs. *Genomics* 29:44–52, 1995.
- [35400] 8952. Mao, S.; Neale, G. A. M.; Goorha, R. M.: T-cell oncogene rhombotin-2 interacts with retinoblastoma-binding protein 2. *Oncogene* 14: 1531–1539, 1997.
- [35401] 8953. Baldi, A.; Boccia, V.; Claudio, P. P.; De Luca, A.; Giordano, A.: Genomic structure of the human retinoblastoma-related Rb2/p130 gene. *Proc. Nat. Acad. Sci.* 93: 4629–4632, 1996.
- [35402] 8954. Mayol, X.; Grana, X.; Baldi, A.; Sang, N.; Hu, Q.; Giordano, A.: Cloning of a new member of the retinoblastoma gene family (pRb2) which binds to the E1A transforming domain. *Oncogene* 8: 2561–2566, 1993.
- [35403] 8955. Yeung, R. S.; Bell, D. W.; Testa, J. R.; Mayol, X.; Baldi, A.; Grana, X.; Klinga-Levan, K.; Knudson, A. G.; Giordano, A.: The retinoblastoma-related gene, RB2, maps to human chromosome 16q12 and rat chromosome 19. *Oncogene* 8:3465–3468, 1993.
- [35404] 8956. Benbrook, D.; Lernhardt, E.; Pfahl, M.: A new retinoic

acid receptor identified from a hepatocellular carcinoma.
(Letter) *Nature* 333:669–672, 1988.

- [35405] 8957.Brand, N.; Petkovich, M.; Krust, A.; Chambon, P.; de The, H.; Marchio, A.; Tiollais, P.; Dejean, A.: Identification of a second human retinoic acid receptor. (Letter) *Nature* 332: 850–853, 1988.
- [35406] 8958.Dejean, A.; Bougueleret, L.; Grzeschik, K.–H.; Tiollais, P.: Hepatitis B virus DNA integration in a sequence homologous to v-erb-A and steroid receptor genes in a hepatocellular carcinoma. *Nature* 322: 70–72, 1986.
- [35407] 8959.de The, H.; del Mar Vivanco–Ruiz, M.; Tiollais, P.; Stunnenberg, H.; Dejean, A.: Identification of a retinoic acid responsive element in the retinoic acid receptor beta gene. *Nature* 343: 177–180, 1990.
- [35408] 8960.Aruffo, A.; Seed, B.: Molecular cloning of a CD28 cDNA by a high–efficiency COS cell expression system. *Proc. Nat. Acad. Sci.* 84: 8573–8577, 1987.
- [35409] 8961.Lafage–Pochitaloff, M.; Costello, R.; Couez, D.; Simonetti, J.; Mannoni, P.; Mawas, C.; Olive, D.: Human CD28 and CTLA–4 Ig superfamily genes are located on chromosome 2 at bands q33–q34. *Immunogenetics* 31:198–201, 1990.
- [35410] 8962.Lee, K. P.; Taylor, C.; Petryniak, B.; Turka, L. A.; June,

C. H.;Thompson, C. B.: The genomic organization of the CD28 gene: implicationsfor the regulation of CD28 mRNA expression and heterogeneity. J.Immun. 145: 344–352, 1990.

[35411] 8963.Lesslauer, W.; Gmunder, H.; Bohlen, P.: Purification and N-terminalamino acid sequence of the human T90/44 (CD28) antigen. Immunogenetics 27:388–391, 1988.

[35412] 8964.Okkenhaug, K.; Wu, L.; Garza, K. M.; La Rose, J.; Khoo, W.; Odermatt,B.; Mak, T. W.; Ohashi, P. S.; Rottapel, R.: A point mutation inCD28 distinguishes proliferative signals from survival signals. NatureImmun. 2: 325–332, 2001.

[35413] 8965.Dear, T. N.; Colledge, W. H.; Carlton, M. B. L.; Lavenir, I.; Larson,T.; Smith, A. J. H.; Warren, A. J.; Evans, M. J.; Sofroniew, M. V.;Rabbitts, T. H.: The Hox11 gene is essential for cell survival duringspleen development. De-velopment 121: 2909–2915, 1995.

[35414] 8966.Dear, T. N.; Sanchez–Garcia, I.; Rabbitts, T. H.: The HOX11 geneencodes a DNA–binding nuclear transcription factor belonging to adistinct family of homeobox genes. Proc. Nat. Acad. Sci. 90: 4431–4435,1993.

[35415] 8967.Dube, I. D.; Kamel–Reid, S.; Yuan, C. C.; Lu, M.; Wu, X.; Corpus,G.; Raimondi, S. C.; Crist, W. M.; Carroll, A. J.;

Minowada, J.; Baker, J. B.: A novel human homeobox gene lies at the chromosome 10 breakpoint in lymphoid neoplasias with chromosomal translocation t(10;14). *Blood* 78:2996–3003, 1991.

[35416] 8968. Hatano, M.; Roberts, C. W. M.; Minden, M.; Crist, W. M.; Korsmeyer, S. J.: Deregulation of a homeobox gene, HOX11, by the t(10;14) in T cell leukemia. *Science* 253: 79–82, 1991.

[35417] 8969. Kagan, J.; Finan, J.; Letofsky, J.; Besa, E. C.; Nowell, P. C.; Croce, C. M.: Alpha-chain locus of the T-cell antigen receptor is involved in the t(10;14) chromosome translocation of T-cell acute lymphocytic leukemia. *Proc. Nat. Acad. Sci.* 84: 4543–4546, 1987.

[35418] 8970. Kennedy, M. A.; Gonzalez-Sarmiento, R.; Kees, U. R.; Lampert, F.; Dear, N.; Boehm, T.; Rabbitts, T. H.: HOX11, a homeobox-containing T-cell oncogene on human chromosome 10q24. *Proc. Nat. Acad. Sci.* 88:8900–8904, 1991.

[35419] 8971. Lu, M.; Gong, Z. Y.; Shen, W. F.; Ho, A. D.: The TCL-3 proto-oncogene altered by chromosomal translocation in T-cell leukemia codes for a homeobox protein. *EMBO J.* 10: 2905–2910, 1991.

[35420] 8972. Roberts, C. W. M.; Shutter, J. R.; Korsmeyer, S. J.:

Hox11 controlsthe genesis of the spleen. Nature 368: 747–750, 1994.

- [35421] 8973.Zutter, M.; Hockett, R. D.; Roberts, C. W. M.; McGuire, E. A.;Bloomstone, J.; Morton, C. C.; Deaven, L. L.; Crist, W. M.; Carroll,A. J.; Korsmeyer, S. J.: The t(10;14)(q24;q11) of T–cell acute lymphoblasticleukemia juxtaposes the delta T–cell receptor with TCL3, a conservedand activated locus at 10q24. Proc. Nat. Acad. Sci. 87: 3161–3165,1990.
- [35422] 8974.Alarcon, B.; Regueiro, J. R.; Arnaiz–Villena, A.; Terhorst, C.: Familial defect in the surface expression of the T–cell receptor–CD3complex. New Eng. J. Med. 319: 1203–1208, 1988.
- [35423] 8975.Caplan, S.; Zeligier, S.; Wang, L.; Baniyash, M.: Cell–surface–expressedT–cell antigen–receptor epsilon chain is associated with the cytoskeleton. Proc.Nat. Acad. Sci. 92: 4768–4772, 1995.
- [35424] 8976.Clevers, H.; Alarcon, B.; Wileman, T.; Terhorst, C.: The T cellreceptor/CD3 complex: a dynamic protein ensemble. Annu. Rev. Immun. 6:629–662, 1988.
- [35425] 8977.Grakoui, A.; Bromley, S. K.; Sumen, C.; Davis, M. M.; Shaw, A.S.; Allen, P. M.; Dustin, M. L.: The immunological synapse: a molecularmachine controlling T cell activation.

Science 285: 221–227, 1999.

- [35426] 8978.Krummel, M. F.; Sjaastad, M. D.; Wulfing, C.; Davis, M. M.: Differentialclustering of CD4 and CD3–zeta during T cell recognition. Science 289:1349–1352, 2000.
- [35427] 8979.Weissman, A. M.; Baniyash, M.; Hou, D.; Samelson, L. E.; Burgess,W. H.; Klausner, R. D.: Molecular cloning of the zeta chain of theT cell antigen receptor. Science 239: 1018–1021, 1988.
- [35428] 8980.Weissman, A. M.; Hou, D.; Orloff, D. G.; Modi, W. S.; Seuanez,H.; O'Brien, S. J.; Klausner, R. D.: Molecular cloning and chromosomallocalization of the human T–cell receptor zeta chain: distinctionfrom the molecular CD3 complex. Proc. Nat. Acad. Sci. 85: 9709–9713,1988.
- [35429] 8981.Weissman, A. M.; Samelson, L. E.; Klausner, R. D.: A new subunitof the human T–cell antigen receptor complex. Nature 324: 480–482,1986.
- [35430] 8982.Zeng, Z.–H.; Castano, A. R.; Segelke, B. W.; Stura, E. A.; Peterson,P. A.; Wilson, I. A.: Crystal structure of mouse CD1: an MHC–likefold with a large hydrophobic binding groove. Science 277: 339–345,1997.
- [35431] 8983.Audhya, T.; Schlesinger, D. H.; Goldstein, G.: Isolation and completeamino acid sequence of human thymopoietin and splenin. Proc. Nat.Acad. Sci. 84:

3545–3549, 1987.

- [35432] 8984.Berger, R.; Theodor, L.; Shoham, J.; Gokkel, E.; Brok-Simoni, F.;Avraham, K. B.; Copeland, N. G.; Jenkins, N. A.; Rechavi, G.; Simon,A. J.: The characterization and localization of the mouse thymopoietin/lamina-associatedpolypeptide 2 gene and its alternatively spliced products. *GenomeRes.* 6: 361–370, 1996.
- [35433] 8985.Harris, C. A.; Andryuk, P. J.; Cline, S. W.; Mathew, S.; Siekierka,J. J.; Goldstein, G.: Structure and mapping of the human thymopoietin(TMPO) gene and relationship of human TMPO beta to rat lamin-associatedpolypeptide 2. *Genomics* 28: 198–205, 1995.
- [35434] 8986.Eschenfeldt, W. H.; Berger, S. L.: The human prothymosin alphagene is polymorphic and induced upon growth stimulation: evidenceusing a cloned cDNA. *Proc. Nat. Acad. Sci.* 83: 9403–9407, 1986.
- [35435] 8987.Goodall, G. J.; Dominguez, F.; Horecker, B. L.: Molecular cloningof cDNA for human prothymosin alpha. *Proc. Nat. Acad. Sci.* 83: 8926–8928,1986.
- [35436] 8988.Haritos, A. A.; Goodall, G. J.; Horecker, B. L.: Prothymosin alpha:isolation and properties of the major immunoreactive form of thymosinalpha-1 in rat thymus. *Proc. Nat. Acad. Sci.* 81: 1008–1011, 1984.

- [35437] 8989.Manrow, R. E.; Leone, A.; Krug, M. S.; Eschenfeldt, W. H.; Berger,S. L.: The human prothymosin alpha gene family contains several processedpseudogenes lacking deleterious lesions. *Genomics* 13: 319–331, 1992.
- [35438] 8990.Szabo, P.; Panneerselvam, C.; Clinton, M.; Frangou-Lazaridis, M.;Weksler, D.; Whittington, E.; Macera, M. J.; Grzeschik, K.–H.; Selvakumar,A.; Horecker, B. L.: Prothymosin alpha–gene in humans: organizationof its promoter region and localization to chromosome 2. *Hum. Genet.* 90:629–634, 1993.
- [35439] 8991.Demczuk, S.; Aledo, R.; Zucman, J.; Delattre, O.; Desmaze, C.;Dauphinot, L.; Jalbert, P.; Rouleau, G. A.; Thomas, G.; Aurias, A.: Cloning of a balanced translocation breakpoint in the DiGeorge syndromecritical region and isolation of a novel potential adhesion receptorgene in its vicinity. *Hum. Molec. Genet.* 4: 551–558, 1995.
- [35440] 8992.Jourdan–Le Saux, C.; Tomsche, A.; Ujfalusi, A.; Jia, L.; Csiszar,K.: Central nervous system, uterus, heart, and leukocyte expressionof the LOXL3 gene, encoding a novel lysyl oxidase–like protein. *Genomics* 74:211–218, 2001.
- [35441] 8993.Maki, J. M.; Kivirikko, K. I.: Cloning and characterization ofa fourth human lysyl oxidase isoenzyme. *Biochem. J.* 355: 381–387,2001.

- [35442] 8994.Jaju, R. J.; Haas, O. A.; Neat, M.; Harbott, J.; Saha, V.; Boulwood,J.; Brown, J. M.; Pirc–Danoewinata, H.; Krings, B. W.; Muller, U.;Morris, S. W.; Wainscoat, J. S.; Kearney, L.: A new recurrent translocation,t(5;11)(q35p15.5), associated with del(5q) in childhood acute myeloidleukemia. Blood 94: 773–780, 1999.
- [35443] 8995.Kurotaki, N.; Harada, N.; Yoshiura, K.; Sugano, S.; Niikawa, N.;Matsumoto, N.: Molecular characterization of NSD1, a human homologueof the mouse Nsd1 gene. Gene 279: 197–204, 2001.
- [35444] 8996.Wang, X.; Yeh, S.; Wu, G.; Hsu, C.–L.; Wang, L.; Chi–ang, T.; Yang,Y.; Guo, Y.; Chang, C.: Identification and characterization of anovel androgen receptor coregulator ARA267–alpha in prostate cancercells. J. Biol. Chem. 276: 40417–40423, 2001.
- [35445] 8997.Riewald, M.; Petrovan, R. J.; Donner, A.; Mueller, B. M.; Ruf,W.: Activation of endothelial cell protease activated receptor 1by the protein C pathway. Science 296: 1880–1882, 2002.
- [35446] 8998.Jankowski, S. A.; De Jong, P.; Meltzer, P. S.: Genomic structureof SAS, a member of the transmembrane 4 superfamily amplified in humansarcomas. Genomics 25: 501–506, 1995.

- [35447] 8999.Meltzer, P. S.; Jankowski, S. A.; Dal Cin, P.; Sandberg, A. A.; Paz, I. B.; Coccia, M. A.; Smith, S. H.: Identification and cloning of a novel amplified DNA sequence in human malignant fibrous histiocytoma derived from a region of chromosome 12 frequently rearranged in soft tissue tumors. (Abstract) Cytogenet. Cell Genet. 58: 1979 only, 1991.
- [35448] 9000.Peschle, C.; Mavilio, F.; Sposi, N. M.; Giampaolo, A.; Care, A.; Bottero, L.; Bruno, M.; Mastroberardino, G.; Gastaldi, R.; Testa, M. G.; Alimena, G.; Amadori, S.; Mandelli, F.: Translocation and rearrangement of c-myc into immunoglobulin alpha heavy chain locus in primary cells from acute lymphocytic leukemia. Proc. Nat. Acad. Sci. 81: 5514–5518, 1984.
- [35449] 9001.Shao, X.; Tarnasky, H. A.; Schalles, U.; Oko, R.; van der Hoorn, F. A.: Interactional cloning of the 84-kDa major outer dense fiber protein Odf84: leucine zippers mediate associations of Odf84 and Odf27. J. Biol. Chem. 272: 6105–6113, 1997.
- [35450] 9002.Shao, X.; van der Hoorn, F. A.: Self-interaction of the major 27-kilodalton outer dense fiber protein is in part mediated by a leucine zipper domain in the rat. Biol. Reprod. 55: 1343–1350, 1996.

- [35451] 9003.Ammer, H.; Henschen, A.; Lee, C.-H.: Isolation and amino-acidsequence analysis of human sperm protamines P1 and P2: occurrenceof two forms of protamine P2. Biol. Chem. Hoppe-Seyler 367: 515-522,1986.
- [35452] 9004.Cho, C.; Willis, W. D.; Goulding, E. H.; Jung-Ha, H.; Choi, Y.-C.;Hecht, N. B.; Eddy, E. M.: Haploinsufficiency of protamine-1 or -2causes infertility in mice. Nature Genet. 28: 82-86, 2001.
- [35453] 9005.Choudhary, S. K.; Wykes, S. M.; Kramer, J. A; Mo-hamed, A. N.; Koppitch,F.; Nelson, J. E.; Krawetz, S. A.: A haploid expressed gene clusterexists as a single chromatin domain in human sperm. J. Biol. Chem. 270:8755-8762, 1995.
- [35454] 9006.Domenjoud, L.; Nussbaum, G.; Adham, I. M.; Greeske, G.; Engel,W.: Genomic sequences of human protamines whose genes, PRM1 and PRM2,are clustered. Genomics 8: 127-133, 1990.
- [35455] 9007.Engel, W.: Personal Communication. Goettingen, Germany 5/17/1990.
- [35456] 9008.Hecht, N. B.; Kleene, K. C.; Yelick, P. C.; Johnson, P. A.; Pravtcheva,D. D.; Ruddle, F. H.: Mapping of haploid expressed genes: genes forboth mouse protamines are located on chromosome 16. Somat. Cell Molec.Genet. 12:

203–208, 1986.

- [35457] 9009.Krawetz, S. A.; Herfort, M. H.; Hamerton, J. L.; Pon, R. T.; Dixon, G. H.: Chromosomal localization and structure of the human P1 protamine gene. *Genomics* 5: 639–645, 1989.
- [35458] 9010.Reeves, R. H.; Gallahan, D.; O'Hara, B. F.; Callahan, R.; Gearhart, J. D.: Genetic mapping of Prm-1, Igl-1, Smst, Mtv-6, Sod-1, and Ets-2 and localization of the Down syndrome region on mouse chromosome 16. *Cytogenet. Cell Genet.* 44: 76–81, 1987.
- [35459] 9011.Reeves, R. H.; Gearhart, J. D.; Hecht, N. B.; Yelick, P.; Johnson, P.; O'Brien, S. J.: The gene encoding protamine 1 is located on chromosome 16 in humans and near the proximal end of mouse chromosome 16, where it is tightly linked to protamine 2. (Abstract) *Cytogenet. Cell Genet.* 46:680 only, 1987.
- [35460] 9012.Reeves, R. H.; Gearhart, J. D.; Hecht, N. B.; Yelick, P.; Johnson, P.; O'Brien, S. J.: The gene encoding protamine 1 is located on human chromosome 16, and near the proximal end of mouse chromosome 16 where it is tightly linked to the gene encoding protamine 2. *J. Hered.* 80:442–446, 1989.
- [35461] 9013.Viguie, F.; Domenjoud, L.; Rousseau-Merck, M.-F.;

Dadoune, J.-P.;Chevaillier, P.: Chromosomal localization of the human protaminegenes, PRM1 and PRM2, to 16p13.3 by in situ hybridization. Hum. Genet. 85:171–174, 1990.

[35462] 9014.de The, H.; Marchio, A.; Tiollais, P.; Dejean, A.: A novel steroidthyroid hormone receptor–related gene inappropriately expressed inhuman hepatocellular carcinoma. Nature 330: 667–670, 1987.

[35463] 9015.Clevers, H. C.; Dunlap, S.; Wileman, T. E.; Terhorst, C.: HumanCD3–epsilon gene contains three miniexons and is transcribed froma non–TATA promoter. Proc. Nat. Acad. Sci. 85: 8156–8160, 1988.

[35464] 9016.DeJarnette, J. B.; Sommers, C. L.; Huang, K.; Woodside, K. J.;Emmons, R.; Katz, K.; Shores, E. W.; Love, P. E.: Specific requirementfor CD3–epsilon in T cell development. Proc. Nat. Acad. Sci. 95:14909–14914, 1998.

[35465] 9017.Gold, D. P.; Puck, J. M.; Pettey, C. L.; Cho, M.; Coligan, J.;Woody, J. N.; Terhorst, C.: Isolation of cDNA clones encoding the20K non–glycosylated polypeptide chain of the human T–cell receptor/T3complex. Nature 321: 431–434, 1986.

[35466] 9018.Le Deist, F.; Thoenes, G.; Corado, J.; Lisowska–GrosPierre, B.;Fischer, A.: Immunodeficiency with low expression of the T cell receptor/CD3complex: effect on T

lymphocyte activation. *Europ. J. Immun.* 21:1641–1647, 1991.

[35467] 9019.Soudais, C.; de Villartay, J.–P.; Le Deist, F.; Fischer, A.; Lisowska–GrosPierre, B.: Independent mutations of the human CD3–epsilon gene resulting in a T cell receptor/CD3 complex immunodeficiency. *Nature Genet.* 3:77–81, 1993.

[35468] 9020.Thoenes, G.; Soudais, C.; Le Deist, F.; Griscelli, C.; Fischer, A.; Lisowska–GrosPierre, B.: Structural analysis of low TCR–CD3 complex expression in T cells of an immunodeficient patient. *J. Biol. Chem.* 267:487–493, 1992.

[35469] 9021.Wong, S.; Moore, S.; Orisio, S.; Millward, A.; Demaine, A. G.: Susceptibility to type I diabetes in women is associated with the CD3 epsilon locus on chromosome 11. *Clin. Exp. Immun.* 83: 69–73, 1991.

[35470] 9022.DeMartino, G. N.; Proske, R. J.; Moomaw, C. R.; Strong, A. A.; Song, X.; Hisamatsu, H.; Tanaka, K.; Slaughter, C. A.: Identification, purification, and characterization of a PA700–dependent activator of the proteasome. *J. Biol. Chem.* 271: 3112–3118, 1996.

[35471] 9023.Ohana, B.; Moore, P. A.; Ruben, S. M.; Southgate, C. D.; Green, M. R.; Rosen, C. A.: The type 1 human immunodeficiency virus Tat binding protein is a transcriptional

activator belonging to an additional family of evolutionarily conserved genes. *Proc. Nat. Acad. Sci.* 90:138–142, 1993.

[35472] 9024. Patel, A.; Rochelle, J. M.; Jones, J. M.; Sumegi, J.; Uhl, G. R.; Seldin, M. F.; Meisler, M. H.; Gregor, P.: Mapping of the taurine transporter gene to mouse chromosome 6 and to the short arm of human chromosome 3. *Genomics* 25: 314–317, 1995.

[35473] 9025. Ramamoorthy, S.; Leibach, F. H.; Mahesh, V. B.; Han, H.; Yang–Feng, T.; Blakely, R. D.; Ganapathy, V.: Functional characterization and chromosomal localization of a cloned taurine transporter from human placenta. *Biochem. J.* 300: 893–900, 1994.

[35474] 9026. Kreczel, W.; Ghyselinck, N.; Samad, T. A.; Dupe, V.; Kastner, P.; Borrelli, E.; Chambon, P.: Impaired locomotion and dopamine signaling in retinoid receptor mutant mice. *Science* 279: 863–867, 1998.

[35475] 9027. Lotan, R.; Xu, X.–C.; Lippman, S. M.; Ro, J. Y.; Lee, J. S.; Lee, J. J.; Hong, W. K.: Suppression of retinoic acid receptor–beta in premalignant oral lesions and its up–regulation by isotretinoin. *New Eng. J. Med.* 332: 1405–1410, 1995.

[35476] 9028. Mattei, M.–G.; de The, H.; Mattei, J.–F.; Marchio, A.; Tiollais, P.; Dejean, A.: Assignment of the human hap

retinoic acid receptorRAR-beta gene to the p24 band of chromosome 3. Hum. Genet. 80: 189-190,1988.

[35477] 9029.Nadeau, J. H.; Compton, J. G.; Giguere, V.; Rossant, J.; Varmuza,S.: Close linkage of retinoic acid receptor genes with homeobox-and keratin-encoding genes on paralogous segments of mouse chromosomes11 and 15. Mammalian Genome 3: 202-208, 1992.

[35478] 9030.Samad, A.; Kreczel, W.; Chambon, P.; Borrelli, E.: Regulationof dopaminergic pathways by retinoids: activation of the D2 receptorpromoter by members of the retinoic acid receptor-retinoid X receptorfamily. Proc. Nat. Acad. Sci. 94: 14349-14354, 1997.

[35479] 9031.Flagiello, D.; Apiou, F.; Gibaud, A.; Poupon, M. F.; Dutirllaux,B.; Malfoy, B.: Assignment of the genes for cellular retinoic acidbinding protein 1 (CRABP1) and 2 (CRABP2) to human chromosome band15q24 and 1q21.3, respectively, by in situ hybridization. Cytogenet.Cell Genet. 76: 17-18, 1997.

[35480] 9032.Geurts van Kessel, A.; de Leeuw, H.; Dekker, E. J.; Rijks, L.;Spurr, N.; Ledbetter, D.; Kootwijk, E.; Vaessen, M. J.: Localizationof the cellular retinoic acid binding protein (CRABP) gene relativeto the acute promyelocytic leukemia-associated breakpoint on humanchromosome 15. Hum.

Genet. 87: 201–204, 1991.

- [35481] 9033. MacGregor, T. M.; Copeland, N. G.; Jenkins, N. A.; Giguere, V.: The murine gene for cellular retinoic acid-binding protein type I: genomic organization, chromosomal localization, and post-transcriptional regulation by retinoic acid. *J. Biol. Chem.* 267: 7777–7783, 1992.
- [35482] 9034. Ong, D. E.: Cellular retinoid-binding proteins. *Arch. Derm.* 123: 1693–1695, 1987.
- [35483] 9035. Vaessen, M. J.; Rijks, L.; Dekker, E. J.; Kootwijk, E.; Bootsma, D.; Westerveld, A.; Geurts van Kessel, A. H. M.: Localization of the human cellular retinoic acid-binding protein (CRABP) on chromosome 15. (Abstract) *Cytogenet. Cell Genet.* 51: 1094 only, 1989.
- [35484] 9036. Elder, J. T.; Astrom, A.; Pettersson, U.; Voorhees, J. J.; Trent, J. M.: Assignment of the human CRABP-II gene to chromosome 1q21 by nonisotopic in situ hybridization. *Hum. Genet.* 89: 487–490, 1992.
- [35485] 9037. Flagiello, D.; Apiou, F.; Gibaud, A.; Poupon, M. F.; Dutrillaux, B.; Malfoy, B.: Assignment of the genes for cellular retinoic acid binding protein 1 (CRABP1) and 2 (CRABP2) to human chromosome bands 15q24 and 1q21.3, respectively, by in situ hybridization. *Cytogenet. Cell Genet.* 76: 17–18, 1997.

- [35486] 9038. Giguere, V.; Lyn, S.; Yip, P.; Siu, C.-H.; Amin, S.: Molecular cloning of cDNA encoding a second cellular retinoic acid-binding protein. *Proc. Nat. Acad. Sci.* 87: 6233–6237, 1990.
- [35487] 9039. Dieterich, W.; Ehnis, T.; Bauer, M.; Donner, P.; Volta, U.; Riecken, E. O.; Schuppan, D.: Identification of tissue transglutaminase as the autoantigen of celiac disease. *Nature Med.* 3: 797–801, 1997.
- [35488] 9040. Gentile, V.; Davies, P. J. A.; Baldini, A.: The human tissue transglutaminase gene maps on chromosome 20q12 by in situ fluorescence hybridization. *Genomics* 20:295–297, 1994.
- [35489] 9041. Gentile, V.; Saydak, M.; Chiocca, E. A.; Akande, O.; Birckbichler, P. J.; Lee, K. N.; Stein, J. P.; Davies, P. J. A.: Isolation and characterization of cDNA clones to mouse macrophage and human endothelial cell tissue transglutaminases. *J. Biol. Chem.* 266: 478–483, 1991.
- [35490] 9042. Lu, S.; Saydak, M.; Gentile, V.; Stein, J. P.; Davies, P. J. A.: Isolation and characterization of the human tissue transglutaminase gene promoter. *J. Biol. Chem.* 270: 9748–9756, 1995.
- [35491] 9043. Wang, M.; Kim, I.-G.; Steinert, P. M.; McBride, O. W.: Assignment of the human transglutaminase 2 (TGM2) and

transglutaminase 3 (TGM3) genes to chromosome 20q11.2. *Genomics* 23: 721–722, 1994.

- [35492] 9044. Dodd, J.; Morton, S. B.; Karagogeos, D.; Yamamoto, M.; Jessell, T. M.: Spatial regulation of axonal glycoprotein expression on subsets of embryonic spinal neurons. *Neuron* 1: 105–116, 1988.
- [35493] 9045. Freigang, J.; Proba, K.; Leder, L.; Diederichs, K.; Sonderegger, P.; Welte, W.: The crystal structure of the ligand binding module of axonin-1/TAG-1 suggests a zipper mechanism for neural cell adhesion. *Cell* 101: 425–433, 2000.
- [35494] 9046. Kenwrick, S.; Leversha, M.; Rooke, L.; Hasler, T.; Sonderegger, P.: Localization of the human TAX-1 gene to 1q32.1: a region implicated in microcephaly and Van der Woude syndrome. *Hum. Molec. Genet.* 2: 1461–1462, 1993.
- [35495] 9047. Kozlov, S. V.; Giger, R. J.; Hasler, T.; Korvatska, E.; Schorderet, D. F.; Sonderegger, P.: The human TAX1 gene encoding the axon-associated cell adhesion molecule TAG-1/axonin-1: genomic structure and basic promoter. *Genomics* 30: 141–148, 1995.
- [35496] 9048. Rickman, D. S.; Tyagi, R.; Zhu, X.-X.; Bobek, M. P.; Song, S.; Blaivas, M.; Miesek, D. E.; Israel, M. A.; Kurnit, D.

M.; Ross, D.A.; Kish, P. E.; Hanash, S. M.: The gene for the axonal cell adhesion molecule TAX-1 is amplified and aberrantly expressed in malignant gliomas. *Cancer Res.* 61: 2162–2168, 2001.

[35497] 9049. Tsiotra, P. C.; Karagogeos, D.; Theodorakis, K.; Michaelisis, T.M.; Modi, W. S.; Furley, A. J.; Jessell, T. M.; Papamatheakis, J.: Isolation of the cDNA and chromosomal localization of the gene (TAX1) encoding the human axonal glycoprotein TAG-1. *Genomics* 18: 562–567, 1993.

[35498] 9050. Artavanis-Tsakonas, S.; Matsuono, K.; Fortini, M.: Notch signaling. *Science* 268:225–232, 1995.

[35499] 9051. Brou, C.; Logeat, F.; Gupta, N.; Bessia, C.; Le Bail, O.; Doedens, J. R.; Cumano, A.; Roux, P.; Black, R. A.; Israel, A.: A novel proteolytic cleavage involved in Notch signaling: the role of the disintegrin-metalloprotease TACE. *Molec. Cell* 5: 207–216, 2000.

[35500] 9052. Bruckner, K.; Perez, L.; Clausen, H.; Cohen, S.: Glycosyltransferase activity of Fringe modulates Notch-Delta interactions. *Nature* 406:411–415, 2000.

[35501] 9053. Ackerman, M. J.; Clapham, D. E.: Ion channels – basic science and clinical disease. *New Eng. J. Med.* 336: 1575–1586, 1997.

[35502] 9054. Ackerman, M. J.; Schroeder, J. J.; Berry, R.; Schaid, D.

J.; Porter,C.-B. J.; Michels, V. V.; Thibodeau, S. N.: A novel mutation in KVLQT1is the molecular basis of inherited long QT syndrome in a near-drowningpatient's family. Pe-
diat. Res. 44: 148-153, 1998.

[35503] 9055.Ackerman, M. J.; Tester, D. J.; Porter, C. J.; Edwards, W. D.:Molecular diagnosis of the inherited long-QT syn-
drome in a woman whodied after near-drowning. New
Eng. J. Med. 341: 1121-1125, 1999.

[35504] 9056.Barlow, D. P.: Box: KVLQT1 complexities in Beck-
with-Wiedeman (sic)syndrome. Nature Genet. 15: 114
only, 1997.

[35505] 9057.Barlow, J. B.; Bosman, C. K.; Cochrane, J. W. C.: Con-
genital cardiacarrhythmia. Lancet II: 531 only, 1964.

[35506] 9058.Baudouy, P.; Andreassian, B.; Attuel, P.; Greze, M.;
Soulie, J.;Fruchaud, J.: Syndrome de Romano-Ward et stel-
lectomie gauche: revuegenerale a propos d'un nouveau
cas. Arch. Mal. Coeur 70: 645-652,1977.

[35507] 9059.Benhorin, J.; Kalman, Y. M.; Medina, A.; Towbin, J.;
Rave-Harel,N.; Dyer, T. D.; Blangero, J.; MacCluer, J. W.;
Kerem, B.: Evidenceof genetic heterogeneity in the long
QT syndrome. Science 260: 1960-1962,1993.

[35508] 9060.Bhandari, A. K.; Scheinman, M.: The long QT syn-
drome. Mod. ConceptsCardiovasc. Dis. 54: 45-50, 1985.

- [35509] 9061. Bonduelle, M.: Personal Communication. Brussels, Belgium 5/30/1993.
- [35510] 9062. Casimiro, M. C.; Knollmann, B. C.; Ebert, S. N.; Vary, J. C., Jr.; Greene, A. E.; Franz, M. R.; Grinberg, A.; Huang, S. P.; Pfeifer, K.: Targeted disruption of the *Kcnq1* gene produces a mouse model of Jervell and Lange-Nielsen syndrome. *Proc. Nat. Acad. Sci.* 98:2526–2531, 2001.
- [35511] 9063. Chen, Q.; Zhang, D.; Gingell, R. L.; Moss, A. J.; Napolitano, C.; Priori, S. G.; Schwartz, P. J.; Kehoe, E.; Robinson, J. L.; Schulze-Bahr, E.; Wang, Q.; Towbin, J. A.: Homozygous deletion in *KVLQT1* associated with Jervell and Lange-Nielsen syndrome. *Circulation* 99: 1344–1347, 1999.
- [35512] 9064. Cleary, M. A.; van Raamsdonk, C. D.; Levorse, J.; Zheng, B.; Bradley, A.; Tilghman, S. M.: Disruption of an imprinted gene cluster by a targeted chromosomal translocation in mice. *Nature Genet.* 29: 78–82, 2001.
- [35513] 9065. Curran, M.; Atkinson, D.; Timothy, K.; Vincent, G. M.; Moss, A. J.; Leppert, M.; Keating, M.: Locus heterogeneity of autosomal dominant long QT syndrome. *J. Clin. Invest.* 92: 799–803, 1993.
- [35514] 9066. DeSilvey, D. L.; Moss, A. J.: Primidone in the treatment of the long QT syndrome: QT shortening and ventric-

ular arrhythmia suppression. Ann.Intern. Med. 93: 53–54, 1980.

[35515] 9067.DiSegni, E.; David, D.; Katzenstein, M.; Klein, H. O.; Kaplinsky,E.; Levy, M. J.: Permanent overdrive pacing for the suppression of recurrent ventricular tachycardia in a newborn with long QT syndrome. J.Electrocardiol. 13: 189–192, 1980.

[35516] 9068.Donger, C.; Denjoy, I.; Berthet, M.; Neyroud, N.; Cruaud, C.;Bennaceur, M.; Chivoret, G.; Schwartz, K.; Coumel, P.; Guicheney,P.: KVLQT1 C-terminal missense mutation causes a forme fruste long-QT syndrome. Circulation 96: 2778–2781, 1997.

[35517] 9069.Engel, J. R.; et al.; et al.: Epigenotype–phenotype correlations in Beckwith–Wiedemann syndrome. J. Med. Genet. 37: 921–926, 2000.

[35518] 9070.Fitzpatrick, G. V.; Soloway, P. D.; Higgins, M. J.: Regional loss of imprinting and growth deficiency in mice with a targeted deletion of KvDMR1. Nature Genet. 9 Sept, 2002. Note: Advance Electronic Publication.

[35519] 9071.Flugelman, M. Y.; Pollack, S.; Hammerman, H.; Riss, E.; Barzilai,D.: Congenital prolongation of Q–T interval: a family study of three generations. Cardiology 69: 170–174, 1982.

- [35520] 9072.Furberg, C.; Hornell, H.: Familial QT prolongation and risk of sudden death. *Acta Paediat. Scand.* 64: 777–782, 1975.
- [35521] 9073.Gale, G. E.; Bosman, C. K.; Tucker, R. B. K.; Barlow, J. B.: Hereditary prolongation of Q–T interval: study of two families. *Brit. Heart J.* 32: 505–509, 1970.
- [35522] 9074.Gamstorp, I.; Nilsen, R.; Westling, H.: Congenital cardiac arrhythmia.(Letter) *Lancet* II: 965 only, 1964.
- [35523] 9075.Garza, L. A.; Vick, R. L.; Nora, J. J.; McNamara, D. G.: Heritable Q–T prolongation without deafness. *Circulation* 41: 39–48, 1970.
- [35524] 9076.Giuffre, R. M.; Hejtmancik, J. F.; McCabe, E. R. B.; Towbin, J.A.: Long QT (Romano–Ward) syndrome: molecular genetic evidence against tight HLA linkage. (Abstract) *Am. J. Hum. Genet.* 47 (suppl.): A180 only, 1990.
- [35525] 9077.Gohl, K.; Feistel, H.; Weikl, A.; Bachmann, K.; Wolf, F.: Congenital myocardial sympathetic dysinnervation (CMSD)—a structural defect of idiopathic long QT syndrome. *PACE* 14: 1544–1553, 1991.
- [35526] 9078.Hashiba, K.: Hereditary QT prolongation syndrome in Japan: genetic analysis and pathological findings of the conduction system. *Jpn.Circ. J.* 42: 1133–1150, 1978.
- [35527] 9079.Horn, C. A.; Beekman, R. H.; Dick, M., II; Lacina, S. J.:

The congenital long QT syndrome: an unusual cause of childhood seizures. *Am.J. Dis. Child.* 140: 659–661, 1986.

[35528] 9080. Itoh, S.; Munemura, S.; Satoh, H.: A study of the inheritance pattern of Romano–Ward syndrome: prolonged Q–T interval, syncope, and sudden death. *Clin. Pediat.* 21: 20–24, 1982.

[35529] 9081. Bulle, F.; Mattei, M. G.; Siegrist, S.; Pawlak, A.; Passage, E.; Chobert, M. N.; Laperche, Y.; Guellaen, G.: Assignment of the human gamma–glutamyl transferase gene to the long arm of chromosome 22. *Hum. Genet.* 76: 283–286, 1987.

[35530] 9082. Miller, W. L.: Phenotypic heterogeneity associated with the splicing mutation in congenital adrenal hyperplasia due to 21–hydroxylase deficiency. *J. Clin. Endocr. Metab.* 82: 1304 only, 1997. 100. Miller, W. L.: Congenital adrenal hyperplasia. (Letter) *New Eng. J. Med.* 317: 1413–1414, 1987. 101. Morel, Y.; Andre, J.; Uring–Lambert, B.; Hauptmann, G.; Betuel, H.; Tossi, M.; Forest, M. G.; David, M.; Bertrand, J.; Miller, W. L.: Rearrangements and point mutations of P450c21 genes are distinguished in five restriction endonuclease haplotypes identified by a new probing strategy in 57 families with congenital adrenal hyperplasia. *J. Clin. Invest.* 83: 527–536,

1989.102. Mornet, E.; Boue, J.; Raux-Demay, M.; Couillin, P.; Oury, J.F.; Dumez, Y.; Dausset, J.; Cohen, D.; Boue, A.: First trimester prenatal diagnosis of 21-hydroxylase deficiency by linkage analysis to HLA-DNA probes and by 17-hydroxyprogesterone determination. *Hum. Genet.* 73: 358-364, 1986.103. Mornet, E.; Crete, P.; Kuttann, F.; Raux-Demay, M.-C.; Boue, J.; White, P. C.; Boue, A.: Distribution of deletions and seven point mutations on CYP21B genes in three clinical forms of steroid 21-hydroxylase deficiency. *Am. J. Hum. Genet.* 48: 79-88, 1991.104. Mulaikal, R. M.; Migeon, C. J.; Rock, J. A.: Fertility rates in female patients with congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *New Eng. J. Med.* 316: 178-182, 1987.105. Murtaza, L.; Sibert, J. R.; Hughes, I.; Balfour, I. C.: Congenital adrenal hyperplasia - a clinical and genetic survey: are we detecting male salt-losers? *Arch. Dis. Child.* 55: 622-625, 1980.106. Murtaza, L. M.; Hughes, I. A.; Sibert, J. R.; Balfour, I. C.: HLA and congenital adrenal hyperplasia. (Letter) *Lancet* II: 524 only, 1978.107. New, M. I.; Dupont, B.; Pang, S.; Pollack, M.; Levine, L. S.: An update of congenital adrenal hyperplasia. *Recent Prog. Horm. Res.* 37: 105-181, 1981.108. New, M. I.; Dupont, B.; Pollack, M. S.; Levine, L. S.: The biochemi-

calbasis for genotyping 21-hydroxylase deficiency. Hum. Genet. 58:123–127, 1981.109. New, M. I.; Levine, L. S.: Recent advances in 21-hydroxylase deficiency. Ann. Rev. Med. 35: 649–663, 1984.110. New, M. I.; Lorenzen, F.; Lerner, A. J.; Kohn, B.; Oberfield, S. E.; Pollack, M. S.; Dupont, B.; Stoner, E.; Levy, D. J.; Pang, S.; Levine, L. S.: Genotyping steroid 21-hydroxylase deficiency: hormonal reference data. J. Clin. Endocr. Metab. 57: 320–326, 1983.111. New, M. I.; Wilson, R. C.: Steroid disorders in children: congenital adrenal hyperplasia and apparent mineralocorticoid excess. Proc. Nat. Acad. Sci. 96: 12790–12797, 1999.112. Nikoshkov, A.; Lajic, S.; Holst, M.; Wedell, A.; Luthman, H.: Synergistic effect of partially inactivating mutations in steroid 21-hydroxylase deficiency. J. Clin. Endocr. Metab. 82: 194–199, 1997.113. Nimkarn, S.; Cerame, B. I.; Wei, J.-Q.; Dunic, M.; Zunec, R.; Brkljacic, L.; Skrabic, V.; New, M. I.; Wilson, R. C.: Congenital adrenal hyperplasia (21-hydroxylase deficiency) without demonstrable genetic mutations. J. Clin. Endocr. Metab. 84: 378–381, 1999.114. Nordenstrom, A.; Marcus, C.; Axelsson, M.; Wedell, A.; Ritzen, E. M.: Failure of cortisone acetate treatment in congenital adrenal hyperplasia because of defective 11-beta-hydroxysteroid dehydroge-

nasereductase activity. J. Clin. Endocr. Metab. 84:
 1210–1213, 1999.115. Nordenstrom, A.; Thilen, A.; Ha-
 genfeldt, L.; Larsson, A.; Wedell, A.: Genotyping is a valu-
 able diagnostic complement to neonatal screening for
 congenital adrenal hyperplasia due to steroid
 21-hydroxylase deficiency. J. Clin. Endocr. Metab. 84:
 1505–1509, 1999.116. Olney, R. C.; Mougey, E. B.; Wang,
 J.; Shulman, D. I.; Sylvester, J. E.: Using real-time, quantita-
 tive PCR for rapid genotyping of the steroid
 21-hydroxylase gene in a north Florida population. J. Clin.
 Endocr. Metab. 87: 735–741, 2002.117. Orta-Flores, Z.;
 Cantu, J. M.; Dominguez, O. V.: Reciprocal interactions of
 progesterone and 17- α -OH-progesterone as exoge-
 nous substrates for rat adrenal 21-hydroxylase. J. Steroid
 Biochem. 7:761–767, 1976.118. Owerbach, D.; Crawford,
 Y. M.; Draznin, M. B.: Direct analysis of CYP21B genes in
 21-hydroxylase deficiency using polymerase chain reac-
 tion amplification. Molec. Endocr. 4: 125–131, 1990.119.
 Owerbach, D.; Sherman, L.; Ballard, A.-L.; Azziz, R.:
 Pro453-to-ser mutation in CYP21 is associated with non-
 classic steroid 21-hydroxylase deficiency. Molec. Endocr.
 6: 1211–1215, 1992.120. Pang, S.; Murphey, W.; Levine,
 L. S.; Spence, D. A.; Leon, A.; LaFranchi, S.; Surve, A. S.;

New, M. I.: A pilot newborn screening program for congenital adrenal hyperplasia in Alaska. *J. Clin. Endocr. Metab.* 55: 413–420, 1982.121. Pang, S.; Pollack, M. S.; Loo, M.; Green, O.; Nussbaum, R.; Clayton, G.; Dupont, B.; New, M. I.: Pitfalls of prenatal diagnosis of 21-hydroxylase deficiency congenital adrenal hyperplasia. *J. Clin. Endocr. Metab.* 61:89–97, 1985.122. Partanen, J.; Campbell, R. D.: Substitution of ile172-to-asn in the steroid 21-hydroxylase B (P450c21B) gene in a Finnish patient with the simple virilizing form of congenital adrenal hyperplasia. *Hum. Genet.* 87: 716–720, 1991.123. Paulino, L. C.; Araujo, M.; Guerra, G., Jr.; Marini, S. H.; DeMello, M. P.: Mutation distribution and CYP21/C4 locus variability in Brazilian families with the classical form of the 21-hydroxylase deficiency. *Acta Paediat.* 88: 275–83, 1999.124. Petersen, F.; Knudsen, F. U.; Nielsen, M. D.; Mikkelsen, M.: Congenital adrenal hyperplasia associated with a balanced 13–18 translocation. *Europ. J. Pediat.* 133: 283–285, 1980.125. Petersen, K. E.; Svejgaard, A.; Nielsen, M. D.; Dissing, J.: Heterozygotes and cryptic patients in families of patients with congenital adrenal hyperplasia (21-hydroxylase deficiency): HLA and glyoxalase typing and hormonal studies. *Hormone Res.* 16:

151–159, 1982.126. Pollack, M. S.; Keenan, B.; Christiansen, F. T.; Cobain, T. J.; Dawkins, R. L.; Clayton, G.: The immunological detection of a 21-OH deficiency mutation HLA supratype. *Am. J. Hum. Genet.* 38: 688–698, 1986.127. Pollack, M. S.; Levine, L. S.; O'Neill, G. J.; Pang, S.; Lorenzen, F.; Kohn, B.; Rondanini, G. F.; Chiumello, G.; New, M. I.; Dupont, B.: HLA linkage and B14, DR1, BfS haplotype association with the genes for late onset and cryptic 21-hydroxylase deficiency. *Am. J. Hum. Genet.* 33: 540–550, 1981.128. Pollack, M. S.; Levine, L. S.; Pang, S.; Owens, R. P.; Nitowsky, H. M.; Maurer, D.; New, M. I.; Duchon, M.; Merkat, I. R.; Sachs, G.; Dupont, B.: Prenatal diagnosis of congenital adrenal hyperplasia (21-hydroxylase deficiency) by HLA typing. *Lancet* I: 1107–1108, 1979.129. Pollack, M. S.; New, M. I.; O'Neill, G. J.; Levine, L. S.; Callaway, C.; Pang, S.; Cacciari, E.; Mantero, F.; Cassio, A.; Scaroni, C.; Chiumello, G.; Rondanini, G. F.; Gargantini, L.; Giovannelli, G.; Viridis, R.; Bartolotta, E.; Migliori, C.; Pintor, C.; Tato, L.; Barboni, F.; Dupont, B.: HLA genotypes and HLA-linked genetic markers in Italian patients with classical 21-hydroxylase deficiency. *Hum. Genet.* 58: 331–337, 1981.130. Prader, A.: Die Häufigkeit des kongenitalen adrenogenitalen Syn-

droms. *Helv. Paediat. Acta* 13: 426–431, 1958.131.

Prader, A.; Anders, G. J. P. A.; Habich, H.: Zur Genetik deskongenitalen adrenogenitalen Syndroms (virilisierende Nebennierenhyperplasia). *Helv. Paediat. Acta* 17: 271–284, 1962.132. Pucholt, V.; Fitzsimmons, J. S.; Gelsthorpe, K.; Reynolds, M.A.; Milner, R. D. G.: Location of the gene for 21-hydroxylase deficiency. *J. Med. Genet.* 17: 447–452, 1980.133. Qazi, Q. H.; Thompson, M. W.: Incidence of salt-losing form of congenital virilizing adrenal hyperplasia. *Arch. Dis. Child.* 47:302–303, 1972.134. Ravichandran, R.; Lafferty, F.; McGinniss, M. J.; Taylor, H.C.: Congenital adrenal hyperplasia presenting as massive adrenal incidentalomas in the sixth decade of life: report of two patients with 21-hydroxylase deficiency. *J. Clin. Endocr. Metab.* 81: 1776–1779, 1996.135. Reindollar, R. H.; Lewis, J. B.; White, P. C.; Fernhoff, P. M.; McDonough, P. G.; Whitney, J. B., III: Prenatal diagnosis of 21-hydroxylase deficiency by the complementary deoxyribonucleic acid probe for cytochrome P-450(C-21OH). *Am. J. Obstet. Gynec.* 158: 545–547, 1988.136. Rodrigues, N. R.; Dunham, I.; Yu, C. Y.; Carroll, M. C.; Porter, R. R.; Campbell, R. D.: Molecular characterization of the HLA-linked steroid 21-hydroxylase B gene from an individual

with congenital adrenalhyperplasia. EMBO J. 6: 1653–1661, 1987.137. Rosenmann, A.; Schumert, Z.; Theodor, R.; Cohen, T.; Brautbar,C.: Amniotic 17–alpha hydroxyprogesterone and HLA typing for theprenatal diagnosis of 21–alpha hydroxylase deficiency–congenitaladrenal hyperplasia. Am. J. Med. Genet. 6: 295–300, 1980.138. Rumsby, G.; Carroll, M. C.; Porter, R. R.; Grant, D. B.; Hjelm,M.: Deletion of the steroid 21–hydroxylase and complement C4 genesin congenital adrenal hyperplasia. J. Med. Genet. 23: 204–209, 1986.139. Schneider, P. M.; Carroll, M. C.; Alper, C. A.; Rittner, C.;Whitehead, A. S.; Yunis, E. J.; Colten, H. R.: Polymorphism of thehuman complement C4 and steroid 21–hydroxylase genes: restrictionfragment length polymorphisms revealing structural deletions, homoduplications,and size variants. J. Clin. Invest. 78: 650–657, 1986.140. Sherman, S. L.; Aston, C. E.; Morton, N. E.; Speiser, P. W.;New, M. I.: A segregation and linkage study of classical and nonclassical21–hydroxylase deficiency. Am. J. Hum. Genet. 42: 830–838, 1988.141. Sinnott, P.; Collier, S.; Costigan, C.; Dyer, P. A.; Harris,R.; Strachan, T.: Genesis by meiotic unequal crossover of a de novodeletion that contributes to a steroid 21–hydroxylase defi–

ciency. *Proc.Nat. Acad. Sci.* 87: 2107–2111, 1990.142.

Sinnott, P. J.; Dyer, P. A.; Price, D. A.; Harris, R.; Strachan, T.: 21-hydroxylase deficiency families with HLA identical affected and unaffected sibs. *J. Med. Genet.* 26: 10–17, 1989.143.

Slominski, A.; Ermak, G.; Mihm, M.: ACTH receptor, CYP11A1, CYP17 and CYP21A2 genes are expressed in skin. *J. Clin. Endocr. Metab.* 81:2746–2749, 1996.144.

Sobel, D. O.; Gutai, J. P.; Jones, J. C.; Wagener, D. D.; Smith, W.: Detection of heterozygote of 21 hydroxylase deficiency. (Letter) *Lancet* I:47 only, 1980.145.

Speiser, P. W.; Dupont, B.; Rubinstein, P.; Piazza, A.; Kastelan, A.; New, M. I.: High frequency of nonclassical steroid 21-hydroxylase deficiency. *Am. J. Hum. Genet.* 37: 650–667, 1985.146.

Speiser, P. W.; Dupont, J.; Zhu, D.; Serrat, J.; Buegeleisen, M.; Tusie-Luna, M.-T.; Lesser, M.; New, M. I.; White, P. C.: Disease expression and molecular genotype in congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *J. Clin. Invest.* 90: 584–595, 1992.147.

Speiser, P. W.; New, M. I.: Genetics of steroid 21-hydroxylase deficiency. *Trends Genet.* 1: 275–278, 1985.148.

Speiser, P. W.; New, M. I.; Tannin, G. M.; Pickering, D.; Yang, S. Y.; White, P. C.: Genotype of Yupik Eskimos with congenital adrenal hyperplasia due to

21-hydroxylase deficiency. Hum. Genet. 88:

647–648,1992.149. Speiser, P. W.; New, M. I.; White, P. C.:

Molecular genetic analysis of nonclassic steroid

21-hydroxylase deficiency associated with HLA-B14,DR1.

New Eng. J. Med. 319: 19–23, 1988.150. Speiser, P. W.;

New, M. I.; White, P. C.: Clinical and genetic characteriza-

tion of nonclassic 21-hydroxylase deficiency. Endocr. Res.

15: 257–276, 1989.151. Spiro, R. P.; Christian, S. L.; Led-

better, D. H.; New, M. I.; Wilson, R. C.; Roizen, N.; Rosen-

field, R. L.: Intrauterine growth retardation associated with

maternal uniparental disomy for chromosome 6 unmasked

by congenital adrenal hyperplasia. Pediat. Res. 46:

510–513,1999.152. Stikkelbroeck, N. M. M. L.; Otten, B.

J.; Pasic, A.; Jager, G. J.; Sweep, C. G. J. (Fred); Noordam, K.;

Hermus, A. R. M. M.: High prevalence of testicular adrenal

rest tumors, impaired spermatogenesis, and Leydig cell

failure in adolescent and adult males with congenital ad-

drenal hyperplasia. J. Clin. Endocr. Metab. 86: 5721–5728,

2001.153. Tajima, T.; Fujieda, K.; Fujii-Kuriyama, Y.: De

novo mutation causes steroid 21-hydroxylase deficiency in

one family of HLA-identical affected and unaffected indi-

viduals. J. Clin. Endocr. Metab. 77:86–89, 1993.154.

Tajima, T.; Fujieda, K.; Nakae, J.; Toyoura, T.; Shimozawa,

K.;Kusuda, S.; Goji, K.; Nagashima, T.; Cutler, G. B., Jr.: Molecularbasis of nonclassical steroid 21-hydroxylase deficiency detected byneonatal mass screening in Japan. J. Clin. Endocr. Metab. 82: 2350–2356,1997.155. Tajima, T.; Fujieda, K.; Nakayama, K.; Fujii–Kuriyama, Y.: Molecular–analysis of patient and carrier genes with congenital steroid 21–hydroxylasedeficiency by using polymerase chain reaction and single strand conformationpolymorphism. J. Clin. Invest. 92: 2182–2190, 1993.156. Thilen, A.; Larsson, A.: Congenital adrenal hyperplasia in Sweden1969–1986: prevalence, symptoms and age at diagnosis. Acta Paediat.Scand. 79: 168–175, 1990.157. Travitz, J.; Metzger, D. L.: Antenatal treatment for classic21–hydroxylase forms of congenital adrenal hyperplasia and the issues. Genet.Med. 1: 224–230, 1999.158. Tusie–Luna, M.–T.; Speiser, P. W.; Dunic, M.; New, M. I.; White,P. C.: A mutation (pro30–to–leu) in CYP21 represents a potentialnonclassic steroid 21–hydroxylase deficiency allele. Molec. Endocr. 5:685–692, 1991.159. Tusie–Luna, M.–T.; White, P. C.: Gene conversions and unequal–crossovers between CYP21 (steroid 21–hydroxylase gene) and CYP21Pinvolve different mechanisms. Proc. Nat. Acad. Sci. 92: 10796–10800,1995.160. Tusie–Luna, M. T.; Trak–

tman, P.; White, P. C.: Determination of functional effects of mutations in the steroid 21-hydroxylase gene (CYP21) using recombinant vaccinia virus. *J. Biol. Chem.* 265: 20916–20922, 1990.161. Urabe, K.; Kimura, A.; Harada, F.; Iwanaga, T.; Sasazuki, T.: Gene conversion in steroid 21-hydroxylase genes. *Am. J. Hum. Genet.* 46:1178–1186, 1990.162. Urban, M. D.; Lee, P. A.; Migeon, C. J.: Adult height and fertility in men with congenital virilizing adrenal hyperplasia. *New Eng. J. Med.* 299: 1392–1396, 1978.163. Warsof, S. L.; Larsen, J. W.; Kent, S. G.; Rosenbaum, K. N.; August, G. P.; Migeon, C. J.; Schulman, J. D.: Prenatal diagnosis of congenital adrenal hyperplasia. *Obstet. Gynec.* 55: 751–754, 1980.164. Webb, T.; Mackintosh, P.; Wells, L. J.: Cytogenetic evidence for the localisation of the gene for congenital adrenal hyperplasia. *Clin. Genet.* 17: 349–354, 1980.165. Wedell, A.: Molecular genetics of congenital adrenal hyperplasia (21-hydroxylase deficiency): implications for diagnosis, prognosis and treatment. *Acta Paediat.* 87: 159–164, 1998.166. Wedell, A.: An update on the molecular genetics of congenital adrenal hyperplasia: diagnostic and therapeutic aspects. *J. Pediatr. Endocr. Metab.* 11: 581–589, 1998.167. Wedell, A.; Luthman, H.: Steroid

21-hydroxylase deficiency: two additional mutations in salt-wasting disease and rapid screening of disease-causing mutations. *Hum. Molec. Genet.* 2: 499-504, 1993.168. Wedell, A.; Ritzen, E. M.; Haglund-Stengler, B.; Luthman, H.: Steroid 21-hydroxylase deficiency: three additional mutated alleles and establishment of phenotype-genotype relationships of common mutations. *Proc. Nat. Acad. Sci.* 89: 7232-7236, 1992.169. Wedell, A.; Thilen, A.; Ritzen, E. M.; Stengler, B.; Luthman, H.: Mutational spectrum of the steroid 21-hydroxylase gene in Sweden: implications for genetic diagnosis and association with disease malformation. *J. Clin. Endocr. Metab.* 78: 1145-1152, 1994.170. Wenzel, U.; Schneider, M.; Zachmann, M.; Knorr-Murset, G.; Weber, A.; Prader, A.: Intelligence of patients with congenital adrenal hyperplasia due to 21-hydroxylase deficiency, their parents and unaffected siblings. *Helv. Paediat. Acta* 33: 11-16, 1978.171. Werder, E. A.; Siebenmann, R. E.; Knorr-Murset, G.; Zimmermann, A.; Sizonenko, P. C.; Theintz, P.; Girard, J.; Zachmann, M.; Prader, A.: The incidence of congenital adrenal hyperplasia in Switzerland--a survey of patients born in 1960 to 1974. *Helv. Paediat. Acta* 35: 5-11, 1980.172. Werkmeister, J. W.; New, M. I.; Dupont, B.;

White, P. C.: Frequent deletion and duplication of the steroid 21-hydroxylase genes. *Am.J. Hum. Genet.* 39: 461–469, 1986.173. White, P. C.; Chaplin, D. D.; Weis, J. H.; Dupont, B.; New, M.I.; Seidman, J. G.: Two steroid 21-hydroxylase genes are located in the murine S region. *Nature* 312: 465–467, 1984.174. White, P. C.; Grossberger, D.; Onufer, B. J.; Chaplin, D. D.; New, M. I.; Dupont, B.; Strominger, J. L.: Two genes encoding steroid 21-hydroxylase are located near the genes encoding the fourth component of complement in man. *Proc. Nat. Acad. Sci.* 82: 1089–1093, 1985.175. White, P. C.; New, M.; Dupont, B.: Congenital adrenal hyperplasia. (Letter) *New Eng. J. Med.* 317: 1414–1415, 1987.176. White, P. C.; New, M. I.; Dupont, B.: Congenital adrenal hyperplasia. *New Eng. J. Med.* 316: 1519–1524, 1987.177. White, P. C.; New, M. I.; Dupont, B.: HLA-linked congenital adrenal hyperplasia results from a defective gene encoding a cytochrome P-450 specific for steroid 21-hydroxylation. *Proc. Nat. Acad. Sci.* 81: 7505–7509, 1984.178. White, P. C.; New, M. I.; Dupont, B.: Structure of human steroid 21-hydroxylase genes. *Proc. Nat. Acad. Sci.* 83: 5111–5115, 1986.179. White, P. C.; Tusie-Luna, M.-T.; New, M. I.; Speiser, P. W.: Mutations in steroid

21-hydroxylase (CYP21). Hum. Mutat. 3: 373–378, 1994.180. White, P. C.; Vitek, A.; Dupont, B.; New, M. I.: Characterization of frequent deletions causing steroid 21-hydroxylase deficiency. Proc. Nat. Acad. Sci. 85: 4436–4440, 1988.181. Winqvist, O.; Karlsson, F. A.; Kampe, O.: 21-Hydroxylase, a major autoantigen in idiopathic Addison's disease. Lancet 339: 1559–1562, 1992.182. Witchel, S. F.; Bhamidipati, D. K.; Hoffman, E. P.; Cohen, J. B.: Phenotypic heterogeneity associated with the splicing mutation in congenital adrenal hyperplasia due to 21-hydroxylase deficiency. J. Clin. Endocr. Metab. 81: 4081–4088, 1996.183. Witchel, S. F.; Lee, P. A.; Suda-Hartman, M.; Trucco, M.; Hoffman, E. P.: Evidence for a heterozygote advantage in congenital adrenal hyperplasia due to 21-hydroxylase deficiency. J. Clin. Endocr. Metab. 82: 2097–2101, 1997.184. Wu, D.-A.; Chung, B.: Mutations of P450c21 (steroid 21-hydroxylase) at cys-428, val-281, and ser-268 result in complete, partial, or no loss of enzymatic activity, respectively. J. Clin. Invest. 88: 519–523, 1991.185. Wudy, S. A.; Dorr, H. G.; Solleder, C.; Djalali, M.; Homoki, J.: Profiling steroid hormones in amniotic fluid of midpregnancy by routine stable isotope dilution/gas chromatography-mass

spectrometry:Reference values and concentrations in fetuses at risk for 21-hydroxylase deficiency. *J. Clin. Endocr. Metab.* 84: 2724–2728, 1999.186. Wudy, S. A.; Homoki, J.; Teller, W. M.: Successful prenatal treatment of congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *Europ. J. Pediatr.* 153: 556–559, 1994.187. Yang, S. Y.; Levine, L. S.; Zachmann, M.; New, M. I.; Prader, A.; Oberfield, S. E.; O'Neill, G. J.; Pollack, M. S.; Dupont, B.: Mapping of the 21-hydroxylase deficiency gene within the HLA linkage group. *Transplant. Proc.* 10: 753–755, 1978.188. Zachmann, M.; Prader, A.: Unusual heterozygotes of congenital adrenal hyperplasia due to 21-hydroxylase deficiency confirmed by HLA tissue typing. *Acta Endocr.* 92: 542–546, 1979.

[35531] 9083. Uchida, S.; Kwon, H. M.; Yamauchi, A.; Preston, A. S.; Marumo, F.; Handler, J. S.: Molecular cloning of the cDNA for an MDCK cell Na(+)- and Cl(-)-dependent taurine transporter that is regulated by hypertonicity. *Proc. Nat. Acad. Sci.* 89: 8230–8234, 1992.

[35532] 9084. Sakaguchi, A. Y.; Zabel, B. U.; Grzeschik, K. H.; Law, M. L.; Ellis, R. W.; Skolnick, E. M.; Naylor, S. L.: Regional localization of two human cellular Kirsten ras genes on chromosomes 6 and 12. *Molec. Cell. Biol.* 4: 989–993, 1984.

- [35533] 9085.Neyroud, N.; Tesson, F.; Denjoy, I.; Leibovici, M.; Donger, C.;Barhanin, J.; Faure, S.; Gary, F.; Coumel, P.; Petit, C.; Schwartz,K.; Guicheney, P.: A novel mutation in the potassium channel geneKVLQT1 causes the Jervell and Lange-Nielsen cardioauditory syndrome. *NatureGenet.* 15: 186–189, 1997.
- [35534] 9086.Pervaiz, S.; Brew, K.: Homology of beta-lactoglobulin, serum retinol-bindingprotein, and protein HC. *Science* 228: 335–337, 1985.
- [35535] 9087.Barber, G. N.; Edelhoff, S.; Katze, M. G.; Disteché, C. M.: Chromosomalassignment of the interferon-inducible double-stranded RNA-dependentprotein kinase (PRKR) to human chromosome 2p21–p22 and mouse chromosome17 E2. *Genomics* 16: 765–767, 1993.
- [35536] 9088.Kuhen, K. L.; Shen, X.; Carlisle, E. R.; Richardson, A.L.; Weier,H.–U. G.; Tanaka, H.; Samuel, C. E.: Structural organization of thehuman gene (PKR) encoding an interferon-inducible RNA-dependent proteinkinase (PKR) and differences from its mouse homolog. *Genomics* 36:197–201, 1996.
- [35537] 9089.Kuhen, K. L.; Shen, X.; Samuel, C. E.: Mechanism of interferonaction: sequence of the human interferon-inducible RNA-dependent proteinkinase (PKR) deduced from

genomic clones. *Gene* 178: 191–193, 1996.

- [35538] 9090.Hansen, R. S.; Wijmenga, C.; Luo, P.; Stanek, A. M.; Canfield, T. K.; Weemaes, C. M. R.; Gartler, S. M.: The DNMT3B DNA methyltransferase gene is mutated in the ICF immunodeficiency syndrome. *Proc. Nat. Acad. Sci.* 96: 14412–14417, 1999.
- [35539] 9091.Hassan, K. M. A.; Norwood, T.; Gimelli, G.; Gartler, S. M.; Hansen, R. S.: Satellite 2 methylation patterns in normal and ICF syndrome cells and association of hypomethylation with advanced replication. *Hum. Genet.* 109: 452–462, 2001.
- [35540] 9092.Shirohzu, H.; Kubota, T.; Kumazawa, A.; Sado, T.; Chijiwa, T.; Inagaki, K.; Suetake, I.; Tajima, S.; Wakui, K.; Miki, Y.; Hayashi, M.; Fukushima, Y.; Sasaki, H.: Three novel DNMT3B mutations in Japanese patients with ICF syndrome. *Am. J. Med. Genet.* 112: 31–37, 2002.
- [35541] 9093.Bonifaci, N.; Moroianu, J.; Radu, A.; Blobel, G.: Karyopherin beta-2 mediates nuclear import of a mRNA binding protein. *Proc. Nat. Acad. Sci.* 94: 5055–5060, 1997.
- [35542] 9094.Pollard, V. W.; Michael, W. M.; Nakielnny, S.; Siomi, M. C.; Wang, F.; Dreyfuss, G.: A novel receptor-mediated nuclear protein import pathway. *Cell* 86: 985–994, 1996.

- [35543] 9095.Salinas, M.; Duprat, F.; Heurteaux, C.; Hugnot, J.-P.; Lazdunski, M.: New modulatory alpha subunits for mammalian Shab K(+) channels. *J.Biol. Chem.* 272: 24371–24379, 1997.
- [35544] 9096.Kas, K.; Roijer, E.; Voz, M.; Meyen, E.; Stenman, G.; Van de Ven, W. J. M.: A 2-Mb YAC contig and physical map covering the chromosome 8q12 breakpoint cluster region in pleomorphic adenomas of the salivary glands. *Genomics* 43: 349–358, 1997.
- [35545] 9097.Kas, K.; Voz, M. L.; Roijer, E.; Astrom, A.-K.; Meyen, E.; Stenman, G.; Van de Ven, W. J. M.: Promoter swapping between the genes for a novel zinc finger protein and beta-catenin in pleomorphic adenomas with t(3;8)(p21;q12) translocations. *Nature Genet.* 15: 170–174, 1997.
- [35546] 9098.Szabo, G.; Dallmann, G.; Muller, G.; Patthy, L.; Soller, M.; Varga, L.: A deletion in the myostatin gene causes the compact (Cmpt) hypermuscular mutation in mice. *Mammalian Genome* 9: 671–672, 1998.
- [35547] 9099.Zimmers, T. A.; Davies, M. V.; Koniaris, L. G.; Haynes, P.; Esquela, A. F.; Tomkinson, K. N.; McPherron, A. C.; Wolfman, N. M.; Lee, S.-J.: Induction of cachexia in mice by systemically administered myostatin. *Science*

296:1486–1488, 2002.

- [35548] 9100.Fransen, M.; Terlecky, S. R.; Subramani, S.: Identification of a human PTS1 receptor docking protein directly required for peroxisomal protein import. *Proc. Nat. Acad. Sci.* 95: 8087–8092, 1998.
- [35549] 9101.Albertini, M.; Rehling, P.; Erdmann, R.; Girzalsky, W.; Kiel, J.A. K.; Veenhuis, M.; Kunau, W.-H.: Pex14p, a peroxisomal membrane protein binding both receptors of the two PTS-dependent import pathways. *Cell* 89:83–92, 1997.
- [35550] 9102.Cohen, P.; Rylatt, D. B.; Nimmo, G. A.: The hormonal control of glycogen metabolism: the amino acid sequence at the phosphorylation site of protein phosphatase inhibitor-1. *FEBS Lett.* 76: 182–186, 1977.
- [35551] 9103.Helps, N. R.; Street, A. J.; Elledge, S. J.; Cohen, P. T.: Cloning of the complete coding region for human protein phosphatase inhibitor 2 using the two hybrid system and expression of inhibitor 2 in *E.coli*. *FEBS Lett.* 340: 93–98, 1994.
- [35552] 9104.Huang, F. L.; Glinsmann, W. H.: Separation and characterization of two phosphorylase phosphatase inhibitors from rabbit skeletal muscle. *Europ.J. Biochem.* 70: 419–426, 1976.

- [35553] 9105.Majer, M.; Mott, D. M.; Mochizuki, H.; Rowles, J. C.; Pedersen,O.; Knowler, W. C.; Bogardus, C.; Prochazka, M.: Association of theglycogen synthase locus on 19q13 with NIDDM in Pima Indians. *Diabetologia* 39:314–321, 1996.
- [35554] 9106.Permana, P. A.; Mott, D. M.: Genetic analysis of human type 1protein phosphatase inhibitor 2 in insulin-resistant Pima Indians. *Genomics* 41:110–114, 1997.
- [35555] 9107.Prochazka, M.; Mochizuki, H.; Baier, L. J.; Cohen, P. T. W.; Bogardus,C.: Molecular and linkage analysis of type–1 protein phosphatasecatalytic beta–subunit gene: lack of evidence for its major role ininsulin resistance in Pima Indians. *Diabetologia* 38: 461–466, 1995.
- [35556] 9108.Sakagami, H.; Kondo, H.: Molecular cloning of the cDNA for ratphosphatase inhibitor–2 and its wide gene expression in the centralnervous system. *J. Chem. Neuroanat.* 8: 259–266, 1995.
- [35557] 9109.Sanseau, P.; Jackson, A.; Alderton, R. P.; Beck, S.; Senger, G.;Sheer, D.; Kelly, A.; Trowsdale, J.: Cloning and characterizationof human phosphatase inhibitor–2 (IPP–2) sequences. *Mammalian Genome* 5:490–496, 1994.
- [35558] 9110.Charest, D. L.; Mordret, G.; Harder, K. W.; Jirik, F.; Pelech,S. L.: Molecular cloning, expression, and characterization of thehuman mitogen–activated protein kinase

p44erk1. *Molec. Cell. Biol.* 13:4679–4690, 1993.

[35559] 9111. Durand, B.; Sperisen, P.; Emery, P.; Barras, E.; Zufferey, M.; Mach, B.; Reith, W.: RFXAP, a novel subunit of the RFX DNA binding complex is mutated in MHC class II deficiency. *EMBO J.* 16: 1045–1055, 1997.

[35560] 9112. Mach, B.: Personal Communication. Geneva, Switzerland 10/28/1998.

[35561] 9113. Nekrep, N.; Jabrane-Ferrat, N.; Peterlin, B. M.: Mutations in the bare lymphocyte syndrome define critical steps in the assembly of the regulatory factor X complex. *Molec. Cell Biol.* 20: 4455–4461, 2000.

[35562] 9114. Schwartz, R. S.: The case of the bare lymphocyte syndrome: tracking down faulty transcription factors. (Editorial) *New Eng. J. Med.* 337:781–783, 1997.

[35563] 9115. Steimle, V.; Durand, B.; Barras, E.; Zufferey, M.; Hadam, M. R.; Mach, B.; Reith, W.: A novel DNA binding-regulatory factor is mutated in primary MHC class II deficiency (bare lymphocyte syndrome). *Genes Dev.* 9: 1021–1032, 1995.

[35564] 9116. Touraine, J. L.; Betuel, H.: Immunodeficiency diseases and expression of HLA antigens. *Hum. Immun.* 2: 147–153, 1981.

[35565] 9117. Villard, J.; Lisowska-Grospierre, B.; van den Elsen, P.;

Fischer,A.; Reith, W.; Mach, B.: Mutation of RFXAP, a regulator of MHC classII genes, in primary MHC class II deficiency. New Eng. J. Med. 337:748–753, 1997.

[35566] 9118.Peijnenburg, A.; Van Eggermond, M. C. J. A.; Van den Berg, R.;Sanal, O.; Vossen, J. M. J. J.; Van den Elsen, P. J.: Molecular analysisof an MHC class II deficiency patient reveals a novel mutation inthe RFX5 gene. Immunogenetics 49: 338–345, 1999.

[35567] 9119.Kim, I.; Kim, J.–H.; Ryu, Y. S.; Jung, S. H.; Nah, J. J.; Koh,G. Y.: Characterization and expression of a novel alternatively splicedhuman angiopoietin–2. J. Biol. Chem. 275: 18550–18556, 2000.

[35568] 9120.Maisonpierre, P. C.; Suri, C.; Jones, P. F.; Bartunkova, S.; Wiegand,S. J.; Radziejewski, C.; Compton, D.; McClain, J.; Aldrich, T. H.;Papadopoulos, N.; Daly, T. J.; Davis, S.; Sato, T. N.; Yancopoulos,G. D.: Angiopoietin–2, a natural antagonist for Tie2 that disruptsin vivo angiogenesis. Science 277: 55–60, 1997.

[35569] 9121.Tanaka, S.; Mori, M.; Sakamoto, Y.; Makuuchi, M.; Sugimachi, K.;Wands, J. R.: Biologic significance of angiopoietin–2 expressionin human hepatocellular carcinoma. J. Clin. Invest. 103: 341–345,1999.

[35570] 9122.Chow, V. T. K.; Quek, H. H.: HEP–COP, a novel hu–

man gene whose product is highly homologous to the alpha-subunit of the yeast coatmer protein complex. Gene 169: 223–227, 1996.

[35571] 9123. Quek, H. H.; Chow, V. T. K.: Molecular and cellular studies of the human homolog of the 160-kD alpha-subunit of the coatmer protein complex. DNA Cell Biol. 16: 275–280, 1997.

[35572] 9124. Quek, H. H.; Chow, V. T. K.: Genomic organization and mapping of the human HEP-COP gene (COPA) to 1q. Cytogenet. Cell Genet. 76:139–143, 1997.

[35573] 9125. Leffers, H.; Nielsen, M. S.; Andersen, A. H.; Honore, B.; Madsen, P.; Vandekerckhove, J.; Celis, J. E.: Identification of two human rho GDP dissociation inhibitor proteins whose overexpression lead to disruption of the actin cytoskeleton. Exp. Cell Res. 209: 165–174, 1993.

[35574] 9126. Wagner, T.; Tommerup, N.; Wirth, J.; Leffers, H.; Zimmer, J.; Back, E.; Weissenbach, J.; Scherer, G.: A somatic cell hybrid panel for distal 17q: GDIA1 maps to 17q25.3. Cytogenet. Cell Genet. 76: 172–175, 1997.

[35575] 9127. Sanicola, M.; Hession, C.; Worley, D.; Carmillo, P.; Ehrenfels, C.; Walus, L.; Robinson, S.; Jaworski, G.; Wei, H.; Tizard, R.; Whitty, A.; Pepinsky, R. B.; Cate, R. L.: Glial cell line-derived neurotrophic factor-dependent RET activation

can be mediated by two different cell-surface accessory proteins. *Proc. Nat. Acad. Sci.* 94: 6238–6243, 1997.

[35576] 9128. Suvanto, P.; Wartiovaara, K.; Lindahl, M.; Arumae, U.; Moshnyakov, M.; Horelli-Kuitunen, N.; Airaksinen, M. S.; Palotie, A.; Sariola, H.; Saarma, M.: Cloning, mRNA distribution and chromosomal localisation of the gene for glial cell line-derived neurotrophic factor receptor beta, a homologue to GDNFR-alpha. *Hum. Molec. Genet.* 6: 1267–1273, 1997.

[35577] 9129. Vanhorne, J. B.; Gimm, O.; Myers, S. M.; Kaushik, A.; von Deimling, A.; Eng, C.; Mulligan, L. M.: Cloning and characterization of the human GFRA2 locus and investigation of the gene in Hirschsprung disease. *Hum. Genet.* 108: 409–415, 2001.

[35578] 9130. Katsanis, N.; Yaspo, M.-L.; Fisher, E. M. C.: Identification and mapping of a novel human gene, HRMT1L1, homologous to the rat protein arginine N-methyltransferase 1 (PRMT1) gene. *Mammalian Genome* 8: 526–529, 1997.

[35579] 9131. Lin, W. J.; Gary, J. D.; Yang, M. C.; Clarke, S.; Herschman, H. R.: The mammalian intermediate-early TIS21 protein and the leukemia-associated BTG1 protein interact with a protein-arginine N-methyltransferase. *J. Biol. Chem.* 271: 15034–15044, 1996.

- [35580] 9132.Scott, H. S.; Antonarakis, S. E.; Lalioti, M. D.; Rossier, C.;Silver, P. A.; Henry, M. F.: Identification and characterization of two putative human arginine methyltransferases (HRMT1L1 and HRMT1L2). *Genomics* 48:330–340, 1998.
- [35581] 9133.Herberg, J. A.; Sgouros, J.; Jones, T.; Copeman, J.; Humphray,S. J.; Sheer, D.; Cresswell, P.; Beck, S.; Trowsdale, J.: Genomic analysis of the Tapasin gene, located close to the TAP loci in the MHC. *Europ. J. Immun.* 28: 459–467, 1998.
- [35582] 9134.Mayer, W. E.; Klein, J.: Is tapasin a modified Mhc class I molecule? *Immunogenetics* 53:719–723, 2001.
- [35583] 9135.Michalova, V.; Murray, B. W.; Sultmann, H.; Klein, J.: A contigmap of the Mhc class I genomic region in the zebrafish reveals ancient synteny. *J. Immun.* 164: 5296–5305, 2000.
- [35584] 9136.Ortmann, B.; Copeman, J.; Lehner, P. J.; Sadasivan, B.; Herberg,J. A.; Granda, A. G.; Riddell, S. R.; Tampe, R.; Spies, T.; Trowsdale,J.; Cresswell, P.: A critical role for tapasin in the assembly and function of multimeric MHC class I–TAP complexes. *Science* 277: 1306–1309,1997.
- [35585] 9137.Sadasivan, B.; Lehner, P. J.; Ortmann, B.; Spies, T.; Cresswell,P.: Roles for calreticulin and a novel glycoprotein, tapasin, in the interaction of MHC class I molecules

with TAP. *Immunity* 5: 103–114, 1996.

[35586] 9138. Teng, M. S.; Stephens, R.; Du Pasquier, L.; Freeman, T.; Lindquist, J. A.; Trowsdale, J.: A human TAPBP (TAPASIN)-related gene, TAPBP-R. *Europ. J. Immun.* 32: 1059–1068, 2002.

[35587] 9139. Yabe, T.; Kawamura, S.; Sato, M.; Kashiwase, K.; Tanaka, H.; Ishikawa, Y.; Asao, Y.; Oyama, J.; Tsuruta, K.; Tokunaga, K.; Tadokoro, K.; Juji, T.: A subject with a novel type I bare lymphocyte syndrome has tapasin deficiency due to deletion of 4 exons by Alu-mediated recombination. *Blood* 100: 1496–1498, 2002.

[35588] 9140. Blank, V.; Kim, M. J.; Andrews, N. C.: Human MAFG is a functional partner for p45 NF- κ B in activating globin gene expression. *Blood* 89: 3925–3935, 1997.

[35589] 9141. Blank, V.; Knoll, J. H. M.; Andrews, N. C.: Molecular characterization and localization of the human MafG gene. *Genomics* 44: 147–149, 1997.

[35590] 9142. Kimura, K.; Ito, M.; Amano, M.; Chihara, K.; Fukata, Y.; Nakafuku, M.; Yamamori, B.; Feng, J.; Nakano, T.; Okawa, K.; Iwamatsu, A.; Kaibuchi, K.: Regulation of myosin phosphatase by Rho and Rho-associated kinase (Rho-kinase). *Science* 273: 245–248, 1996.

[35591] 9143. Takahashi, N.; Ito, M.; Tanaka, J.; Nakano, T.;

Kaibuchi, K.; Odai, H.; Takemura, K.: Localization of the gene coding for myosin phosphatase, target subunit 1 (MYPT1) to human chromosome 12q15–q21. *Genomics* 44:150–152, 1997.

[35592] 9144. Ishikawa, S.; Takahashi, T.; Ogawa, M.; Nakamura, Y.: Genomic structure of the human PLCD1 (phospholipase C delta 1) locus on 3p22–p21.3. *Cytogenet. Cell Genet.* 78: 58–60, 1997.

[35593] 9145. Brown, A. M.; Willi, S. M.; Argyropoulos, G.; Garvey, W. T.: A novel missense mutation, R70W, in the human uncoupling protein 3 gene in a family with type 2 diabetes. (Abstract) *Hum. Mutat.* 13: 508 only, 1999. Note: Full article online.

[35594] 9146. Sato, N.; Arai, K.; Masai, H.: Human and *Xenopus* cDNAs encoding budding yeast Cdc7–related kinases: in vitro phosphorylation of MCM subunits by a putative human homologue of Cdc7. *EMBO J.* 16: 4340–4351, 1997.

[35595] 9147. Liu, J.; Dalmau, J.; Szabo, A.; Rosenfeld, M.; Huber, J.; Furneaux, H.: Paraneoplastic encephalomyelitis antigens bind to the AU-rich elements of mRNA. *Neurology* 45: 544–550, 1995.

[35596] 9148. Sakai, K.; Gofuku, M.; Kitagawa, Y.; Ogasawara, T.; Hirose, G.; Yamazaki, M.; Koh, C.-S.; Yanagisawa, N.;

Steinman, L.: A hippocampal protein associated with paraneoplastic neurologic syndrome and small cell lung carcinoma. *Biochem. Biophys. Res. Commun.* 199: 1200–1208, 1994.

[35597] 9149. Van Tine, B. A.; Knops, J. F.; Butler, A.; Deloukas, P.; Shaw, G. M.; King, P. H.: Localization of HuC (ELAVL3) to chromosome 19p13.2 by fluorescence in situ hybridization utilizing a novel tyramide labeling technique. *Genomics* 53: 296–299, 1998.

[35598] 9150. Whitmore, T. E.; Maurer, M. F.; Sexson, S.; Raymond, F.; Conklin, D.; Deisher, T. A.: Assignment of fibroblast growth factor 18 (FGF18) to human chromosome 5q34 by use of radiation hybrid mapping and fluorescence in situ hybridization. *Cytogenet. Cell Genet.* 90: 231–233, 2000.

[35599] 9151. Sherrington, R.; Rogaev, E. I.; Liang, Y.; Rogaeva, E. A.; Levesque, G.; Ikeda, M.; Chi, H.; Lin, C.; Li, G.; Holman, K.; Tsuda, T.; Mar.; and 21 others: Cloning of a gene bearing missense mutations in early-onset familial Alzheimer's disease. *Nature* 375: 754–760, 1995.

[35600] 9152. Verdi, J. M.; Bashirullah, A.; Goldhawk, D. E.; Kubu, C. J.; Jamali, M.; Meakin, S. O.; Lipshitz, H. D.: Distinct human NUMB isoforms regulate differentiation vs. proliferation in the neuronal lineage. *Proc. Nat. Acad. Sci.* 96:

10472–10476, 1999.

- [35601] 9153. Zhong, W.; Feder, J. N.; Jiang, M.-M.; Jan, L. Y.; Jan, Y. N.: Asymmetric localization of a mammalian Numb homolog during mouse cortical neurogenesis. *Neuron* 17: 43–53, 1996.
- [35602] 9154. Thresher, R. J.; Vitaterna, M. H.; Miyamoto, Y.; Kazantsev, A.; Hsu, D. S.; Petit, C.; Selby, C. P.; Dawut, L.; Smithies, O.; Takahashi, J. S.; Sancar, A.: Role of mouse cryptochrome blue-light photoreceptor in circadian photoresponses. *Science* 282: 1490–1494, 1998.
- [35603] 9155. Strehl, S.; LaSalle, J. M.; Lalande, M.: High-resolution analysis of DNA replication domain organization across an R/G-band boundary. *Molec. Cell. Biol.* 17: 6157–6166, 1997.
- [35604] 9156. Stathakis, D. G.; Lee, D.; Bryant, P. J.: Fine-scale physical map of the 11q21 region surrounding the human DLG2 locus, the gene encoding Chapsyn-110. *Genomics* 54: 186–188, 1998.
- [35605] 9157. Chow, V. T. K.; Lee, S. S.: DENN, a novel human gene differentially expressed in normal and neoplastic cells. *DNA Seq.* 6: 263–273, 1996.
- [35606] 9158. Chow, V. T. K.; Lim, K. M.; Lim, D.: The human DENN gene: genomic organization, alternative splicing, and lo-

calization to chromosome11p11.21–p11.22. *Genome* 41: 543–552, 1998.

[35607] 9159.Peyrard, M.; Seroussi, E.; Sandberg–Nordqvist, A.–C.; Xie, Y.–G.; Han, F.–Y.; Fransson, I.; Collins, J.; Dunham, I.; Kost–Alimova, M.; Imreh, S.; Dumanski, J. P.: The human LARGE gene from 22q12.3–q13.1 is a new, distinct member of the glycosyltransferase gene family. *Proc.Nat. Acad. Sci.* 96: 598–603, 1999.

[35608] 9160.Gerlai, R.; Roder, J. C.; Hampson, D. R.: Altered spatial learning and memory in mice lacking the mGluR4 subtype of metabotropic glutamate receptor. *Behav. Neurosci.* 112: 525–532, 1998.

[35609] 9161.Chatterjee, T. K.; Eapen, A.; Kanis, A. B.; Fisher, R. A.: Genomic organization, 5–prime–flanking region, and chromosomal localization of the human RGS3 gene. *Genomics* 45: 429–433, 1997.

[35610] 9162.Druey, K. M.; Blumer, K. J.; Kang, V. H.; Kehrl, J. H.: Inhibition of G–protein–mediated MAP kinase activation by a new mammalian gene family. *Nature* 379: 742–746, 1996.

[35611] 9163.Oettgen, P.; Alani, R. M.; Barcinski, M. A.; Brown, L.; Akbarali, Y.; Boltax, J.; Kunsch, C.; Munger, K.; Liebermann, T. A.: Isolation and characterization of a novel ep–

ithelium-specific transcriptionfactor, ESE-1, a member of the Ets family. *Molec. Cell. Biol.* 17:4419–4433, 1997.

[35612] 9164.Oettgen, P.; Barcinski, M.; Boltax, J.; Stolt, P.; Akbarali, Y.;Libermann, T. A.: Genomic organization of the human ELF3 (ESE-1/ESX)gene, a member of the Ets transcription factor family, and identificationof a functional promoter. *Genomics* 55: 358–362, 1999.

[35613] 9165.Oettgen, P.; Carter, K. C.; Augustus, M.; Barcinski, M.; Boltax,J.; Kunsch, C.; Libermann, T. A.: The novel epithelial-specific Etstranscription factor gene ESX maps to human chromosome 1q32.1. *Genomics* 45:456–457, 1997.

[35614] 9166.Lunn, C. A.; Fan, X.; Dalie, B.; Miller, K.; Zavodny, P. J.; Narula,S. K.; Lundell, D.: Purification of ADAM 10 from bovine spleen asa TNFalpha convertase. *FEBS Lett.* 400: 333–335, 1997.

[35615] 9167.Tymms, M. J.; Ng, A. Y. N.; Thomas, R. S.; Schutte, B. C.; Zhou,J.; Eyre, H. J.; Sutherland, G. R.; Seth, A.; Rosenberg, M.; Papas,T.; Debouck, C.; Kola, I.: A novel epithelial-expressed ETS gene,ELF3: human and murine cDNA sequences, murine genomic organization,human mapping to 1q32.2 and expression in tissues and cancer. *Onco-gene* 15:2449–2462, 1997.

- [35616] 9168. Wolfsberg, T. G.; Primakoff, P.; Myles, D. G.; White, J. M.: ADAM, a novel family of membrane proteins containing a disintegrin and metalloprotease domain: multipotential functions in cell–cell and cell–matrix interactions. *J. Cell Biol.* 131: 275–278, 1995.
- [35617] 9169. Yamazaki, K.; Mizui, Y.; Tanaka, I.: Radiation hybrid mapping of human ADAM10 gene to chromosome 15. *Genomics* 45: 457–459, 1997.
- [35618] 9170. Choi, D.-S.; Handa, M.; Young, H.; Gordon, A. S.; Diamond, I.; Messing, R. O.: Genomic organization and expression of the mouse equilibrative, nitrobenzylthioinosine-sensitive nucleoside transporter1 (ENT1) gene. *Biochem. Biophys. Res. Commun.* 277: 200–208, 2000.
- [35619] 9171. Coe, I. R.; Griffiths, M.; Young, J. D.; Baldwin, S. A.; Cass, C. E.: Assignment of the human equilibrative nucleoside transporter (hENT1) to 6p21.1–p21.2. *Genomics* 45: 459–460, 1997.
- [35620] 9172. Griffiths, M.; Beaumont, N.; Yao, S. Y. M.; Sundaram, M.; Boumah, C. E.; Davies, A.; Kwong, F. Y. P.; Coe, I.; Cass, C. E.; Young, J. D.; Baldwin, S. A.: Cloning of a human nucleoside transporter implicated in the cellular uptake of adenosine and chemotherapeutic drugs. *Nature Med.* 3: 89–94, 1997.

- [35621] 9173.Hu, S.-I.; Carozza, M.; Klein, M.; Nantermet, P.; Luk, D.; Crowl, R. M.: Human HtrA, an evolutionarily conserved serine protease identified as a differentially expressed gene product in osteoarthritic cartilage. *J.Biol. Chem.* 273: 34406–34412, 1998.
- [35622] 9174.Zumbrunn, J.; Trueb, B.: Localization of the gene for a serine protease with IGF-binding domain (PRSS11) to human chromosome 10q25.3–q26.2. *Genomics* 45:461–462, 1997.
- [35623] 9175.Zumbrunn, J.; Trueb, B.: Primary structure of a putative serine protease specific for IGF-binding proteins. *FEBS Lett.* 398: 187–192, 1996.
- [35624] 9176.D'Esposito, M.; Strazzullo, M.; Cuccurese, M.; Spalluto, C.; Rocchi, M.; D'Urso, M.; Ciccodicola, A.: Identification and assignment of the human transient receptor potential channel 6 gene TRPC6 to chromosome 11q21–q22. *Cytogenet. Cell Genet.* 83: 46–47, 1998.
- [35625] 9177.Hofmann, T.; Obukhov, A. G.; Schaefer, M.; Harteneck, C.; Gudermann, T.; Schultz, G.: Direct activation of human TRPC6 and TRPC3 channels by diacylglycerol. *Nature* 397: 259–263, 1999.
- [35626] 9178.Garcia, C. K.; Brown, M. S.; Pathak, R. K.; Goldstein, J. L.: cDNA cloning of MCT2, a second monocarboxylate

transporter expressed in different cells than MCT1. J. Biol. Chem. 270: 1843–1849, 1995.

[35627] 9179. Lin, R.-Y.; Vera, J. C.; Chaganti, R. S. K.; Golde, D. W.: Human monocarboxylate transporter 2 (MCT2) is a high affinity pyruvate transporter. J. Biol. Chem. 273: 28959–28965, 1998.

[35628] 9180. Ong, O. C.; Hu, K.; Rong, H.; Lee, R. H.; Fung, B. K.-K.: Gene structure and chromosome localization of the G-gamma-c subunit of human cone G-protein (GNGT2). Genomics 44: 101–109, 1997.

[35629] 9181. Ong, O. C.; Yamane, H. K.; Phan, K. B.; Fong, H. K.; Bok, D.; Lee, R. H.; Fung, B. K.-K.: Molecular cloning and characterization of the G protein gamma subunit of cone photoreceptors. J. Biol. Chem. 270: 8495–8500, 1995.

[35630] 9182. Tasheva, E. S.; Pettenati, M.; Von Kap-Her, C.; Conrad, G. W.: Assignment of mimecan gene (OGN) to human chromosome band 9q22 by in situ hybridization. Cytogenet. Cell Genet. 88: 326–327, 2000.

[35631] 9183. Park, S. H.; Ryu, S. H.; Suh, P. G.; Kim, H.: Assignment of human PLD2 to chromosome band 17p13.1 by fluorescence in situ hybridization. Cytogenet. Cell Genet. 82: 225 only, 1998.

[35632] 9184. Pucharcos, C.; Fuentes, J.-J.; Casas, C.; de la Luna,

S.; Alcantara,S.; Arbones, M. L.; Soriano, E.; Estivill, X.; Prichard, M.: Alu-splicecloning of human intersectin (ITSN), a putative multivalent bindingprotein expressed in proliferating and differentiating neurons andoverexpressed in Down syndrome. *Europ. J. Hum. Genet.* 7: 704–712,1999.

[35633] 9185.Fumagalli, P.; Accarino, M.; Egeo, A.; Scartezzini, P.; Rappazzo,G.; Pizzuti, A.; Avvantaggiato, V.; Simeone, A.; Arrigo, G.; Zuffardi,O.; Ottolenghi, S.; Taramelli, R.: Human NRD convertase: a highlyconserved metalloendopeptidase expressed at specific sites duringdevelopment and in adult tissues. *Genomics* 47: 238–245, 1998.

[35634] 9186.Kanezaki, R.; Toki, T.; Yokoyama, M.; Yomogida, K.; Sugiyama, K.;Yamamoto, M.; Igarashi, K.; Ito, E.: Transcription factor BACH1 isrecruited to the nucleus by its novel alternative spliced isoform. *J.Biol. Chem.* 276: 7278–7284, 2001.

[35635] 9187.Katz, P.; Whalen, G.; Kehrl, J. H.: Differential expression ofa novel protein kinase in human B lymphocytes: preferential localizationin the germinal center. *J. Biol. Chem.* 269: 16802–16809, 1994.

[35636] 9188.Pombo, C. M.; Kehrl, J. H.; Sanchez, I.; Katz, P.; Avruch, J.;Zon, L. I.; Woodgett, J. R.; Force, T.; Kyriakis, J.

M.: Activation of the SAPK pathway by the human STE20 homologue germinal centre kinase. *Nature* 377:750–754, 1995.

[35637] 9189. Ren, M.; Zeng, J.; De Lemos–Chiarandini, C.; Rosenfeld, M.; Adesnik, M.; Sabatini, D. D.: In its active form, the GTP-binding protein rab8 interacts with a stress-activated protein kinase. *Proc. Nat. Acad. Sci.* 93: 5151–5155, 1996.

[35638] 9190. Bonthron, D. T.; Hayward, B. E.; Moran, V.; Strain, L.: Characterization of TH1 and CTSZ, two non-imprinted genes downstream of GNAS1 in chromosome 20q13. *Hum. Genet.* 107: 165–175, 2000.

[35639] 9191. Santamaria, I.; Velasco, G.; Pendas, A. M.; Fueyo, A.; Lopez–Otin, C.: Cathepsin Z, a novel human cysteine proteinase with a short propeptide domain and a unique chromosomal location. *J. Biol. Chem.* 273: 16816–16823, 1998.

[35640] 9192. Jacquemin, P.; Chen, Z.; Martial, J. A.; Davidson, I.: Genomic structure and chromosomal mapping of the mouse transcription factor TEF–5 (Tead3) gene. *Mammalian Genome* 10: 632–634, 1999.

[35641] 9193. Sparks, A. B.; Hoffman, N. G.; McConnell, S. J.; Fowlkes, D. M.; Kay, B. K.: Cloning of ligand targets: systematic isolation of SH3 domain-containing proteins. *Na–*

ture Biotech. 14: 741–744, 1996.

- [35642] 9194. Blau, N.; Niederwieser, A.; Shmerling, D. H.: Peptiduria presumably caused by aminopeptidase-P deficiency: a new inborn error of metabolism. *J. Inher. Metab. Dis.* 11 (suppl): 240–242, 1988.
- [35643] 9195. Cottrell, G. S.; Hooper, N. M.; Turner, A. J.: Cloning, expression, and characterization of human cytosolic aminopeptidase P: a single manganese(II)-dependent enzyme. *Biochemistry* 39: 15121–15128, 2000.
- [35644] 9196. Sprinkle, T. J.; Caldwell, C.; Ryan, J. W.: Cloning, chromosomal sublocalization of the human soluble aminopeptidase P gene (XPNPEP1) to 10q25.3 and conservation of the putative proton shuttle and metal ligand binding sites with XPNPEP2. *Arch. Biochem. Biophys.* 378: 51–56, 2000.
- [35645] 9197. Vanhoof, G.; Goossens, F.; Juliano, M. A.; Juliano, L.; Hendriks, D.; Schatteman, K.; Lin, A. H.; Scharpe, S.: Isolation and sequence analysis of a human cDNA clone (XPNPEPL) homologous to X-prolyl aminopeptidase (aminopeptidase P). *Cytogenet. Cell Genet.* 78: 275–280, 1997.
- [35646] 9198. Vergas Romero, C.; Neudorfer, I.; Mann, K.; Schafer, W.: Purification and amino acid sequence of aminopepti-

dase P from pig kidney. *Europ.J. Biochem.* 229: 262–269, 1995.

[35647] 9199.Hegele, R. A.; Connelly, P. W.; Scherer, S. W.; Hanley, A. J. G.; Harris, S. B.; Tsui, L.-C.; Zinman, B.: Paraoxonase-2 gene (PON2)G148 variant associated with elevated fasting plasma glucose in noninsulin-dependent diabetes mellitus. *J. Clin. Endocr. Metab.* 82: 3373–3377, 1997.

[35648] 9200.Heinecke, J. W.; Lusi, A. J.: Paraoxonase-gene polymorphisms associated with coronary heart disease: support for the oxidative damage hypothesis? (Letter) *Am. J. Hum. Genet.* 62: 20–24, 1998.

[35649] 9201.Primo-Parmo, S. L.; Hsu, C.; Law, D. J.; La Du, B. N.: Location and arrangement of three paraoxonase genes: PON1, PON2, and PON3, on human chromosome 7. (Abstract) *Am. J. Hum. Genet.* 59 (suppl.):A406, 1996.

[35650] 9202.Primo-Parmo, S. L.; Sorenson, R. C.; Teiber, L.; La Du, B. N.: The human serum paraoxonase/arylesterase gene (PON1) is one member of multigene family. *Genomics* 33: 498–507, 1996.

[35651] 9203.Prochazka, M.; Thompson, D. B.; Scherer, S. W.; Tsui, L.-C.; Knowler, W. C.; Bennett, P. H.; Bogardus, C.: Linkage and association of insulin resistance and NIDDM with markers at 7q21.3–q22.1 in the Pima Indi-

ans.(Abstract) Diabetes 44 (suppl.): 42A, 1995.

[35652] 9204.Sanghera, D. K.; Aston, C. E.; Saha, N.; Kamboh, M. I.: DNA polymorphisms in two paraoxonase genes (PON1 and PON2) are associated with the risk of coronary heart disease. Am. J. Hum. Genet. 62: 36–44, 1998.

[35653] 9205.Chang, H. Y.; Nishitoh, H.; Yang, X.; Ichijo, H.; Baltimore, D.: Activation of apoptosis signal-regulating kinase 1 (ASK1) by the adapter protein Daxx. Science 281: 1860–1863, 1998.

[35654] 9206.Gelezianus, R.; Xu, W.; Takeda, K.; Ichijo, H.; Greene, W. C.: HIV-1 Nef inhibits ASK1-dependent death signalling providing a potential mechanism for protecting the infected host cell. Nature 410: 834–838, 2001.

[35655] 9207.Ichijo, H.; Nishida, E.; Irie, K.; ten Dijke, P.; Saitoh, M.; Moriguchi, T.; Takagi, M.; Matsumoto, K.; Miyazono, K.; Gotoh, Y.: Induction of apoptosis by ASK1, a mammalian MAPKKK that activates SAPK/JNK and p38 signaling pathways. Science 275: 90–94, 1997.

[35656] 9208.Nishitoh, H.; Saitoh, M.; Mochida, Y.; Takeda, K.; Nakano, H.; Rothe, M.; Miyazono, K.; Ichijo, H.: ASK1 is essential for JNK/SAPK activation by TRAF2. Molec. Cell 2: 389–395, 1998.

[35657] 9209.Fuchs, M.; Muller, T.; Lerch, M. M.; Ullrich, A.: Asso-

ciation of human protein-tyrosine phosphatase kappa with members of the armadillo family. *J. Biol. Chem.* 271: 16712–16719, 1996.

[35658] 9210. Yang, Y.; Gil, M. C.; Choi, E. Y.; Park, S. H.; Pyun, K. H.; Ha, H.: Molecular cloning and chromosomal localization of a human gene homologous to the murine R-PTP-kappa, a receptor-type protein tyrosine phosphatase. *Gene* 186: 77–82, 1997.

[35659] 9211. Zhang, Y.; Siebert, R.; Matthiesen, P.; Yang, Y.; Ha, H.; Schlegelberger, B.: Cytogenetical assignment and physical mapping of the human R-PTP-kappa gene (PTPRK) to the putative tumor suppressor gene region 6q22.2–q22.3 *Genomics* 51:309–311, 1998.

[35660] 9212. Hospital, V.; Prat, A.; Joulie, C.; Cherif, D.; Day, R.; Cohen, P.: Human and rat testis express two mRNA species encoding variants of NRD convertase, a metalloendopeptidase of the insulinase family. *Biochem. J.* 327: 773–779, 1997.

[35661] 9213. Pierotti, A. R.; Prat, A.; Chesneau, V.; Gaudoux, F.; Leseney, A.-M.; Foulon, T.; Cohen, P.: N-Arginine dibasic convertase, a metalloendopeptidase as a prototype of a class of processing enzymes. *Proc. Nat. Acad. Sci.* 91: 6078–6082, 1994.

- [35662] 9214. Tomasetto, C.; Regnier, C.; Moog-Lutz, C.; Mattei, M. G.; Chenard, M. P.; Lidereau, R.; Basset, P.; Rio, M. C.: Identification of four novel human genes amplified and overexpressed in breast carcinoma and localized to the q11-q21.3 region of chromosome 17. *Genomics* 28:367-376, 1995.
- [35663] 9215. Xu, Z.-H.; Otterness, D. M.; Freimuth, R. R.; Carlini, E. J.; Wood, T. C.; Mitchell, S.; Moon, E.; Kim, U.-J.; Xu, J.-P.; Siciliano, M. J.; Weinshilboum, R. M.: Human 3-prime-phosphoadenosine 5-prime-phosphosulfate synthetase 1 (PAPSS1) and PAPSS2: gene cloning, characterization and chromosomal localization. *Biochem. Biophys. Res. Commun.* 268: 437-444, 2000.
- [35664] 9216. Chalmers, I. J.; Hofler, H.; Atkinson, M. J.: Mapping of a cadherin gene cluster to a region of chromosome 5 subject to frequent allelic loss in carcinoma. *Genomics* 57: 160-163, 1999.
- [35665] 9217. Suzuki, S.; Sano, K.; Tanihara, H.: Diversity of the cadherin family: evidence for eight new cadherins in nervous tissue. *Cell Regul.* 2: 261-270, 1991.
- [35666] 9218. Shimoyama, Y.; Gotoh, M.; Terasaki, T.; Kitajima, M.; Hirohashi, S.: Isolation and sequence analysis of human

cadherin-6 complementaryDNA for the full coding sequence and its expression in human carcinomacells. Cancer Res. 55: 2206–2211, 1995.

- [35667] 9219. Anderson, L. V. B.; Davison, K.; Moss, J. A.; Young, C.; Cullen, M. J.; Walsh, J.; Johnson, M. A.; Bashir, R.; Britton, S.; Keers, S.; Argov, Z.; Mahjneh, I.; Fougrousse, F.; Beckmann, J. S.; Bushby, K. M. D.: Dysferlin is a plasma membrane protein and is expressed early in human development. Hum. Molec. Genet. 8: 855–861, 1999.
- [35668] 9220. Gong, B.; Almasan, A.: Genomic organization and transcriptional regulation of human Apo2/TRAIL gene. Biochem. Biophys. Res. Commun. 278: 747–752, 2000.
- [35669] 9221. Ohira, M.; Seki, N.; Nagase, T.; Ishikawa, K.; Nomura, N.; Ohara, O.: Characterization of a human homolog (BACH1) of the mouse Bach1 gene encoding a BTB–basic leucine zipper transcription factor and its mapping to chromosome 21q22.1. Genomics 47: 300–306, 1998.
- [35670] 9222. Oyake, T.; Itoh, K.; Motohashi, H.; Hayashi, N.; Hoshino, H.; Nishizawa, M.; Yamamoto, M.; Igarashi, K.: Bach proteins belong to a novel family of BTB–basic leucine zipper transcription factors that interact with MafK and regulate transcription through the NF-E2 site. Molec. Cell. Biol. 16: 6083–6095, 1996.

- [35671] 9223.Johnson, K. R.; Smith, L.; Johnson, D. K.; Rhodes, J.; Rinchik,E. M.; Thayer, M.; Lewis, E. J.: Mapping of the ARIX homeodomain gene to mouse chromosome 7 and human chromosome 11q13. *Genomics* 33:527–531, 1996.
- [35672] 9224.Merscher, S.; Bekri, S.; de Leeuw, B.; Pedeutour, F.; Grosgeorge,J.; Show, S. T. D.; Mullenbach, R.; et al.: A 5,5 megabase high resolution integrated map of distal 11q13. *Genomics* 39: 340–347, 1997.
- [35673] 9225.Morin, X.; Cremer, H.; Hirsch, M.–R.; Kapur, R. P.; Goridis, C.;Brunet, J.–F.: Defects in sensory and autonomic ganglia and absence of locus coeruleus in mice deficient for the homeobox gene Phox2a. *Neuron* 18:411–423, 1997.
- [35674] 9226.Pattyn, A.; Morin, X.; Goridis, C.; Brunet, J. F.: Expression and interaction of the two closely related homeobox genes PHOX2a and PHOX2b during neurogenesis. *Development* 124: 4065–4075, 1997.
- [35675] 9227.Zellmer, E.; Zhang, Z.; Greco, D.; Rhodes, J.; Cassel, S.; Lewis,E. J.: A homeodomain protein selectively expressed in noradrenergic tissue regulates transcription of neurotransmitter biosynthetic genes. *J.Neurosci.* 15: 8109–8120, 1995.
- [35676] 9228.Ghanshani, S.; Coleman, M.; Gustavsson, P.; Wu, A.

C.-L.; Gargus, J. J.; Gutman, G. A.; Dahl, N.; Mohrenweiser, H.; Chandy, K. G.: Human calcium-activated potassium channel gene KCNN4 maps to chromosome 19q13.2 in the region deleted in Diamond-Blackfan anemia. *Genomics* 51:160-161, 1998.

[35677] 9229. Ishii, T. M.; Silvia, C.; Hirschberg, B.; Bond, C. T.; Adelman, J. P.; Maylie, J.: A human intermediate conductance calcium-activated potassium channel. *Proc. Nat. Acad. Sci.* 94: 11651-11656, 1997.

[35678] 9230. Joiner, W. J.; Wang, L.-Y.; Tang, M. D.; Kaczmarek, L. K.: hSK4, a member of a novel subfamily of calcium-activated potassium channels. *Proc. Nat. Acad. Sci.* 94: 11013-11018, 1997.

[35679] 9231. Logsdon, N. J.; Kang, J.; Togo, J. A.; Christian, E. P.; Aiyar, J.: A novel gene, hKCa4, encodes the calcium-activated potassium channel in human T lymphocytes. *J. Biol. Chem.* 272: 32723-32726, 1997.

[35680] 9232. Matsuoka, H.; Iwata, N.; Ito, M.; Shimoyama, M.; Nagata, A.; Chihara, K.; Takai, S.; Matsui, T.: Expression of a kinase-defective Eph-like receptor in the normal human brain. *Biochem. Biophys. Res. Commun.* 235:487-492, 1997.

[35681] 9233. Tang, X. X.; Zhao, H.; Robinson, M. E.; Cohen, B.;

Cnaan, A.; London, W.; Cohn, S. L.; Cheung, N.-K. V.; Brodeur, G. M.; Evans, A. E.; Ikegaki, N.: Implications of EPHB6, EFNB2, and EFNB3 expressions in human neuroblastoma. *Proc. Nat. Acad. Sci.* 97: 10936–10941, 2000.

[35682] 9234. Meyers, R.; Cantley, L. C.: Cloning and characterization of a wortmannin-sensitive human phosphatidylinositol 4-kinase. *J. Biol. Chem.* 272: 4384–4390, 1997.

[35683] 9235. Nakanishi, S.; Catt, K. J.; Balla, T.: A wortmannin-sensitive phosphatidylinositol 4-kinase that regulates hormone-sensitive pools of inositol phospholipids. *Proc. Nat. Acad. Sci.* 92: 5317–5321, 1995.

[35684] 9236. Saito, T.; Seki, N.; Ishii, H.; Ohira, M.; Hayashi, A.; Kozuma, S.; Hori, T.: Complementary DNA cloning and chromosomal mapping of a novel phosphatidylinositol kinase gene. *DNA Res.* 4: 301–305, 1997.

[35685] 9237. Suzuki, K.; Hirano, H.; Okutomi, K.; Suzuki, M.; Kuga, Y.; Fujiwara, T.; Kanemoto, N.; Isono, K.; Horie, M.: Identification and characterization of a novel human phosphatidylinositol 4-kinase. *DNA Res.* 4: 273–280, 1997.

[35686] 9238. Miura, Y.; Miyake, K.; Yamashita, Y.; Shimazu, R.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Inazawa, J.; Abe, T.; Kimoto, M.: Molecular cloning of a human RP105 homologue and chromosomal localization of the mouse

and human RP105 genes (Ly64 and LY64). *Genomics* 38:299–304, 1996.

[35687] 9239.Miura, Y.; Shimazu, R.; Miyake, K.; Akashi, S.; Ogata, H.; Yamashita, Y.; Narisawa, Y.; Kimoto, M.: RP105 is associated with MD-1 and transmits an activation signal in human B cells. *Blood* 92: 2815–2822, 1998.

[35688] 9240.Castrop, J.; van Norren, K.; Clevers, H.: A gene family of HMG-box transcription factors with homology to TCF-1. *Nucleic Acids Res.* 20:611 only, 1992.

[35689] 9241.Duval, A.; Busson-Leconiat, M.; Berger, R.; Hamelin, R.: Assignment of the TCF-4 (TCF7L2) to human chromosome band 10q25.3. *Cytogenet. Cell Genet.* 88: 264–265, 2000.

[35690] 9242.Duval, A.; Gayet, J.; Zhou, X.-P.; Iacopetta, B.; Thomas, G.; Hamelin, R.: Frequent frameshift mutations of the TCF-4 gene in colorectal cancers with microsatellite instability. *Cancer Res.* 59: 4213–4215, 1999.

[35691] 9243.Duval, A.; Rolland, S.; Tubacher, E.; Bui, H.; Thomas, G.; Hamelin, R.: The human T-cell transcription factor-4 gene: structure, extensive characterization of alternative splicings, and mutational analysis in colorectal cancer cell lines. *Cancer Res.* 60: 3872–3879, 2000.

- [35692] 9244.Korinek, V.; Barker, N.; Moerer, P.; van Donselaar, E.; Huls, G.;Peters, P. J.; Clevers, H.: Depletion of epithelial stem-cell compartmentsin the small intestine of mice lacking Tcf-4. *Nature Genet.* 19:379–383, 1998.
- [35693] 9245.Ando, A.; Kikuti, Y. Y.; Abe, K.; Shigenari, A.; Kawata, H.; Ikemura,T.; Kimura, M.; Inoko, H.: cDNA cloning, northern hybridization,and mapping analysis of a putative GDS-related protein gene at thecentromeric ends of the human and mouse MHC regions. *Immunogenetics* 49:354–356, 1999.
- [35694] 9246.Isomura, M.; Okui, K.; Fujiwara, T.; Shin, S.; Nakamura, Y.: Isolationand mapping of RAB2L, a human cDNA that encodes a protein homologousto RalGDS. *Cytogenet. Cell Genet.* 74: 263–265, 1996.
- [35695] 9247.Pirozzi, G.; McConnell, S. J.; Uveges, A. J.; Carter, J. M.; Sparks,A. B.; Kay, B. K.; Fowlkes, D. M.: Identification of novel humanWW domain-containing proteins by cloning of ligand targets. *J. Biol.Chem.* 272: 14611–14616, 1997.
- [35696] 9248.Chambers, J. A.; Gardner, E.; Hauptmann, R.; Ponder, B. A.; Mulligan,L. M.: TaqI polymorphisms at the annexin VIII locus (ANX8). *Hum.Molec. Genet.* 1: 550 only, 1992.
- [35697] 9249.Chang, K.-S.; Wang, G.; Freireich, E. J.; Daly, M.;

Naylor, S.L.; Trujillo, J. M.; Stass, S. A.: Specific expression of the annexinVIII gene in acute promyelocytic leukemia. Blood 79: 1802–1810,1992.

- [35698] 9250.Fernandez, M. P.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N.A.; Morgan, R. O.: The genetic origin of mouse annexin VIII. MammalianGenome 9: 8–14, 1998.
- [35699] 9251.Sarkar, A.; Yang, P.; Fan, Y.–H.; Mu, Z. M.; Hauptmann, R.; Adolf,G. R.; Stass, S. A.; Chang, K.–S.: Regulation of the expression ofannexin VIII in acute promyelocytic leukemia. Blood 84: 279–286,1994.
- [35700] 9252.Waterham, H. R.; Koster, J.; Romeijn, G. J.; Hennekam, R. C. M.;Vreken, P.; Andersson, H. C.; FitzPatrick, D. R.; Kelley, R. I.; Wanders,R. J. A.: Mutations in the 3–beta–hydroxysterol delta–24–reductasegene cause desmosterolosis, an autosomal recessive disorder of cholesterolbiosynthesis. Am. J. Hum. Genet. 69: 685–694, 2001.
- [35701] 9253.Lechner, C.; Zahalka, M. A.; Giot, J.–F.; Moller, N. P. H.; Ullrich,A.: ERK6, a mitogen–activated protein kinase involved in C2C12 myoblastdifferentiation. Proc. Nat. Acad. Sci. 93: 4355–4359, 1996.
- [35702] 9254.Li, Z.; Jiang, Y.; Ulevitch, R. J.; Han, J.: The primary structureof p38–gamma: a new member of p38 group of

MAP kinases. *Biochem.Biophys. Res. Commun.* 228: 334–340, 1996.

[35703] 9255.Hibi, M.; Lin, A.; Smeal, T.; Minden, A.; Karin, M.: Identification of an oncoprotein- and UV-responsive protein kinase that binds and potentiates the c-Jun activation domain. *Genes Dev.* 7: 2135–2148, 1993.

[35704] 9256.Kallunki, T.; Su, B.; Tsigelny, I.; Sluss, H. K.; Derijard, B.; Moore, G.; Davis, R.; Karin, M.: JNK2 contains a specificity-determining region responsible for efficient c-Jun binding and phosphorylation. *Genes Dev.* 8: 2996–3007, 1994.

[35705] 9257.Sluss, H. K.; Barrett, T.; Derijard, B.; Davis, R. J.: Signal transduction by tumor necrosis factor mediated by JNK protein kinases. *Molec.Cell Biol.* 14: 8376–8384, 1994.

[35706] 9258.Mohit, A. A.; Martin, J. H.; Miller, C. A.: p49(3F12) kinase: a novel MAP kinase expressed in a subset of neurons in the human nervous system. *Neuron* 14: 67–78, 1995.

[35707] 9259.Yang, D. D.; Kuan, C. Y.; Whitmarsh, A. J.; Rincon, M.; Zheng, T. S.; Davis, R. J.; Rakic, P.; Flavell, R. A.: Absence of excitotoxicity-induced apoptosis in the hippocampus of mice lacking the Jnk3 gene. *Nature*

389:865–870, 1997.

- [35708] 9260. Yoshida, S.; Fukino, K.; Harada, H.; Nagai, H.; Imoto, I.; Inazawa, J.; Takahashi, H.; Teramoto, A.; Emi, M.: The c-Jun NH2-terminal kinase 3 (JNK3) gene: genomic structure, chromosomal assignment, and loss of expression in brain tumors. *J. Hum. Genet.* 46: 182–187, 2001.
- [35709] 9261. Carpenter, N. J.; Filipovich, A.; Blaese, R. M.; Carey, T. L.; Berkel, A. I.: Variable immunodeficiency with abnormal condensation of the heterochromatin of chromosomes 1, 9, and 16. *J. Pediatr.* 112: 757–760, 1988.
- [35710] 9262. Bunger, M. K.; Wilsbacher, L. D.; Moran, S. M.; Clendenin, C.; Radcliffe, L. A.; Hogenesch, J. B.; Simon, M. C.; Takahashi, J. S.; Bradfield, C. A.: Mop3 is an essential component of the master circadian pacemaker in mammals. *Cell* 103: 1009–1017, 2000.
- [35711] 9263. Ikeda, M.; Nomura, M.: cDNA cloning and tissue-specific expression of a novel basic helix–loop–helix/PAS protein (BMAL1) and identification of alternatively spliced variants with alternative translation initiation site usage. *Biochem. Biophys. Res. Commun.* 233: 258–264, 1997.
- [35712] 9264. Shaw, P. J.; Tononi, G.; Greenspan, R. J.; Robinson, D. F.: Stress response genes protect against lethal effects of sleep deprivation in *Drosophila*. *Nature* 417: 287–291,

2002.

- [35713] 9265. Wolting, C. D.; McGlade, C. J.: Cloning and chromosomal localization of a new member of the bHLH/PAS transcription factor family. *Mammalian Genome* 9: 463–468, 1998.
- [35714] 9266. Faucheu, C.; Diu, A.; Chan, A. W. E.; Blanchet, A.-M.; Miossec, C.; Herve, F.; Collard-Dutilleul, V.; Gu, Y.; Aldape, R. A.; Lippke, J. A.; Rocher, C.; Su, M. S.-S.; Livingston, D. J.; Hercend, T.; Lalanne, J.-L.: A novel human protease similar to the interleukin-1-beta converting enzyme induces apoptosis in transfected cells. *EMBO J.* 14: 1914–1922, 1995.
- [35715] 9267. Kamens, J.; Paskind, M.; Hugunin, M.; Talanian, R. V.; Allen, H.; Banach, D.; Bump, N.; Hackett, M.; Johnston, C. G.; Li, P.; Mankovich, J. A.; Terranova, M.; Ghayur, T.: Identification and characterization of ICH-2, a novel member of the interleukin-1-beta-converting enzyme family of cysteine proteases. *J. Biol. Chem.* 270: 15250–15256, 1995.
- [35716] 9268. Munday, N. A.; Vaillancourt, J. P.; Ali, A.; Casano, F. J.; Miller, D. K.; Molineaux, S. M.; Yamin, T.-T.; Yu, V. L.; Nicholson, D. W.: Molecular cloning and pro-apoptotic activity of ICE(rel)II and ICE(rel)III, members of the ICE/CED-3

family of cysteine proteases. J. Biol. Chem.

270:15870–15876, 1995.

[35717] 9269.Wang, S.; Miura, M.; Jung, Y.; Zhu, H.; Li, E.; Yuan, J.: Murine caspase-11, an ICE-interacting protease, is essential for the activation of ICE. Cell 92: 501–509, 1998.

[35718] 9270.Liburd, N.; Ghosh, M.; Riazuddin, S.; Naz, S.; Khan, S.; Ahmed, Z.; Riazuddin, S.; Liang, Y.; Menon, P. S. N.; Smith, T.; Smith, A.C. M.; Chen, K.-S.; Lupski, J. R.; Wilcox, E. R.; Potocki, L.; Friedman, T. B.: Novel mutations of MYO15A associated with profound deafness in consanguineous families and moderately severe hearing loss in a patient with Smith–Magenis syndrome. Hum. Genet. 109: 535–541, 2001.

[35719] 9271.Carlomagno, F.; Chang–Claude, J.; Dunning, A. M.; Ponder, B. A.J.: Determination of the frequency of the common 675del5 Nijmegen breakage syndrome mutation in the German population: no association with risk of breast cancer. Genes Chromosomes Cancer 25: 393–395, 1999.

[35720] 9272.Kleier, S.; Herrmann, M.; Wittwer, B.; Varon, R.; Reis, A.; Horst, J.: Clinical presentation and mutation identification in the NBS1 gene in a boy with Nijmegen breakage syndrome. Clin. Genet. 57:384–387, 2000.

- [35721] 9273.Lim, D.-S.; Kim, S.-T.; Xu, B.; Maser, R. S.; Lin, J.; Petrini, J. H. J.; Kastan, M. B.: ATM phosphorylates p95/nbs1 in an S-phase checkpoint pathway. *Nature* 404: 613–617, 2000.
- [35722] 9274.Lombard, D. B.; Guarente, L.: Nijmegen breakage syndrome disease protein and MRE11 at PML nuclear bodies and meiotic telomeres. *Cancer Res.* 60: 2331–2334, 2000.
- [35723] 9275.Maser, R. S.; Zinkel, R.; Petrini, J. H. J.: An alternative mode of translation permits production of a variant NBS1 protein from the common Nijmegen breakage syndrome allele. *Nature Genet.* 27: 417–421, 2001.
- [35724] 9276.Matsuura, S.; Tauchi, H.; Nakamura, A.; Kondo, N.; Sakamoto, S.; Endo, S.; Smeets, D.; Solder, B.; Belohradsky, B. H.; Der Kaloustian, V. M.; Oshimura, M.; Isomura, M.; Nakamura, Y.; Komatsu, K.: Positional cloning of the gene for Nijmegen breakage syndrome. *Nature Genet.* 19:179–181, 1998.
- [35725] 9277.Skelly, R. H.; Korbonits, M.; Grossman, A.; Besser, G. M.; Monson, J. P.; Geddes, J. F.; Burrin, J. M.: Expression of the pituitary transcription factor Ptx-1, but not that of the trans-activating factor Prop-1, is reduced in human corticotroph adenomas and is associated with decreased alpha-

subunit secretion. *J. Clin. Endocr. Metab.* 85:2537–2542, 2000.

[35726] 9278.Morgan, R. O.; Bell, D. W.; Testa, J. R.; Fernandez, M.–P.: Humanannexin 31 genetic mapping and origin. *Gene* 227: 33–38, 1999.

[35727] 9279.Morgan, R. O.; Fernandez, M.–P.: Expression profile and structuraldivergence of novel human annexin 31. *FEBS Lett.* 434: 300–304, 1998.

[35728] 9280.Nadler, M. J. S.; Hermosura, M. C.; Inabe, K.; Perraud, A.–L.;Zhu, Q.; Stokes, A. J.; Kurosaki, T.; Kinet, J.–P.; Penner, R.; Scharenberg,A. M.; Fleig, A.: LTRPC7 is a Mg–ATP–regulated divalent cation channelrequired for cell viability. *Nature* 411: 590–595, 2001. Note: Erratum *Nature* 412:660 only, 2001.

[35729] 9281.Runnels, L. W.; Yue, L.; Clapham, D. E.: TRP–PLIK, a bifunctionalprotein with kinase and ion channel activities. *Science* 291: 1043–1047,2001.

[35730] 9282.Schlingmann, K. P.; Weber, S.; Peters, M.; Nejsun, L. N.; Vitzthum,H.; Klingel, K.; Kratz, M.; Haddad, E.; Ristoff, E.; Dinour, D.; Syrrou,M.; Nielsen, S.; Sassen, M.; Waldegger, S.; Seyberth, H. W.; Konrad,M.: Hypomagnesemia with secondary hypocalcemia is caused by mutationsin TRPM6, a new member of the TRPM gene family. *Nature Genet.*

31:166–170, 2002.

- [35731] 9283. Walder, R. Y.; Shalev, H.; Brennan, T. M. H.; Carmi, R.; Elbedour, K.; Scott, D. A.; Hanauer, A.; Mark, A. L.; Patil, S.; Stone, E. M.; Sheffield, V. C.: Familial hypomagnesemia maps to chromosome 9q, not to the X chromosome: genetic linkage mapping and analysis of a balanced translocation breakpoint. *Hum. Molec. Genet.* 6: 1491–1497, 1997.
- [35732] 9284. Brohmann, H.; Pinnecke, S.; Hoyer-Fender, S.: Identification and characterization of new cDNAs encoding outer dense fiber proteins of rat sperm. *J. Biol. Chem.* 272: 10327–10332, 1997.
- [35733] 9285. Scott, A. F.: Personal Communication. Baltimore, Md. 9/10/1997.
- [35734] 9286. Shao, X.; Murthy, S.; Demetrick, D. J.; van der Hoorn, F. A.: Human outer dense fiber gene, ODF2, localizes to chromosome 9q34. *Cytogenet. Cell Genet.* 83: 221–223, 1998.
- [35735] 9287. Trachtulec, Z.; Hamvas, R. M. J.; Forejt, J.; Lehrach, H. R.; Vincek, V.; Klein, J.: Linkage of TATA-binding protein and proteasome subunit C5 genes in mice and humans reveals synteny conserved between mammals and invertebrates. *Genomics* 44: 1–7, 1997.

- [35736] 9288.Calabrese, G.; Sallese, M.; Stornaiuolo, A.; Stuppia, L.; Palka,G.; De Blasi, A.: Chromosome mapping of the human arrestin (SAG),beta-arrestin 2 (ARRB2), and beta-adrenergic receptor kinase 2 (ADRBK2)genes. *Genomics* 23: 286–288, 1994.
- [35737] 9289.Porteous, S.; Torban, E.; Cho, N.–P.; Cunliffe, H.; Chua, L.;McNoe, L.; Ward, T.; Souza, C.; Gus, P.; Giugliani, R.; Sato, T.;Yun, K.; Favor, J.; Sicotte, M.; Goodyer, P.; Eccles, M.: Primaryrenal hypoplasia in humans and mice with PAX2 mutations: evidenceof increased apoptosis in fetal kidneys of Pax2(1Neu) +/– mutant mice. *Hum.Molec. Genet.* 9: 1–11, 2000.
- [35738] 9290.Johnson, L.; Mercer, K.; Greenbaum, D.; Bronson, R. T.; Crowley,D.; Tuveson, D. A.; Jacks, T.: Somatic activation of the K–ras oncogenecauses early onset lung cancer in mice. *Nature* 410: 1111–1116, 2001.
- [35739] 9291.Kozma, S. C.; Bogaard, M. E.; Buser, K.; Saurer, S. M.; Bos, J.L.; Groner, B.; Hynes, N. E.: The human c–Kirsten ras gene is activatedby a novel mutation in codon 13 in the breast carcinoma cell lineMDA–MB231. *Nucleic Acids Res.* 15: 5963–5971, 1987.
- [35740] 9292.Laghi, L.; Orbetegli, O.; Bianchi, P.; Zerbi, A.; Di Carlo, V.;Boland, C. R.; Malesci, A.: Common occurrence of

multiple K-RAS mutations in pancreatic cancers with associated precursor lesions and in biliary cancers. *Oncogene* 21: 4301–4306, 2002.

[35741] 9293. Lee, K.-H.; Lee, J.-S.; Suh, C.; Kim, S.-W.; Kim, S.-B.; Lee, J.-H.; Lee, M.-S.; Park, M.-Y.; Sun, H.-S.; Kim, S.-H.: Clinicopathologic significance of the K-ras gene codon 12 point mutation in stomach cancer: an analysis of 140 cases. *Cancer* 75: 2794–2801, 1995.

[35742] 9294. Liu, E.; Hjelle, B.; Morgan, R.; Hecht, F.; Bishop, J. M.: Mutations of the Kirsten-ras proto-oncogene in human preleukaemia. *Nature* 330:186–188, 1987.

[35743] 9295. McBride, O. W.; Swand, D. C.; Tronick, S. R.; Gol, R.; Klimanis, D.; Moore, D. E.; Aaronson, S. A.: Regional chromosomal localization of N-ras, K-ras-1, K-ras-2 and myb oncogenes in human cells. *Nucleic Acids Res.* 11: 8221–8236, 1983.

[35744] 9296. McCoy, M. S.; Toole, J. J.; Cunningham, J. M.; Chang, E. H.; Lowy, D. R.; Weinberg, R. A.: Characterization of a human colon/lung carcinoma oncogene. *Nature* 302: 79–81, 1983.

[35745] 9297. McGrath, J. P.; Capon, D. J.; Smith, D. H.; Chen, E. Y.; Seeburg, P. H.; Goeddel, D. V.; Levinson, A. D.: Structure and organization of the human Ki-ras proto-oncogene and

a related processed pseudogene. *Nature* 304:501–506, 1983.

[35746] 9298.Motojima, K.; Urano, T.; Nagata, Y.; Shiku, H.; Tsurifune, T.; Kanematsu, T.: Detection of point mutations in the Kirsten-ras oncogene provides evidence for the multicentricity of pancreatic carcinoma. *Ann.Surg.* 217:

138–143, 1993.

[35747] 9299.Muller, R.; Slamon, D. J.; Adamson, E. D.; Tremblay, J. M.; Muller, D.; Cline, M. J.; Verma, I. M.: Transcription of c-onc genes c-ras(Ki) and c-fms during mouse development. *Molec. Cell. Biol.* 3: 1062–1069, 1983.

[35748] 9300.Nakano, H.; Yamamoto, F.; Neville, C.; Evans, D.; Mizuno, T.; Perucho, M.: Isolation of transforming sequences of two human lung carcinomas: structural and functional analysis of the activated c-K-ras oncogenes. *Proc. Nat. Acad. Sci.* 81: 71–75, 1984.

[35749] 9301.O'Brien, S. J.; Nash, W. G.; Goodwin, J. L.; Lowry, D. R.; Chang, E. H.: Dispersion of the ras family of transforming genes to four different chromosomes in man. *Nature* 302: 839–842, 1983.

[35750] 9302.O'Connell, P.; Leppert, M.; Hoff, M.; Kumlin, E.; Thomas, W.; Cai, G.; Law, M.; White, R.: A linkage map for human chromosome 12.(Abstract) *Am. J. Hum. Genet.* 37:

A169 only, 1985.

- [35751] 9303.Ohashi, H.; Ishikiriya, S.; Fukushima, Y.: New diagnostic method for Pallister-Killian syndrome: detection of i(12p) in interphasenuclei of buccal mucosa by fluorescence in situ hybridization. *Am.J. Med. Genet.* 45: 123-128, 1993.
- [35752] 9304.Otori, K.; Oda, Y.; Sugiyama, K.; Hasebe, T.; Mukai, K.; Fujii, T.; Tajiri, H.; Yoshida, S.; Fukushima, S.; Esumi, H.: High frequency of K-ras mutations in human colorectal hyperplastic polyps. *Gut* 40:660-663, 1997.
- [35753] 9305.Peltomaki, P.; Knuutila, S.; Ritvanen, A.; Kaitila, I.; de laChapelle, A.: Pallister-Killian syndrome: cytogenetic and molecular studies. *Clin. Genet.* 31: 399-405, 1987.
- [35754] 9306.Pfeifer, G. P.: A new verdict for an old convict. *Nature Genet.* 29:3-4, 2001.
- [35755] 9307.Porta, M.; Malats, N.; Jarrod, M.; Grimalt, J. O.; Rifa, J.; Carrato, A.; Guarner, L.; Salas, A.; Santiago-Silva, M.; Corominas, J. M.; Andreu, M.; Real, F. X.: Serum concentrations of organochlorine compounds and K-ras mutations in exocrine pancreatic cancer. *Lancet* 354: 2125-2129, 1999.
- [35756] 9308.Pulciani, S.; Santos, E.; Lauver, A. V.; Long, L. K.; Aaronson, S. A.; Barbacid, M.: Oncogene in solid human

tumors. *Nature* 300:539–542, 1982.

- [35757] 9309.Rodenhuis, S.; van de Wetering, M. L.; Mooi, W. J.; Evers, S.G.; van Zandwijk, N.; Bos, J. L.: Mutational activation of the K-RAS oncogene: a possible pathogenetic factor in adenocarcinoma of the lung. *New Eng. J. Med.* 317: 929–935, 1987.
- [35758] 9310.Bertazzoni, U.; Scovassi, A. I.; Brun, G. M.: Chick-embryo DNA polymerase gamma: identity of gamma-polymerases purified from nuclei and mitochondria. *Europ. J. Biochem.* 81: 237–248, 1977.
- [35759] 9311.Chang, L.-S.; Zhao, L.; Zhu, L.; Chen, M.-L.; Lee, M. Y. W. T.: Structure of the gene for the catalytic subunit of human DNA polymerase delta (POLD1). *Genomics* 28: 411–419, 1995.
- [35760] 9312.Chung, D. W.; Zhang, J.; Tan, C.-K.; Davie, E. W.; So, A. G.; Downey, K. M.: Primary structure of the catalytic subunit of human DNA polymerase delta and chromosomal location of the gene. *Proc. Nat. Acad. Sci.* 88:11197–11201, 1991.
- [35761] 9313.Dresler, S. L.; Gowans, B. J.; Robinson-Hill, R. M.; Hunting, D.J.: Involvement of DNA polymerase delta in DNA repair synthesis in human fibroblasts at late times after ultraviolet irradiation. *Biochemistry* 27:6379–6383,

1988.

- [35762] 9314.Goldsby, R. E.; Singh, M.; Preston, B. D.: Mouse DNA polymerase delta gene (Pold1) maps to chromosome 7. *Mammalian Genome* 9: 92–93, 1998.
- [35763] 9315.Kamath–Loeb, A. S.; Johansson, E.; Burgers, P. M. J.; Loeb, L.A.: Functional interaction between the Werner syndrome protein and DNA polymerase delta. *Proc. Nat. Acad. Sci.* 97: 4603–4608, 2000.
- [35764] 9316.Kemper, R. R.; Ahn, E. R.; Zhang, P.; Lee, M. Y. W. T.; Rabin, M.: Human DNA polymerase delta gene maps to region 19q13.3–q13.4 by in situ hybridization. *Genomics* 14: 205–206, 1992.
- [35765] 9317.Lee, M. Y. W. T.: Isolation of multiple forms of DNA polymerase delta: evidence of proteolytic modification during isolation. *Biochemistry* 27: 5188–5193, 1988.
- [35766] 9318.Lee, M. Y. W. T.; Toomey, N. L.: Human placental DNA polymerase delta: identification of a 170–kilodalton polypeptide by activity staining and immunoblotting. *Biochemistry* 26: 1076–1085, 1987.
- [35767] 9319.Lestienne, P.: Evidence for a direct role of the DNA polymerase gamma in the replication of the human mitochondrial DNA in vitro. *Biochem. Biophys. Res. Commun.* 146: 1146–1153, 1987.

- [35768] 9320.Nishida, C.; Reinhard, P.; Linn, S.: DNA repair synthesis inhuman fibroblasts requires DNA polymerase delta. J. Biol. Chem. 263:501–510, 1988.
- [35769] 9321.Syvaoja, J.; Linn, S.: Characterization of a large form of DNAPolymerase delta from HeLa cells that is insensitive to proliferatingcell nuclear antigen. J. Biol. Chem. 264: 2489–2497, 1989.
- [35770] 9322.Syvaoja, J.; Suomensaaari, S.; Nishida, C.; Goldsmith, J. S.; Chui,G. S. J.; Jain, S.; Linn, S.: DNA polymerases alpha, delta, and epsilon:three distinct enzymes from HeLa cells. Proc. Nat. Acad. Sci. 87:6664–6668, 1990.
- [35771] 9323.Yang, C.–L.; Chang, L.–S.; Zhang, P.; Hao, H.; Zhu, L.; Toomey,N. L.; Lee, M. Y. W. T.: Molecular cloning of the cDNA for the catalyticsubunit of human DNA polymerase delta. Nucleic Acids Res. 20: 735–745,1992.
- [35772] 9324.Zhao, L.; Chang, L.–S.: The human POLD1 gene: identificationof an upstream activator sequence, activation by Sp1 and Sp3, andcell cycle regulation. J. Biol. Chem. 272: 4869–4882, 1997.
- [35773] 9325.Lind, B.; van Solinge, W. W.; Schwartz, M.; Thorsen, S.: Splicesite mutation in the human protein C gene associated with venous thrombosis:demonstration of exon skipping by ectopic transcript analysis. Blood

82:2423–2432, 1993.

- [35774] 9326.Domenjoud, L.; Fronia, C.; Uhde, F.; Engel, W.: Sequence of humanprotamine 2 cDNA. Nucleic Acids Res. 16: 7733 only, 1988.
- [35775] 9327.Funke, B.; Epstein, J. A.; Kochilas, L. K.; Lu, M. M.; Pandita,R. K.; Liao, J.; Bauerndistel, R.; Schuler, T.; Schorle, H.; Brown,M. C.; Adams, J.; Morrow, B. E.: Mice overexpressing genes from the22q11 region deleted in velo-cardio-facial syndrome/DiGeorge syndromehave middle and inner ear defects. Hum. Molec. Genet. 10: 2549–2556,2001.
- [35776] 9328.Halford, S.; Wilson, D. I.; Daw, S. C. M.; Roberts, C.; Wadey,R.; Kamath, S.; Wickremasinghe, A.; Burn, J.; Goodship, J.; Mattei,M.–G.; Moorman, A. F. M.; Scambler, P. J.: Isolation of a gene expressedduring early embryogenesis from the region of 22q11 commonly deletedin DiGeorge syndrome.. Hum. Molec. Genet. 2: 1577–1582, 1993.
- [35777] 9329.Jerome, L. A.; Papaioannou, V. E.: DiGeorge syndrome phenotypein mice mutant for the T-box gene, Tbx1. Nature Genet. 27: 286–291,2001.
- [35778] 9330.Santos, E.; Martin-Zanca, D.; Reddy, E. P.; Pierotti, M. A.; DellaPorta, G.; Barbacid, M.: Malignant activation of a K-ras oncogenein lung carcinoma but not in normal tis–

sue of the same patient. Science 223:661–664, 1984.

[35779] 9331.Schinzel, A.: Tetrasomy 12p (Pallister–Killian syndrome). J.Med. Genet. 28: 122–125, 1991.

[35780] 9332.Shimizu, K.; Birnbaum, D.; Ruley, M. A.; Fasano, O.; Suard, Y.;Edlund, L.; Taparowsky, E.; Goldfarb, M.; Wigler, M.: Structure ofthe Ki–ras gene of the human lung carcinoma cell line Calu–1. Nature 304:497–500, 1983.

[35781] 9333.Sidransky, D.; Tokino, T.; Hamilton, S. R.; Kinzler, K. W.; Levin,B.; Frost, P.; Vogelstein, B.: Identification of RAS oncogene mutationsin the stool of patients with curable colorectal tumors. Science 256:102–105, 1992.

[35782] 9334.Smit, V. T. H. B. M.; Boot, A. J. M.; Smits, A. M. M.; Fleuren,G. J.; Cornelisse, C. J.; Bos, J. L.: KRAS codon 12 mutations occurvery frequently in pancreatic adenocarcinomas. Nucleic Acids Res. 16:7773–7782, 1988.

[35783] 9335.Soukup, S.; Neidich, K.: Prenatal diagnosis of Pallister–Killiansyndrome. Am. J. Med. Genet. 35: 526–528, 1990.

[35784] 9336.Speleman, F.; Leroy, J. G.; Van Roy, N.; De Paepe, A.; Suijkerbuijk,R.; Brunner, H.; Looijenga, L.; Verschraegen–Spae, M.–R.; Orye, E.: Pallister–Killian syndrome: characterization of the isochromosome12p by fluorescent in situ hybridization. Am. J. Med. Genet. 41:381–387, 1991.

- [35785] 9337. Takeda, S.; Ichii, S.; Nakamura, Y.: Detection of K-ras mutation in sputum by mutant-allele-specific amplification (MASA). *Hum. Mutat.* 2:112-117, 1993.
- [35786] 9338. Weinberg, R. A.: Fewer and fewer oncogenes. *Cell* 30: 3-4, 1982.
- [35787] 9339. Wenger, S. L.; Boone, L. Y.; Steele, M. W.: Mosaicism in Pallister(12p) syndrome. *Am. J. Med. Genet.* 35: 523-525, 1990.
- [35788] 9340. Wingo, P. A.; Ries, L. A. G.; Giovino, G. A.; Miller, D. S.; Rosenberg, H. M.; Shopland, D. R.; Thun, M. J.; Edwards, B. K.: Annual report to the nation on the status of cancer, 1973-1996, with a special section on lung cancer and tobacco smoking. *J. Nat. Cancer Inst.* 91: 675-690, 1999.
- [35789] 9341. Yanez, L.; Groffen, J.; Valenzuela, D. M.: c-K-ras mutations in human carcinomas occur preferentially in codon 12. *Oncogene* 1:315-318, 1987.
- [35790] 9342. Zakowski, M. F.; Wright, Y.; Ricci, A., Jr.: Pericardial agenesis and focal aplasia cutis in tetrasomy 12p (Pallister-Killian syndrome). *Am. J. Med. Genet.* 42: 323-325, 1992.
- [35791] 9343. Zhang, J.; Marynen, P.; Devriendt, K.; Fryns, J.-P.; Van den Berghe, H.; Cassiman, J.-J.: Molecular analysis of the isochromosome 12p in the Pallister-Killian syndrome:

construction of a mouse–human hybridcell line containing an i(12p) as the sole human chromosome. Hum.Genet. 83: 359–363, 1989.

[35792] 9344.Zhang, Z.; Wang, Y.; Vikis, H. G.; Johnson, L.; Liu, G.; Li, J.;Anderson, M. W.; Sills, R. C.; Hong, H. L.; Devereux, T. R.; Jacks,T.; Guan, K.–L.; You, M.: Wildtype Kras2 can inhibit lung carcinogenesis in mice. Nature Genet. 29: 25–33, 2001.

[35793] 9345.Adams, J. M.; Gerondakis, S.; Webb, E.; Corcoran, L. M.; Cory,S.: Cellular myc oncogene is altered by chromosome translocation to an immunoglobulin locus in murine plasmacytomas and is rearranged similarly in human Burkitt lymphomas. Proc. Nat. Acad. Sci. 80:1982–1986, 1983.

[35794] 9346.Alitalo, K.; Schwab, M.; Lin, C. C.; Varmus, H. E.; Bishop, J.M.: Homogeneously staining chromosomal regions contain amplified copies of an abundantly expressed cellular oncogene (c–myc) in malignant neuroendocrine cells from a human colon carcinoma. Proc. Nat. Acad.Sci. 80: 1707–1711, 1983.

[35795] 9347.Atchley, W. R.; Fitch, W. M.: Myc and Max: molecular evolution of a family of proto–oncogene products and their dimerization partner. Proc.Nat. Acad. Sci. 92:

10217–10221, 1995.

- [35796] 9348. Battey, J.; Moulding, C.; Taub, R.; Murphy, W.; Stewart, T.; Potter, H.; Lenoir, G.; Leder, P.: The human c-myc oncogene: structural consequences of translocation into the IgH locus in Burkitt lymphoma. *Cell* 34:779–787, 1983.
- [35797] 9349. Beimling, P.; Benter, T.; Sander, T.; Moelling, K.: Isolation and characterization of the human cellular myc gene product. *Biochemistry* 24:6349–6355, 1985.
- [35798] 9350. Bernard, O.; Cory, S.; Gerondakis, S.; Webb, E.; Adams, J. M.: Sequence of the murine and human cellular myc oncogenes and two modes of myc transcription resulting from chromosome translocation in B lymphoid tumours. *EMBO J.* 2: 2375–2383, 1983.
- [35799] 9351. Bernheim, A.; Berger, R.; Lenoir, G.: Cytogenetic studies on African Burkitt's lymphoma cell lines: t(8;14), t(2;8) and t(8;22) translocations. *Cancer Genet. Cytogenet.* 3: 307–315, 1981.
- [35800] 9352. Bhatia, K.; Huppi, K.; Spangler, G.; Siwarski, D.; Iyer, R.; Magrath, I.: Point mutations in the c-Myc transactivation domain are common in Burkitt's lymphoma and mouse plasmacytomas. *Nature Genet.* 5:56–61, 1993.
- [35801] 9353. Colby, W. W.; Chen, E. Y.; Smith, D. H.; Levinson, A.

D.: Identification and nucleotide sequence of a human locus homologous to the v-myc oncogene of avian myelocytomatosis virus MC29. *Nature* 301: 722–725, 1983.

[35802] 9354. Cole, M. D.: The myc oncogene: its role in transformation and differentiation. *Annu. Rev. Genet.* 20: 361–384, 1986.

[35803] 9355. Collins, S.; Groudine, M.: Amplification of endogenous myc-related DNA sequences in a human myeloid leukaemia cell line. *Nature* 298: 679–681, 1982.

[35804] 9356. Croce, C. M.; Thierfelder, W.; Erikson, J.; Nishikura, K.; Finan, J.; Lenoir, G. M.; Nowell, P. C.: Transcriptional activation of an unrearranged and untranslocated c-myc oncogene by translocation of a C-lambda locus in Burkitt lymphoma cells. *Proc. Nat. Acad. Sci.* 80: 6922–6926, 1983.

[35805] 9357. Dalla-Favera, R.; Bregni, M.; Erikson, J.; Patterson, D.; Gallo, R. C.; Croce, C. M.: Human c-myc oncogene is located on the region of chromosome 8 that is translocated in Burkitt lymphoma cells. *Proc. Nat. Acad. Sci.* 79: 7824–7827, 1982.

[35806] 9358. Dalla-Favera, R.; Gelmann, E. P.; Martinotti, S.; Franchini, G.; Papas, T. S.; Gallo, R. C.; Wong-Staal, F.: Cloning and characterization of different human sequences related

to the onc gene (v-myc) of avianmyelocytomatosis virus (MC29). Proc. Nat. Acad. Sci. 79: 6497–6501,1982.

[35807] 9359.Duesberg, P. H.: Cancer genes: rare recombinants instead of activatedoncogenes (a review). Proc. Nat. Acad. Sci. 84: 2117–2124, 1987.

[35808] 9360.Dyson, E.; Sucov, H. M.; Kubalak, S. W.; Schmid-Schonbein, G. W.;DeLano, F. A.; Evans, R. M.; Ross, J., Jr.; Chien, K. R.: Atrial-likephenotype is associated with embryonic ventricular failure in retinoidX receptor alpha -/- mice. Proc. Nat. Acad. Sci. 92: 7386–7390,1995.

[35809] 9361.Gampe, R. T., Jr.; Montana, V. G.; Lambert, M. H.; Miller, A. B.;Bledsoe, R. K.; Milburn, M. V.; Kliewer, S. A.; Willson, T. M.; Xu,H. E.: Asymmetry in the PPAR-gamma/RXR-alpha crystal structure revealsthe molecular basis of heterodimerization among nuclear receptors. Molec.Cell 5: 545–555, 2000.

[35810] 9362.Gruber, P. J.; Kubalak, S. W.; Pexieder, T.; Sucov, H. M.; Evans,R. M.; Chien, K. R.: RXR-alpha deficiency confers genetic susceptibilityfor aortic sac, conotruncal, atrioventricular cushion, and ventricular muscle defects in mice. J. Clin. Invest. 98: 1332–1343, 1996.

[35811] 9363.Heyman, R. A.; Mangelsdorf, D. J.; Dyck, J. A.; Stein, R. B.; Eichele,G.; Evans, R. M.; Thaller, C.: 9-cis retinoic

acid is a high affinity ligand for the retinoid X receptor.

Cell 68: 397–406, 1992.

- [35812] 9364. Hoopes, C. W.; Taketo, M.; Ozato, K.; Liu, Q.; Howard, T. A.; Linney, E.; Seldin, M. F.: Mapping the mouse Rxr loci encoding nuclear retinoid X receptors Rxr-alpha, Rxr-beta, and Rxr-gamma. Genomics 14: 611–617, 1992.
- [35813] 9365. Jones, K. A.; Fitzgibbon, J.; Woodward, K. J.; Goudie, D.; Ferguson-Smith, M. A.; Povey, S.; Wolfe, J.; Solomon, E.: Localization of the retinoid X receptor alpha gene (RXRA) to chromosome 9q34. Ann. Hum. Genet. 57: 195–201, 1993.
- [35814] 9366. Li, M.; Indra, A. K.; Warot, X.; Brocard, J.; Messaddeq, N.; Kato, S.; Metzger, D.; Chambon, P.: Skin abnormalities generated by temporally controlled RXR-alpha mutations in mouse epidermis. Nature 407: 633–636, 2000.
- [35815] 9367. Lu, T. T.; Makishima, M.; Repa, J. J.; Schoonjans, K.; Kerr, T. A.; Auwerx, J.; Mangelsdorf, D. J.: Molecular basis for feedback regulation of bile acid synthesis by nuclear receptors. Molec. Cell 6: 507–515, 2000.
- [35816] 9368. Mangelsdorf, D. J.; Ong, E. S.; Dyck, J. A.; Evans, R. M.: Nuclear receptor that identifies a novel retinoic acid response pathway. Nature 345: 224–229, 1990.
- [35817] 9369. Mangelsdorf, D. J.; Umesono, K.; Kliewer, S. A.;

Borgmeyer, U.; Ong, E. S.; Evans, R. M.: A direct repeat in the cellular retinol-binding protein type II gene confers differential regulation by RXR and RAR. *Cell* 66:555–561, 1991.

[35818] 9370. Repa, J. J.; Turley, S. D.; Lobaccaro, J.-M. A.; Medina, J.; Li, L.; Lustig, K.; Shan, B.; Heyman, R. A.; Dletschy, J. M.; Mangelsdorf, D. J.: Regulation of absorption and ABC1-mediated efflux of cholesterol by RXR heterodimers. *Science* 289: 1524–1529, 2000.

[35819] 9371. Tontonoz, P.; Hu, E.; Spiegelman, B. M.: Stimulation of adipogenesis in fibroblasts by PPAR- γ -2, a lipid-activated transcription factor. *Cell* 79:1147–1156, 1994.

[35820] 9372. Willy, P. J.; Umesono, K.; Ong, E. S.; Evans, R. M.; Heyman, R. A.; Mangelsdorf, D. J.: LXR, a nuclear receptor that defines a distinct retinoid response pathway. *Genes Dev.* 9: 1033–1045, 1995.

[35821] 9373. Wan, Y.-J. Y.; An, D.; Cai, Y.; Repa, J. J.; Chen, T. H.-P.; Flores, M.; Postic, C.; Magnuson, M. A.; Chen, J.; Chien, K. R.; French, S.; Mangelsdorf, D. J.; Sucov, H. M.: Hepatocyte-specific mutation establishes retinoid X receptor alpha as a heterodimeric integrator of multiple physiological processes in the liver. *Molec. Cell. Biol.* 20: 4436–4444, 2000.

- [35822] 9374. Zhou, M. D.; Sucov, H. M.; Evans, R. M.; Chien, K. R.: Retinoid-dependent pathways suppress myocardial cell hypertrophy. *Proc. Nat. Acad. Sci.* 92:7391–7395, 1995.
- [35823] 9375. Fitzgibbon, J.; Gillett, G. T.; Woodward, K. J.; Boyle, J. M.; Wolfe, J.; Povey, S.: Mapping of RXRB to human chromosome 6p21.3. *Ann. Hum. Genet.* 57: 203–209, 1993.
- [35824] 9376. Fleischhauer, K.; McBride, O. W.; DiSanto, J. P.; Ozato, K.; Yang, S. Y.: Cloning and chromosome mapping of human retinoid X receptor beta: selective amino acid sequence conservation of a nuclear hormone receptor in mammals. *Hum. Genet.* 90: 505–510, 1993.
- [35825] 9377. Nagata, T.; Weiss, E. H.; Abe, K.; Kitagawa, K.; Ando, A.; Yara-Kikuti, Y.; Seldin, M. F.; Ozato, K.; Inoko, H.; Taketo, M.: Physical mapping of the retinoid X receptor B gene in mouse and human. *Immunogenetics* 41:83–90, 1995.
- [35826] 9378. Yu, V. C.; Delsert, C.; Andersen, B.; Holloway, J. M.; Devary, O. V.; Naar, A. M.; Kim, S. Y.; Boutin, J.-M.; Glass, C. K.; Rosenfeld, M. G.: RXR-beta: a coregulator that enhances binding of retinoic acid, thyroid hormone, and vitamin D receptors to their cognate response elements. *Cell* 67: 1251–1266, 1991.
- [35827] 9379. Graycar, J. L.; Miller, D. A.; Arrick, B. A.; Lyons, R. M.;

Moses, H. L.; Derynck, R.: Human transforming growth factor- β -3: recombinant expression, purification, and biological activities in comparison with transforming growth factors- β -1 and β -2. *Molec. Endocr.* 3:1977–1986, 1989.

[35828] 9380. Kaartinen, V.; Voncken, J. W.; Shuler, C.; Warburton, D.; Bu, D.; Heisterkamp, N.; Groffen, J.: Abnormal lung development and cleft palate in mice lacking TGF- β -3 indicates defects of epithelial-mesenchymal interaction. *Nature Genet.* 11: 415–421, 1995.

[35829] 9381. Lee, B.-S.; Nowak, R. A.: Human leiomyoma smooth muscle cells show increased expression of transforming growth factor- β -3 (TGF- β -3) and altered responses to the antiproliferative effects of TGF- β . *J. Clin. Endocr. Metab.* 86: 913–920, 2001.

[35830] 9382. Moren, A.; Ichijo, H.; Miyazono, K.: Molecular cloning and characterization of the human and porcine transforming growth factor- β type III receptors. *Biochem. Biophys. Res. Commun.* 189: 356–362, 1992.

[35831] 9383. Proetzel, G.; Pawlowski, S. A.; Wiles, M. V.; Yin, M.; Boivin, G. P.; Howles, P. N.; Ding, J.; Ferguson, M. W. J.; Doetschman, T.: Transforming growth factor- β -3 is required for secondary palate fusion. *Nature Genet.* 11:

409–414, 1995.

- [35832] 9384.ten Dijke, P.; Geurts van Kessel, A. H. M.; Foulkes, J. G.; LeBeau, M. M.: Transforming growth factor type beta-3 maps to human chromosome 14, region q23–q24. *Oncogene* 3: 721–724, 1988.
- [35833] 9385.ten Dijke, P.; Hansen, P.; Iwata, K. K.; Pieler, C.; Foulkes, J.G.: Identification of another member of the transforming growth factor type beta gene family. *Proc. Nat. Acad. Sci.* 85: 4715–4719, 1988.
- [35834] 9386.Engel, W.; Keime, S.; Kremling, H.; Hameister, H.; Schluter, G.: The genes for protamine 1 and 2 (PRM1 and PRM2) and transition protein 2 (TNP2) are closely linked in the mammalian genome. *Cytogenet. Cell Genet.* 61: 158–159, 1992.
- [35835] 9387.Nelson, J. E.; Krawetz, S. A.: Linkage of human spermatid-specific basic nuclear protein genes: definition and evolution of the P1-to-P2-to-TP2 locus. *J. Biol. Chem.* 268: 2932–2936, 1993.
- [35836] 9388.Schluter, G.; Kremling, H.; Engel, W.: The gene for human transition protein 2: nucleotide sequence, assignment to the protamine gene cluster, and evidence for its low expression. *Genomics* 14: 377–383, 1992.
- [35837] 9389.Clapham, J. C.; Arch, J. R. S.; Chapman, H.; Haynes,

A.; Lister,C.; Moore, G. B. T.; Piercy, V.; Carter, S. A.;
Lehner, I.; Smith,S. A.; Beeley, L. J.; Godden, R. J.; and 15
others: Mice overexpressinghuman uncoupling protein-3
in skeletal muscle are hyperphagic andlean. *Nature* 406:
415-418, 2000.

[35838] 9390.Dalgaard, L. T.; Sorensen, T. I. A.; Drivsholm, T.;
Borch-Johnsen,K.; Andersen, T.; Hansen, T.; Pedersen, O.:
A prevalent polymorphismin the promoter of the UCP3
gene and its relationship to body massindex and long
term body weight change in the Danish population. *J.Clin.*
Endocr. Metab. 86: 1398-1402, 2001.

[35839] 9391.Kim, P. K. M.; Dutra, A. S.; Chandrasekharappa, S.
C.; Puck, J.M.: Genomic structure and mapping of human
FADD, an intracellularmediator of lymphocyte apoptosis. *J.*
Immun. 157: 5461-5466, 1996.

[35840] 9392.Lavedan, C.; Leroy, E.; Torres, R.; Dehejia, A.; Dutra,
A.; Buchholtz,S.; Nussbaum, R. L.; Polymeropoulos, M. H.:
Genomic organizationand expression of the human beta-
synuclein gene (SNCB) *Genomics* 54:173-175, 1998.

[35841] 9393.Lorenz, B.; Migliaccio, C.; Lichtner, P.; Meyer, C.;
Strom, T.M.; D'Urso, M.; Becker, J.; Ciccodicola, A.;
Meitinger, T.: Cloningand gene structure of the rod cGMP
phosphodiesterase delta subunitgene (PDED) in man and

mouse. *Europ. J. Hum. Genet.* 6: 283–290,1998.

- [35842] 9394.Hymowitz, S. G.; Christinger, H. W.; Fuh, G.; Ultsch, M.; O'Connell,M.; Kelley, R. F.; Ashkenazi, A.; de Vos, A. M.: Triggering celldeath: the crystal structure of Apo2L/TRAIL in a complex with deathreceptor 5. *Molec. Cell* 4: 563–571, 1999.
- [35843] 9395.Nitsch, R.; Bechmann, I.; Deisz, R. A.; Haas, D.; Lehmann, T. N.;Wendling, U.; Zipp, F.: Human brain–cell death induced by tumour–necrosis–factor–relatedapoptosis–inducing ligand (TRAIL). *Lancet* 356: 827–828, 2000.
- [35844] 9396.Pitti, R. M.; Marsters, S. A.; Ruppert, S.; Donahue, C. J.; Moore,A.; Ashkenazi, A.: Induction of apoptosis by Apo–2 ligand, a newmember of the tumor necrosis factor cytokine family. *J. Biol. Chem.* 271:12687–12690, 1996.
- [35845] 9397.Wiley, S. R.; Schooley, K.; Smolak, P. J.; Din, W. S.; Huang, C.–P.;Nicholl, J. K.; Sutherland, G. R.; Smith, T. D.; Rauch, C.; Smith,C. A.; Goodwin, R. G.: Identification and characterization of a newmember of the TNF family that induces apoptosis. *Immunity* 3: 673–682,1995.
- [35846] 9398.Lai, C. S. L.; Fisher, S. E.; Hurst, J. A.; Levy, E. R.; Hodgson,S.; Fox, M.; Jeremiah, S.; Povey, S.; Jamison, D. C.; Green, E. D.;Vargha–Khadem, F.; Monaco, A. P.: The

SPCH1 region on human 7q31:genomic characterization of the critical interval and localizationof translocations associated with speech and language disorder. *Am.J. Hum. Genet.* 67: 357–368, 2000.

[35847] 9399.Lai, C. S. L.; Fisher, S. E.; Hurst, J. A.; Vargha-Khadem, F.;Monaco, A. P.: A forkhead-domain gene is mutated in a severe speechand language disorder. *Nature* 413: 519–523, 2001.

[35848] 9400.Lyu, M. S.; Park, D. J.; Rhee, S. G.; Kozak, C. A.: Genetic mappingof the human and mouse phospholipase C genes. *Mammalian Genome* 7:501–504, 1996.

[35849] 9401.Shimohama, S.; Kamiya, S.; Fujii, M.; Ogawa, T.; Kanamori, M.;Kawamata, J.; Imura, T.; Taniguchi, T.; Yagisawa, H.: Mutation inthe pleckstrin homology domain of the human phospholipase C-delta-1gene is associated with loss of function. *Biochem. Biophys. Res.Comm.* 245: 722–728, 1998.

[35850] 9402.Iwabuchi, K.; Bartel, P. L.; Li, B.; Marraccino, R.; Fields, S.: Two cellular proteins that bind to wild-type but not mutant p53. *Proc.Nat. Acad. Sci.* 91: 6098–6102, 1994.

[35851] 9403.Iwabuchi, K.; Li, B.; Massa, H. F.; Trask, B. J.; Date, T.; Fields,S.: Stimulation of p53-mediated transcriptional

activation by the p53-binding proteins, 53BP1 and 53BP2. J. Biol. Chem. 273: 26061–26068, 1998.

[35852] 9404. Naumovski, L.; Cleary, M. L.: The p53-binding protein 53BP2 also interacts with Bcl2 and impedes cell cycle progression at G2/M. Molec. Cell Biol. 16: 3884–3892, 1996.

[35853] 9405. Samuels-Lev, Y.; O'Connor, D. J.; Bergamaschi, D.; Trigiante, G.; Hsieh, J.-K.; Zhong, S.; Campargue, I.; Naumovski, L.; Crook, T.; Lu, X.: ASPP proteins specifically stimulate the apoptotic function of p53. Molec. Cell 8: 781–794, 2001.

[35854] 9406. Yang, J.-P.; Ono, T.; Sonta, S.; Kawabe, T.; Okamoto, T.: Assignment of p53 binding protein (TP53BP2) to human chromosome band 1q42.1 by in situ hybridization. Cytogenet. Cell Genet. 78: 61–62, 1997.

[35855] 9407. Crawford, M. J.; Lanctot, C.; Tremblay, J. J.; Jenkins, N.; Gilbert, D.; Copeland, N.; Beatty, B.; Drouin, J.: Human and murine PTX1/Ptx1 gene maps to the region for Treacher Collins syndrome. Mammalian Genome 8: 841–845, 1997.

[35856] 9408. Lamonerie, T.; Tremblay, J. J.; Lanctot, C.; Thierrien, M.; Gauthier, Y.; Drouin, J.: Ptx1, a bicoid-related homeobox transcription factor involved in transcription of the

pro-opiomelanocortin gene. *GenesDev.* 10: 1284–1295, 1996.

[35857] 9409. Shang, J.; Li, X.; Ring, H. Z.; Clayton, D. A.; Francke, U.: Backfoot, a novel homeobox gene, maps to human chromosome 5 (BFT) and mouse chromosome 13 (Bft). *Genomics* 40: 108–113, 1997.

[35858] 9410. Szeto, D. P.; Rodriguez-Esteban, C.; Ryan, A. K.; O'Connell, S.M.; Liu, F.; Kioussi, C.; Gleiberman, A. S.; Izpisua-Belmonte, J.C.; Rosenfeld, M. G.: Role of the Bicoid-related homeodomain factor Pitx1 in specifying hindlimb morphogenesis and pituitary development. *GenesDev.* 13: 484–494, 1999.

[35859] 9411. Cohen, M. E.; Yin, M.; Paznekas, W. A.; Schertzer, M.; Wood, S.; Jabs, E. W.: Human SLUG gene organization, expression, and chromosomal location on 8q. *Genomics* 51: 468–471, 1998.

[35860] 9412. Inukai, T.; Inoue, A.; Kurosawa, H.; Goi, K.; Shinjyo, T.; Ozawa, K.; Mao, M.; Inaba, T.; Look, A. T.: SLUG, a ccs-1-related zinc finger transcription factor gene with anti-apoptotic activity, is a downstream target of the E2A-HLF oncoprotein. *Molec. Cell* 4: 343–352, 1999.

[35861] 9413. Nieto, A. M.; Sargent, M. G.; Wilkinson, D. G.; Cooke, J.: Control of cell behavior during vertebrate development

by Slug, a zinc finger gene. *Science* 264: 835–839, 1994.

[35862] 9414. Glover, M. T.; Atherton, D. J.: Transient zinc deficiency in two full-term breast-fed siblings associated with low maternal breastmilk zinc concentration. *Pediatr. Derm.* 5: 10–13, 1988.

[35863] 9415. Kuramoto, Y.; Igarashi, Y.; Tagami, H.: Acquired zinc deficiency in breast-fed infants. *Semin. Derm.* 10: 309–312, 1991.

[35864] 9416. Kury, S.; Devilder, M.-C.; Avet-Loiseau, H.; Dreno, B.; Moisan, J.-P.: Expression pattern, genomic structure and evaluation of the human SLC30A4 gene as a candidate for acrodermatitis enteropathica. *Hum. Genet.* 109: 178–185, 2001.

[35865] 9417. Nakano, A.; Nakano, H.; Hanada, K.; Nomura, K.; Uitto, J.: ZNT4 gene is not responsible for acrodermatitis enteropathica in Japanese families. (Letter) *Hum. Genet.* 110: 201–202, 2002.

[35866] 9418. Agulnick, A. D.; Taira, M.; Breen, J. J.; Tanaka, T.; Dawid, I. B.; Westphal, H.: Interactions of the LIM-domain-binding factor Ldb1 with LIM homeodomain proteins. *Nature* 384: 270–272, 1996.

[35867] 9419. Bach, I.; Carriere, C.; Ostendorff, H. P.; Andersen, B.; Rosenfield, M. G.: A family of LIM domain-associated co-

factors confer transcriptional synergism between LIM and Otx homeodomain proteins. *Genes Dev.* 11:1370–1380, 1997.

[35868] 9420. Jurata, L. W.; Gill, G. N.: Functional analysis of the nuclear LIM domain interactor NL1. *Molec. Cell. Biol.* 17: 5688–5698, 1997.

[35869] 9421. Jurata, L. W.; Kenny, D. A.; Gill, G. N.: Nuclear LIM interactor, a rhombotin and LIM homeodomain interacting protein, is expressed early in neuronal development. *Proc. Nat. Acad. Sci.* 93: 11693–11698, 1996.

[35870] 9422. Semina, E. V.; Altherr, M. R.; Murray, J. C.: Cloning and chromosomal localization of two novel human genes encoding LIM-domain binding factors CLIM1 and CLIM2/LDB1/NL1. *Mammalian Genome* 9: 921–924, 1998.

[35871] 9423. Yamashita, T.; Agulnick, A. D.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Westphal, H.: Genomic structure and chromosomal localization of the mouse LIM domain-binding protein 1 gene, *Ldb1*. *Genomics* 48:87–92, 1998.

[35872] 9424. Ahmad, M.; Srinivasula, S. M.; Wang, L.; Talanian, R. V.; Litwack, G.; Fernandes-Alnemri, T.; Alnemri, E. S.: CRADD, a novel human apoptotic adaptor molecule for caspase-2, and FasL/tumor necrosis factor receptor–

interacting protein RIP. *Cancer Res.* 57: 615–619, 1997.

[35873] 9425. Duan, H.; Dixit, V. M.: RAIDD is a new 'death' adaptor molecule. *Nature* 385:86–89, 1997.

[35874] 9426. Horvat, S.; Medrano, J. F.: A 500-kb YAC and BAC contig encompassing the high-growth deletion in mouse chromosome 10 and identification of the murine Raidd/Cradd gene in the candidate region. *Genomics* 54:159–164, 1998.

[35875] 9427. Foley, B. T.; Moehring, J. M.; Moehring, T. J.: Mutations in the elongation factor 2 gene which confer resistance to diphtheria toxin and *Pseudomonas* exotoxin A: genetic and biochemical analyses. *J. Biol. Chem.* 270: 23218–23225, 1995.

[35876] 9428. Mattheakis, L. C.; Sor, F.; Collier, R. J.: Diphthamide synthesis in *Saccharomyces cerevisiae*: structure of the DPH2 gene. *Gene* 132:149–154, 1993.

[35877] 9429. Schultz, D. C.; Balasara, B. R.; Testa, J. R.; Godwin, A. K.: Cloning and localization of a human diphthamide biosynthesis-like protein-2 gene, DPH2L2. *Genomics* 52: 186–191, 1998.

[35878] 9430. Abe, R.; Sakashita, E.; Yamamoto, K.; Sakamoto, H.: Two different RNA binding activities for the AU-rich element and the poly(A) sequence of the mouse neuronal

protein mHuC. *Nucleic Acids Res.* 24: 4895–4901,1996.

[35879] 9431.Rhim, H.; Savagner, P.; Thibaudieu, G.; Thiery, J. P.; Pavan, W.J.: Localization of a neural crest transcription factor, Slug, to mouse chromosome 16 and human chromosome 8. *Mammalian Genome* 8:872–873, 1997.

[35880] 9432.Savagner, P.; Yamada, K. M.; Thiery, J. P.: The zinc-finger protein Slug causes desmosome dissociation, an initial and necessary step for growth factor-induced epithelial-mesenchymal transition. *J. Cell Biol.* 137: 1403–1419, 1997.

[35881] 9433.Winter, H.; Rogers, M. A.; Gebhardt, M.; Wollina, U.; Boxall, L.; Chitayat, D.; Babul-Hirji, R.; Stevens, H. P.; Zlotogorski, A.; Schweizer, J.: A new mutation in the type II hair cortex keratin hHb1 involved in the inherited hair disorder monilethrix. *Hum. Genet.* 101: 165–169,1997.

[35882] 9434.Buckanovich, R. J.; Posner, J. B.; Darnell, R. B.: Nova, the paraneoplastic Ri antigen, is homologous to an RNA-binding protein and is specifically expressed in the developing motor system. *Neuron* 11: 657–672, 1993.

[35883] 9435.Buckanovich, R. J.; Yang, Y. Y.; Darnell, R. B.: The onconeural antigen Nova-1 is a neuron-specific RNA-binding protein, the activity of which is inhibited by paraneoplastic antibodies. *J. Neurosci.* 16:1114–1122, 1996.

- [35884] 9436.Prestigiacomo, C. J.; Balmaceda, C.; Dalmau, J.: Anti-Ri-associatedparaneoplastic opsoclonus-ataxia syndrome in a man with transitionalcell carcinoma: a case report. Cancer 91: 1423-1428, 2001.
- [35885] 9437.Anguita, J.; Chalfant, M. L.; Civan, M. M.; Coca-Prados, M.: Molecularcloning of the human volume-sensitive chloride conductance regulatoryprotein, pl(Cln), from ocular ciliary epithelium. Biochem. Biophys.Res. Commun. 208: 89-95, 1995.
- [35886] 9438.Buyse, G.; De Greef, C.; Raeymaekers, L.; Droogmans, G.; Nilius,B.; Eggermont, J.: The ubiquitously expressed pl(Cln) protein formshomomeric complexes in vitro. Biochem. Biophys. Res. Commun. 218:822-827, 1996.
- [35887] 9439.Nagl, U. O.; Erdel, M.; Schmarda, A.; Seri, M.; Pinggera, G. M.;Gschwentner, M.; Duba, C.; Galietta, L. J. V.; Deetjen, P.; Utermann,G.; Paulmichl, M.: Chromosomal localization of the genes (CLNS1Aand CLNS1B) coding for the swelling-dependent chloride channel I(Cln). Genomics 38:438-441, 1996.
- [35888] 9440.Schwartz, R. S.; Rybicki, A. C.; Nagel, R. L.: Molecular cloningand expression of a chloride channel-associated protein pl(Cln) inhuman young red blood cells: association

with actin. *Biochem. J.* 327:609–616, 1997.

- [35889] 9441. Muzio, M.; Ni, J.; Feng, P.; Dixit, V. M.: IRAK (Pelle) family member IRAK-2 and MyD88 as proximal mediators of IL-1 signaling. *Science* 278:1612–1615, 1997.
- [35890] 9442. Takeuchi, O.; Hoshino, K.; Akira, S.: Cutting edge: TLR2-deficient and MyD88-deficient mice are highly susceptible to *Staphylococcus aureus* infection. *J. Immun.* 165: 5392–5396, 2000.
- [35891] 9443. Eggertsen, G.; Olin, M.; Andersson, U.; Ishida, H.; Kubota, S.; Hellman, U.; Okuda, K.-I.; Bjorkhem, I.: Molecular cloning and expression of rabbit sterol 12- α -hydroxylase. *J. Biol. Chem.* 271: 32269–32275, 1996.
- [35892] 9444. Gafvels, M.; Olin, M.; Chowdhary, B. P.; Raudsepp, T.; Andersson, U.; Persson, B.; Jansson, M.; Bjorkhem, I.; Eggertsen, G.: Structure and chromosomal assignment of the sterol 12- α -hydroxylase gene (CYP8B1) in human and mouse: eukaryotic cytochrome P-450 gene devoid of introns. *Genomics* 56: 184–196, 1999.
- [35893] 9445. Zhang, M.; Chiang, J. Y. L.: Transcriptional regulation of the human sterol 12- α -hydroxylase gene (CYP8B1): roles of hepatocyte nuclear factor 4- α in mediating bile acid repression. *J. Biol. Chem.* 276:

41690–41699, 2001.

- [35894] 9446.Brinkman, A.; van der Flier, S.; Kok, E. M.; Dorssers, L. C. J.: BCAR1, a human homologue of the adapter protein p130Cas, and antiestrogenresistance in breast cancer cells. J. Nat. Cancer Inst. 92: 112–120,2000.
- [35895] 9447.Brugge, J. S.: Casting light on focal adhesions. Nature Genet. 19:309–311, 1998.
- [35896] 9448.Dorssers, L. C. J.; van Agthoven, T.; Dekker, A.; van Agthoven,T. L. A.; Kok, E. M.: Induction of antiestrogen resistance in humanbreast cancer cells by random insertional mutagenesis using defectiveretroviruses: identification of bcar–1, a common integration site. Molec.Endocr. 7: 870–878, 1993.
- [35897] 9449.Honda, H.; Oda, H.; Nakamoto, T.; Honda, Z.; Sakai, R.; Suzuki,T.; Saito, T.; Nakamura, K.; Nakao, K.; Ishikawa, T.; Katsuki, M.;Yazaki, Y.; Hirai, H.: Cardiovascular anomaly, impaired actin bundlingand resistance to Src-induced transformation in mice lacking p130–Cas. NatureGenet. 19: 361–365, 1998.
- [35898] 9450.Kanner, S. B.; Reynolds, A. B.; Wang, H. C.; Vines, R. R.; Parsons,J. T.: The SH2 and SH3 domains of pp60src direct stable associationwith tyrosine phosphorylated proteins p130 and p110. EMBO J. 10:1689–1698, 1991.

- [35899] 9451.Kuo, C. T.; Morrissey, E. E.; Anandappa, R.; Sigrist, K.; Lu, M.M.; Parmacek, M. S.; Soudais, C.; Leiden, J. M.: GATA4 transcriptionfactor is required for ventral morphogenesis and heart tube formation. *GenesDev.* 11: 1048–1060, 1997.
- [35900] 9452.Reynolds, A. B.; Kanner, S. B.; Wang, H. C.; Parsons, J. T.: Stableassociation of activated pp60src with two tyrosine–phosphorylatedcellular proteins. *Molec. Cell. Biol.* 9: 3951–3958, 1989.
- [35901] 9453.Sakai, R.; Iwamatsu, A.; Hirano, N.; Ogawa, S.; Tanaka, T.; Mano,H.; Yazaki, Y.; Hirai, H.: A novel signaling molecule, p130, formsstable complexes in vivo with v–Crk and v–Src in a tyrosine phosphorylation–dependentmanner. *EMBO J.* 13: 3748–3756, 1994.
- [35902] 9454.van der Flier, S.; Brinkman, A.; Look, M. P.; Kok, E. M.; Meijer–vanGelder, M. E.; Klijn, J. G. M.; Dorssers, L. C. J.; Foekens, J. A.: Bcar1/p130Cas protein and primary breast cancer: prognosis and responseto tamoxifen treatment. *J. Nat. Cancer Inst.* 92: 120–127, 2000.
- [35903] 9455.Yoshida, K.; Taga, T.; Saito, M.; Suematsu, S.; Kumanogoh, A.;Tanaka, T.; Fujiwara, H.; Hirata, M.; Yamagami, T.; Nakahata, T.;Hirabayashi, T.; Yoneda, Y.; Tanaka, K.; Wang, W. Z.; Mori, C.; Shiota,K.; Yoshida, N.;

Kishimoto, T.: Targeted disruption of gp130, a common signal transducer for the interleukin 6 family of cytokines, leads to myocardial and hematological disorders. *Proc. Nat. Acad. Sci.* 93:407–411, 1996.

[35904] 9456. Hirose, T.; Smith, R. J.; Jetten, A. M.: ROR-gamma: the third member of ROR/RZR orphan receptor subfamily that is highly expressed in skeletal muscle. *Biochem. Biophys. Res. Commun.* 205: 1976–1983, 1994.

[35905] 9457. Kurebayashi, S.; Ueda, E.; Sakaue, M.; Patel, D. D.; Medvedev, A.; Zhang, F.; Jetten, A. M.: Retinoid-related orphan receptor gamma (ROR-gamma) is essential for lymphoid organogenesis and controls apoptosis during thymopoiesis. *Proc. Nat. Acad. Sci.* 97: 10132–10137, 2000.

[35906] 9458. Medvedev, A.; Chistokhina, A.; Hirose, T.; Jetten, A. M.: Genomic structure and chromosomal mapping of the nuclear orphan receptor ROR-gamma (RORC) gene. *Genomics* 46: 93–102, 1997.

[35907] 9459. Soengas, M. S.; Capodice, P.; Polsky, D.; Mora, J.; Esteller, M.; Opitz-Araya, X.; McCombie, R.; Herman, J. G.; Gerald, W. L.; Lazebnik, Y. A.; Cordon-Cardo, C.; Lowe, S. W.: Inactivation of the apoptosis effector Apaf-1 in malignant melanoma. *Nature* 409: 207–211, 2001.

[35908] 9460. Srinivasula, S. M.; Ahmad, M.; Fernandes-Alnemri,

T.; Alnemri, E. S.: Autoactivation of procaspase-9 by Apaf-1-mediated oligomerization. *Molec. Cell* 1: 949-957, 1998.

- [35909] 9461. Yoshida, H.; Kong, Y.-Y.; Yoshida, R.; Elia, A. J.; Hakem, A.; Hakem, R.; Penninger, J. M.; Mak, T. W.: Apaf1 is required for mitochondrial pathways of apoptosis and brain development. *Cell* 94: 739-750, 1998.
- [35910] 9462. Zou, H.; Henzel, W. J.; Liu, X.; Lutschg, A.; Wang, X.: APAF-1, a human protein homologous to *C. elegans* CED-4, participates in cytochrome c-dependent activation of caspase-3. *Cell* 90: 405-413, 1997.
- [35911] 9463. Biervert, C.; Schroeder, B. C.; Kubisch, C.; Berkovic, S. F.; Propping, P.; Jentsch, T. J.; Steinlein, O. K.: A potassium channel mutation in neonatal human epilepsy. *Science* 279: 406-409, 1998.
- [35912] 9464. Biervert, C.; Steinlein, O. K.: Structural and mutational analysis of KCNQ2, the major gene locus for benign familial neonatal convulsions. *Hum. Genet.* 104: 234-240, 1999.
- [35913] 9465. Dedek, K.; Kunath, B.; Kananura, C.; Reuner, U.; Jentsch, T. J.; Steinlein, O. K.: Myokymia and neonatal epilepsy caused by a mutation in the voltage sensor of the KCNQ2 K(+) channel. *Proc. Nat. Acad. Sci.* 98:

12272–12277, 2001.

- [35914] 9466.del Giudice, E. M.; Coppola, G.; Scuccimarra, G.; Cirillo, G.; Bellini, G.; Pascotto, A.: Benign familial neonatal convulsions (BFNC) resulting from mutation of the KCNQ2 voltage sensor. *Europ. J. Hum. Genet.* 8: 994–997, 2000.
- [35915] 9467.Niederreither, K.; Abu–Abed, S.; Schuhbaur, B.; Petkovich, M.; Chambon, P.; Dolle, P.: Genetic evidence that oxidative derivatives of retinoic acid are not involved in retinoid signaling during mouse development. *Nature Genet.* 31: 84–88, 2002.
- [35916] 9468.White, J. A.; Beckett, B.; Scherer, S. W.; Herbrick, J.–A.; Petkovich, M.: P450RAI (CYP26A) maps to human chromosome 10q23–q24 and mouse chromosome 19C2–3. *Genomics* 48: 270–272, 1998.
- [35917] 9469.White, J. A.; Beckett–Jones, B.; Guo, Y.–D.; Dilworth, F. J.; Bonasoro, J.; Jones, G.; Petkovich, M.: cDNA cloning of human retinoic acid–metabolizing enzyme (hP450RAI) identifies a novel family of cytochromes P450 (CYP26). *J. Biol. Chem.* 272: 18538–18541, 1997.
- [35918] 9470.Chardin, P.; Paris, S.; Antonny, B.; Robineau, S.; Beraud–Dufour, S.; Jackson, C. L.; Chabre, M.: A human exchange factor for ARF contains Sec7– and pleckstrin–homology domains. *Nature* 384: 481–484, 1996.

- [35919] 9471.Perletti, L.; Talarico, D.; Trecca, D.; Ronchetti, D.; Fracchiolla, N. S.; Maiolo, A. T.; Neri, A.: Identification of a novel gene, PSD, adjacent to NFKB2/lyt-10, which contains Sec7 and pleckstrin-homology domains. *Genomics* 46: 251-259, 1997.
- [35920] 9472.Biunno, I.; Appierto, V.; Cattaneo, M.; Leone, B. E.; Balzano, G.; Socci, C.; Saccone, S.; Letizia, A.; Valle, G. D.; Sgaramella, V.: Isolation of a pancreas-specific gene located on human chromosome 14q31: expression analysis in human pancreatic ductal carcinomas. *Genomics* 46:284-286, 1997.
- [35921] 9473.Donoviel, D. B.; Bernstein, A.: SEL-1L maps to human chromosome 14, near the insulin-dependent diabetes mellitus locus 11. *Genomics* 56:232-233, 1999.
- [35922] 9474.Grant, B.; Greenwald, I.: The *Caenorhabditis elegans* sel-1 gene, a negative regulator of lin-12 and glp-1, encodes a predicted extracellular protein. *Genetics* 143: 237-247, 1996.
- [35923] 9475.Grant, B.; Greenwald, I.: Structure, function and expression of SEL-1, a negative regulator of LIN-12 and GLP-1 in *C. elegans*. *Development* 124:637-644, 1997.
- [35924] 9476.Frohling, S.; Nakabayashi, K.; Scherer, S. W.; Dohner, H.; Dohner, K.: Mutation analysis of the origin recognition

complex subunit 5(ORC5L) gene in adult patients with myeloid leukemias exhibiting deletionsof chromosome band 7q22. Hum. Genet. 108: 304–309, 2001.

- [35925] 9477.Ishiai, M.; Dean, F. B.; Okumura, K.; Abe, M.; Moon, K.-Y.; Amin,A. A.; Kagotani, K.; Taguchi, H.; Murakami, Y.; Hanaoka, F.; O'Donnell,M.; Hurwitz, J.; Eki, T.: Isolation of human and fission yeast homologuesof the budding yeast origin recognition complex subunit ORC5: humanhomo-
logue (ORC5L) maps to 7q22. Genomics 46: 294–298, 1997.
- [35926] 9478.Quintana, D. G.; Thome, K. C.; Hou, Z.; Ligon, A. H.; Morton, C.C.; Dutta, A.: ORC5L, a new member of the hu-
man origin recognitioncomplex, is deleted in uterine leiomyomas and malignant myeloid diseases. J.Biol. Chem. 273: 27137–27145, 1998.
- [35927] 9479.Ben Porath, I.; Kozak, C. A.; Benvenisty, N.: Chromo-
somal mappingof Tmp (Emp1), Xmp (Emp2), and Ymp (Emp3), genes encoding membraneproteins related to Pmp22. Genomics 49: 443–447, 1998.
- [35928] 9480.Chen, Y.; Medvedev, A.; Ruzanov, P.; Marvin, K. W.; Jetten, A.M.: cDNA cloning, genomic structure, and chro-
mosome mapping of thehuman epithelial membrane pro-
tein CL-20 gene (EMP1), a member of thePMP22 family.

Genomics 41: 40–48, 1997.

- [35929] 9481.Liehr, T.; Kuhlenthal, G.; Wulf, P.; Taylor, V.; Suter, U.; VanBroeckhoven, C.; Lupski, J. R.; Claussen, U.; Rautenstrauss, B.:Regional localization of the human epithelial membrane protein genes1, 2, and 3 (EMP1, EMP2, EMP3) to 12p12.3, 16p13.2, and 19q13.3. Genomics 58:106–108, 1999.
- [35930] 9482.Marvin, K. W.; Fujimoto, W.; Jetten, A. M.: Identification and characterization of a novel squamous cell-associated gene related to PMP22. J. Biol. Chem. 270: 28910–28916, 1995.
- [35931] 9483.Ruegg, C. L.; Wu, H.; Fagnoni, F. F.; Engleman, E. G.; Laus, R.: B4B, a novel growth-arrest gene, is expressed by a subset of progenitor/pre-B lymphocytes negative for cytoplasmic mu-chain. J. Immun. 157: 72–80, 1996.
- [35932] 9484.Taylor, V.; Suter, U.: Epithelial membrane protein-2 and epithelial membrane protein-3: two novel members of the peripheral myelin protein22 gene family. Gene 175: 115–120, 1996.
- [35933] 9485.Masiakowski, P.; Carroll, R. D.: A novel family of cell surface receptors with tyrosine kinase-like domain. J. Biol. Chem. 267:26181–26190, 1992.
- [35934] 9486.Adelmant, G.; Begue, A.; Stehelin, D.; Laudet, V.: A

functional Rev-erb- α responsive element located in the human Rev-erb- α promoter mediates a repressing activity. Proc. Nat. Acad. Sci. 93:3553–3558, 1996.

- [35935] 9487. Chawla, A.; Lazar, M. A.: Induction of Rev-ErbA α , an orphan receptor encoded on the opposite strand of the α -thyroid hormone receptor gene, during adipocyte differentiation. J. Biol. Chem. 268:16265–16269, 1993.
- [35936] 9488. Lazar, M. A.; Hodin, R. A.; Darling, D. S.; Chin, W. W.: A novel member of the thyroid/steroid hormone receptor family is encoded by the opposite strand of the rat c-erbA- α transcriptional unit. Molec. Cell. Biol. 9: 1128–1136, 1989.
- [35937] 9489. Miyajima, N.; Horiuchi, R.; Shibuya, S.; Matsubara, K.; Toyoshima, K.; Yamamoto, T.: Two erbA homologs encoding proteins with different T(3) binding capacities are transcribed from opposite DNA strands of the same genetic locus. Cell 57: 31–39, 1989.
- [35938] 9490. Preitner, N.; Damiola, F.; Lopez-Molina, L.; Zakany, J.; Duboule, D.; Albrecht, U.; Schibler, U.: The orphan nuclear receptor REV-ERB- α controls circadian transcription within the positive limb of the mammalian circadian oscillator. Cell 110: 251–260, 2002.

- [35939] 9491. Zhao, Q.; Khorasanizadeh, S.; Miyoshi, Y.; Lazar, M. A.; Rastinejad, F.: Structural elements of an orphan nuclear receptor–DNA complex. *Molec. Cell* 1: 849–861, 1998.
- [35940] 9492. Gregorini, A.; Sahin, F. I.; Lillington, D. M.; Meerabux, J.; Saha, V.; McCullagh, P.; Bocci, M.; Menevse, S.; Papa, S.; Young, B. D.: Gene BR140, which is related to AF10 and AF17, maps to chromosome band 3p25. *Genes Chromosomes Cancer* 17: 269–272, 1996.
- [35941] 9493. Thompson, K. A.; Wang, B.; Argraves, W. S.; Giannotti, F. G.; Schranck, D. P.; Ruoslahti, E.: BR140, a novel zinc–finger protein with homology to the TAF250 subunit of TFIID. *Biochem. Biophys. Res. Commun.* 1143–1152, 1994.
- [35942] 9494. Saito, T.; Seki, N.; Yamauchi, M.; Tsuji, S.; Hayashi, A.; Kozuma, S.; Hori, T.: Structure, chromosomal location, and expression profile of EXTR1 and EXTR2, new members of the multiple exostoses gene family. *Biochem. Biophys. Res. Commun.* 243: 61–66, 1998.
- [35943] 9495. Wuyts, W.; Van Hul, W.: Characterization and genomic localization of the mouse *Extl2* gene. *Cytogenet. Cell Genet.* 89: 185–188, 2000.
- [35944] 9496. Wuyts, W.; Van Hul, W.; Hendrickx, J.; Speleman, F.; Wauters, J.; De Boulle, K.; Van Roy, N.; Van Agtmael, T.;

Bossuyt, P.; Willems, P. J.: Identification and characterization of a novel member of the EXT gene family, EXTL2. *Eur. J. Hum. Genet.* 5: 382–389, 1997.

[35945] 9497. Dick, T.; Ray, K.; Salz, H. K.; Chia, W.: Cytoplasmic dynein (ddlc1) mutations cause morphogenetic defects and apoptotic cell death in *Drosophila melanogaster*. *Molec. Cell. Biol.* 16: 1966–1977, 1996.

[35946] 9498. Matsushita, M.; Endo, Y.; Taira, S.; Sato, Y.; Fujita, T.; Ichikawa, N.; Nakata, M.; Misuochi, T.: A novel human lectin with collagen- and fibrinogen-like domains which functions as an opsonin. *J. Biol. Chem.* 271: 2448–2454, 1996.

[35947] 9499. Haag, F.; Koch-Nolte, F.; Kuhl, M.; Lorenzen, S.; Thiele, H.-G.: Premature stop codons inactivate the RT6 genes of the human and chimpanzee species. *J. Molec. Biol.* 243: 537–546, 1994.

[35948] 9500. Koch-Nolte, F.; Haag, F.; Kuhl, M.; van Heyningen, V.; Hoovers, J.; Grzeschik, K.-H.; Singh, S.; Thiele, H.-G.: Assignment of the human RT6 gene to 11q13 by PCR screening of somatic cell hybrids and in situ hybridization. *Genomics* 18: 404–406, 1993.

[35949] 9501. Koch-Nolte, F.; Kuhl, M.; Haag, F.; Cetkovic-Cvrlje, M.; Leiter, E. H.; Thiele, H.-G.: Assignment of the human

and mouse genes formuscle ecto mono
(ADPriboseyl)transferase to a conserved linkage group on
human chromosome 11p15 and mouse chromosome 7.
Genomics 36: 215–216,1996.

[35950] 9502.Okazaki, I. J.; Zolkiewska, A.; Nightingale, M. S.;
Moss, J.:Immunological and structural conservation of
mammalian skeletal muscleglycosylphosphatidylinositol-
linked ADP-ribosyltransferases. Biochemistry
33:12828–12836, 1994.

[35951] 9503.Prochazka, M.; Gaskins, H. R.; Leiter, E. H.; Koch-
Nolte, F.; Haag,F.; Thiele, H.-G.: Chromosomal localiza-
tion, DNA polymorphism andexpression of Rt-6, the
mouse homologue of rat T-lymphocyte differentiation-
marker RT6. Immunogenetics 33: 152–156, 1991.

[35952] 9504.Banfi, S.; Borsani, G.; Rossi, E.; Bernard, L.; Guffanti,
A.; Rubboli,F.; Marchitello, A.; Giglio, S.; Coluccia, E.;
Zollo, M.; Zuffardi,O.; Ballabio, A.: Identification and map-
ping of human cDNAs homologousto Drosophila mutant
genes through EST database searching. NatureGenet. 13:
167–174, 1996.

[35953] 9505.Duncan, M. K.; Kos, L.; Jenkins, N. A.; Gilbert, D. J.;
Copeland,N. G.; Tomarev, S. I.: Eyes absent: a gene family
found in severalmetazoan phyla. Mammalian Genome 8:

479–485, 1997. Note: Erratum:Mammalian Genome 8:
877 only, 1997.

- [35954] 9506.Xu, P.-X.; Cheng, J.; Epstein, J. A.; Maas, R. L.: Mouse Eya genes are expressed during limb tendon development and encode a transcriptional activation function. *Proc. Nat. Acad. Sci.* 94: 11974–11979, 1997.
- [35955] 9507.Zimmerman, J. E.; Bui, Q. T.; Steingrimsson, E.; Nagle, D. L.; Fu, W.; Genin, A.; Spinner, N. B.; Copeland, N. G.; Jenkins, N. A.; Bucan, M.; Bonini, N. M.: Cloning and characterization of two vertebrate homologs of the *Drosophila* eyes absent gene. *Genome Res.* 7: 128–141, 1997.
- [35956] 9508.Morrissey, E. E.; Tang, Z.; Sigrist, K.; Lu, M. M.; Jiang, F.; Ip, H. S.; Parmacek, M. S.: GATA6 regulates HNF4 and is required for differentiation of visceral endoderm in the mouse embryo. *Genes Dev.* 12:3579–3590, 1998.
- [35957] 9509.Suzuki, E.; Evans, T.; Lowry, J.; Truong, L.; Bell, D. W.; Testa, J. R.; Walsh, K.: The human GATA-6 gene: structure, chromosomal location, and regulation of expression by tissue-specific and mitogen-responsive signals. *Genomics* 38: 283–290, 1996.
- [35958] 9510.Meurs, E.; Chong, K.; Galabru, J.; Shaun, N.; Thomas, S. B.; Kerr, I. M.; Williams, B. R. G.; Hovanesian, A. G.: Molecular cloning and characterization of the human dou-

ble-stranded RNA-activated protein kinase induced by interferon. *Cell* 62: 379–390, 1990.

[35959] 9511. Dunnick, W.; Shell, B. E.; Dery, C.: DNA sequences near the site of reciprocal recombination between a c-myc oncogene and an immunoglobulin switch region. *Proc. Nat. Acad. Sci.* 80: 7269–7273, 1983.

[35960] 9512. Bashir, R.; Strachan, T.; Keers, S.; Stephenson, A.; Mahjneh, I.; Marconi, G.; Nashef, L.; Bushby, K. M. D.: A gene for autosomal recessive limb-girdle muscular dystrophy maps to chromosome 2p. *Hum. Molec. Genet.* 3: 455–457, 1994.

[35961] 9513. Levy, G. G.; Nichols, W. C.; Lian, E. C.; Foroud, T.; McClintick, J. N.; McGee, B. M.; Yang, A. Y.; Slemieniak, D. R.; Stark, K. R.; Gruppo, R.; Sarode, R.; Shurin, S. B.; Chandrasekaran, V.; Stabler, S. P.; Sabio, H.; Bouhassira, E. E.; Upshaw, J. D., Jr.; Ginsburg, D.; Tsai, H.-M.: Mutations in a member of the ADAMTS gene family cause thrombotic thrombocytopenic purpura. *Nature* 413: 488–494, 2001.

[35962] 9514. Amrani, A.; Verdaguer, J.; Serra, P.; Tafuro, S.; Tan, R.; Santamaria, P.: Progression of autoimmune diabetes driven by avidity maturation of a T-cell population. *Nature* 406: 739–742, 2000.

[35963] 9515. Bowcock, A. M.; Kavathas, P.; Margolskee, R. F.;

Herzenberg, L.;Cavalli-Sforza, L. L.: An RFLP associated with pcDLeu2-14, a humanT-cell differentiation antigen CD8 (Leu2) cDNA mapped to 2p12. NucleicAcids Res. 14: 7817 only, 1986.

[35964] 9516.Bruns, G.; Kavathas, P.; Shiloh, Y.; Sakai, K.; Schwaber, J.; Latt,S. A.; Herzenberg, L. A.: The human T cell antigen Leu-2 (T8) is encoded on chromosome 2. Hum. Genet. 70: 311-314, 1985.

[35965] 9517.Giblin, P.; Ledbetter, J. A.; Kavathas, P.: A secreted form of the human lymphocyte cell surface molecule CD8 arises from alternative splicing. Proc. Nat. Acad. Sci. 86: 998-1002, 1989.

[35966] 9518.Kavathas, P.; Sukhatme, V. P.; Herzenberg, L. A.; Parnes, J. R.: Isolation of the gene encoding the human T-lymphocyte differentiation antigen Leu-2 (T8) by gene transfer and cDNA subtraction. Proc. Nat.Acad. Sci. 81: 7688-7692, 1984.

[35967] 9519.Ledbetter, J. A.; Evans, R. L.; Lipinski, M.; Cunningham-Rundles,C.; Good, R. A.; Herzenberg, L. A.: Evolutionary conservation of surface molecules that distinguish T lymphocyte helper-inducer and cytotoxic-suppressor subpopulations in mouse and man. J. Exp. Med. 153:310-323, 1981.

- [35968] 9520.Ledbetter, J. A.; Seaman, W. E.; Tsu, T. T.; Herzenberg, L. A.: Lyt-2 and Lyt-3 antigens are on two different polypeptide subunits linked by disulfide bonds: relationship of subunits to T cell cytolytic activity. *J. Exp. Med.* 153: 1503-1516, 1981.
- [35969] 9521.Leishman, A. J.; Naidenko, O. V.; Attinger, A.; Konig, F.; Lena, C. J.; Xiong, Y.; Chang, H.-C.; Reinherz, E.; Kronenberg, M.; Cheroutre, H.: T cell responses modulated through interaction between CD8-alpha-alpha and the nonclassical MHC class I molecule, TL. *Science* 294: 1936-1939, 2001.
- [35970] 9522.Littman, D. R.: The structure of the CD4 and CD8 genes. *Annu. Rev. Immun.* 5: 561-584, 1987.
- [35971] 9523.Littman, D. R.; Thomas, Y.; Maddon, P. J.; Chess, L.; Axel, R.: The isolation and sequence of the gene encoding T8: a molecule defining functional classes of T lymphocytes. *Cell* 40: 237-246, 1985.
- [35972] 9524.Mecucci, C.; Van Den Berghe, H.: OKT8-positive T-cell lymphoma associated with a chromosome rearrangement t(2;17) possibly involving the T8 locus. (Letter) *New Eng. J. Med.* 313: 185-186, 1985.
- [35973] 9525.Nakauchi, H.; Nolan, G. P.; Hsu, C.; Huang, H. S.; Kavathas, P.; Herzenberg, L. A.: Molecular cloning of Lyt-2, a

membrane glycoprotein marking a subset of mouse T lymphocytes: molecular homology to its human counterpart, Leu-2/T8, and to immunoglobulin variable regions.

Proc.Nat. Acad. Sci. 82: 5126–5130, 1985.

- [35974] 9526.Lindsay, E. A.; Vitelli, F.; Su, H.; Morishima, M.; Huynh, T.; Pramparo, T.; Jurecic, V.; Ogunrinu, G.; Sutherland, H. F.; Scambler, P. J.; Bradley, A.; Baldini, A.: Tbx1 haploinsufficiency in the DiGeorges syndrome region causes aortic arch defects in mice. *Nature* 410:97–101, 2001.
- [35975] 9527.Merscher, S.; Funke, B.; Epstein, J. A.; Heyer, J.; Puech, A.; Lu, M. M.; Xavier, R. J.; Demay, M. B.; Russell, R. G.; Factor, S.; Tokooya, K.; St. Jore, B.; and 12 others: TBX1 is responsible for cardiovascular defects in velo-cardio-facial/DiGeorge syndrome. *Cell* 104:619–629, 2001.
- [35976] 9528.Erikson, J.; Finger, L.; Sun, L.; ar-Rushdi, A.; Nishikura, K.; Minowada, J.; Finan, J.; Emanuel, B. S.; Nowell, P. C.; Croce, C.M.: Deregulation of c-myc by translocation of the alpha-locus of the T-cell receptor in T-cell leukemias. *Science* 232: 884–886, 1986.
- [35977] 9529.Erikson, J.; Nishikura, K.; ar-Rushdi, A.; Finan, J.; Emanuel, B.; Lenoir, G.; Nowell, P. C.; Croce, C. M.: Translocation of an immunoglobulin kappa locus to a region 3-prime of an unrearranged c-myc oncogene en-

hances c-myc transcription. Proc. Nat. Acad. Sci.
80:7581–7585, 1983.

- [35978] 9530. Feng, X.-H.; Liang, Y.-Y.; Liang, M.; Zhai, W.; Lin, X.: Direct interaction of c-Myc with Smad2 and Smad3 to inhibit TGF-beta-mediated induction of the CDK inhibitor p15(Ink4B). Molec. Cell 9: 133–143, 2002.
- [35979] 9531. Grandori, C.; Mac, J.; Siebelt, F.; Ayer, D. E.; Eisenman, R.N.: Myc-Max heterodimers activate a DEAD box gene and interact with multiple E box-related sites in vivo. EMBO J. 15: 4344–4357, 1996.
- [35980] 9532. Hamlyn, P. H.; Rabbitts, T. H.: Translocation joins c-myc and immunoglobulin gamma-1 genes in a Burkitt lymphoma revealing a third exon in the c-myc oncogene. Nature 304: 135–139, 1983.
- [35981] 9533. Hayday, A. C.; Gillies, S. D.; Saito, H.; Wood, C.; Wiman, K.; Hayward, W. S.; Tonegawa, S.: Activation of a translocated human c-myc gene by an enhancer in the immunoglobulin heavy-chain locus. Nature 307: 334–340, 1984.
- [35982] 9534. He, T.-C.; Sparks, A. B.; Rago, C.; Hermeking, H.; Zawel, L.; da Costa, L. T.; Morin, P. J.; Vogelstein, B.; Kinzler, K. W.: Identification of c-MYC as a target of the APC pathway. Science 281: 1509–1512, 1998.

- [35983] 9535.Heim, S.; Mitelman, F.: Nineteen of 26 cellular oncogenes precisely localized in the human genome map to one of the 83 bands involved in primary cancer-specific rearrangements. *Hum. Genet.* 75: 70–72, 1987.
- [35984] 9536.Henderson, A.; Ripley, S.; Heller, M.; Kieff, E.: Chromosome site for Epstein–Barr virus DNA in a Burkitt tumor cell line and in lymphocytes growth-transformed in vitro. *Proc. Nat. Acad. Sci.* 80:1987–1991, 1983.
- [35985] 9537.Jain, M.; Arvanitis, C.; Chu, K.; Dewey, W.; Leonhardt, E.; Trinh, M.; Sundberg, C. D.; Bishop, J. M.; Felsher, D. W.: Sustained loss of a neoplastic phenotype by brief inactivation of MYC. *Science* 297:102–104, 2002.
- [35986] 9538.Leder, P.: Personal Communication. Boston, Mass. 10/1/1982.
- [35987] 9539.Lee, T. C.; Li, L.; Philipson, L.; Ziff, E. B.: Myc represses transcription of the growth arrest gene *gas1*. *Proc. Nat. Acad. Sci.* 94:12886–12891, 1997.
- [35988] 9540.Leven, D.: Disentangling the MYC web. (Commentary) *Proc. Nat. Acad. Sci.* 99: 5757–5759, 2002.
- [35989] 9541.Lin, C. S.; Goldthwait, D. A.; Samols, D.: Identification of Alu transposition in human lung carcinoma cells. *Cell* 54: 153–159, 1988.
- [35990] 9542.Mager, D. L.; Goodchild, N. L.: Homologous recom-

ination between the LTRs of a human retrovirus-like element causes a 5-kb deletion in two siblings. *Am. J. Hum. Genet.* 45: 848–854, 1989.

- [35991] 9543. Magrath, I.; Erikson, J.; Whang-Peng, J.; Sieverts, H.; Armstrong, G.; Benjamin, D.; Triche, T.; Alabaster, O.; Croce, C. M.: Synthesis of kappa light chains by cell lines containing an 8;22 chromosomal translocation derived from a male homosexual with Burkitt's lymphoma. *Science* 222:1094–1098, 1983.
- [35992] 9544. Maguire, R. T.; Robins, T. S.; Thorgeirsson, S. S.; Heilman, C. A.: Expression of cellular myc and mos genes in undifferentiated B cell lymphomas of Burkitt and non-Burkitt types. *Proc. Nat. Acad. Sci.* 80: 1947–1950, 1983.
- [35993] 9545. Manolov, G.; Manolova, Y.: Marker band in one chromosome 14 from Burkitt lymphomas. *Nature* 237: 33–34, 1972.
- [35994] 9546. Marcu, K. B.; Harris, L. J.; Stanton, L. W.; Erikson, J.; Watt, R.; Croce, C. M.: Transcriptionally active c-myc oncogene is contained within NIARD, a DNA sequence associated with chromosome translocations in B-cell neoplasia. *Proc. Nat. Acad. Sci.* 80: 519–523, 1983.
- [35995] 9547. Menssen, A.; Hermeking, H.: Characterization of the c-MYC-regulated transcriptome by SAGE: identification

and analysis of c-MYC targetgenes. Proc. Nat. Acad. Sci. 99: 6274–6279, 2002.

[35996] 9548.Mitelman, F.: Catalog of Chromosome Aberrations in Cancer. New York: Alan R. Liss (pub.) (2nd ed.): 1985.

[35997] 9549.Morse, B.; Rotherg, P. G.; South, V. J.; Spandorfer, J. M.; Astrin,S. M.: Insertional mutagenesis of the myc locus by a LINE-1 sequencein a human breast carcinoma. Nature 333: 87–90, 1988.

[35998] 9550.Murphy, W.; Sarid, J.; Taub, R.; Vasicek, T.; Battey, J.; Lenoir,G.; Leder, P.: A translocated human c-myc onco-gene is altered ina conserved coding sequence. Proc. Nat. Acad. Sci. 83: 2939–2943,1986.

[35999] 9551.Nishikura, K.; ar-Rushdi, A.; Erikson, J.; Watt, R.; Rovera, G.;Croce, C. M.: Differential expression of the normal and of the translocatedhuman c-myc oncogenes in B cells. Proc. Nat. Acad. Sci. 80: 4822–4826,1983.

[36000] 9552.Pasqualucci, L.; Neumeister, P.; Goossens, T.; Nan-jangud, G.;Chaganti, R. S. K.; Kuppers, R.; Dalla-Favera, R.: Hypermutationof multiple proto-oncogenes in B-cell diffuse large-cell lymphomas. Nature 412:341–346, 2001.

[36001] 9553.Pelengaris, S.; Khan, M.; Evan, G. I.: Suppression of Myc-inducedapoptosis in beta cells exposes multiple oncogenic properties of Mycand triggers carcinogenic

progression. *Cell* 109: 321–334, 2002.

[36002] 9554. Persson, H.; Hennighausen, L.; Taub, R.; DeGrado, W.; Leder, P.: Antibodies to human c-myc oncogene product: evidence of an evolutionarily conserved protein induced during cell proliferation. *Science* 225:687–693, 1984.

[36003] 9555. Persson, H.; Leder, P.: Nuclear localization and DNA binding properties of a protein expressed by human c-myc oncogene. *Science* 225:718–721, 1984.

[36004] 9556. Peukert, K.; Staller, P.; Schneider, A.; Carmichael, G.; Hanel, F.; Eilers, M.: An alternative pathway for gene regulation by Myc. *EMBO J.* 16: 5672–5686, 1997.

[36005] 9557. Saito, H.; Hayday, A. C.; Wiman, K.; Hayward, W. S.; Tonegawa, S.: Activation of the c-myc gene by translocation: a model for translational control. *Proc. Nat. Acad. Sci.* 80: 7476–7480, 1983.

[36006] 9558. Brookman, K. W.; Tebbs, R. S.; Allen, S. A.; Tucker, J. D.; Swiger, R. R.; Lamerdin, J. E.; Carrano, A. V.; Thompson, L. H.: Isolation and characterization of mouse Xrcc1, a DNA repair gene affecting ligation. *Genomics* 22:180–188, 1994.

[36007] 9559. Lamerdin, J. E.; Montgomery, M. A.; Stilwagen, S. A.; Scheidecker, L. K.; Tebbs, R. S.; Brookman, K. W.; Thomp-

son, L. H.; Carrano, A.V.: Genomic sequence comparison of the human and mouse XRCC1 DNA repair gene regions. *Genomics* 25: 547–554, 1995.

[36008] 9560. Thompson, L. H.; Bachinski, L. L.; Stallings, R. L.; Dolf, G.; Weber, C. A.; Westerveld, A.; Siciliano, M. J.: Complementation of repair gene mutations on the hemizygous chromosome 9 in CHO: a third repair gene on human chromosome 19. *Genomics* 5: 670–679, 1989.

[36009] 9561. Whitehouse, C. J.; Taylor, R. M.; Thistlethwaite, A.; Zhang, H.; Karimi-Busheri, F.; Lasko, D. D.; Weinfeld, M.; Caldecott, K. W.: XRCC1 stimulates human polynucleotide kinase activity at damaged DNA termini and accelerates DNA single-strand break repair. *Cell* 104:107–117, 2001.

[36010] 9562. Gao, Y.; Sun, Y.; Frank, K. M.; Dikkes, P.; Fujiwara, Y.; Seidl, K. J.; Sekiguchi, J. M.; Rathbun, G. A.; Swat, W.; Wang, J.; Bronson, R. T.; Malynn, B. A.; Bryans, M.; Zhu, C.; Chaudhuri, J.; Davidson, L.; Ferrini, R.; Stamato, T.; Orkin, S. H.; Greenberg, M. E.; Alt, F. W.: A critical role for DNA end-joining proteins in both lymphogenesis and neurogenesis. *Cell* 95: 891–902, 1998.

[36011] 9563. Giaccia, A. J.; Denko, N.; MacLaren, R.; Mirman, D.; Waldren, C.; Hart, I.; Stamato, T. D.: Human chromosome 5 complements the DNA double-strand break-repair defi-

ciency and gamma-ray sensitivity of the XR-1 hamster variant. *Am. J. Hum. Genet.* 47: 459–469, 1990.

- [36012] 9564. Li, Z.; Otevrel, T.; Gao, Y.; Cheng, H.-L.; Seed, B.; Stamato, T. D.; Taccioli, G. E.; Alt, F. W.: The XRCC4 gene encodes a novel protein involved in DNA double-strand break repair and V(D)J recombination. *Cell* 83:1079–1089, 1995.
- [36013] 9565. Otevrel, T.; Stamato, T. D.: Regional localization of the XRCC4 human radiation repair gene. *Genomics* 27: 211–214, 1995.
- [36014] 9566. Blunt, T.; Taccioli, G. E.; Priestley, A.; Hafezparast, M.; McMillan, T.; Liu, J.; Cole, C. C.; White, J.; Alt, F. W.; Jackson, S. P.; Schurr, E.; Lehmann, A. R.; Jeggo, P. A.: A YAC contig encompassing the XRCC5(Ku80) DNA repair gene and complementation defective cells by YAC protoplast fusion. *Genomics* 30: 320–328, 1995.
- [36015] 9567. Fuentes, J.-J.; Pritchard, M. A.; Planas, A. M.; Bosch, A.; Ferrer, I.; Estivill, X.: A new human gene from the Down syndrome critical region encodes a proline-rich protein highly expressed in fetal brain and heart. *Hum. Molec. Genet.* 4: 1935–1944, 1995.
- [36016] 9568. Magnussen, K.: Beitrag zur Genetik und Histologie eines isolierten Augenalbinismus beim Kaninchen. *Z.*

Morph. Anthropol. 44: 127–135, 1952.

- [36017] 9569. Sukhatme, V. P.; Sizer, K. C.; Vollmer, A. C.; Hunkapiller, T.; Parnes, J. R.: The T cell differentiation antigen leu-2/T8 is homologous to immunoglobulin and T cell receptor variable regions. *Cell* 40:591–597, 1985.
- [36018] 9570. Sakaguchi, A. Y.; Lalley, P. A.; Naylor, S. L.: Human and mouse cellular myc protooncogenes reside on chromosomes involved in numerical and structural aberrations in cancer. *Somat. Cell Genet.* 9: 391–405, 1983.
- [36019] 9571. Chen, D. J.; Marrone, B. L.; Nguyen, T.; Stackhouse, M.; Zhao, Y.; Siciliano, M. J.: Regional assignment of a human DNA repair gene (XRCC5) to 2q35 by x-ray hybrid mapping. *Genomics* 21: 423–427, 1994.
- [36020] 9572. Kirchgessner, C. U.; Patil, C. K.; Evans, J. W.; Cuomo, C. A.; Fried, L. M.; Carter, T.; Oettinger, M. A.; Brown, J. M.: DNA-dependent kinase (p350) as a candidate gene for the murine SCID defect. *Science* 267:1178–1183, 1995.
- [36021] 9573. Oakley, C. E.; Oakley, B. R.: Identification of gamma-tubulin, a new member of the tubulin superfamily encoded by mipA gene of *Aspergillus nidulans*. *Nature* 338: 662–664, 1989.
- [36022] 9574. Rommens, J. M.; Durocher, F.; McArthur, J.; Tonin, P.; LeBlanc, J.-F.; Allen, T.; Samson, C.; Ferri, L.; Narod, S.;

Morgan, K.; Simard, J.: Generation of a transcription map at the HSD17B locus centromeric to BRCA1 at 17q21. *Genomics* 28: 530–542, 1995.

[36023] 9575. Simerly, C.; Wu, G.-J.; Zoran, S.; Ord, T.; Rawlins, R.; Jones, J.; Navara, C.; Gerrity, M.; Rinehart, J.; Binor, Z.; Asch, R.; Schatten, G.: The paternal inheritance of the centrosome, the cell's microtubule-organizing center, in humans, and the implications for infertility. *Nature Med.* 1:47–52, 1995.

[36024] 9576. Stearns, T.; Evans, L.; Kirschner, M.: Gamma-tubulin is a highly conserved component of the centrosome. *Cell* 65: 825–836, 1991.

[36025] 9577. Wise, D. O.; Krahe, R.; Oakley, B. R.: The gamma-tubulin gene family in humans. *Genomics* 67: 164–170, 2000.

[36026] 9578. Zheng, Y.; Jung, M. K.; Oakley, B. R.: Gamma-tubulin is present in *Drosophila melanogaster* and *Homo sapiens* and is associated with the centrosome. *Cell* 65: 817–823, 1991.

[36027] 9579. Bettaieb, A.; Fromont, P.; Rodet, M.; Godeau, B.; Duedari, N.; Bierling, P.: Br(b), a platelet alloantigen involved in neonatal alloimmune thrombocytopenia. *Vox Sang.* 60: 230–234, 1991.

- [36028] 9580. Carlsson, L. E.; Santoso, S.; Spitzer, C.; Kessler, C.; Greinacher, A.: The alpha-2 gene coding sequence T807/A873 of the platelet collagen receptor integrin alpha-2/beta-1 might be a genetic risk factor for the development of stroke in younger patients. *Blood* 93: 3583–3586, 1999.
- [36029] 9581. Chen, D. J.; Park, M. S.; Campbell, E.; Oshimura, M.; Liu, P.; Zhao, Y.; White, B. F.; Siciliano, M. J.: Assignment of a human DNA double-strand break repair gene (XRCC5) to chromosome 2. *Genomics* 13:1088–1094, 1992.
- [36030] 9582. Difilippantonio, M. J.; Zhu, J.; Chen, H. T.; Meffre, E.; Nussenzweig, M. C.; Max, E. E.; Ried, T.; Nussenzweig, A.: DNA repair protein Ku80 suppresses chromosomal aberrations and malignant transformation. *Nature* 404:510–514, 2000.
- [36031] 9583. Jeggo, P. A.; Hafezparast, M.; Thompson, A. F.; Broughton, B. C.; Kaur, G. P.; Zdzienicka, M. Z.; Athwal, R. S.: Localization of a DNA repair gene (XRCC5) involved in double-strand-break rejoining to human chromosome 2. *Proc. Nat. Acad. Sci.* 89: 6423–6427, 1992.
- [36032] 9584. Taccioli, G. E.; Gottlieb, T. M.; Blunt, T.; Priestley, A.; Demengeot, J.; Mizuta, R.; Lehmann, A. R.; Alt, F. W.; Jackson, S. P.; Jeggo, P. A.: Ku80: product of the XRCC5 gene

and its role in DNA repair and V(D)J recombination. *Science* 265: 1442–1445, 1994.

- [36033] 9585. Ohkuchi, A.; Shiraishi, H.; Minakami, H.; Eguchi, Y.; Izumi, A.; Sato, I.: Fetus with long QT syndrome manifested by tachyarrhythmia: a case report. *Prenat. Diag.* 19: 990–992, 1999.
- [36034] 9586. Pacia, S. V.; Devinsky, O.; Luciano, D. J.; Vazquez, B.: The prolonged QT syndrome presenting as epilepsy: a report of two cases and literature review. *Neurology* 44: 1408–1410, 1994.
- [36035] 9587. Piippo, K.; Swan, H.; Pasternack, M.; Chapman, H.; Paavonen, K.; Viitasalo, M.; Toivonen, L.; Kontula, K.: A founder mutation of the potassium channel KCNQ1 in long QT syndrome: implications for estimation of disease prevalence and molecular diagnostics. *J. Am. Coll. Cardiol.* 37: 562–568, 2001.
- [36036] 9588. Pony, J. C.; Matheyses, M.; Daubert, J. C.; Fourdilis, M.; Gouffault, J.: Le syndrome QT long–syncope familial: deux observations de syndrome de Romano et Ward. *Arch. Mal. Coeur* 70: 1105–1114, 1977.
- [36037] 9589. Priori, S. G.; Schwartz, P. J.; Napolitano, C.; Bianchi, L.; Dennis, A.; De Fusco, M.; Brown, A. M.; Casari, G.: A recessive variant of the Romano–Ward Long–QT syndrome?

Circulation 97: 2420–2425, 1998.

- [36038] 9590.Romano, C.: Congenital cardiac arrhythmia. (Letter) Lancet I:658–659, 1965.
- [36039] 9591.Romano, C.; Gemme, G.; Pongiglione, R.: Aritmie cardiache rare dell' eta pediatrica. II. Accessi sincopali per fibrillazione ventricolare parossistica. (Presentazione del primo caso della letteratura pediatrica italiana.) Clin. Pediat. 45: 656–683, 1963.
- [36040] 9592.Roy, N.; Kahlem, P.; Dausse, E.; Bennaceur, M.; Faure, S.; Weissenbach, J.; Komajda, M.; Denjoy, I.; Coumel, P.; Schwartz, K.; Guicheney, P.: Exclusion of HRAS from long QT locus. (Letter) Nature Genet. 8:113–114, 1994.
- [36041] 9593.Roy, P. R.; Emanuel, R.; Ismail, S. A.; Tayib, M. H.: Hereditary prolongation of the Q–T interval: genetic observations and management in three families with 12 affected members. Am. J. Cardiol. 37:237–243, 1976.
- [36042] 9594.Russell, M. W.; Dick, M., II; Collins, F. S.; Brody, L. C.: KVLQT1 mutations in three families with familial or sporadic long QT syndrome. Hum. Molec. Genet. 5: 1319–1324, 1996.
- [36043] 9595.Schmitt, N.; Schwarz, M.; Peretz, A.; Abitbol, I.; Attali, B.; Pongs, O.: A recessive C-terminal Jervell and Lange-Nielsen mutation of the KCNQ1 channel impairs

subunit assembly. EMBO J. 19: 332–340,2000.

- [36044] 9596.Schwartz, P. J.: Cardiac sympathetic innervation and the sudden infant death syndrome: a possible pathogenetic link. Am. J. Med. 60:167–172, 1976.
- [36045] 9597.Schwartz, P. J.; Priori, S. G.; Bloise, R.; Napolitano, C.; Ronchetti, E.; Piccinini, A.; Goj, C.; Breithardt, G.; Schulze-Bahr, E.; Wedekind, H.; Nastoli, J.: Molecular diagnosis in a child with sudden infant death syndrome. (Letter) Lancet 358: 1342–1343, 2001.
- [36046] 9598.Schwartz, P. J.; Stramba-Badiale, M.; Segantini, A.; Austoni, P.; Bosi, G.; Giorgetti, R.; Grancini, F.; Marni, E. D.; Perticone, F.; Rosti, D.; Salice, P.: Prolongation of the QT interval and the sudden infant death syndrome. New Eng. J. Med. 338: 1709–1714, 1998.
- [36047] 9599.Shalaby, F. Y.; Levesque, P. C.; Yang, W.-P.; Little, W. A.; Conder, M. L.; Jenkins-West, T.; Blannar, M. A.: Dominant-negative KvLQT1 mutations underlie the LQT1 form of long QT syndrome. Circulation 96:1733–1736, 1997.
- [36048] 9600.Shimizu, W.; Kurita, T.; Matsuo, K.; Suyama, K.; Aihara, N.; Kamakura, S.; Towbin, J. A.; Shimomura, K.: Improvement of repolarization abnormalities by a K⁺ channel opener in the LQT1 form of congenital long-QT syndrome. Circulation 97:1581–1588, 1998.

- [36049] 9601.Singer, P. A.; Crampton, R. S.; Bass, N. H.: Familial Q-T prolongationsyndrome: convulsive seizures and paroxysmal ventricular fibrillation. Arch.Neurol. 31: 64-66, 1974.
- [36050] 9602.Smilinich, N. J.; Day, C. D.; Fitzpatrick, G. V.; Caldwell, G.M.; Lossie, A. C.; Cooper, P. R.; Smallwood, A. C.; Joyce, J. A.;Schofield, P. N.; Reik, W.; Nicholls, R. D.; Weksberg, R.; Driscoll,D. J.; Maher, E. R.; Shows, T. B.; Higgins, M. J.: A maternally methylatedCpG island in KvLQT1 is associated with an antisense paternal transcriptand loss of imprinting in Beckwith-Wiedemann syndrome. Proc. Nat.Acad. Sci. 96: 8064-8069, 1999.
- [36051] 9603.Splawski, I.; Timothy, K. W.; Vincent, G. M.; Atkinson, D. L.;Keating, M. T.: Molecular basis of the long-QT syndrome associatedwith deafness. New Eng. J. Med. 336: 1562-1567, 1997.
- [36052] 9604.Campbell, H. D.; Webb, G. C.; Young, I. G.: A human homologueof the Drosophila melanogaster sluggish-A (proline oxidase) gene mapsto 22q11.2, and is a candidate gene for type-I hyperprolinaemia. Hum.Genet. 101: 69-74, 1997.
- [36053] 9605.Slominski, A.; Ermak, G.; Mihm, M.: ACTH receptor, CYP11A1, CYP17and CYP21A2 genes are expressed in

skin. J. Clin. Endocr. Metab. 81:2746–2749, 1996.

- [36054] 9606. Touraine, J. L.; Betuel, H.; Souillet, G.; Jeune, M.: Combined immunodeficiency disease associated with absence of cell-surface HLA-A and -B antigens. J. Pediatr. 93: 47–51, 1978.
- [36055] 9607. Wolf, H. M.; Hauber, I.; Gulle, H.; Thon, V.; Eggensbauer, H.; Fischer, M. B.; Fiala, S.; Eibl, M. M.: Brief report: Twin boys with major histocompatibility complex class II deficiency but inducible immune responses. New Eng. J. Med. 332: 86–90, 1995.
- [36056] 9608. Dupuis, S.; Dargemont, C.; Fieschi, C.; Thomassin, N.; Rosenzweig, S.; Harris, J.; Holland, S. M.; Schreiber, R. D.; Casanova, J.-L.: Impairment of mycobacterial but not viral immunity by a germline human STAT1 mutation. Science 293: 300–303, 2001.
- [36057] 9609. Findlay, K. A. B.; Kaptein, E.; Visser, T. J.; Burchell, B.: Characterization of the uridine diphosphate-glucuronosyltransferase-catalyzing thyroid hormone glucuronidation in man. J. Clin. Endocr. Metab. 85: 2879–2883, 2000.
- [36058] 9610. Wooster, R.; Sutherland, L.; Ebner, T.; Clarke, D.; Da Cruz e Silva, O.; Burchell, B.: Cloning and stable expression of a new member of the human liver phenol/bilirubin-

bin:UDP-glucuronosyltransferase cDNAfamily. *Biochem. J.* 278: 465–469, 1991.

[36059] 9611.Tanaka, S.; Yamashita, S.; Hosaka, K.: Cloning and expressionof human cDNA encoding phosphatidylinositol transfer protein beta. *Biochim.Biophys. Acta* 1259: 199–202, 1995.

[36060] 9612.Choi, Y.–H.; Kim, K.–B.; Kim, H.–H.; Hong, G.–S.; Kwon, Y.–K.;Chung, C.–W.; Park, Y.–M.; Shen, Z.–J.; Kim, B. J.; Lee, S.–Y.; Jung,Y.–K.: FLASH coordinates NF–kappa–B activity via TRAF2. *J. Biol.Chem.* 276: 25073–25077, 2001.

[36061] 9613.Imai, Y.; Kimura, T.; Murakami, A.; Yajima, N.; Sakamaki, K.; Yonehara,S.: The CED–4–homologous protein FLASH is involved in Fas–mediatedactivation of caspase–8 during apoptosis. *Nature* 398: 777–785, 1999.

[36062] 9614.Cossee, M.; Durr, A.; Schmitt, M.; Dahl, N.; Trouillas, P.; Allinson,P.; Kostrzewa, M.; Nivelon–Chevallier, A.; Gustavson, K.–H.; Kohlschutter,A.; Muller, U.; Mandel, J.–L.; and 11 others: Friedreich's ataxia:point mutations and clinical presentation of compound heterozygotes. *Ann.Neurol.* 45: 200–206, 1999.

[36063] 9615.De Castro, M.; Garcia–Planells, J.; Monros, E.; Canizares, J.;Vazquez–Manrique, R.; Vilchez, J. J.; Urtasun,

M.; Lucus, M.; Navarro, G.; Izquierdo, G.; Molto, M. D.; Palau, F.: Genotype and phenotype analysis of Friedreich's ataxia compound heterozygous patients. *Hum. Genet.* 106: 86–92, 2000.

[36064] 9616. Delatycki, M. B.; Paris, D.; Gardner, R. J. M.; Forshaw, K.; Nicholson, G. A.; Nassif, N.; Williamson, R.; Forrest, S. M.: Sperm DNA analysis in a Friedreich ataxia premutation carrier suggests both meiotic and mitotic expansion in the *FRDA* gene. *J. Med. Genet.* 35: 713–716, 1998.

[36065] 9617. Duclos, F.; Boschert, U.; Sirugo, G.; Mandel, J.-L.; Hen, R.; Koenig, M.: Gene in the region of the Friedreich ataxia locus encodes a putative transmembrane protein expressed in the nervous system. *Proc. Nat. Acad. Sci.* 90: 109–113, 1993.

[36066] 9618. Doi, A.; Shiosaka, T.; Takaoka, Y.; Yanagisawa, K.; Fujita, S.: Molecular cloning of the cDNA encoding A+U-rich element RNA binding factor. *Biochim. Biophys. Acta* 1396: 51–56, 1998.

[36067] 9619. Kamei, D.; Tsuchiya, N.; Yamazaki, M.; Meguro, H.; Yamada, M.: Two forms of expression and genomic structure of the human heterogeneous nuclear ribonucleoprotein D-like JKTBP gene (*HNRPDL*). *Gene* 228: 13–22, 1999.

[36068] 9620. Tsuchiya, N.; Kamei, D.; Takano, A.; Matsui, T.; Ya-

mada, M.: Cloning and characterization of a cDNA encoding a novel heterogeneous nuclear ribonucleoprotein-like protein and its expression in myeloid leukemia cells. *J. Biochem.* 123: 499–507, 1998.

[36069] 9621. Ma, Y. H.; Betard, C.; Roy, M.; Davignon, J.; Kessling, A. M.: Identification of a second 'French Canadian' LDL receptor gene deletion and development of a rapid method to detect both deletions. *Clin. Genet.* 36: 219–228, 1989.

[36070] 9622. Mandelshtam, M.; Chakir, K.; Shevtsov, S.; Golubkov, V.; Skobeleva, N.; Lipovetsky, B.; Konstantinov, V.; Denisenko, A.; Gaitskhoki, V.; Schwartz, E.: Prevalence of Lithuanian mutation among St. Petersburg Jews with familial hypercholesterolemia. *Hum. Mutat.* 12: 255–258, 1998.

[36071] 9623. Meiner, V.; Landsberger, D.; Berkman, N.; Reshef, A.; Segal, P.; Seftel, H. C.; van der Westhuyzen, D. R.; Jeenah, M. S.; Coetzee, G. A.; Leitersdorf, E.: A common Lithuanian mutation causing familial hypercholesterolemia in Ashkenazi Jews. *Am. J. Hum. Genet.* 49: 443–449, 1991.

[36072] 9624. Miyake, Y.; Tajima, S.; Funahashi, T.; Yamamura, T.; Yamamoto, A.: A point mutation of low-density-lipoprotein receptor causing rapid degradation of the receptor. *Europ. J. Biochem.* 210: 1–7, 1992.

[36073] 9625. Moorjani, S.; Roy, M.; Torres, A.; Betard, C.; Gagne,

C.; Lambert,M.; Brun, D.; Davignon, J.; Lupien, P.: Mutations of low-density-lipoprotein-receptorgene, variation in plasma cholesterol, and expression of coronaryheart disease in homozygous familial hypercholesterolaemia. Lancet 341:1303-1306, 1993.

[36074] 9626.Oppenheim, A.; Friedlander, Y.; Dann, E. J.; Berkman, N.; Schwartz,S. P.; Leitersdorf, E.: Hypercholesterolemia in five Israeli Christian-Arabkindreds is caused by the 'Lebanese' allele at the low density lipoproteinreceptor gene locus and by an additional independent major factor. Hum.Genet. 88: 75-84, 1991.

[36075] 9627.Pisciotta, L.; Cantafora, A.; De Stefano, F.; Langheim, S.; Calandra,S.; Bertolini, S.: A 'de novo' mutation of the LDL-receptor geneas the cause of familial hypercholesterolemia. Biochim. Biophys.Acta 1587: 7-11, 2002.

[36076] 9628.Rodningen, O. K.; Rosby, O.; Tonstad, S.; Ose, L.; Berg, K.; Leren,T. P.: A 9.6 kilobase deletion in the low density lipoprotein receptorgene in Norwegian familial hypercholesterolemia subjects. Clin. Genet. 42:288-295, 1992.

[36077] 9629.Rubinsztein, D. C.; Coetzee, G. A.; Marais, A. D.; Leitersdorf,E.; Seftel, H. C.; van der Westhuyzen, D. R.: Identification andproperties of the proline-644-to-leucine

mutant LDL receptor in SouthAfricans of Indian origin. J. Lipid Res. 33: 1647–1655, 1992.

- [36078] 9630.Rudiger, N. S.; Heinsvig, E. M.; Hansen, F. A.; Faergeman, O.;Bolund, L.; Gregersen, N.: DNA deletions in the low density lipoprotein(LDL) receptor gene in Danish families with familial hypercholesterolemia. Clin.Genet. 39: 451–462, 1991.
- [36079] 9631.Ruffner, D. E.; Sprung, C. N.; Minghetti, P. P.; Gibbs, P. E.M.; Dugaiczky, A.: Invasion of the human albumin–alpha–fetoproteingene family by Alu, Kpn, and two novel repetitive DNA elements. Molec.Biol. Evol. 4: 1–9, 1987.
- [36080] 9632.Russell, D. W.; Lehrman, M. A.; Sudhof, T. C.; Yamamoto, T.; Davis,C. G.; Hobbs, H. H.; Brown, M. S.; Goldstein, J. L.: The LDL receptorin familial hypercholesterolemia: use of human mutations to dissecta membrane protein. Cold Spring Harbor Symp. Quant. Biol. 51: 811–819,1986.
- [36081] 9633.Russell, D. W.; Schneider, W. J.; Yamamoto, T.; Luskey, K. L.;Brown, M. S.; Goldstein, J. L.: Domain map of the LDL receptor: sequencehomology with the epidermal growth factor precursor. Cell 37: 577–585,1984.
- [36082] 9634.Schuster, H.; Manke, C.; Fischer, J.; Keller, C.; Wolfram, G.;Zollner, N.: Identification of the valine 408 to

methionine mutation in the LDL receptor in a Greek patient with homozygous familial hypercholesterolemia.

Clin.Genet. 48: 90–92, 1995.

[36083] 9635.Schuster, H.; Ostwald, P.; Keller, P.; Wolfram, G.;

Keller, C.: Identification of the serine–156 to leucine mutation in the low–density lipoprotein receptor in a German family with familial hypercholesterolemia. Clin.Investig. 71: 172–175, 1993.

[36084] 9636.Slagel, V.; Flemington, E.; Traina–Dorge, V.; Bradshaw, H.; Deininger, P.: Clustering and subfamily relationships of the Alu family in the human genome. Molec. Biol. Evol. 4: 19–29, 1987.100. Soutar, A. K.; Knight, B. L.; Patel, D. D.: Identification of a point mutation in growth repeat C of the low density lipoprotein–receptor gene in a patient with homozygous familial hypercholesterolemia that affects ligand binding and intracellular movement of receptors. Proc.Nat. Acad. Sci. 86: 4166–4170, 1989.101. Soutar, A. K.; McCarthy, S. N.; Seed, M.; Knight, B. L.: Relationship between apolipoprotein(a) phenotype, lipoprotein(a) concentration in plasma, and low density lipoprotein receptor function in a large kindred with familial hypercholesterolemia due to the pro664–to–leu mutation in the LDL receptor gene. J. Clin. Invest. 88:

483–492,1991.102. Steyn, K.; Weight, M. J.; Dando, B. R.; Christopher, K. J.; Rossouw, J. E.: The use of low density lipoprotein receptor activity of lymphocytes to determine the prevalence of familial hypercholesterolaemia in a rural South African community. *J. Med. Genet.* 26: 32–36, 1989.103. Sudhof, T. C.; Goldstein, J. L.; Brown, M. S.; Russell, D. W.: The LDL receptor gene: a mosaic of exons shared with different proteins. *Science* 228:815–822, 1985.104. Sudhof, T. C.; Russell, D. W.; Goldstein, J. L.; Brown, M. S.; Sanchez-Pescador, R.; Bell, G. I.: Cassette of eight exons shared by genes for LDL receptor and EGF precursor. *Science* 228: 893–895, 1985.105. Sun, X.-M.; Patel, D. D.; Bhatnager, D.; Knight, B. L.; Soutar, A. K.: Characterization of a splice-site mutation in the gene for the LDL receptor associated with an unpredictably severe clinical phenotype in English patients with heterozygous FH. *Arterioscler. Thromb. Vasc. Biol.* 15: 219–227, 1995.106. Takahashi, M.; Ikeda, U.; Takahashi, S.; Hattori, H.; Iwasaki, T.; Ishihara, M.; Egashira, T.; Honma, S.; Asano, Y.; Shimada, K.: A novel mutation in exon 2 of the low-density lipoprotein-receptor gene in a patient with homozygous familial hypercholesterolemia. (Letter) *Clin. Genet.* 59: 290–292, 2001.107. Taylor, R.; Bryant, J.;

Gudnason, V.; Sigurdsson, G.; Humphries, S.: A study of familial hypercholesterolaemia in Iceland using RFLPs. *J. Med. Genet.* 26: 494–498, 1989.108. Thiart, R.; Scholtz, C. L.; Vergotine, J.; Hoogendijk, C. F.; de Villiers, J. N. P.; Nissen, H.; Brusgaard, K.; Gaffney, D.; Hoffs, M. S.; Vermaak, W. J. H.; Kotze, M. J.: Predominance of a 6 bp deletion in exon 2 of the LDL receptor gene in Africans with familial hypercholesterolaemia. *J. Med. Genet.* 37: 514–519, 2000.109. Tolleshaug, H.; Goldstein, J. L.; Schneider, W. J.; Brown, M. S.: Posttranslational processing of the LDL receptor and its genetic disruption in familial hypercholesterolemia. *Cell* 30: 715–724, 1982.110. Top, B.; Koeleman, B. P. C.; Gevers Leuven, J. A.; Havekes, L. M.; Frants, R. R.: Rearrangements in the LDL receptor gene in Dutch-familial hypercholesterolemic patients and the presence of a common 4 kb deletion. *Atherosclerosis* 83: 127–136, 1990.111. Top, B.; Uitterlinden, A. G.; van der Zee, A.; Kastelein, J. J. P.; Gevers Leuven, J. A.; Havekes, L. M.; Frants, R. R.: Absence of mutations in the promoter region of the low density lipoprotein receptor gene in a large number of familial hypercholesterolaemia patients as revealed by denaturing gradient gel electrophoresis. *Hum. Genet.* 89: 561–565, 1992.112. Torrington, M.;

Botha, J. L.: Familial hypercholesterolaemia and church affiliation. (Letter) *Lancet* II: 1120 only, 1981.113. Ullu, E.; Tschudi, C.: Alu sequences are processed 7SL RNA genes. *Nature* 312:171–172, 1984.114. Varret, M.; Rabes, J.-P.; Collod-Beroud, G.; Junien, C.; Boileau, C.; Beroud, C.: Software and database for the analysis of mutations in the human LDL receptor gene. *Nucleic Acids Res.* 25: 172–180, 1997.115. Vergopoulos, A.; Bajari, T.; Jouma, M.; Knoblauch, H.; Aydin, A.; Bähring, S.; Mueller-Myhsok, B.; Dresel, A.; Joubran, R.; Luft, F. C.; Schuster, H.: A xanthomatosis-susceptibility gene may exist in a Syrian family with familial hypercholesterolemia. *Europ. J. Hum. Genet.* 5: 315–323, 1997.116. Vergotine, J.; Thiart, R.; Langenhoven, E.; Hillermann, R.; DeJong, G.; Kotze, M. J.: Prenatal diagnosis of familial hypercholesterolemia: importance of DNA analysis in the high-risk South African population. *Genet. Counsel.* 12: 121–127, 2001.117. Vuorio, A. F.; Turtola, H.; Piilahti, K.-M.; Repo, P.; Kanninen, T.; Kontula, K.: Familial hypercholesterolemia in the Finnish North Karelia: a molecular, clinical, and genealogical study. *Arterioscler. Thromb. Vasc. Biol.* 17: 3127–3138, 1997.118. Wilson, D. J.; Gahan, M.; Haddad, L.; Heath, K.; Whittall, R. A.; Williams, R. R.; Humphries, S. E.; Day, I. N.

M.: A World WideWeb site for low-density lipoprotein receptor gene mutations in familialhypercholesterolemia: sequence-based, tabular, and direct submissiondata handling. Am. J. Cardiol. 81: 1509–1511, 1998.119. Yamakawa, K.; Okafuji, T.; Iwamura, Y.; Yuzawa, K.; Satoh, J.;Hattori, N.; Yamanouchi, Y.; Yanagi, H.; Kawai, K.; Tsuchiya, S.;Russell, D. W.; Hamaguchi, H.: Taql polymorphism in the LDL receptorgene and a Taql 1.5-kb band associated with familial hypercholesterolemia. Hum.Genet. 80: 1–5, 1988.120. Yamakawa, K.; Takada, K.; Yanagi, H.; Tsuchiya, S.; Kawai, K.;Nakagawa, S.; Kajiyama, G.; Hamaguchi, H.: Three novel partial deletionsof the low-density lipoprotein (LDL) receptor gene in familial hypercholesterolemia. Hum.Genet. 82: 317–321, 1989.121. Yamamoto, T.; Davis, C. G.; Brown, M. S.; Schneider, W. J.; Casey,M. L.; Goldstein, J. L.; Russell, D. W.: The human LDL receptor:a cysteine-rich protein with multiple Alu sequences in its mRNA. Cell 39:27–38, 1984.122. Zuliani, G.; Hobbs, H. H.: Personal Communication. Dallas,Tex. 1990.

[36085] 9637.Ishibashi, K.; Suzuki, M.; Sasaki, S.; Imai, M.: Identificationof a new multigene four transmembrane family (MS4A) related to CD20,HTm4 and beta subunit of the

high affinity IgE receptor. *Gene* 264:87–93, 2001.

[36086] 9638.Liang, Y.; Tedder, T. F.: Identification of a CD20–, Fc–epsilon–RI–beta–, and HTm4–related gene family: sixteen new MS4A family members expressed in human and mouse. *Genomics* 72: 119–127, 2001.

[36087] 9639.Nagase, T.; Kikuno, R.; Hattori, A.; Kondo, Y.; Okumura, K.; Ohara, O.: Prediction of the coding sequences of unidentified human genes. XIX. The complete sequences of 100 new cDNA clones from brain which code for large proteins in vitro. *DNA Res.* 7: 347–355, 2000.

[36088] 9640.Gingras, M. C.; Lapillonne, H.; Margolin, J. F.: CFFM4: a new member of the CD20/Fc–epsilon–RI–beta family. *Immunogenetics* 53:468–476, 2001.

[36089] 9641.O'Carroll, D.; Scherthan, H.; Peters, A. H. F. M.; Opravil, S.; Haynes, A. R.; Laible, G.; Rea, S.; Schmid, M.; Lebersorger, A.; Jerratsch, M.; Sattler, L.; Mattei, M. G.; Denny, P.; Brown, S. D. M.; Schweizer, D.; Jenuwein, T.: Isolation and characterization of Suv39h2, a second histone H3 methyltransferase gene that displays testis-specific expression. *Molec. Cell. Biol.* 20: 9423–9433, 2000.

[36090] 9642.Davis, R. S.; Wang, Y.–H.; Kubagawa, H.; Cooper, M. D.: Identification of a family of Fc receptor homologs with

preferential B cell expression. Proc.Nat. Acad. Sci. 98: 9772–9777, 2001.

[36091] 9643.Xu, M.; Zhao, R.; Zhao, Z. J.: Molecular cloning and characterization of SPAP1, an inhibitory receptor. Biochem. Biophys. Res. Commun. 280:768–775, 2001.

[36092] 9644.Drewes, G.; Ebner, A.; Preuss, U.; Mandelkow, E. M.; Mandelkow, E.: MARK, a novel family of protein kinases that phosphorylate microtubule-associated proteins and trigger microtubule disruption. Cell 89: 297–308, 1997.

[36093] 9645.Nagase, T.; Kikuno, R.; Ishikawa, K.; Hirose, M.; Ohara, O.: Prediction of the coding sequences of unidentified human genes. XVII. The complete sequences of 100 new cDNA clones from brain which code for large proteins in vitro. DNA Res. 7: 143–150, 2000.

[36094] 9646.Yoshikawa, T.; Sanders, A. R.; Esterling, L. E.; Detera-Wadleigh, S. D.: Multiple transcriptional variants and RNA editing in C18orf1, a novel gene with LDLRA and transmembrane domains on 18p11.2. Genomics 47:246–257, 1998.

[36095] 9647.Yoshikawa, T.; Sanders, A. R.; Esterling, L. E.; Overhager, J.; Barnes, J. A.; Lennon, G.; Grewal, R.; Detera-Wadleigh, S. D.: Isolation of chromosome 18-specific brain transcripts as positional candidates for bipolar disorder.

Am. J. Med. Genet. (Neuropsych. Genet.) 74:140–149, 1997.

- [36096] 9648. Anneren, C.; Reedquist, K. A.; Bos, J. L.; Welsh, M.: GTK, a Src-related tyrosine kinase, induces nerve growth factor-independent neurite outgrowth in PC12 cells through activation of the Rap1 pathway: relationship to Shb tyrosine phosphorylation and elevated levels of focal adhesion kinase. J. Biol. Chem. 275: 29153–29161, 2000.
- [36097] 9649. Cance, W. G.; Craven, R. J.; Bergman, M.; Xu, L.; Altitalo, K.; Liu, E. T.: Rak, a novel nuclear tyrosine kinase expressed in epithelial cells. Cell Growth Differ. 5: 1347–1355, 1994.
- [36098] 9650. Cance, W. G.; Craven, R. J.; Weiner, T. M.; Liu, E. T.: Novel protein kinases expressed in human breast cancer. Int. J. Cancer 54:571–577, 1993.
- [36099] 9651. Lee, J.; Wang, Z.; Luoh, S.-M.; Wood, W. I.; Scadden, D. T.: Cloning of FRK, a novel human intracellular SRC-like tyrosine kinase-encoding gene. Gene 138: 247–251, 1994.
- [36100] 9652. Scott, A. F.: Personal Communication. Baltimore, Md. 12/19/2001.
- [36101] 9653. Cao, L.; Zhang, L.; Ruiz-Lazano, P.; Yang, Q.; Chien, K. R.; Graham, R. M.; Zhou, M.: A novel putative protein-tyrosine phosphatase contains a BRO1-like domain and

suppresses Ha-ras-mediated transformation. J.Biol. Chem. 273: 21077–21083, 1998.

[36102] 9654. Toyooka, S.; Ouchida, M.; Jitsumori, Y.; Tsukuda, K.; Sakai, A.; Nakamura, A.; Shimizu, N.; Shimizu, K.: HD-PTP: a novel protein tyrosinephosphatase gene on human chromosome 3p21.3. Biochem. Biophys. Res. Commun. 278: 671–678, 2000.

[36103] 9655. Betz, A.; Thakur, P.; Junge, H. J.; Ashery, U.; Rhee, J.-S.; Scheuss, V.; Rosenmund, C.; Rettig, J.; Brose, N.: Functional interaction of the active zone proteins Munc13–1 and RIM1 in synaptic vesicle priming. Neuron 30: 183–196, 2001.

[36104] 9656. Castillo, P. E.; Schoch, S.; Schmitz, F.; Sudhof, T. C.; Malenka, R. C.: RIM1–alpha is required for presynaptic long-term potentiation. Nature 415: 327–330, 2002.

[36105] 9657. Coppola, T.; Magnin-Luthi, S.; Perret-Menoud, V.; Gattesco, S.; Schiavo, G.; Regazzi, R.: Direct interaction of the Rab3 effector RIM with Ca(2+) channels, SNAP–25, and synaptotagmin. J. Biol. Chem. 276: 32756–32762, 2001.

[36106] 9658. Schoch, S.; Castillo, P. E.; Jo, T.; Mukherjee, K.; Geppert, M.; Wang, Y.; Schmitz, F.; Malenka, R. C.; Sudhof, T. C.: RIM1–alpha forms a protein scaffold for regulating neurotransmitter release at the active zone. Nature 415:

321–326, 2002.

- [36107] 9659.Wang, Y.; Sugita, S.; Sudhof, T. C.: The RIM/NIM family of neuronal C-2 domain proteins: interactions with Rab3 and a new class of Src homology 3 domain proteins. *J. Biol. Chem.* 275: 20033–20044, 2000.
- [36108] 9660.Cha, S. H.; Sekine, T.; Kusuvara, H.; Yu, E.; Kim, J. Y.; Kim, D. K.; Sugiyama, Y.; Kanai, Y.; Endou, H.: Molecular cloning and characterization of multispecific organic anion transporter 4 expressed in the placenta. *J. Biol. Chem.* 275: 4507–4512, 2000.
- [36109] 9661.Vreugde, S.; Erven, A.; Kros, C. J.; Marcotti, W.; Fuchs, H.; Kurima, K.; Wilcox, E. R.; Friedman, T. B.; Griffith, A. J.; Balling, R.; de Angelis, M. H.; Avraham, K. B.; Steel, K. P.: Beethoven, a mouse model for dominant, progressive hearing loss DFNA36. *Nature Genet.* 30: 257–258, 2002.
- [36110] 9662.Grunder, S.; Geissler, H.-S.; Bassler, E.-L.; Ruppersberg, J. P.: A new member of acid-sensing ion channels from pituitary gland. *Neuroreport* 11:1607–1611, 2000.
- [36111] 9663.Tatarelli, C.; Linnenbach, A.; Mimori, K.; Croce, C. M.: Characterization of the human TESTIN gene localized in the FRA7G region at 7q31.2. *Genomics* 68:1–12, 2000.
- [36112] 9664.Tobias, E. S.; Hurlstone, A. F. L.; MacKenzie, E.; Mc-

Farlane, R.; Black, D. M.: The TES gene at 7q31.1 is methylated in tumours and encodes a novel growth-suppressing LIM domain protein. *Oncogene* 20:2844–2853, 2001.

[36113] 9665. Mizuno, Y.; Thompson, T. G.; Guyon, J. R.; Lidov, H. G. W.; Brosius, M.; Imamura, M.; Ozawa, E.; Watkins, S. C.; Kunkel, L. M.: Desmuslin, an intermediate filament protein that interacts with alpha-dystrobrevin and desmin. *Proc. Nat. Acad. Sci.* 98: 6156–6161, 2001.

[36114] 9666. Ferdinandusse, S.; Mulders, J.; IJlst, L.; Denis, S.; Dacremont, G.; Waterham, H. R.; Wanders, R. J. A.: Molecular cloning and expression of human carnitine octanoyl-transferase: evidence for its role in the peroxisomal beta-oxidation of branched-chain fatty acids. *Biochem. Biophys. Res. Commun.* 263: 213–218, 1999.

[36115] 9667. Nestel, F. P.; Colwill, K.; Harper, S.; Pawson, T.; Anderson, S. K.: RS cyclophilins: identification of an NK-TR(1)-related cyclophilin. *Gene* 180:151–155, 1996.

[36116] 9668. Gaynor, E. C.; Mondesert, G.; Grimme, S. J.; Reed, S. I.; Orlean, P.; Emr, S. D.: MCD4 encodes a conserved endoplasmic reticulum membrane protein essential for glycosylphosphatidylinositol anchor synthesis in yeast. *Molec. Biol. Cell* 10: 627–648, 1999.

[36117] 9669. Hong, Y.; Maeda, Y.; Watanabe, R.; Ohishi, K.;

Mishkind, M.; Riezman, H.; Kinoshita, T.: Pig-n, a mammalian homologue of yeast Mcd4p, is involved in transferring phosphoethanolamine to the first mannose of the glycosylphosphatidylinositol. *J. Biol. Chem.* 274: 35099–35106, 1999.

[36118] 9670. Gao, J.; Yu, L.; Zhang, P.; Jiang, J.; Chen, J.; Peng, J.; Wei, Y.; Zhao, S.: Cloning and characterization of human and mouse mitochondria elongation factor G, GFM and Gfm, and mapping of GFM to human chromosome 3q25.1–q26.2. *Genomics* 74: 109–114, 2001.

[36119] 9671. Clark, K. L.; Zeng, Z.; Langford, A. L.; Bowen, S. M.; Todd, S. C.: PGRL is a major CD81-associated protein on lymphocytes and distinguishes a new family of cell surface proteins. *J. Immun.* 167: 5115–5121, 2001.

[36120] 9672. Stipp, C. S.; Kolesnikova, T. V.; Hemler, M. E.: EWI-2 is a major CD9 and CD81 partner and member of a novel Ig protein subfamily. *J. Biol. Chem.* 276: 40545–40554, 2001.

[36121] 9673. Miura, K.; Jacques, K. M.; Stauffer, S.; Kubosaki, A.; Zhu, K.; Hirsch, D. S.; Resau, J.; Zheng, Y.; Randazzo, P. A.: ARAP1: a point of convergence for Arf and Rho signaling. *Molec. Cell* 9: 109–119, 2002.

[36122] 9674. Dumoutier, L.; Lejeune, D.; Colau, D.; Renauld, J. C.:

Cloning and characterization of IL-22 binding protein, a natural antagonist of IL-10-related T cell-derived inducible factor/IL 22. *J. Immun.* 166:7090–7095, 2001.

[36123] 9675. Kotenko, S. V.; Izotova, L. S.; Mirochnitchenko, O. V.; Esterova, E.; Dickensheets, H.; Donnelly, R. P.; Pestka, S.: Identification, cloning, and characterization of a novel soluble receptor that binds IL-22 and neutralizes its activity. *J. Immun.* 166: 7096–7103, 2001.

[36124] 9676. Xu, W.; Presnell, S. R.; Parrish–Novak, J.; Kindsvogel, W.; Jaspers, S.; Chen, Z.; Dillon, S. R.; Gao, Z.; Gilbert, T.; Madden, K.; Schlutsmeyer, S.; Yao, L.; and 11 others: A soluble class II cytokine receptor, IL-22RA2, is a naturally occurring IL-22 antagonist. *Proc. Nat. Acad. Sci.* 98: 9511–9516, 2001.

[36125] 9677. Hicar, M. D.; Liu, Y.; Allen, C. E.; Wu, L.–C.: Structure of the human zinc finger protein HIVEP3: molecular cloning, expression, exon–intron structure, and comparison with paralogous genes HIVEP1 and HIVEP2. *Genomics* 71:89–100, 2001.

[36126] 9678. Bates, E. E. M.; Fridman, W. H.; Mueller, C. G. F.: The ADAMDEC1 (decysin) gene structure: evolution by duplication in a metalloprotease gene cluster on chromosome 8p12. *Immunogenetics* 54: 96–105, 2002.

- [36127] 9679. Bridges, L. C.; Tani, P. H.; Hanson, K. R.; Roberts, C. M.; Judkins, M. B.; Bowditch, R. D.: The lymphocyte metalloprotease MDC-L (ADAM28) is a ligand for the integrin α -4/ β -1. *J. Biol. Chem.* 277:3784–3792, 2002.
- [36128] 9680. Jury, J. A.; Perry, A. C. F.; Hall, L.: Identification, sequence analysis and expression of transcripts encoding a putative metalloproteinase, eMDC II, in human and macaque epididymis. *Molec. Hum. Reprod.* 5:1127–1134, 1999.
- [36129] 9681. Roberts, C. M.; Tani, P. H.; Bridges, L. C.; Laszik, Z.; Bowditch, R. D.: MDC-L, a novel metalloprotease disintegrin cysteine-rich protein family member expressed by human lymphocytes. *J. Biol. Chem.* 274:29251–29259, 1999.
- [36130] 9682. Koontz, J. I.; Soreng, A. L.; Nucci, M.; Kuo, F. C.; Pauwels, P.; van den Berghe, H.; Cin, P. D.; Fletcher, J. A.; Sklar, J.: Frequent fusion of the JAZF1 and JJAZ1 genes in endometrial stromal tumors. *Proc. Nat. Acad. Sci.* 98: 6348–6353, 2001.
- [36131] 9683. Fitzgerald, K. A.; Palsson-McDermott, E. M.; Bowie, A. G.; Jefferies, C. A.; Mansell, A. S.; Brady, G.; Brint, E.; Dunne, A.; Gray, P.; Harte, M. T.; McMurray, D.; Smith, D. E.; Sims, J. E.; Bird, T. A.; O'Neill, L. A. J.: Mal

(MyD88–adapter–like) is required for Toll–likereceptor–4 signal transduction. *Nature* 413: 78–83, 2001.

[36132] 9684.Horng, T.; Barton, G. M.; Medzhitov, R.: TIRAP: an adapter molecule in the Toll signaling pathway. *Nature Immun.* 2: 835–841, 2001.

[36133] 9685.Mirzayans, F.; Pearce, W. G.; MacDonald, I. M.; Walter, M. A.: Mutation of the PAX6 gene in patients with autosomal dominant keratitis. *Am.J. Hum. Genet.* 57: 539–548, 1995.

[36134] 9686.Dose, A. C.; Burnside, B.: Cloning and chromosomal localization of a human class III myosin. *Genomics* 67: 333–342, 2000.

[36135] 9687.Walsh, T.; Walsh, V.; Vreugde, S.; Hertzano, R.; Shahin, H.; Haika, S.; Lee, M. K.; Kanaan, M.; King, M.–C.; Avraham, K. B.: From flies' eyes to our ears: mutations in a human class III myosin cause progressive nonsyndromic hearing loss DFNB30. *Proc. Nat. Acad. Sci.* 99: 7518–7523, 2002.

[36136] 9688.Baron, M.: Genetics of schizophrenia and the new millennium: progress and pitfalls. *Am. J. Hum. Genet.* 68: 299–312, 2001.

[36137] 9689.Chakravarti, A.: A compelling genetic hypothesis for a complex disease: PRODH2/DGCR6 variation leads to

schizophrenia susceptibility. *Proc.Nat. Acad. Sci.* 99: 4755–4756, 2002.

- [36138] 9690.Gogos, J. A.; Santha, M.; Takacs, Z.; Beck, K. D.; Luine, V.; Lucas, L. R.; Nadler, J. V.; Karayiorgou, M.: The gene encoding prolinedehydrogenase modulates sensorimotor gating in mice. *Nature Genet.* 21:434–439, 1999.
- [36139] 9691.Liu, H.; Heath, S. C.; Sobin, C.; Roos, J. L.; Galke, B. L.; Blundell, M. L.; Lenane, M.; Robertson, B.; Wijsman, E. M.; Rapoport, J. L.; Gogos, J. A.; Karayiorgou, M.: Genetic variation at the 22q11 *PRODH2/DGCR6* locus presents an unusual pattern and increases susceptibility to schizophrenia. *Proc. Nat. Acad. Sci.* 99: 3717–3722, 2002.
- [36140] 9692.Scott, A.: Personal Communication. Baltimore, Md. 4/2002.
- [36141] 9693.Plager, D. A.; Loegering, D. A.; Weiler, D. A.; Checkel, J. L.; Wagner, J. M.; Clarke, N. J.; Naylor, S.; Page, S. M.; Thomas, L.L.; Akerblom, I.; Cocks, B.; Stuart, S.; Gleich, G. J.: A novel and highly divergent homolog of human eosinophil granule major basic protein. *J.Biol. Chem.* 274: 14464–14473, 1999.
- [36142] 9694.Leo, C. P.; Hsu, S. Y.; McGee, E. A.; Salanova, M.; Hsueh, A. J.W.: DEFT, a novel death effector domain–

containing molecule predominantly expressed in testicular germ cells. *Endocrinology* 139: 4839–4848, 1998.

[36143] 9695. Stegh, A. H.; Schickling, O.; Ehret, A.; Scaffidi, C.; Peterhansel, C.; Hofmann, T. G.; Grummt, I.; Krammer, P. H.; Peter, M. E.: DEDD, a novel death effector domain-containing protein, targeted to the nucleolus. *EMBO J.* 17: 5974–5986, 1998.

[36144] 9696. Charest, A.; Lane, K.; McMahon, K.; Housman, D. E.: Association of a novel PDZ domain-containing peripheral Golgi protein with the Q-SNARE (Q-soluble N-ethylmaleimide-sensitive fusion protein (NSF) attachment protein receptor) protein syntaxin 6. *J. Biol. Chem.* 276: 29456–29465, 2001.

[36145] 9697. Cheng, J.; Moyer, B. D.; Milewski, M.; Loffing, J.; Ikeda, M.; Mickle, J. E.; Cutting, G. R.; Li, M.; Stanton, B. A.; Guggino, W. B.: A Golgi-associated PDZ domain protein modulates cystic fibrosis transmembrane regulator plasma membrane expression. *J. Biol. Chem.* 277: 3520–3529, 2002.

[36146] 9698. Neudauer, C. L.; Joberty, G.; Macara, I. G.: PIST: a novel PDZ/coiled-coil domain binding partner for the Rho-family GTPase TC10. *Biochem. Biophys. Res. Commun.* 280: 541–547, 2001.

- [36147] 9699.Yao, R.; Maeda, T.; Takada, S.; Noda, T.: Identification of aPDZ domain containing Golgi protein, GOPC, as an interaction partner of frizzled. *Biochem. Biophys. Res. Commun.* 286: 771–778, 2001.
- [36148] 9700.Kelter, A.–R.; Herchenbach, J.; Wirth, B.: The transcription factor–like nuclear regulator (TFNR) contains a novel 55–amino–acid motif repeated nine times and maps closely to SMN1. *Genomics* 70: 315–326, 2000.
- [36149] 9701.Schramm, L.; Pendergrast, P. S.; Sun, Y.; Hernandez, N.: Different human TFIIIB activities direct RNA polymerase III transcription from TATA–containing and TATA–less promoters. *Genes Dev.* 14: 2650–2663, 2000.
- [36150] 9702.Cabart, P.; Murphy, S.: BRFU, a TFIIIB–like factor, is directly recruited to the TATA–box of polymerase III small nuclear RNA gene promoters through its interaction with TATA–binding protein. *J. Biol.Chem.* 276: 43056–43064, 2001.
- [36151] 9703.Schramm, L.; Pendergrast, P. S.; Sun, Y.; Hernandez, N.: Different human TFIIIB activities direct RNA polymerase III transcription from TATA–containing and TATA–less promoters. *Genes Dev.* 14: 2650–2663, 2000.
- [36152] 9704.Kobayashi, K.; Kuroda, S.; Fukata, M.; Nakamura, T.; Nagase, T.; Nomura, N.; Matsuura, Y.; Yoshida–Kubomura,

N.; Iwamatsu, A.; Kaibuchi, K.: p140Sra-1 (specifically Rac1-associated protein) is a novel specific target for Rac1 small GTPase. *J. Biol. Chem.* 273: 291–295, 1998.

[36153] 9705. Schenck, A.; Bardoni, B.; Moro, A.; Bagni, C.; Mandel, J. L.: A highly conserved protein family interacting with the fragile X mental retardation protein (FMRP) and displaying selective interactions with FMRP-related proteins FXR1P and FXR2P. *Proc. Nat. Acad. Sci.* 98: 8844–8849, 2001.

[36154] 9706. Lopez-Rios, J.; Gallardo, M. E.; Rodriguez de Cordoba, S.; Bovolenta, P.: Six9 (Optx2), a new member of the Six gene family of transcription factors, is expressed at early stages of vertebrate ocular and pituitary development. *Mech. Dev.* 83: 155–159, 1999.

[36155] 9707. Oukka, M.; Kim, S. T.; Lugo, G.; Sun, J.; Wu, L.-C.; Glimcher, L. H.: A mammalian homolog of *Drosophila* schnurri, KRC, regulates TNF receptor-driven responses and interacts with TRAF2. *Molec. Cell* 9: 121–131, 2002.

[36156] 9708. Walder, R. Y.; Landau, D.; Meyer, P.; Shalev, H.; Tsolia, M.; Borochowitz, Z.; Boettger, M. B.; Beck, G. E.; Englehardt, R. K.; Carmi, R.; Sheffield, V. C.: Mutation of TRPM6 causes familial hypomagnesemia with secondary hypocalcemia. *Nature Genet.* 31: 171–174, 2002.

[36157] 9709. Liang, J. C.; Chang, K. S.; Schroeder, W.; Siciliano,

M.; Trujillo, J.; Stass, S.: The human myeloperoxidase gene locates on chromosome 17q22–24 and is translocated in acute promyelocytic leukemia. (Abstract) *Am. J. Hum. Genet.* 41: A226, 1987.

[36158] 9710. Liang, J. C.; Chang, K. S.; Schroeder, W. T.; Freireich, E. J.; Stass, S. A.; Trujillo, J. M.: The myeloperoxidase gene is translocated from chromosome 17 to 15 in a patient with acute promyelocytic leukemia. *Cancer Genet. Cytogenet.* 30: 103–107, 1988.

[36159] 9711. Miki, T.; Weil, S. C.; Rosner, G. L.; Reid, M. S.; Kidd, K. K.: An MPO cDNA clone identifies an RFLP with *Pst*I. *Nucleic Acids Res.* 16:1649, 1988.

[36160] 9712. Morishita, K.; Kubota, N.; Asano, S.; Kaziro, Y.; Nagata, S.: Molecular cloning and characterization of cDNA for human myeloperoxidase. *J. Biol. Chem.* 262: 3844–3851, 1987.

[36161] 9713. Murao, S.-I.; Stevens, F. J.; Ito, A.; Huberman, E.: Myeloperoxidase: a myeloid cell nuclear antigen with DNA-binding properties. *Proc. Nat. Acad. Sci.* 85: 1232–1236, 1988.

[36162] 9714. Nauseef, W.; Cogley, M.; McCormick, S.: Effect of the R569W missense mutation on the biosynthesis of myeloperoxidase. *J. Biol. Chem.* 271:9546–9549, 1996.

- [36163] 9715.Nauseef, W. M.; Olsson, I.; Arnljots, K.: Biosynthesis and processing of myeloperoxidase--a marker for myeloid cell differentiation. *Europ.J. Haemat.* 40: 97-110, 1988.
- [36164] 9716.Reynolds, W. F.; Hiltunen, M.; Pirskanen, M.; Mannermaa, A.; Helisalmi, S.; Lehtovirta, M.; Alafuzoff, I.; Soininen, H.: MPO and APOE epsilon-4 polymorphisms interact to increase risk for AD in Finnish males. *Neurology* 55:1284-1290, 2000.
- [36165] 9717.Robinson, T. J.; Morris, D. J.; Ledbetter, D. H.: Chromosomal assignment and regional localization of myeloperoxidase in the mouse. *Cytogenet.Cell Genet.* 53: 83-86, 1990.
- [36166] 9718.Romano, M.; Dri, P.; Dadalt, L.; Patriarca, P.; Baralle, F. E.: Biochemical and molecular characterization of hereditary myeloproliferative deficiency. *Blood* 90: 4126-4134, 1997.
- [36167] 9719.van Tuinen, P.; Johnson, K. R.; Ledbetter, S. A.; Nussbaum, R.L.; Rovera, G.; Ledbetter, D. H.: Localization of myeloperoxidase to the long arm of human chromosome 17: relationship to the 15;17 translocation of acute promyelocytic leukemia. *Oncogene* 1: 319-322, 1987.
- [36168] 9720.Weil, S. C.; Rosner, G. L.; Reid, M. S.; Chisholm, R. L.; Farber, N. M.; Spitznagel, J. K.; Swanson, M. S.: cDNA

cloning of human myeloperoxidase:decrease in myeloperoxidase mRNA upon induction of HL-60 cells. *Proc.Nat. Acad. Sci.* 84: 2057–2061, 1987.

[36169] 9721.Weil, S. C.; Rosner, G. L.; Reid, M. S.; Chisholm, R. L.; Lemons,R. S.; Swanson, M. S.; Carrino, J. J.; Diaz, M. O.; Le Beau, M. M.: Translocation and rearrangement of myeloperoxidase gene in acute promyelocytic leukemia. *Science* 240: 790–792, 1988.

[36170] 9722.Yamada, M.; Hur, S.-J.; Hashinaka, K.; Tsuneoka, K.; Saeki, T.;Nishio, C.; Sakiyama, F.; Tsunasawa, S.: Isolation and characterization of a cDNA coding for human myeloperoxidase. *Arch. Biochem. Biophys.* 255:147–155, 1987.

[36171] 9723.Zaki, S. R.; Austin, G. E.; Chan, W. C.; Conaty, A. L.; Trusler,S.; Trappier, S.; Lindsey, R. B.; Swan, D. C.: Chromosomal localization of the human myeloperoxidase gene by in situ hybridization using oligonucleotide probes. *Genes Chromosomes Cancer* 2: 266–270, 1990.

[36172] 9724.Saiardi, A.; Erdjument-Bromage, H.; Snowman, A. M.; Tempst, P.;Snyder, S. H.: Synthesis of diphosphoinositol pentakisphosphate by a newly identified family of higher inositol polyphosphate kinases. *Curr.Biol.* 9: 1323–1326, 1999.

- [36173] 9725.Saiardi, A.; Nagata, E.; Luo, H. R.; Snowman, A. M.; Snyder, S.H.: Identification and characterization of a novel inositol hexakisphosphatekinase. *J. Biol. Chem.* 276: 39179–39185, 2001.
- [36174] 9726.Morrison, B. H.; Bauer, J. A.; Hu, J.; Grane, R. W.; Ozdemir, A.M.; Chawla–Sarkar, M.; Gong, B.; Almasan, A.; Kalvakolanu, D. V.;Lindner, D. J.: Inositol hexakisphosphate kinase 2 sensitizes ovariancarcinoma cells to multiple cancer therapeutics. *Oncogene* 21: 1882–1889,2002.
- [36175] 9727.Gasdaska, P. Y.; Berggren, M. M.; Berry, M. J.; Powis, G.: Cloning,sequencing, and functional expression of a novel human thioredoxinreductase. *FEBS Lett.* 442: 105–111, 1999.
- [36176] 9728.Kawai, H.; Ota, T.; Suzuki, F.; Tatsuka, M.: Molecular cloningof mouse thioredoxin reductases. *Gene* 242: 321–330, 2000.
- [36177] 9729.Miranda–Vizuite, A.; Damdimopoulos, A. E.; Spyrou, G.: cDNA cloning,expression and chromosomal localization of the mouse mitochondrialthioredoxin reductase gene. *Biochim. Biophys. Acta* 1447: 113–118,1999.
- [36178] 9730.Miranda–Vizuite, A.; Damdimopoulos, A. E.; Pedrajas, J. R.; Gustafsson,J.–A.; Spyrou, G.: Human mitochondrial thioredoxin reductase: cDNAcloning, expression, and

genomic organization. *Europ. J. Biochem.* 261:405–412, 1999.

[36179] 9731. Nishida, K.; Yoshida, Y.; Itoh, M.; Fukada, T.; Ohtani, T.; Shirogane, T.; Atsumi, T.; Takahashi–Tezuka, M.; Ishihara, K.; Hibi, M.; Hirano, T.: Gab–family adapter proteins act downstream of cytokine and growthfactor receptors and T– and B–cell antigen receptors. *Blood* 93:1809–1816, 1999.

[36180] 9732. Zhao, C.; Yu, D.–H.; Shen, R.; Feng, G.–S.: Gab2, a new pleckstrin homology domain–containing adapter protein, acts to uncouple signaling from ERK kinase to Elk–1. *J. Biol. Chem.* 274: 19649–19654, 1999.

[36181] 9733. Brakenhoff, R. H.; Gerretsen, M.; Knippels, E. M. C.; van Dijk, M.; van Essen, H.; Weghuis, D. O.; Sinke, R. J.; Snow, G. B.; van Dongen, G. A. M. S.: The human E48 antigen, highly homologous to the murine Ly–6 antigen ThB, is a GPI–anchored molecule apparently involved in keratinocyte cell–cell adhesion. *J. Cell Biol.* 129:1677–1689, 1995.

[36182] 9734. Brakenhoff, R. H.; van Dijk, M.; Rood–Knippels, E. M. C.; Snow, G. B.: A gain of novel tissue specificity in the human Ly–6 gene E48. *J. Immun.* 159: 4879–4886, 1997.

[36183] 9735. Loewen, S. K.; Ng, A. M. L.; Yao, S. Y. M.; Cass, C. E.;

Baldwin, S. A.; Young, J. D.: Identification of amino acid residues responsible for the pyrimidine and purine nucleoside specificities of human concentrative Na⁺ nucleoside cotransporters hCNT1 and hCNT2. *J. Biol. Chem.* 274:24475–24484, 1999.

[36184] 9736. Ritzel, M. W. L.; Yao, S. Y. M.; Huang, M.-Y.; Elliott, J. F.; Cass, C. E.; Young, J. D.: Molecular cloning and functional expression of cDNAs encoding a human Na(+)-nucleoside cotransporter (hCNT1). *Am. J. Physiol.* 272: C707–C714, 1997.

[36185] 9737. Ritzel, M. W. L.; Yao, S. Y. M.; Ng, A. M. L.; Mackey, J. R.; Cass, C. E.; Young, J. D.: Molecular cloning, functional expression and chromosomal localization of a cDNA encoding a human Na⁺/nucleoside cotransporter (hCNT2) selective for purine nucleosides and uridine. *Molec. Membrane Biol.* 15: 203–211, 1998.

[36186] 9738. Lescure, A.; Gautheret, D.; Carbon, P.; Krol, A.: Novel selenoproteins identified in silico and in vivo by using a conserved RNA structural motif. *J. Biol. Chem.* 274: 38147–38154, 1999.

[36187] 9739. Berghs, S.; Aggujaro, D.; Dirkx, R., Jr.; Maksimova, E.; Stabach, P.; Hermel, J.-M.; Zhang, J.-P.; Philbrick, W.; Slepnev, V.; Ort, T.; Slimena, M.: Beta-IV spectrin, a new

spectrin localized at axoninitial segments and nodes of Ranvier in the central and peripheralnervous system. J. Cell Biol. 151: 985–1001, 2000.

[36188] 9740.Bock, G. R.; Frank, M. P.; Steel, K. P.; Deol, M. S.: The quiveringmutant mouse: hereditary deafness of central origin. Acta Otolaryng. 96:371–377, 1983.

[36189] 9741.Deol, M. S.; Frank, M. P.; Steel, K. P.; Bock, G. R.: Geneticdeafness of central origin. Brain Res. 258: 177–179, 1983.

[36190] 9742.Parkinson, N. J.; Olsson, C. L.; Hallows, J. L.; McKee-Johnson,J.; Keogh, B. P.; Noben-Trauth, K.; Kujawa, S. G.; Tempel, B. L.:Mutant beta-spectrin 4 causes auditory and motor neuropathies in quiveringmice. Nature Genet. 29: 61–65, 2001.

[36191] 9743.Tse, W. T.; Tang, J.; Jin, O.; Korsgren, C.; John, K. M.; Kung,A. L.; Gwynn, B.; Peters, L. L.; Lux, S. E.: A new spectrin, beta-IV,has a major truncated isoform that associates with promyelocytic leukemia protein nuclear bodies and the nuclear matrix. J. Biol. Chem. 276:23974–23985, 2001.

[36192] 9744.Yoon, C. H.; Les, E. P.: Quivering, a new first chromosome mutationin mice. J. Hered. 48: 176–180, 1957.

[36193] 9745.Petek, E.; Windpassinger, C.; Egger, H.; Kroisel, P.

M.; Wagner,K.: Localization of the human anterior gradient-2 gene (AGR2) to chromosome band 7p21.3 by radiation hybrid mapping and fluorescence in situ hybridisation. *Cytogenet. Cell Genet.* 89: 141–142, 2000.

[36194] 9746. Thompson, D. A.; Weigel, R. J.: hAG-2, the human homologue of the *Xenopus laevis* cement gland gene XAG-2, is coexpressed with estrogen receptor in breast cancer cell lines. *Biochem. Biophys. Res. Commun.* 251:111–116, 1998.

[36195] 9747. Saitoh, T.; Katoh, M.: Molecular cloning and characterization of human WNT5B on chromosome 12p13.3 region. *Int. J. Oncol.* 19:347–351, 2001.

[36196] 9748. Miyazaki, K.; Matsuda, S.; Ichigotani, Y.; Takenouchi, Y.; Hayashi, K.; Fukuda, Y.; Nimura, Y.; Hamaguchi, M.: Isolation and characterization of a novel human gene (NESH) which encodes a putative signaling molecule similar to e3B1 protein. *Biochim. Biophys. Acta* 1493: 237–241, 2000.

[36197] 9749. Nosaka, K.; Onozuka, M.; Kakazu, N.; Hibi, S.; Nishimura, H.; Nishino, H.; Abe, T.: Isolation and characterization of a human thiamine pyrophosphokinase cDNA. *Biochem. Biophys. Acta* 1517: 293–297, 2001.

[36198] 9750. Nosaka, K.; Onozuka, M.; Nishino, H.; Nishimura, H.;

Kawasaki,Y.; Ueyama, H.: Molecular cloning and expression of a mouse thiaminpyrophosphokinase cDNA. J. Biol. Chem. 274: 34129–34133, 1999.

[36199] 9751.Zhao, R.; Gao, F.; Goldman, I. D.: Molecular cloning of humanthiamin pyrophosphokinase. Biochim. Biophys. Acta 1517: 320–322,2001.

[36200] 9752.Reboul, J.; Gardiner, K.; Monneron, D.; Uze, G.; Lutfalla, G.:Comparative genomic analysis of the interferon/interleukin–10 receptorgene cluster. Genome Res. 9: 242–250, 1999.

[36201] 9753.Chen, Q.; Ghilardi, N.; Wang, H.; Baker, T.; Xie, M.–H.; Gurney,A.; Grewal, I. S.; de Sauvage, F. J.: Development of Th1–type immuneresponses requires the type 1 cytokine receptor TCCR. Nature 407:916–920, 2000.

[36202] 9754.Sprecher, C. A.; Grant, F. J.; Baumgartner, J. W.; Presnell, S.R.; Schrader, S. K.; Yamagiwa, T.; Whitmore, T. E.; O'Hara, P. J.;Foster, D. F.: Cloning and characterization of a novel class I cytokinereceptor. Biochem. Biophys. Res. Comm. 246: 82–90, 1998.

[36203] 9755.Yoshida, H.; Hamano, S.; Senaldi, G.; Covey, T.; Faggioni, R.;Mu, S.; Xia, M.; Wakeham, A. C.; Nishina, H.; Potter, J.; Saris, C.J. M.; Mak, T. W.: WSX–1 is required for the initiation of Th1 responsesand resistance to L. major in–

fection. *Immunity* 15: 569–578, 2001.

[36204] 9756. Autieri, M. V.; Carbone, C. J.: 14-3-3-Gamma interacts with and is phosphorylated by multiple protein kinase C isoforms in PDGF-stimulated human vascular smooth muscle cells. *DNA Cell Biol.* 18: 555–564, 1999.

[36205] 9757. Yeh, W.-C.; de la Pompa, J. L.; McCurrach, M. E.; Shu, H.-B.; Elia, A. J.; Shahinian, A.; Ng, M.; Wakeham, A.; Khoo, W.; Mitchell, K.; El-Deiry, W. S.; Lowe, S. W.; Goeddel, D. V.; Mak, T. W.: FADD: essential for embryo development and signaling from some, but not all, inducers of apoptosis. *Science* 279: 1954–1958, 1998.

[36206] 9758. Zhang, J.; Cado, D.; Chen, A.; Kabra, N. H.; Winoto, A.: Fas-mediated apoptosis and activation-induced T-cell proliferation are defective in mice lacking FADD/Mort1. *Nature* 392: 296–300, 1998.

[36207] 9759. Torres, R.; Polymeropoulos, M. H.: Genomic organization and localization of the human CRMP-1 gene. *DNA Res.* 5: 393–395, 1998.

[36208] 9760. Koyama, K.; Sudo, K.; Nakamura, Y.: Isolation of 115 human chromosome 8-specific expressed-sequence tags by exon amplification. *Genomics* 26: 245–253, 1995.

[36209] 9761. Katahira, J.; Sugiyama, H.; Inoue, N.; Horiguchi, Y.; Matsuda, M.; Sugimoto, N.: *Clostridium perfringens* en-

terotoxin utilizes two structurally related membrane proteins as functional receptors *in vivo*. *J. Biol. Chem.* 272: 26652–26658, 1997.

- [36210] 9762. Furuse, M.; Fujita, K.; Hiragi, T.; Fujimoto, K.; Tsukita, S.: Claudin-1 and -2: novel integral membrane proteins localizing at tight junctions with no sequence similarity to occludin. *J. Cell Biol.* 141: 1539–1550, 1998.
- [36211] 9763. Peacock, R. E.; Keen, T. J.; Inglehearn, C. F.: Analysis of a human gene homologous to rat ventral prostate.1 protein. *Genomics* 46:443–449, 1997.
- [36212] 9764. Black, J. L., III; Lennon, V. A.: Identification and cloning of putative human neuronal voltage-gated calcium channel γ -2 and γ -3 subunits: neurologic implications. *Mayo Clin. Proc.* 74:357–361, 1999.
- [36213] 9765. Chen, L.; Chetkovich, D. M.; Petralla, R. S.; Sweeney, N. T.; Kawasaki, Y.; Wenthold, R. J.; Brecht, D. S.; Nicoli, R. A.: Stargazin regulates synaptic targeting of AMPA receptors by two distinct mechanisms. *Nature* 408:936–943, 2000.
- [36214] 9766. Letts, V. A.; Felix, R.; Biddlecome, G. H.; Arikath, J.; Mahaffey, C. L.; Valenzuela, A.; Bartlett, F. S., II; Mori, Y.; Campbell, K. P.; Frankel, W. N.: The mouse stargazer gene encodes a neuronal Ca^{2+} -channel γ subunit. *Na-*

ture Genet. 19: 340–347, 1998.

- [36215] 9767. Biffo, S.; Sanvito, F.; Costa, S.; Preve, L.; Pignatelli, R.; Spinardi, L.; Marchisio, P. C.: Isolation of a novel beta-4 integrin-binding protein (p27BBP) highly expressed in epithelial cells. J. Biol. Chem. 272:30314–30321, 1997.
- [36216] 9768. Sanvito, F.; Arrigo, G.; Zuffardi, O.; Agnelli, M.; Marchisio, P. C.; Biffo, S.: Localization of p27 beta-4 binding protein gene (ITGB4BP) to human chromosome region 20q11.2. Genomics 52: 111–112, 1998.
- [36217] 9769. Si, K.; Chaudhuri, J.; Chevesich, J.; Maitra, U.: Molecular cloning and functional expression of a human cDNA encoding translation initiation factor 6. Proc. Nat. Acad. Sci. 94: 14285–14290, 1997.
- [36218] 9770. Liu, Z.; Shen, J.; Carbrey, J. M.; Mukhopadhyay, R.; Agre, P.; Rosen, B. P.: Arsenite transport by mammalian aquaglyceroporins AQP7 and AQP9. Proc. Nat. Acad. Sci. 99: 6053–6058, 2002.
- [36219] 9771. Nuber, U.; Schwarz, S.; Kaiser, P.; Schneider, R.; Scheffner, M.: Cloning of human ubiquitin-conjugating enzymes UbCH6 and UbCH7 (E2-F1) and characterization of their interaction with E6-AP and RSP5. J. Biol. Chem. 271: 2795–2800, 1996.
- [36220] 9772. Fuentes, J. J.; Pritchard, M. A.; Estivill, X.: Genomic

organization, alternative splicing, and expression patterns of the DSCR1 (Down syndrome candidate region 1) gene. Genomics 44: 358–361, 1997.

- [36221] 9773. Kingsbury, T. J.; Cunningham, K. W.: A conserved family of calcineurin regulators. Genes Dev. 14: 1595–1604, 2000.
- [36222] 9774. Rothermel, B.; Vega, R. B.; Yang, J.; Wu, H.; Bassel-Duby, R.; Williams, R. S.: A protein encoded within the Down syndrome critical region is enriched in striated muscles and inhibits calcineurin signaling. J. Biol. Chem. 275: 8719–8725, 2000.
- [36223] 9775. Pata, I.; Tensing, K.; Metspalu, A.: A human cDNA encoding the homologue of NADH:ubiquinone oxidoreductase subunit B13. Biochim. Biophys. Acta 1350: 115–118, 1997.
- [36224] 9776. Russell, M. W.; du Manoir, S.; Collins, F. S.; Brody, L. C.: Cloning of the human NADH:ubiquinone oxidoreductase subunit B13: localization to chromosome 7q32 and identification of a pseudogene on 11p15. Mammalian Genome 8: 60–61, 1997.
- [36225] 9777. Perez Jurado, L. A.; Wang, Y.-K.; Peoples, R.; Coloma, A.; Cruces, J.; Francke, U.: A duplicated gene in the breakpoint regions of the 7q11.23 Williams–Beuren

syndrome deletion encodes the initiator binding protein
TFII-I and BAP-135, a phosphorylation target of BTK.
Hum.Molec. Genet. 7: 325-334, 1998.

[36226] 9778. Yang, W.; Desiderio, S.: BAP-135, a target for Bruton's tyrosine kinase in response to B cell receptor engagement. Proc. Nat. Acad. Sci. 94: 604-609, 1997.

[36227] 9779. Lee, J. W.; Choi, H.-S.; Gyuris, J.; Brent, R.; Moore, D. D.: Two classes of proteins dependent on either the presence or absence of thyroid hormone for interaction with the thyroid hormone receptor. Molec. Endocr. 9: 243-254, 1995.

[36228] 9780. Burnatowska-Hledin, M. A.; Spielman, W. S.; Smith, W. L.; Shi, P.; Meyer, J. M.; Dewitt, D. L.: Expression cloning of an AVP-activated, calcium-mobilizing receptor from rabbit kidney medulla. Am. J. Physiol. 268: F1198-F1210, 1995.

[36229] 9781. Byrd, P. J.; Stankovic, T.; McConville, C. M.; Smith, A. D.; Cooper, P. R.; Taylor, A. M. R.: Identification and analysis of expression of human VACM-1, a cullin gene family member located on chromosome 11q22-23. Genome Res. 7: 71-75, 1997.

[36230] 9782. Venturini, L.; You, J.; Stadler, M.; Galien, R.; Lallemand, V.; Koken, M. H. M.; Mattei, M. G.; Ganser, A.;

Chambon, P.; Losson, R.; de The, H.: TIF1-gamma, a novel member of the transcriptional intermediary factor 1 family. *Oncogene* 18: 1209–1217, 1999.

[36231] 9783. Lesage, F.; Guillemare, E.; Fink, M.; Duprat, F.; Lazdunski, M.; Romey, G.; Barhanin, J.: TWIK-1, a ubiquitous human weakly inward rectifying K⁺ channel with a novel structure. *EMBO J.* 15: 1004–1011, 1996.

[36232] 9784. Lesage, F.; Mattei, M.-G.; Fink, M.; Barhanin, J.; Lazdunski, M.: Assignment of the human weak inward rectifier K⁺ channel TWIK-1 gene to chromosome 1q42–q43. *Genomics* 34: 153–155, 1996.

[36233] 9785. Holzmann, K.; Poltl, A.; Sauermann, G.: A novel spliced transcript of human CLAPS2 encoding a protein alternative to clathrin adaptor protein AP17. *Gene* 220: 39–44, 1998.

[36234] 9786. Winterpacht, A.; Ende, S.; Enklaar, T.; Fuhry, M.; Zabel, B.: Human CLAPS2 encoding AP17, a small chain of the clathrin-associated protein complex: cDNA cloning and chromosomal assignment to 19q13.2–q13.3. *Cytogenet. Cell Genet.* 75: 132–135, 1996.

[36235] 9787. Rodriguez, A. M.; Rodin, D.; Nomura, H.; Morton, C. C.; Weremowicz, S.; Schneider, M. C.: Identification, localization, and expression of two novel human genes similar

to deoxyribonuclease I. *Genomics* 42:507–513, 1997.

[36236] 9788.Zeng, Z.; Parmelee, D.; Hyaw, H.; Coleman, T. A.; Su, K.; Zhang, J.; Gentz, R.; Ruben, S.; Rosen, C.; Li, Y.: Cloning and characterization of a novel human DNase. *Biochem. Biophys. Res. Commun.* 231: 499–504, 1997.

[36237] 9789.Alderson, M. R.; Smith, C. A.; Tough, T. W.; Davis-Smith, T.; Armitage, R. J.; Falk, B.; Roux, E.; Baker, E.; Sutherland, G. R.; Din, W. S.; Goodwin, R. G.: Molecular and biological characterization of human 4–1BB and its ligand. *Europ. J. Immun.* 24: 2219–2227, 1994.

[36238] 9790.Kwon, B. S.; Weissman, S. M.: cDNA sequences of two inducible T-cell genes. *Proc. Nat. Acad. Sci.* 86: 1963–1967, 1989.

[36239] 9791.Loo, D. T.; Chalupny, N. J.; Bajorath, J.; Shuford, W. W.; Mittler, R. S.; Aruffo, A.: Analysis of 4–1BBL and laminin binding to murine 4–1BB, a member of the tumor necrosis factor receptor superfamily, and comparison with human 4–1BB. *J. Biol. Chem.* 272: 6448–6456, 1997.

[36240] 9792.Schwarz, H.; Arden, K.; Lotz, M.: CD137, a member of the tumor necrosis factor receptor family, is located on chromosome 1p36, in a cluster of related genes, and colocalizes with several malignancies. *Biochem. Biophys. Res. Commun.* 235: 699–703, 1997.

- [36241] 9793.Schwarz, H.; Blanco, F. J.; von Kempis, J.; Valbracht, J.; Lotz,M.: ILA, a member of the human nerve growth factor/tumor necrosisfactor receptor family, regulates T-lymphocyte proliferation and survival. *Blood* 87:2839–2845, 1996.
- [36242] 9794.Schwarz, H.; Tuckwell, J.; Lotz, M.: A receptor induced by lymphocyteactivation (ILA): a new member of the human nerve-growth-factor/tumor-necrosis-factorreceptor family. *Gene* 134: 295–298, 1993.
- [36243] 9795.Deere, M.; Johnson, J.; Garza, S.; Harrison, W. R.; Yoon, S.-J.;Elder, F. F. B.; Kucherlapati, R.; Hook, M.; Hecht, J. T.: Characterizationof human DSPG3, a small dermatan sulfate proteoglycan. *Genomics* 38:399–404, 1996.
- [36244] 9796.Shinomura, T.; Kimata, K.; Oike, Y.; Noro, A.; Hirose, N.; Tanabe,K.; Suzuki, S.: The occurrence of three different proteoglycan speciesin chick embryo cartilage: Isolation and characterization of a secondproteoglycan (PG-Lb) and its precursor form. *J. Biol. Chem.* 258:9314–9322, 1983.
- [36245] 9797.Feuchter-Murthy, A. E.; Freeman, J. D.; Mager, D. L.: Splicingof a human endogenous retrovirus to a novel

phospholipase A2 related gene. *Nucleic Acids Res.* 21: 135–143, 1993.

[36246] 9798. Kowalski, P. E.; Freeman, J. D.; Mager, D. L.: Inter-genic splicing between a HERV-H endogenous retrovirus and two adjacent human genes. *Genomics* 57:371–379, 1999.

[36247] 9799. Baylin, S. B.; Herman, J. G.; Graff, J. R.; Vertino, P. M.; Issa, J. P.: Alterations in DNA methylation: a fundamental aspect of neoplasia. *Adv. Cancer Res.* 72: 141–196, 1998.

[36248] 9800. Lei, H.; Oh, S. P.; Okano, M.; Juttermann, R.; Goss, K. A.; Jaenisch, R.; Li, E.: De novo DNA cytosine methyltransferase activities in mouse embryonic stem cells. *Development* 122: 3195–3205, 1996.

[36249] 9801. Okano, M.; Xie, S.; Li, E.: Cloning and characterization of a family of novel mammalian DNA (cytosine-5) methyltransferases. (Letter) *Nature Genet.* 19: 219–220, 1998.

[36250] 9802. Robertson, K. D.; Uzvolgyi, E.; Liang, G.; Talmadge, C.; Sumegi, J.; Gonzales, F. A.; Jones, P. A.: The human DNA methyltransferases (DNMTs) 1, 3a and 3b: coordinate mRNA expression in normal tissues and overexpression in tumors. *Nucleic Acids Res.* 27: 2291–2298, 1999.

[36251] 9803. Xie, S.; Wang, Z.; Okano, M.; Nogami, M.; Li, Y.; He,

W.-W.; Okumura,K.; Li, E.: Cloning, expression and chromosome locations of the humanDNMT3 gene family. *Gene* 236: 87–95, 1999.

[36252] 9804.Jiang, R.; Lan, Y.; Chapman, H. D.; Shawber, C.; Norton, C. R.;Serreze, D. V.; Weinmaster, G.; Gridley, T.: Defects in limb, craniofacial,and thymic development in Jagged2 mutant mice. *Genes Dev.* 12: 1046–1057,1998.

[36253] 9805.Lan, Y.; Jiang, R.; Shawber, C.; Weinmaster, G.; Gridley, T.:The Jagged2 gene maps to chromosome 12 and is a candidate for theIgl and sm mutations. *Mammalian Genome* 8: 875–876, 1997.

[36254] 9806.Lanford, P. J.; Lan, Y.; Jiang, R.; Lindsell, C.; Weinmaster, G.;Gridley, T.; Kelley, M. W.: Notch signalling pathway mediates haircell development in mammalian cochlea. *Nature Genet.* 21: 289–292,1999.

[36255] 9807.Luo, B.; Aster, J. C.; Hasserjian, R. P.; Kuo, F.; Sklar, J.:Isolation and functional analysis of a cDNA for human Jagged2, a geneencoding a ligand for the Notch1 receptor. *Molec. Cell. Biol.* 17:6057–6067, 1997.

[36256] 9808.Misaki, Y.; Pruijn, G. J. M.; van der Kemp, A. W. C. M.; van Venrooij,W. J.: The 56K autoantigen is identical to human annexin XI. *J.Biol. Chem.* 269: 4240–4246, 1994.

[36257] 9809.Morgan, R. O.; Bell, D. W.; Testa, J. R.; Fernandez, M.

P.: Genomiclocations of ANX11 and ANX13 and the evolutionary genetics of humanannexins. *Genomics* 48: 100–110, 1998.

[36258] 9810.Ono, T.; Kawabe, T.; Sonta, S.; Okamoto, T.: Assignment of MARK3alias KP78 to human chromosome band 14q32.3 by in situ hybridization. *Cytogenet.Cell Genet.* 79: 101–102, 1997.

[36259] 9811.Parsa, I.: Loss of Mr 78,000 marker in chemically induced transplantablecarcinomas and primary carcinoma of human pancreas. *Cancer Res.* 48:2265–2272, 1988.

[36260] 9812.Peng, C.–Y.; Graves, P. R.; Ogg, S.; Thoma, R. S.; Byrnes, M. J.,III; Wu, Z.; Stephenson, M. T.; Piwnica–Worms, H.: C–TAK1 proteinkinase phosphorylates human Cdc25C on serine 216 and promotes 14–3–3protein binding. *Cell Growth Differ.* 9: 197–208, 1998.

[36261] 9813.Brubaker, K.; Cowley, S. M.; Huang, K.; Loo, L.; Yochum, G. S.;Ayer, D. E.; Eisenman, R. N.; Radhakrishnan, I.: Solution structureof the interacting domains of the Mad–Sin3 complex: implications forrecruitment of a chromatin–modifying complex. *Cell* 103: 655–665,2000.

[36262] 9814.Jin, D.–Y.; Kozak, C. A.; Pangilinan, F.; Spencer, F.; Green, E.D.; Jeang, K.–T.: Mitotic checkpoint locus MAD1L1 maps to human chromosome7p22 and mouse

chromosome 5. *Genomics* 55: 363–364, 1999.

- [36263] 9815. Jin, D.-Y.; Spencer, F.; Jeang, K.-T.: Human T cell leukemia virus type 1 oncoprotein Tax targets the human mitotic checkpoint protein MAD1. *Cell* 93: 81–91, 1998.
- [36264] 9816. Tsukasaki, K.; Miller, C. W.; Greenspun, E.; Es-haghian, S.; Kawabata, H.; Fujimoto, T.; Tomonaga, M.; Sawyers, C.; Said, J. W.; Koeffler, H. P.: Mutations in the mitotic check point gene, MAD1L1, in human cancers. *Oncogene* 20: 3301–3305, 2001.
- [36265] 9817. Kimura, M.; Kotani, S.; Hattori, T.; Sumi, N.; Yoshioka, T.; Todokoro, T.; Okano, Y.: Cell cycle-dependent expression and spindle pole localization of a novel human protein kinase Aik, related to Aurora of *Drosophila* and yeast Ipl1. *J. Biol. Chem.* 272: 13766–13771, 1997.
- [36266] 9818. Goustin, A. S.: Personal Communication. Detroit, Mich. 8/17/1998.
- [36267] 9819. Harrington, L.; McPhail, T.; Mar, V.; Zhou, W.; Oulton, R.; Bass, M. B.; Arruda, I.; Robinson, M. O.: A mammalian telomerase-associated protein. *Science* 275: 973–976, 1997.
- [36268] 9820. Saito, T.; Matsuda, Y.; Suzuki, T.; Hayashi, A.; Yuan, X.; Saito, M.; Nakayama, J.; Hori, T.; Ishikawa, F.: Comparative gene mapping of the human and mouse TEP1 genes,

which encode one protein component of telomerases. *Genomics* 46: 46–50, 1997.

[36269] 9821. Coggins, K. G.; Latour, A.; Nguyen, M. S.; Audoly, L.; Coffman, T. M.; Koller, B. H.: Metabolism of PGE₂ by prostaglandin dehydrogenase is essential for remodeling the ductus arteriosus. (Letter) *Nature Med.* 8: 91–92, 2002.

[36270] 9822. Ensor, C. M.; Yang, J. Y.; Okita, R. T.; Tai, H. H.: Cloning and sequence analysis of the cDNA for human placental NAD(+)-dependent 15-hydroxyprostaglandin dehydrogenase. *J. Biol. Chem.* 265: 14888–14891, 1990.

[36271] 9823. Krook, M.; Marekov, L.; Jornvall, H.: Purification and structural characterization of placental NAD(+)-linked 15-hydroxyprostaglandin dehydrogenase: the primary structure reveals the enzyme to belong to the short-chain alcohol dehydrogenase family. *Biochemistry* 29: 738–743, 1990.

[36272] 9824. Chadwick, B. P.; Frischauf, A.-M.: The CD39-like gene family: identification of three new human members (CD39L2, CD39L3, and CD39L4), their murine homologues, and a member of the gene family from *Drosophila melanogaster*. *Genomics* 50: 357–367, 1998.

[36273] 9825. Enjyoji, K.; Sevigny, J.; Lin, Y.; Frenette, P. S.; Christie, P. D.; van Esch, J. S., II; Imai, M.; Edelberg, J. M.;

Rayburn, H.; Lech, M.; Beeler, D. L.; Csizmadia, E.; Wagner, D. D.; Robson, S. C.; Rosenberg, R. D.: Targeted disruption of cd39/ATP diphosphohydrolase results in disordered hemostasis and thromboregulation. *Nature Med.* 5: 1010–1017, 1999.

[36274] 9826. Granstein, R. D.: The skinny on CD39 in immunity and inflammation. *Nature Med.* 8: 336–338, 2002.

[36275] 9827. Gray, I. C.; Fallowfield, J.; Ford, S.; Nobile, C.; Volpi, E. V.; Spurr, N. K.: An integrated physical and genetic map spanning chromosome band 10q24. *Genomics* 43: 85–88, 1997.

[36276] 9828. Kaczmarek, E.; Koziak, K.; Seigny, J.; Siegel, J. B.; Anrather, J.; Beaudoin, A. R.; Bach, F. H.; Robson, S. C.: Identification and characterization of CD39/vascular ATP diphosphohydrolase. *J. Biol. Chem.* 271: 33116–33122, 1996.

[36277] 9829. Maliszewski, C. R.; Delespesse, G. L.; Schoenborn, M. A.; Armitage, R. J.; Fanslow, W. C.; Nakajima, T.; Baker, E.; Sutherland, G. R.; Poindexter, K.; Birks, C.; Alpert, A.; Friend, D.; Gimpel, S. D.; Gayle, R. B., III: The CD39 lymphoid cell activation antigen. Molecular cloning and structural characterization. *J. Immun.* 153: 3574–3583, 1994.

[36278] 9830. Mizumoto, N.; Kumamoto, T.; Robson, S. C.; Sevi-

gny, J.; Matsue, H.; Enjoji, K.; Takashima, A.: CD39 is the dominant Langerhans cell-associated ecto-NTPDase: modulatory roles in inflammation and immune responsiveness. *Nature Med.* 8: 358–365, 2002.

[36279] 9831. Wolff, K.; Winkelmann, R. K.: Ultrastructural localization of nucleoside triphosphatase in Langerhans cells. *J. Invest. Derm.* 48:50–54, 1967.

[36280] 9832. Kieffer, L. J.; Seng, T. W.; Li, W.; Osterman, D. G.; Handschumacher, R. E.; Bayney, R. M.: Cyclophilin-40, a protein with homology to the P59 component of the steroid receptor complex: cloning of the cDNA and further characterization. *J. Biol. Chem.* 268: 12303–12310, 1993.

[36281] 9833. Kieffer, L. J.; Thalhammer, T.; Handschumacher, R. E.: Isolation and characterization of a 40-kDa cyclophilin-related protein. *J. Biol. Chem.* 267: 5503–5507, 1992.

[36282] 9834. Ratajczak, T.; Woollatt, E.; Kumar, P.; Ward, B. K.; Minchin, R. F.; Baker, E.: Cyclophilin 40 (PPID) gene map position 4q31.3. *Chromosome Res.* 5: 151 only, 1997.

[36283] 9835. Yokoi, H.; Shimizu, Y.; Anazawa, H.; Lefebvre, C. A.; Korneluk, R. G.; Ikeda, J.-E.: The structure and complete nucleotide sequence of the human cyclophilin 40 (PPID) gene. *Genomics* 35: 448–455, 1996.

[36284] 9836. Pizutti, A.; Novelli, G.; Ratti, A.; Amati, F.; Mari, A.;

Calabrese,G.; Nicolis, S.; Silani, V.; Marino, B.; Scarlato, G.; Ottolenghi,S.; Dallapiccola, B.: UFD1L, a developmentally expressed ubiquitination gene, is deleted in CATCH 22 syndrome. Hum. Molec. Genet. 6: 259–265,1997.

[36285] 9837.Yamagishi, H.; Garg, V.; Matsuoka, R.; Thomas, T.; Srivastava,D.: A molecular pathway revealing a genetic basis for human cardiac and craniofacial defects. Science 283: 1158–1161, 1999.

[36286] 9838.Gong, W.; Emanuel, B. S.; Collins, J.; Kim, D. H.; Wang, Z.; Chen,F.; Zhang, G.; Roe, B.; Budarf, M. L.: A transcription map of the DiGeorge and velo–cardio–facial syndrome minimal critical region on 22q11. Hum. Molec. Genet. 5: 789–800, 1996.

[36287] 9839.Gong, W.; Emanuel, B. S.; Galili, N.; Kim, D. H.; Roe, B.; Driscoll,D. A.; Budarf, M. L.: Structural and mutational analysis of a conserved gene (DGSI) from the minimal DiGeorge syndrome critical region. Hum.Molec. Genet. 6: 267–276, 1997.

[36288] 9840.Lindsay, E. A.; Botta, A.; Jurecic, V.; Carattini–Rivera, S.; Cheah,Y.–C.; Rosenblatt, H. M.; Bradley, A.; Baldini, A.: Congenital heart disease in mice deficient for the DiGeorge syndrome region. Nature 401:379–383, 1999.

[36289] 9841.Lindsay, E. A.; Harvey, E. L.; Scambler, P. J.; Baldini,

A.: ES2, a gene deleted in DiGeorge syndrome, encodes a nuclear protein and is expressed during early mouse development, where it shares an expression domain with a Goosecoid-like gene. *Hum. Molec. Genet.* 7: 629–635, 1998.

[36290] 9842. Fukunaga, R.; Hunter, T.: MNK1, a new MAP kinase-activated protein kinase, isolated by a novel expression screening method for identifying protein kinase substrates. *EMBO J.* 16: 1921–1933, 1997.

[36291] 9843. Pages, G.; Guerin, S.; Grall, D.; Bonino, F.; Smith, A.; Anjuere, F.; Auberger, P.; Pouyssegur, J.: Defective thymocyte maturation in p44 MAP kinase (Erk 1) knockout mice. *Science* 286: 1374–1378, 1999.

[36292] 9844. Stefanovsky, V. Y.; Pelletier, G.; Hannan, R.; Gagnon-Kugler, T.; Rothblum, L. I.; Moss, T.: An immediate response of ribosomal transcription to growth factor stimulation in mammals is mediated by ERK phosphorylation of UBF. *Molec. Cell* 8: 1063–1073, 2001.

[36293] 9845. Mengus, G.; May, M.; Carre, L.; Chambon, P.; Davidson, I.: Human TAF(II)135 potentiates transcriptional activation by the AF-2s of the retinoic acid, vitamin D3, and thyroid hormone receptors in mammalian cells. *Genes Dev.* 11: 1381–1395, 1997.

- [36294] 9846. Shimohata, T.; Nakajima, T.; Yamada, M.; Uchida, C.; Onodera, O.; Naruse, S.; Kimura, T.; Koide, R.; Nozaki, K.; Sano, Y.; Ishiguro, H.; Sakoe, K.; and 16 others: Expanded polyglutamine stretches interact with TAF(II)130, interfering with CREB-dependent transcription. *Nature Genet.* 26: 29–36, 2000.
- [36295] 9847. Ray, M. E.; Su, Y. A.; Meltzer, P. S.; Trent, J. M.: Isolation and characterization of genes associated with chromosome-6 mediated tumor suppression in human malignant melanoma. *Oncogene* 12: 2527–2533, 1996.
- [36296] 9848. Ray, M. E.; Wistow, G.; Su, Y. A.; Meltzer, P. S.; Trent, J. M.: AIM1, a novel non-lens member of the beta-gamma-crystallin superfamily, is associated with the control of tumorigenicity in human malignant melanoma. *Proc. Nat. Acad. Sci.* 94: 3229–3234, 1997.
- [36297] 9849. Teichmann, U.; Ray, M. E.; Ellison, J.; Graham, C.; Wistow, G.; Meltzer, P. S.; Trent, J. M.; Pavan, W. J.: Cloning and tissue expression of the mouse ortholog of AIM1, a beta-gamma-crystallin superfamily member. *Mammalian Genome* 9: 715–720, 1998.
- [36298] 9850. Trent, J. M.; Stanbridge, E. J.; McBride, H. L.; Meese, E. U.; Casey, G.; Araujo, D. E.; Witkowski, C. M.; Nagle, R. B.: Tumorigenicity in human melanoma cell lines controlled

by introduction of human chromosome 6. *Science* 247: 568–571, 1990.

[36299] 9851. Nakamura, Y.; Miura, K.; Fujino, Y.; Iwao, H.; Ogita, S.; Yamanaka, S.: Evolution, structure, and expression of GNPI/oscillin orthologous genes. *Genomics* 68: 179–186, 2000.

[36300] 9852. Rogers, M. J.; Ohgi, T.; Plumbridge, J.; Soll, D.: Nucleotide sequences of the *E. coli* *nagE* and *nagB* genes: the structural genes for the N-acetylglucosamine transport protein of the bacterial phosphoenolpyruvate:sugar phosphotransferase system and for glucosamine 6-phosphate deaminase. *Gene* 62:197–207, 1988.

[36301] 9853. Shevchenko, V.; Hogben, M.; Ekong, R.; Parrington, J.; Lai, F.A.: The human glucosamine-6-phosphate deaminase gene: cDNA cloning and expression, genomic organization and chromosomal localization. *Gene* 216:31–38, 1998.

[36302] 9854. Weidanz, J. A.; Campbell, P.; DeLucas, L. J.; Jin, J.; Moore, D.; Roden, L.; Yu, H.; Heilmann, E.; Vezza, A. C.: Glucosamine 6-phosphate deaminase in normal human erythrocytes. *Brit. J. Haemat.* 91: 72–79, 1995.

[36303] 9855. Bladergroen, B. A.; Strik, M. C. M.; Bovenschen, N.; van Berkum, O.; Scheffer, G. L.; Meijer, C. J. L. M.; Hack, C.

E.; Kummer, J.A.: The granzyme B inhibitor, protease inhibitor 9, is mainly expressed by dendritic cells and at immune-privileged sites. *J. Immun.* 166:3218–3225, 2001.

[36304] 9856. Eyre, H. J.; Sun, J.; Sutherland, G. R.; Bird, P.: Chromosomal mapping of the gene (PI9) encoding the intracellular serpin proteinase inhibitor 9 to 6p25 by fluorescence in situ hybridization. *Genomics* 37:406–408, 1996.

[36305] 9857. Krieg, S. A.; Krieg, A. J.; Shapiro, D. J.: A unique downstream estrogen responsive unit mediates estrogen induction of proteinase inhibitor-9, a cellular inhibitor of IL-1-beta-converting enzyme (caspase 1). *Molec. Endocr.* 15: 1971–1982, 2001.

[36306] 9858. Sun, J.; Bird, C. H.; Sutton, V.; McDonald, L.; Coughlin, P. B.; De Jong, T. A.; Trapani, J. A.; Bird, P. I.: A cytosolic granzyme B inhibitor related to the viral apoptotic regulator cytokine response modifier A is present in cytotoxic lymphocytes. *J. Biol. Chem.* 271:27802–27809, 1996.

[36307] 9859. Steimle, V.; Durand, B.; Barras, E.; Zuffrey, M.; Hadam, M. R.; Mach, B.; Reith, W.: A novel DNA binding regulatory factor is mutated in primary MHC class II deficiency (bare lymphocyte syndrome). *Genes Dev.* 9: 1021–1032, 1995.

[36308] 9860. Teumer, J.; Tseng, H.; Green, H.: The human ba-

sonuclin gene. *Gene* 188:1–7, 1997.

- [36309] 9861. Tseng, H.; Green, H.: Basonuclin: a keratinocyte protein with multiple paired zinc fingers. *Proc. Nat. Acad. Sci.* 89: 10311–10315, 1992.
- [36310] 9862. Tseng, H.; Green, H.: Association of basonuclin with ability of keratinocytes to multiply and with absence of terminal differentiation. *J. Cell Biol.* 126: 495–506, 1994.
- [36311] 9863. Gibson, L.; Holmgreen, S. P.; Huang, D. C. S.; Bernard, O.; Copeland, N. G.; Jenkins, N. A.; Sutherland, G. R.; Baker, E.; Adams, J. M.; Cory, S.: bcl-w, a novel member of the bcl-2 family, promotes cell survival. *Oncogene* 13: 665–675, 1996.
- [36312] 9864. Ross, A. J.; Waymire, K. G.; Moss, J. E.; Parlow, A. F.; Skinner, M. K.; Russell, L. D.; MacGregor, G. R.: Testicular degeneration in Bclw-deficient mice. *Nature Genet.* 18: 251–256, 1998.
- [36313] 9865. Freedman, M. S.; Lucas, R. J.; Soni, B.; von Schantz, M.; Munoz, M.; David-Gray, Z.; Foster, R.: Regulation of mammalian circadian behavior by non-rod, non-cone, ocular photoreceptors. *Science* 284:502–504, 1999.
- [36314] 9866. Griffin, E. A., Jr.; Staknis, D.; Weitz, C. J.: Light-independent role of CRY1 and CRY2 in the mammalian circadian clock. *Science* 286:768–771, 1999.

- [36315] 9867.Hsu, D. S.; Zhao, X.; Zhao, S.; Kazantsev, A.; Wang, R.-P.; Todo,T.; Wei, Y.-F.; Sancar, A.: Putative human blue-light photoreceptorshCRY1 and hCRY2 are flavoproteins. *Biochemistry* 35: 13871–13877,1996.
- [36316] 9868.Kobayashi, K.; Kanno, S.; Smit, B.; van der Horst, G. T. J.; Takao,M.; Yasui, A.: Characterization of photolyase/blue-light receptorhomologs in mouse and human cells. *Nucleic Acids Res.* 26: 5086–5092,1998.
- [36317] 9869.Kume, K.; Zylka, M. J.; Sriram, S.; Shearman, L. P.; Weaver, D.R.; Jin, X.; Maywood, E. S.; Hastings, M. H.; Reppert, S. M.: mCRY1and mCRY2 are essential components of the negative limb of the circadianclock feedback loop. *Cell* 98: 193–205, 1999.
- [36318] 9870.Lucas, R. J.; Freedman, M. S.; Munoz, M.; Garcia-Fernandez, J.-M.;Foster, R. G.: Regulation of the mammalian pineal by non-rod, non-cone,ocular photoreceptors. *Science* 284: 505–507, 1999.
- [36319] 9871.Okamura, H.; Miyake, S.; Sumi, Y.; Yamaguchi, S.; Yasui, A.; Muijtjens,M.; Hoeijmakers, J. H. J.; van der Horst, G. T. J.: Photic inductionof mPer1 and mPer2 in Cry-deficient mice lacking a biological clock. *Science* 286:2531–2534, 1999.
- [36320] 9872.Reick, M.; Garcia, J. A.; Dudley, C.; McKnight, S. L.:

NPAS2:an analog of clock operative in the mammalian forebrain. Science 293:506–509, 2001.

- [36321] 9873. Todo, T.; Ryo, H.; Yamamoto, K.; Toh, H.; Inui, T.; Ayaki, H.; Nomura, T.; Ikenaga, M.: Similarity among the Drosophila (6–4)photolyase, a human photolyase homolog, and the DNA photolyase--blue-light photoreceptor family. Science 272: 109–112, 1996.
- [36322] 9874. van der Horst, G. T. J.; Muijtjens, M.; Kobayashi, K.; Takano, R.; Kanno, S.; Takao, M.; de Wit, J.; Verkerk, A.; Eker, A. P. M.; van Leenen, D.; Buijs, R.; Bootsma, D.; Hoeijmakers, J. H. J.; Yasui, A.: Mammalian Cry1 and Cry2 are essential for maintenance of circadian rhythms. Nature 398: 627–630, 1999.
- [36323] 9875. van der Spek, P. J.; Kobayashi, K.; Bootsma, D.; Takao, M.; Eker, A. P. M.; Yasui, A.: Cloning, tissue expression, and mapping of a human photolyase homolog with similarity to plant blue-light receptors. Genomics 37:177–182, 1996.
- [36324] 9876. Yagita, K.; Tamanini, F.; van der Horst, G. T. J.; Okamura, H.: Molecular mechanisms of the biological clock in cultured fibroblasts. Science 292:278–281, 2001.
- [36325] 9877. Villard, J.; Reith, W.; Barras, E.; Gos, A.; Morris, M. A.; Antonarakis, S. E.; Van den Elsen, P. J.; Mach, B.: Analy-

sis of mutations and chromosomal localisation of the gene encoding RFX5, a novel transcription factor affected in major histocompatibility complex class II deficiency.

Hum.Mutat. 10: 430–435, 1997.

[36326] 9878. Sipila, L.; Szatanik, M.; Vainionpää, H.; Ruotsalainen, H.; Myllylä, R.; Guenet, J.–L.: The genes encoding mouse lysyl hydroxylase isoforms map to chromosomes 4, 5, and 9. Mammalian Genome 11: 1132–1134, 2000.

[36327] 9879. Szpirer, C.; Szpirer, J.; Riviere, M.; Vanvooren, P.; Valtavaara, M.; Myllylä, R.: Localization of the gene encoding a novel isoform of lysyl hydroxylase. Mammalian Genome 8: 707–708, 1997.

[36328] 9880. Valtavaara, M.; Papponen, H.; Pirttilä, A.–M.; Hiltunen, K.; Helander, H.; Myllylä, R.: Cloning and characterization of a novel human lysyl hydroxylase isoform highly expressed in pancreas and muscle. J. Biol.Chem. 272: 6831–6834, 1997.

[36329] 9881. Hellborg, F.; Qian, W.; Mendez–Vidal, C.; Asker, C.; Kost–Alimova, M.; Wilhelm, M.; Imreh, S.; Wiman, K. G.: Human wig–1, a p53 target gene that encodes a growth inhibitory zinc finger protein. Oncogene 20: 5466–5474, 2001.

[36330] 9882. Varmeh–Ziaie, S.; Okan, I.; Wang, Y.; Magnusson, K.

P.; Warthoe,P.; Strauss, M.; Wiman, K. G.: Wig-1, a new p53-induced gene encoding a zinc finger protein. *Oncogene* 15: 2699-2704, 1997.

- [36331] 9883.Modregger, J.; Ritter, B.; Witter, B.; Paulsson, M.; Plomann, M.: All three PACSIN isoforms bind to endocytic proteins and inhibit endocytosis. *J. Cell Sci.* 113: 4511-4521, 2000.
- [36332] 9884.Fardaei, M.; Rogers, M. T.; Thorpe, H. M.; Larkin, K.; Hamshire, M. G.; Harper, P. S.; Brook, J. D.: Three proteins, MBNL, MBLL and MBXL, co-localize in vivo with nuclear foci of expanded-repeat transcripts in DM1 and DM2 cells. *Hum. Molec. Genet.* 11: 805-814, 2002.
- [36333] 9885.Miller, J. W.; Urbinati, C. R.; Teng-umnuay, P.; Stenberg, M. G.; Byrne, B. J.; Thornton, C. A.; Swanson, M. S.: Recruitment of human muscleblind proteins to (CUG)_n expansions associated with myotonic dystrophy. *EMBO J.* 19: 4439-4448, 2000.
- [36334] 9886.Kutty, R. K.; Kutty, G.; Samuel, W.; Duncan, T.; Bridges, C. C.; El-Sherbeeney, A.; Nagineni, C. N.; Smith, S. B.; Wiggert, B.: Molecular characterization and developmental expression of NORPEG, a novel gene induced by retinoic acid. *J. Biol. Chem.* 276: 2831-2840, 2001.
- [36335] 9887.Aapola, U.; Shibuya, K.; Scott, H. S.; Ollila, J.; Vihi-

nen, M.;Heino, M.; Shintani, A.; Kawasaki, K.; Minoshima, S.; Krohn, K.; Antonarakis,S. E.; Shimizu, N.; Kudoh, J.; Peterson, P.: Isolation and initial characterization of a novel zinc finger gene, DNMT3L, on 21q22.3,related to the cysteine-5-methyltransferase 3 gene family. *Genomics* 65:293-298, 2000.

[36336] 9888.Bourc'his, D.; Xu, G.-L.; Lin, C.-S.; Bollman, B.; Bestor, T. H.: Dnmt3L and the establishment of maternal genomic imprints. *Science* 294:2536-2539, 2001.

[36337] 9889.Monney, L.; Sabatos, C.; Gaglia, J. L.; Ryu, A.; Waldner, H.; Chernova,T.; Manning, S.; Greenfield, E. A.; Coyle, A. J.; Sobel, R. A.; Freeman,G. J.; Kuchroo, V. K.: Th1-specific cell surface protein regulates macrophage activation and severity of an autoimmune disease. *Nature* 415:536-541, 2002.

[36338] 9890.Jourdan-Le Saux, C.; Le Saux, O.; Donlon, T.; Boyd, C. D.; Csiszar,K.: The human lysyl oxidase-related gene (LOXL2) maps between markersD8S280 and D8S278 on chromosome 8p21.2-p21.3. *Genomics* 51: 305-307,1998.

[36339] 9891.Jourdan-Le Saux, C.; Tronecker, H.; Bogic, L.; Bryant-Greenwood,G. D.; Boyd, C. D.; Csiszar, K.: The LOXL2 gene encodes a new lysyloxidase-like protein and

is expressed at high levels in reproductivetissues. J. Biol. Chem. 274: 12939–12944, 1999.

- [36340] 9892.Ikeda, K.; Sato, M.; Tsutsumi, O.; Tsuchiya, F.; Tsuneizumi, M.;Emi, M.; Imoto, I.; Inazawa, J.; Muramatsu, M.; Inoue, S.: Promoteranalysis and chromosomal mapping of human EBAG9 gene. Biochem. Biophys.Res. Commun. 273: 654–660, 2000.
- [36341] 9893.Nakashima, M.; Sonoda, K.; Watanabe, T.: Inhibition of cell growthand induction of apoptotic cell death by the human tumor-associatedantigen RCAS1. Nature Med. 5: 938–942, 1999.
- [36342] 9894.Kim, H.–S.; Choi, J.–Y.; Jung, A.–R.; Jang, K.–L.; Lee, W.–H.;Choi, W.–C.; Crow, T. J.; Hyun, B.–H.: Assignment of the human RhoHP1(ARHD) to chromosome 11q14.3 by radiation hybrid mapping. Cytogenet.Cell Genet. 89: 53 only, 2000.
- [36343] 9895.Shimizu, F.; Watanabe, T. K.; Okuno, S.; Omori, Y.; Fujiwara, T.;Takahashi, E.; Nakamura, Y.: Isolation of a novel human cDNA (rhoHP1)homologous to rho genes. Biochim. Biophys. Acta 1351: 13–16, 1997.
- [36344] 9896.Nagasaki, K.; Maass, N.; Manabe, T.; Hanzawa, H.; Tsukada, T.;Kikuchi, K.; Yamaguchi, K.: Identification of a novel gene, DAM1,amplified at chromosome 1p13.3–21

region in human breast cancer celllines. *Cancer Lett.* 140: 219–226, 1999.

[36345] 9897. Neubauer, G.; King, A.; Rappsilber, J.; Calvio, C.; Watson, M.; Ajuh, P.; Sleeman, J.; Lamond, A.; Mann, M.: Mass spectrometry and EST–database searching allows characterization of the multi–protein spliceosome complex. *Nature Genet.* 20: 46–50, 1998.

[36346] 9898. Rader, K.; Boyer, A. D.; Farquhar, M. G.; Arden, K. C.: Assignment of ankyrin repeat, family A (RFXANK–like) 2 (ANKRA2) to human chromosome 5q12–q13 by radiation hybrid mapping and somatic cell hybrid PCR. *Cytogenet. Cell Genet.* 89: 164–165, 2000.

[36347] 9899. Rader, K.; Orlando, R. A.; Lou, X.; Farquhar, M. G.: Characterization of ANKRA, a novel ankyrin repeat protein that interacts with the cytoplasmic domain of megalin. *J. Am. Soc. Nephrol.* 11: 2167–2178, 2000.

[36348] 9900. Nakagawa, H.; Koyama, K.; Murata, Y.; Morito, M.; Akiyama, T.; Nakamura, Y.: EB3, a novel member of the EB1 family preferentially expressed in the central nervous system, binds to a CNS–specific APC homologue. *Oncogene* 19: 210–216, 2000.

[36349] 9901. Dunbar, D. R.; Shibasaki, Y.; Dobbie, L.; Andersson, B.; Brookes, A. J.: In situ hybridisation mapping of genomic

clones for five human respiratory chain complex I genes. Cytogenet. Cell Genet. 78: 21–24, 1997.

[36350] 9902. Emahazion, T.; Brookes, A. J.: Mapping of the ND-UFA2, NDUFA6, NDUFA7, NDUFB8, and NDUF58 electron transport chain genes by intron-based radiation hybrid mapping. Cytogenet. Cell Genet. 82: 114 only, 1998.

[36351] 9903. Ton, C.; Hwang, D. M.; Dempsey, A. A.; Liew, C.-C.: Identification and primary structure of five human NADH-ubiquinone oxidoreductase subunits. Biochem. Biophys. Res. Commun. 241: 589–594, 1997.

[36352] 9904. Walker, J. E.; Arizmendi, J. M.; Dupuis, A.; Fearnley, I. M.; Finel, M.; Medd, S. M.; Pilkington, S. J.; Runswick, M. J.; Skehel, J. M.: Sequences of 20 subunits of NADH:ubiquinone oxidoreductase from bovine heart mitochondria. J. Molec. Biol. 226: 1051–1072, 1992.

[36353] 9905. de Sury, R.; Martinez, P.; Procaccio, V.; Lunardi, J.; Issartel, J.-P.: Genomic structure of the human NDUF58 gene coding for the iron-sulfur TYKY subunit of the mitochondrial NADH:ubiquinone oxidoreductase. Gene 215: 1–10, 1998.

[36354] 9906. Loeffen, J.; Smeitink, J.; Triepels, R.; Smeets, R.; Schuelke, M.; Sengers, R.; Trijbels, F.; Hamel, B.; Mullaart, R.; van den Heuvel, L.: The first nuclear-encoded complex I

mutation in a patient with Leigh syndrome. *Am. J. Hum. Genet.* 63: 1598–1608, 1998.

[36355] 9907. Procaccio, V.; Depetris, D.; Soularue, P.; Mattei, M.-G.; Lunardi, J.; Issartel, J.-P.: cDNA sequence and chromosomal localization of the NDUF58 human gene coding for the 23 kDa subunit of the mitochondrial complex I.

Biochim. Biophys. Acta 1351: 37–41, 1997.

[36356] 9908. Cheng, H. F.; Jiang, M. J.; Chen, C. L.; Liu, S. M.; Wong, L. P.; Lomasney, J. W.; King, K.: Cloning and identification of amino acid residues of human phospholipase C delta 1 essential for catalysis. *J. Biol. Chem.* 270:

5495–5505, 1995.

[36357] 9909. Inhorn, R. C.; Aster, J. C.; Roach, S. A.; Slapak, C. A.; Soiffer, R.; Tantravani, R.; Stone, R. M.: A syndrome of lymphoblastic lymphoma, eosinophilia, and myeloid hyperplasia/malignancy associated with

t(8;13)(p11;q11): description of a distinctive clinicopathologic entity. *Blood* 85:1881–1887, 1995.

[36358] 9910. Michaux, L.; Mecucci, C.; Pereira Velloso, E. R.; Dierlamm, J.; Criel, A.; Louwagie, A.; van Orshoven, A.; Van den Berghe, H.: About the t(8;13)(p11;q12) clinico-pathologic entity. (Letter) *Blood* 87:1658–1659, 1996.

[36359] 9911. Reiter, A.; Sohal, J.; Kulkarni, S.; Chase, A.; Macdon-

ald, D. H.C.; Aguiar, R. C. T.; Goncalves, C.; Hernandez, J. M.; Jennings, B.A.; Goldman, J. M.; Cross, N. C. P.: Consistent fusion of ZNF198 to the fibroblast growth factor receptor-1 in the t(8;13)(p11;q12) myeloproliferative syndrome. *Blood* 92: 1735–1742, 1998.

[36360] 9912. Smedley, D.; Hamoudi, R.; Clark, J.; Warren, W.; Abdul-Rauf, M.; Somers, G.; Venter, D.; Fagan, K.; Cooper, C.; Shipley, J.: The t(8;13)(p11;q11–12) rearrangement associated with an atypical myeloproliferative disorder fuses the fibroblast growth factor receptor 1 gene to a novel gene R-AMP. *Hum. Molec. Genet.* 7: 637–642, 1998.

[36361] 9913. Still, I. H.; Cowell, J. K.: The t(8;13) atypical myeloproliferative disorder: further analysis of the ZNF198 gene and lack of evidence for multiple genes disrupted on chromosome 13. (Letter) *Blood* 92: 1456–1458, 1998.

[36362] 9914. Rousseau, D.; Gingras, A.-C.; Pause, A.; Sonenberg, N.: The eIF4E-binding proteins 1 and 2 are negative regulators of cell growth. *Oncogene* 13: 2415–2420, 1996.

[36363] 9915. Tsukiyama-Kohara, K.; Vidal, S. M.; Gingras, A.-C.; Glover, T.W.; Hanash, S. M.; Heng, H.; Sonenberg, N.: Tissue distribution, genomic structure, and chromosome mapping of mouse and human eukaryotic initiation factor 4E-binding proteins 1 and 2. *Genomics* 38:

353–363,1996.

- [36364] 9916.Meluh, P. B.; Koshland, D.: Suppressors of MIF2, a putative centromereprotein gene in *Saccharomyces cerevisiae*. (Abstract) *Molec. Biol.Cell* 6 (supp.): 360a only, 1995.
- [36365] 9917.Meluh, P. B.; Koshland, D.: Evidence that the MIF2 gene of *Saccharomycescerevisiae* encodes a centromere protein with homology to the mammaliancentromere protein CENP–C. *Molec. Biol. Cell* 6: 793–807, 1995.
- [36366] 9918.Cooper, E. C.; Aldape, K. D.; Abosch, A.; Barbaro, N. M.; Berger,M. S.; Peacock, W. S.; Jan, Y. N.; Jan, L. Y.: Colocalization andcoassembly of two human brain M-type potassium channel subunits thatare mutated in epilepsy. *Proc. Nat. Acad. Sci.* 97: 4914–4919, 2000.
- [36367] 9919.Wang, H.–S.; Pan, Z.; Shi, W.; Brown, B. S.; Wymore, R. S.; Cohen,I. S.; Dixon, J. E.; McKinnon, D.: KCNQ2 and KCNQ3 potassium channelsubunits: molecular correlates of the M–channel. *Science* 282: 1890–1893,1998.
- [36368] 9920.Yang, W.–P.; Levesque, P. C.; Little, W. A.; Conder, M. L.; Ramakrishnan,P.; Neubauer, M. G.; Blana, M. A.: Functional expression of twoKvLQT1–related potassium channels responsible for an inherited idiopathic epilepsy. *J. Biol. Chem.* 273: 19419–19423, 1998.

- [36369] 9921.Cecconi, F.; Alvarez-Bolado, G.; Meyer, B. I.; Roth, K. A.; Gruss,P.: Apaf1 (CED-4 homolog) regulates programmed cell death in mammaliandevlopment. Cell 94: 727-737, 1998.
- [36370] 9922.Hahn, C.; Hirsch, B.; Jahnke, D.; Durkop, H.; Stein, H.: Threenew types of Apaf-1 in mammalian cells. Biochem. Biophys. Res. Commun. 261:746-749, 1999.
- [36371] 9923.Honarpour, N.; Du, C.; Richardson, J. A.; Hammer, R. E.; Wang,X.; Herz, J.: Adult Apaf-1-deficient mice exhibit male infertility. Dev.Biol. 218: 248-258, 2000.
- [36372] 9924.Honarpour, N.; Gilbert, S. L.; Lahn, B. T.; Wang, X.; Herz, J.: Apaf-1 deficiency and neural tube closure defects are found in fogmice. Proc. Nat. Acad. Sci. 98: 9683-9687, 2001.
- [36373] 9925.Kim, H.; Jung, Y. K.; Kwon, Y. K.; Park, S. H.: Assign-ment ofapoptotic protease activating factor-1 gene (APAF1) to human chromosomeband 12q23 by fluo-escence in situ hybridization. Cytogenet. CellGenet. 87: 252-253, 1999.
- [36374] 9926.Kawai, J.; Suzuki, H.; Hara, A.; Hirose, K.; Watanabe, S.: Humanand mouse chromosomal mapping of Stac, a neuron-specific protein withan SH3 domain. Genomics 47: 140-142, 1998.

- [36375] 9927.Suzuki, H.; Kawai, J.; Taga, C.; Yaoi, T.; Hara, A.; Hirose, K.; Hayashizaki, Y.; Watanabe, S.: Stac, a novel neuron-specific protein with cysteine-rich and SH3 domains. *Biochem. Biophys. Res. Commun.* 229:902–909, 1996.
- [36376] 9928.Bradshaw, T.; Graves, T.; Biewald, T.: Personal Communication. St.Louis, Mo. 1/29/1998.
- [36377] 9929.Daga, A.; Micol, V.; Hess, D.; Aebersold, R.; Attardi, G.: Molecular characterization of the transcription termination factor from human mitochondria. *J. Biol. Chem.* 268: 8123–8130, 1993.
- [36378] 9930.Fernandez-Silva, P.; Martinez-Azorin, F.; Micol, V.; Attardi, G.: The human mitochondrial transcription termination factor (mTERF) is a multizipper protein but binds to DNA as a monomer, with evidence pointing to intramolecular leucine zipper interactions. *EMBO J.* 16:1066–1079, 1997.
- [36379] 9931.Hugnot, J. P.; Pedeutour, F.; Le Calvez, C.; Grosgeorge, J.; Passage, E.; Fontes, M.; Lazdunski, M.: The human inward rectifying K(+) channel Kir 2.2 (KCNJ12) gene: gene structure, assignment to chromosome 17p11.1, and identification of a simple tandem repeat polymorphism. *Genomics* 39:113–116, 1997.
- [36380] 9932.Namba, N.; Mori, R.; Tanaka, H.; Kondo, I.; Narahara,

K.; Seino,Y.: The inwardly rectifying potassium channel subunit Kir2.2v (KCNJN1)maps to 17p11.2–p11.1. Cyto-genet. Cell Genet. 79: 85–87, 1997.

[36381] 9933.Wible, B. A.; De Biasi, M.; Majumder, K.; Taglialatela, M.; Brown,A. M.: Cloning and functional expression of an inwardly rectifyingK(+) channel from human atrium. Circ. Res. 76: 343–350, 1995.

[36382] 9934.Imataka, H.; Olsen, H. S.; Sonenberg. N.: A new translationalregulator with homology to eukaryotic trans-lation initiation factor4G. EMBO J. 16: 817–825, 1997.

[36383] 9935.Levy–Strumpf, N.; Deiss, L. P.; Berissi, H.; Kimchi, A.: DAP–5,a novel homolog of eukaryotic translation initiation factor 4G isolatedas a putative modulator of gamma inter-feron–induced programmed cellddeath. Molec. Cell. Biol. 17: 1615–1625, 1997.

[36384] 9936.Shaughnessy, J. D., Jr.; Jenkins, N. A.; Copeland, N. G.: cDNAcloning, expression analysis, and chromosomal localization of a genewith high homology to wheat eIF–(iso)4F and mammalian eIF–4G. Genomics 39:192–197, 1997.

[36385] 9937.Yamanaka, S.; Poksay, K. S.; Arnold, K. S.; Innerarity, T. L.:A novel translational repressor mRNA is edited ex-tensively in liverscontaining tumors caused by the trans–

gene expression of the apoB mRNA-editing enzyme. *Genes Dev.* 11: 321–333, 1997.

- [36386] 9938. Fan, W.; Christensen, M.; Eichler, E.; Zhang, X.; Lennon, G.: Cloning, sequencing, gene organization, and localization of the human ribosomal protein RPL23A gene. *Genomics* 46: 234–239, 1997.
- [36387] 9939. Nomi, M.; Oishi, I.; Kani, S.; Suzuki, H.; Matsuda, T.; Yoda, A.; Kitamura, M.; Itoh, K.; Takeuchi, S.; Takeda, K.; Akira, S.; Ikeya, M.; Takada, S.; Minami, Y.: Loss of mRor1 enhances the heart and skeletal abnormalities in mRor2-deficient mice: redundant and pleiotropic functions of mRor1 and mRor2 receptor tyrosine kinases. *Molec. Cell. Biol.* 21: 8329–8335, 2001.
- [36388] 9940. Westendorf, J. M.; Rao, P. N.; Gerace, L.: Cloning of cDNAs for M-phase phosphoproteins recognized by the MPM2 monoclonal antibody and determination of the phosphorylated epitope. *Proc. Nat. Acad. Sci.* 91: 714–718, 1994.
- [36389] 9941. Wang, X. S.; Diener, K.; Jannuzzi, D.; Trollinger, D.; Tan, T.-H.; Lichenstein, H.; Zukowski, M.; Yao, Z.: Molecular cloning and characterization of a novel protein kinase with a catalytic domain homologous to mitogen-activated protein kinase kinase kinase. *J. Biol. Chem.* 271:

31607–31611, 1996.

[36390] 9942.Chuaqui, R. F.; Englert, C. R.; Strup, S. E.; Vocke, C. D.; Zhuang,Z.; Duray, P. H.; Bostwick, D. G.; Linehan, W. M.; Liotta, L. A.;Emmert–Buck, M. R.: Identification of a novel transcript up–regulatedin a clinically aggressive prostate carcinoma. *Urology* 50: 302–307,1997.

[36391] 9943.Cole, K. A.; Chuaqui, R. F.; Katz, K.; Pack, S.; Zhuang, Z.; Cole,C. E.; Lyne, J. C.; Linehan, W. M.; Liotta, L. A.; Emmert–Buck, M.R.: cDNA sequencing and analysis of POV1 (PB39): a novel gene up–regulatedin prostate cancer. *Genomics* 51: 282–287, 1998.

[36392] 9944.Bono, P.; Salmi, M.; Smith, D. J.; Leppanen, I.; Horelli–Kuitunen,N.; Palotie, A.; Jalkanen, S.: Isolation, structural characterization,and chromosomal mapping of the mouse vascular adhesion protein–1 geneand promoter. *J. Immun.* 161: 2953–2960, 1998.

[36393] 9945.Morris, N. J.; Ducret, A.; Aebersold, R.; Ross, S. A.; Keller,S. R.; Lienhard, G. E.: Membrane amine oxidase cloning and identificationas a major protein in the adipocyte plasma membrane. *J. Biol. Chem.* 272:9388–9392, 1997.

[36394] 9946.Bromme, D.; Rossi, A. B.; Smeekens, S. P.; Anderson, D. C.; Payan,D. G.: Human bleomycin hydrolase: molecular

cloning, sequencing, functional expression, and enzymatic characterization. *Biochemistry* 35:6706–6714, 1996.

- [36395] 9947. Cloos, J.; Nieuwenhuis, E. J. C.; Boomsma, D. I.; Kuik, D. J.; van der Sterre, M. L. T.; Arwert, F.; Snow, G. B.; Braakhuis, B. J. M.: Inherited susceptibility to bleomycin-induced chromatid breaks in cultured peripheral blood lymphocytes. *J. Nat. Cancer Inst.* 91:1125–1130, 1999.
- [36396] 9948. Farrer, L. A.; Abraham, C. R.; Haines, J. L.; Rogaeva, E. A.; Song, Y.; McGraw, W. T.; Brindle, N.; Premkumar, S.; Scott, W. K.; Yamaoka, L. H.; Saunders, A. M.; Roses, A. D.; Auerbach, S. A.; Sorbi, S.; Duara, R.; Pericak-Vance, M. A.; St. George-Hyslop, P. H.: Association between bleomycin hydrolase and Alzheimer's disease in Caucasians. *Ann. Neurol.* 44: 808–811, 1998.
- [36397] 9949. Ferrando, A. A.; Pendas, A. M.; Llano, E.; Velasco, G.; Lidereau, R.; Lopez-Otin, C.: Gene characterization, promoter analysis, and chromosomal localization of human bleomycin hydrolase. *J. Biol. Chem.* 272:33298–33304, 1997.
- [36398] 9950. Ferrando, A. A.; Velasco, G.; Campo, E.; Lopez-Otin, C.: Cloning and expression analysis of human bleomycin hydrolase, a cysteine proteinase involved in chemotherapy resistance. *Cancer Res.* 56: 1746–1750, 1996.

- [36399] 9951.Haston, C. K.; Amos, C. I.; King, T. M.; Travis, E. L.: Inheritance of susceptibility to bleomycin-induced pulmonary fibrosis in the mouse. *Cancer Res.* 56: 2596–2601, 1996.
- [36400] 9952.Hsu, T. C.; Johnston, D. A.; Cherry, L. M.; Ramkissoon, D.; Schantz, S. P.; Jessup, J. M.; Winn, R. J.; Shirley, L.; Furlong, C.: Sensitivity to genotoxic effects of bleomycin in humans: possible relationship to environmental carcinogenesis. *Int. J. Cancer* 43: 403–409, 1989.
- [36401] 9953.Lazo, J. S.; Humphreys, C. J.: Lack of metabolism as the biochemical basis of bleomycin-induced pulmonary toxicity. *Proc. Nat. Acad. Sci.* 80:3064–3068, 1983.
- [36402] 9954.Montoya, S. E.; Aston, C. E.; DeKosky, S. T.; Kamboh, M. I.; Lazo, J. S.; Ferrell, R. E.: Bleomycin hydrolase is associated with risk of sporadic Alzheimer's disease. (Letter) *Nature Genet.* 18: 211–212, 1998. Note: Erratum: *Nature Genet.* 19: 404 only, 1998.
- [36403] 9955.Montoya, S. E.; Ferrell, R. E.; Lazo, J. S.: Genomic structure and genetic mapping of the human neutral cysteine protease bleomycin hydrolase. *Cancer Res.* 57: 4191–4195, 1997.
- [36404] 9956.Zheng, W.; Johnston, S. A.; Joshua-Tor, L.: The unusual active site of Gal6/bleomycin hydrolase can act as a

carboxypeptidase, aminopeptidase, and peptide ligase.

Cell 93: 103–109, 1998.

- [36405] 9957. Xu, X.-N.; Screaton, G. R.; Gotch, F. M.; Dong, T.; Tan, R.; Almond, N.; Walker, B.; Stebbings, R.; Kent, K.; Nagata, S.; Stott, J. E.; McMichael, A. J.: Evasion of cytotoxic T lymphocyte (CTL) responses by Nef-dependent induction of Fas ligand (CD95L) expression on simian immunodeficiency virus-infected cells. *J. Exp. Med.* 186: 7–16, 1997.
- [36406] 9958. Huang, L. J.; Durick, K.; Weiner, J. A.; Chun, J.; Taylor, S. S.: Identification of a novel protein kinase A anchoring protein that binds both type I and type II regulatory subunits. *J. Biol. Chem.* 272: 8057–8064, 1997.
- [36407] 9959. Lin, R.-Y.; Moss, S. B.; Rubin, C. S.: Characterization of S-AKAP84, a novel developmentally regulated A kinase anchor protein of male germ cells. *J. Biol. Chem.* 270: 27804–27811, 1995.
- [36408] 9960. Trendelenburg, G.; Hummel, M.; Riecken, E.-O.; Hanski, C.: Molecular characterization of AKAP149, a novel A kinase anchor protein with a KH domain. *Biochem. Biophys. Res. Commun.* 225: 313–319, 1996.
- [36409] 9961. Moshous, D.; Callebaut, I.; de Chasseval, R.; Corneo, B.; Cavazzana-Calvo, M.; Le Deist, F.; Tezcan, I.; Sanal, O.; Bertrand, Y.; Philippe, N.; Fischer, A.; de Villartay, J.-P.:

Artemis, a novel DNA double-strandbreak repair/V(D)J recombination protein, is mutated in human severe combined immune deficiency. *Cell* 105: 177–186, 2001.

[36410] 9962.Chang, K.; Hanaoka, K.; Kumada, M.; Takuwa, Y.: Molecular cloning and functional analysis of a novel P2 nucleotide receptor. *J. Biol.Chem.* 270: 26152–26158, 1995.

[36411] 9963.Communi, D.; Parmentier, M.; Boeynaems, J.–M.: Cloning, functional expression and tissue distribution of the human P2Y6 receptor. *Biochem.Biophys. Res. Commun.* 222: 303–308, 1996.

[36412] 9964.Maier, R.; Glatz, A.; Mosbacher, J.; Bilbe, G.: Cloning of P2Y6 cDNAs and identification of a pseudogene: comparison of P2Y receptor subtype expression in bone and brain tissues. *Biochem. Biophys. Res.Comm.* 237: 297–302, 1997.

[36413] 9965.Pidlaoan, L. V.; Jin, J.; Sandhu, A. K.; Athwal, R. S.; Kunapuli, S. P.: Colocalization of P2Y2 and P2Y6 receptor genes at human chromosome 11q13.3–14.1. *Somat. Cell Molec. Genet.* 23: 291–296, 1997.

[36414] 9966.Wang, B.; Kishihara, K.; Zhang, D.; Hara, H.; Nomoto, K.: Molecular cloning and characterization of a novel human receptor protein tyrosine phosphatase gene, hPTP-J: down-regulation of gene expression by PMA and calcium

ionophore in Jurkat T lymphoma cells. *Biochem. Biophys. Res. Commun.* 231: 77–81, 1997.

[36415] 9967. Wang, H.; Lian, Z.; Lerch, M. M.; Chen, Z.; Xie, W.; Ullrich, A.: Characterization of PCP-2, a novel receptor protein tyrosine phosphatase of the MAM domain family. *Oncogene* 12: 2555–2562, 1996.

[36416] 9968. Chinnaiyan, A. M.; O'Rourke, K.; Tewari, M.; Dixit, V. M.: FADD, a novel death domain-containing protein, interacts with the death domain of Fas and initiates apoptosis. *Cell* 81: 505–512, 1995.

[36417] 9969. Kabra, N. H.; Kang, C.; Hsing, L. C.; Zhang, J.; Winoto, A.: T cell-specific FADD-deficient mice: FADD is required for early T cell development. *Proc. Nat. Acad. Sci.* 98: 6307–6312, 2001.

[36418] 9970. Yanagisawa, K.; Sakakibara, Y.; Suiko, M.; Takami, Y.; Nakayama, T.; Nakajima, H.; Takayanagi, K.; Natori, Y.; Liu, M.-C.: cDNA cloning, expression, and characterization of the human bifunctional ATP sulfurylase/adenosine 5'-prime-phosphosulfate kinase enzyme. *Biosci. Biotech. Biochem.* 62: 1037–1040, 1998.

[36419] 9971. Sahin, U.; Tureci, O.; Schmitt, H.; Cochlovius, B.; Johannes, T.; Schmits, R.; Stenner, F.; Luo, G.; Schobert, I.; Pfreundschuh, M.: Human neoplasms elicit multiple spe-

cific immune responses in the autologous host. *Proc. Nat. Acad. Sci.* 92: 11810–11813, 1995.

[36420] 1992. Tureci, O.; Sahin, U.; Vollmar, E.; Siemer, S.; Gottert, E.; Seitz, G.; Parkkila, A. K.; Shah, G. N.; Grubb, J. H.; Pfrendrich, M.; Sly, W. S.: Human carbonic anhydrase XII: cDNA cloning, expression, and chromosomal localization of a carbonic anhydrase gene that is overexpressed in some renal cell cancers. *Proc. Nat. Acad. Sci.* 95:7608–7613, 1998.

[36421] 1993. Herrscher, R. F.; Kaplan, M. H.; Lelsz, D. L.; Das, C.; Scheuermann, R.; Tucker, P. W.: The immunoglobulin heavy-chain matrix-associating regions are bound by Bright: a B cell-specific trans-activator that describes a new DNA-binding protein family. *Genes Dev.* 9: 3067–3082, 1995.

[36422] 1994. Kortschak, R. D.; Reimann, H.; Zimmer, M.; Eyre, H. J.; Saint, R.; Jenne, D. E.: The human dead ringer/bright homolog, DRIL1: cDNA cloning, gene structure, and mapping to D19S886, a marker on 19p13.3 that is strictly linked to the Peutz-Jeghers syndrome. *Genomics* 51:288–292, 1998.

[36423] 1995. Kamei, M.; Webb, G. C.; Young, I. G.; Campbell, H. D.: SOLH, a human homologue of the *Drosophila*

melanogaster small optic lobes gene is a member of the calpain and zinc-finger gene families and maps to human chromosome 16p13.3 near CATM (cataract with microphthalmia). Genomics 51:197–206, 1998.

[36424] 1996. Eriksson, I.; Sandback, D.; Ek, B.; Lindahl, U.; Kjellen, L.: cDNA cloning and sequencing of mouse mastocytoma glucosaminyl N-deacetylase/N-sulfotransferase, an enzyme involved in the biosynthesis of heparin. J. Biol. Chem. 269:10438–10443, 1994.

[36425] 1997. Forsberg, E.; Pejler, G.; Ringvall, M.; Lunderius, C.; Tomasini-Johansson, B.; Kusche-Gullberg, M.; Eriksson, I.; Ledin, J.; Hellman, L.; Kjellen, L.: Abnormal mast cells in mice deficient in a heparin-synthesizing enzyme. Nature 400: 773–776, 1999.

[36426] 1998. Humphries, D. E.; Lanciotti, J.; Karlinsky, J. B.: cDNA cloning, genomic organization and chromosomal localization of human heparan glucosaminyl N-deacetylase/N-sulphotransferase-2. Biochem. J. 332:303–307, 1998.

[36427] 1999. Humphries, D. E.; Wong, G. W.; Friend, D. S.; Gurish, M. F.; Qiu, W.-T.; Huang, C.; Sharpe, A. H.; Stevens, R. L.: Heparin is essential for the storage of specific granule proteases in mast cells. Nature 400:769–772, 1999.

- [36428] 9980.Orellana, A.; Hirschberg, C. B.; Wei, Z.; Swiedler, S. J.; Ishihara,M.: Molecular cloning and expression of a glycosaminoglycan N-acetylglucosaminylN-deacetylase/N-sulfotransferase from a heparin-producing cell line. *J.Biol. Chem.* 269: 2270-2276, 1994.
- [36429] 9981.Zehnder, J. L.; Galli, S. J.: Mast-cell heparin demystified. *Nature* 400:714-715, 1999.
- [36430] 9982.Lavedan, C.; Buchholtz, S.; Auburger, G.; Albin, R. L.; Athanassiadou,A.; Blancato, J.; Burguera, J. A.; Ferrell, R. E.; Kostic, V.; Leroy,E.; Leube, B.; Mota-Vieira, L.; and 9 others: Absence of mutationin the beta- and gamma-synuclein genes in familial autosomal dominantParkinson's disease. *DNA Research* 5: 401-402, 1998.
- [36431] 9983.Leek, J. P.; Hamlin, P. J.; Wilton, J.; Lench, N. J.: Assignmentof the Rab13 gene (RAB13) to human chromosome band 12q13 by in situhybridization. *Cytogenet. Cell Genet.* 79: 210-211, 1997.
- [36432] 9984.Novick, P.; Zerial, M.: The diversity of Rab proteins in vesicletransport. *Curr. Opin. Cell Biol.* 9: 496-504, 1997.
- [36433] 9985.Salminen, A.; Novick, P. J.: A ras-like protein is required fora post-Golgi event in yeast secretion. *Cell* 49:

527–538, 1987.

- [36434] 9986.Zahraoui, A.; Joberty, G.; Arpin, M.; Fontaine, J. J.; Hellio,R.; Tavitian, A.; Louvard, D.: A small rab GTPase is distributedin cytoplasmic vesicles in nonpolarized cells but colocalizes withthe tight junction marker ZO–1 in polarized epithelial cells. *J.Cell. Biol.* 124: 101–115, 1994.
- [36435] 9987.Liu, X.–L.; Wazer, D. E.; Watanabe, K.; Band, V.: Identificationof a novel serine protease–like gene, the expression of which is down–regulatedduring breast cancer progression. *Cancer Res.* 56: 3371–3379, 1996.
- [36436] 9988.Polikoff, D.; Kuo, W.–L.; Cochran, J. F.; Wernick, M.; Kowbel,D.; Myambo, K.; Collins, C. C.: Assignment of protease, serine–like1 (PRSSL1) to human chromosome 19q13 by in situ hybridization andradiation hybrid mapping. *Cytogenet. Cell Genet.* 79: 147–148, 1997.
- [36437] 9989.Aurich–Costa, J.; Cadel, S.; Gouzy, C.; Foulon, T.; Cherif, D.;Cohen, P.: Assignment of the aminopeptidase B gene (RNPEP) to humanchromosome 1 band q32 by in situ hybridization. *Cytogenet. Cell Genet.* 79:143–144, 1997.
- [36438] 9990.Cadel, S.; Foulon, T.; Viron, A.; Balogh, A.; Midol–Monnet, S.;Noel, N.; Cohen, P.: Aminopeptidase B from the rat testis is a bifunctionalenzyme structurally related to leukotriene–A4 hydrolase. *Proc. Nat.Acad. Sci.* 94:

2963–2968, 1997.

- [36439] 9991.Cadel, S.; Pierotti, A. R.; Foulton, T.; Creminon, C.; Barre, N.;Segretain, D.; Cohen, P.: Aminopeptidase–B in the rat testes: isolation,functional properties and cellular localization in the seminiferoustubules. *Molec. Cell. Endocr.* 110: 149–160, 1995.
- [36440] 9992.Hopsu, V. K.; Kantonen, U. M.; Glenner, G. G.: A peptidase fromrat tissues selectively hydrolyzing N–terminal arginine and lysineresidues. *Life Sci.* 3: 1449–1453, 1964.
- [36441] 9993.Ershova, G.; Derre, J.; Chetelin, S.; Nancy, V.; Berger, R.; Kaplan,J.; Munnich, A.; de Gunzburg, J.: cDNA sequence, genomic organizationand mapping of PDE6D, the human gene encoding the delta subunit ofthe cGMP phosphodiesterase of retinal rod cells to chromosome 2q36. *Cytogenet.Cell Genet.* 79: 139–141, 1997.
- [36442] 9994.Florio, S. K.; Prusti, R. K.; Beavo, J. A.: Solubilization ofmembrane–bound rod phosphodiesterase by the rod phosphodiesteraserecombinant delta subunit. *J. Biol. Chem.* 271: 24036–24047, 1996.
- [36443] 9995.Li, N.; Florio, S. K.; Pettenati, M. J.; Rao, P. N.; Beavo, J.A.; Baehr, W.: Characterization of human and mouse rod cGMP phosphodiesterasedelta subunit (PDE6D) and chro–

mosomal localization of the human gene. Genomics 49:76–82, 1998.

- [36444] 1996.Jacquemin, P.; Martial, J. A.; Davidson, I.: Human TEF-5 is preferentially expressed in placenta and binds to multiple functional elements of the human chorionic somatomammotropin-B gene enhancer. J. Biol. Chem. 272:12928–12937, 1997.
- [36445] 1997.Jiang, S.-W.; Wu, K.; Eberhardt, N. L.: Human placental TEF-5 transactivates the human chorionic somatomammotropin gene enhancer. Molec.Endocr. 13: 879–889, 1999.
- [36446] 1998.Enattah, N. S.; Sahi, T.; Savilahti, E.; Terwilliger, J. D.; Peltonen, L.; Jarvela, I.: Identification of a variant associated with adult-type hypolactasia. Nature Genet. 30: 233–237, 2002.
- [36447] 1999.Jarvela, I.; Enattah, N. S.; Kokkonen, J.; Varilo, T.; Savilahti, E.; Peltonen, L.: Assignment of the locus for congenital lactase deficiency to 2q21, in the vicinity of but separate from the lactase-phlorizin hydrolase gene. Am. J. Hum. Genet. 63: 1078–1085, 1998.
- [36448] 2000.Kotaka, M.; Kostin, S.; Ngai, S.; Chan, K.; Lau, Y.; Lee, S. M.Y.; Li, H.; Ng, E. K. O.; Schaper, J.; Tsui, S. K. W.; Fung, K.; Lee, C.; Waye, M. M. Y.: Interaction of hCLIM1, an

Enigma family protein,with alpha-actinin 2. J. Cell.

Biochem. 78: 558–565, 2000.

[36449] 10001.Kotaka, M.; Ngai, S.-M.; Garcia-Barcelo, M.; Tsui, S. K. W.; Fung,K.-P.; Lee, C.-Y.; Waye, M. M. Y.: Characteri-
zation of the human36-kDa carboxyl terminal LIM domain
protein (hCLIM1). J. Cell. Biochem. 72:279–285, 1999.

[36450] 10002.Bach, I.: The LIM domain: regulation by association.
Mech. Dev. 91:5–17, 2000.

[36451] 10003.Guy, P. M.; Kenny, D. A.; Gill, G. N.: The PDZ do-
main of the LIMprotein enigma binds to beta-
tropomyosin. Molec. Biol. Cell 10:1973–1984, 1999.

[36452] 10004.Kuroda, S.; Tokunaga, C.; Kiyohara, Y.; Higuchi, O.;
Konishi, H.;Mizuno, K.; Gill, G. N.; Kikkawa, U.: Protein-
protein interactionof zinc finger LIM domains with protein
kinase C. J. Biol. Chem. 271:31029–31032, 1996.

[36453] 10005.Wu, R.-Y.; Gill, G. N.: LIM domain recognition of a
tyrosine-containingtight turn. J. Biol. Chem. 269:
25085–25090, 1994.

[36454] 10006.Rudnick, A.; Ling, T. Y.; Odagiri, H.; Rutter, W. J.;
German, M.S.: Pancreatic beta cells express a diverse set
of homeobox genes. Proc.Nat. Acad. Sci. 91:
12203–12207, 1994.

[36455] 10007.ten Berge, D.; Brouwer, A.; El Bahi, S.; Guenet, J.-L.;

Robert,B.; Meijlink, F.: Mouse *Alx3*: an *aristaless*-like homeobox gene expressed during embryogenesis in ectomesenchyme and lateral plate mesoderm. *Dev.Biol.* 199: 11–25, 1998.

[36456] 10008.Khoja, H.; Wang, G.; Ng, C.-T. L.; Tucker, J.; Brown, T.; Shyamala,V.: Cloning of CCRL1, an orphan seven transmembrane receptor related to chemokine receptors, expressed abundantly in the heart. *Gene* 246:229–238, 2000.

[36457] 10009.Schweickart, V. L.; Epp, A.; Raport, C. J.; Gray, P. W.: CCR11 is a functional receptor for the monocyte chemoattractant protein family of chemokines. *J. Biol. Chem.* 275: 9550–9556, 2000. Note:Erratum: *J. Biol. Chem.* 276: 856 only, 2001.

[36458] 10010.Launay, P.; Fleig, A.; Perraud, A.-L.; Scharenberg, A. M.; Penner,R.; Kinet, J.-P.: TRPM4 is a Ca^{2+} -activated nonselective cation channel mediating cell membrane depolarization. *Cell* 109: 397–407,2002.

[36459] 10011.Xu, X.-Z. S.; Moebius, F.; Gill, D. L.; Montell, C.: Regulation of melastatin, a TRP-related protein, through interaction with a cytoplasmic isoform. *Proc. Nat. Acad. Sci.* 98: 10692–10697, 2001.

[36460] 10012.Astrin, K. H.; Warner, C. A.; Yoo, H.-W.; Goodfel-

low, P. J.; Tsai, S.-F.; Desnick, R. J.: Regional assignment of the human uroporphyrinogen III synthase (UROS) gene to chromosome 10q25.2-q26.3. *Hum. Genet.* 87:18-22, 1991.

[36461] 10013. Boulechfar, S.; Da Silva, V.; Deybach, J.-C.; Nordmann, Y.; Grandchamp, B.; de Verneuil, H.: Heterogeneity of mutations in the uroporphyrinogen III synthase gene in congenital erythropoietic porphyria. *Hum. Genet.* 88:320-324, 1992.

[36462] 10014. Frank, J.; Wang, X.; Lam, H.-M.; Aita, V. M.; Jugert, F. K.; Goerz, G.; Merk, H. F.; Poh-Fitzpatrick, M. B.; Christiano, A. M.: C73R is a hotspot mutation in the uroporphyrinogen III synthase gene in congenital erythropoietic porphyria. *Ann. Hum. Genet.* 62: 225-230, 1998.

[36463] 10015. Shady, A. A.; Colby, B. R.; Cunha, L. F.; Astrin, K. H.; Bishop, D. F.; Desnick, R. J.: Congenital erythropoietic porphyria: identification and expression of eight novel mutations in the uroporphyrinogen III synthase gene. *Brit. J. Haemat.* 117: 980-987, 2002.

[36464] 10016. Tanigawa, K.; Takamura, N.; Yamashita, S.: Congenital erythropoietic porphyria. *Nippon Rinsho* 53: 1422-1426, 1995.

[36465] 10017. Tsai, S.-F.; Bishop, D. F.; Desnick, R. J.: Human

uroporphyrinogenIII synthase: molecular cloning, nucleotide sequence, and expression of a full-length cDNA. Proc. Nat. Acad. Sci. 85: 7049–7053, 1988.

- [36466] 10018. Warner, C. A.; Yoo, H.-W.; Roberts, A. G.; Desnick, R. J.: Congenital erythropoietic porphyria: identification and expression of exonic mutations in the uroporphyrinogen III synthase gene. J. Clin. Invest. 89:693–700, 1992.
- [36467] 10019. Warner, C. A.; Yoo, H. W.; Tsai, S.-F.; Roberts, A. G.; Desnick, R. J.: Congenital erythropoietic porphyria: characterization of the genomic structure and identification of mutations in the uroporphyrinogenIII synthase gene. (Abstract) Am. J. Hum. Genet. 47 (suppl.): A83 only, 1990.
- [36468] 10020. Lapsys, N. M.; Layfield, R.; Baker, E.; Callen, D. F.; Sutherland, G. R.; Abedinia, M.; Nixon, P. F.; Mattick, J. S.: Chromosomal location of the human transketolase gene. Cytogenet. Cell Genet. 61: 274–275, 1992.
- [36469] 10021. Gladyshev, V. N.; Liu, A.; Novoselov, S. V.; Krysan, K.; Sun, Q.-A.; Kryukov, V. M.; Kryukov, G. V.; Lou, M. F.: Identification and characterization of a new mammalian glutaredoxin (thioltransferase), Grx2. J. Biol. Chem. 276: 30374–30380, 2001.
- [36470] 10022. Lundberg, M.; Johansson, C.; Chandra, J.; Enoks-

son, M.; Jacobsson, G.; Ljung, J.; Johansson, M.; Holmgren, A.: Cloning and expression of a novel human glutaredoxin (Grx2) with mitochondrial and nuclear isoforms. *J. Biol. Chem.* 276: 26269–26275, 2001.

- [36471] 10023. Ungar, D.; Oka, T.; Brittle, E. E.; Vasile, E.; Lupashin, V. V.; Chatterton, J. E.; Heuser, J. E.; Krieger, M.; Waters, M. G.: Characterization of a mammalian Golgi-localized protein complex, COG, that is required for normal Golgi morphology and function. *J. Cell Biol.* 157: 405–415, 2002.
- [36472] 10024. Walter, D. M.; Paul, K. S.; Waters, M. G.: Purification and characterization of a novel 13 S hetero oligomeric protein complex that stimulates *in vitro* Golgi transport. *J. Biol. Chem.* 273: 29565–29576, 1998.
- [36473] 10025. Baloh, R. H.; Tansey, M. G.; Golden, J. P.; Creedon, D. J.; Heuckeroth, R. O.; Keck, C. L.; Zimonjic, D. B.; Popescu, N. C.; Johnson, E. M., Jr.; Milbrandt, J.: TrnR2, a novel receptor that mediates neurturin and GDNF signaling through Ret. *Neuron* 18: 793–802, 1997.
- [36474] 10026. Meuwissen, R. L. J.; Meerts, I.; Hoovers, J. M. N.; Leschot, N. J.; Heyting, C.: Human synaptonemal complex protein 1 (SCP1): isolation and characterization of the cDNA and chromosomal localization of the gene. *Genomics*

39: 377–384, 1997.

- [36475] 10027. Meuwissen, R. L. J.; Offenberg, H. H.; Dietrich, A. J. J.; Riesewijk, A.; van Iersel, M.; Heyting, C.: A coiled-coil related protein specific for synapsed regions of meiotic prophase chromosomes. *EMBO J.* 11:5091–5100, 1992.
- [36476] 10028. Levedakou, E. N.; He, M.; Baptist, E. W.; Craven, R. J.; Cance, W. G.; Welch, P. L.; Simmons, A.; Naylor, S. L.; Leach, R. L.; Lewis, T. B.; Bowcock, A.; Liu, E. T.: Two novel human serine/threonine kinases with homologies to the cell cycle regulating *Xenopus* MO15, and NIMA kinases: cloning and characterization of their expression pattern. *Oncogene* 9: 1977–1988, 1994.
- [36477] 10029. Kondoh, N.; Nishina, Y.; Tsuchida, J.; Koga, M.; Tanaka, H.; Uchida, K.; Inazawa, J.; Taketo, M.; Nozaki, M.; Nojima, H.; Matsumiya, K.; Namiki, M.; Okuyama, A.; Nishimune, Y.: Assignment of synaptonemal complex protein 1 (SCP1) to human chromosome 1p13 by fluorescence in situ hybridization and its expression in the testis. *Cytogenet. Cell Genet.* 78: 103–104, 1997.
- [36478] 10030. Blondel, O.; Vandecasteele, G.; Gastineau, M.; Leclerc, S.; Dahmoune, Y.; Langlois, M.; Fischmeister, R.: Molecular and functional characterization of a 5-HT(4) receptor cloned from human atrium. *FEBS Lett.*

412:465–474, 1997.

- [36479] 10031.Claeysen, S.; Faye, P.; Sebben, M.; Lemaire, S.; Bockaert, J.; Dumuis, A.; Taviaux, S.: Assignment of 5–hydroxytryptamine receptor(HTR4) to human chromosome 5 bands q31–to–q33 by in situ hybridization. *Cytogenet.Cell Genet.* 78: 133–134, 1997.
- [36480] 10032.Dumuis, A.; Bouhelal, R.; Sebben, M.; Cory, R.; Bockaert, J.:A nonclassical 5–hydroxytryptamine receptor positively coupled withadenylate cyclase in the central nervous system. *Molec. Pharm.* 34:880–887, 1988.
- [36481] 10033.Eglen, R. M.; Wong, E. H. F.; Dumuis, A.; Bockaert, J.: Central5–HT4 receptors. *Trends Pharm. Sci.* 16: 391–398, 1995.
- [36482] 10034.Isomura, T.; Tamiya–Koizumi, K.; Suzuki, M.; Yoshida, S.; Taniguchi,M.; Matsuyama, M.; Ishigaki, T.; Sakuma, S.; Takahashi, M.: RFP isa DNA binding protein associated with the nuclear matrix. *NucleicAcids Res.* 20: 5305–5310, 1992.
- [36483] 10035.Szpirer, C.; Szpirer, J.; Riviere, M.; Tazi, R.; Pontarotti, P.: Mapping of the Olf89 and Rfp genes to the rat genome: comparisonwith the mouse and human and new insights into the evolution of therodent genome. *Cytogenet. Cell Genet.* 78: 137–139, 1997.

- [36484] 10036.Lim, D.-S.; Kirsch, D. G.; Canman, C. E.; Ahn, J.-H.; Ziv, Y.; Newman, L. S.; Darnell, R. B.; Shiloh, Y.; Kastan, M. B.: ATM binds to beta-adaptin in cytoplasmic vesicles. *Proc. Nat. Acad. Sci.* 95:10146–10151, 1998.
- [36485] 10037.Newman, L. S.; McKeever, M. O.; Okano, H. J.; Darnell, R. B.:Beta-NAP, a cerebellar degeneration antigen, is a neuron-specific vesicle coat protein. *Cell* 82: 773–783, 1995.
- [36486] 10038.Simpson, F.; Bright, N. A.; West, M. A.; Newman, L. S.; Darnell, R. B.; Robinson, M. S.: A novel adaptor-related protein complex. *J. Cell. Biol.* 133: 749–760, 1996.
- [36487] 10039.Luo, J.; Sladek, R.; Bader, J. A.; Matthysen, A.; Rossant, J.; Giguere, V.: Placental abnormalities in mouse embryos lacking orphan nuclear receptor ERR-beta. *Nature* 388: 778–782, 1997.
- [36488] 10040.Adachi, O.; Kawai, T.; Takeda, K.; Matsumoto, M.; Tsutsui, H.; Sakagami, M.; Nakanishi, K.; Akira, S.: Targeted disruption of the MyD88 gene results in loss of IL-1- and IL-18-mediated function. *Immunity* 143–150, 1998.
- [36489] 10041.Bonnert, T. P.; Garaka, K. E.; Parnet, P.; Sonoda, G.; Testa, J.R.; Sims, J. E.: The cloning and characterization of human MyD88: a member of an IL-1 receptor related fam-

ily. FEBS Lett. 402: 81–84,1997.

- [36490] 10042.Hardiman, G.; Jenkins, N. A.; Copeland, N. G.; Gilbert, D. J.;Garcia, D. K.; Naylor, S. L.; Kastelein, R. A.; Bazan, J. F.: Geneticstructure and chromosomal mapping of MyD88. Genomics 45: 332–339,1997.
- [36491] 10043.Hayashi, F.; Smith, K. D.; Ozinsky, A.,; Hawn, T. R.; Yi, E. C.;Goodlett, D. R.; Eng, J. K.; Akira, S.; Underhill, D. M.; Aderem,A.: The innate immune response to bacterial flagellin is mediatedby Toll–like receptor 5. Nature 410: 1099–1103, 2001.
- [36492] 10044.Kawai, T.; Adachi, O.; Ogawa, T.; Takeda, K.; Akira, S.: Unresponsivenessof MyD88–deficient mice to endo–toxin. Immunity 11: 115–122, 1999.
- [36493] 10045.Lord, K. A.; Hoffman–Liebermann, B.; Liebermann, D. A.: Complexityof the immediate early response of myeloid cells to terminal differentiationand growth arrest includes ICAM–1, Jun–B and histone variants. Oncogene 5:387–396, 1990.
- [36494] 10046.Medzhitov, R.; Preston–Hurlburt, P.; Kopp, E.; Staden, A.; Chen,C.; Ghosh, S.; Janeway, C. A., Jr.: MyD88 is an adaptor protein inthe hToll/Il–1 receptor family signaling pathways. Molec. Cell 2:253–258, 1998.
- [36495] 10047.Ge, K.; Guermah, M.; Yuan, C.–X.; Ito, M.; Wallberg,

A. E.; Spiegelman, B. M.; Roeder, R. G.: Transcription coactivator TRAP220 is required for PPAR-gamma-2-stimulated adipogenesis. *Nature* 417: 563–567, 2002.

[36496] 10048. Greene, M. E.; Blumberg, B.; McBride, O. W.; Yi, H. F.; Kronquist, K.; Kwan, K.; Hsieh, L.; Greene, G.; Nimer, S. D.: Isolation of the human peroxisome proliferator activated receptor gamma cDNA: expression in hematopoietic cells and chromosomal mapping. *Gene Expr.* 4: 281–299, 1995.

[36497] 10049. Hara, M.; Alcoser, S. Y.; Qadir, A.; Beiswenger, K. K.; Cox, N. J.; Ehrmann, D. A.: Insulin resistance is attenuated in women with polycystic ovary syndrome with the Pro12Ala polymorphism in the PPAR-gamma gene. *J. Clin. Endocr. Metab.* 87: 772–775, 2002.

[36498] 10050. Harris, S. G.; Phipps, R. P.: Prostaglandin D2, its metabolite 15-d-PGJ2, and peroxisome proliferator activated receptor-gamma agonists induce apoptosis in transformed, but not normal, human T lineage cells. *Immunology* 105: 23–34, 2002.

[36499] 10051. Hasstedt, S. J.; Ren, Q.-F.; Teng, K.; Elbein, S. C.: Effect of the peroxisome proliferator-activated receptor-gamma-2 Pro12Ala variant on obesity, glucose homeosta-

sis, and blood pressure in members of familial type 2 diabetic kindreds. *J. Clin. Endocr. Metab.* 86:536–541, 2001.

[36500] 10052. Dabovic, B.; Chen, Y.; Colarossi, C.; Obata, H.; Zambuto, L.; Perle, M. A.; Rifkin, D. B.: Bone abnormalities in latent TGF- β binding protein (Ltbp)-3-null mice indicate a role for Ltbp-3 in modulating TGF- β bioavailability. *J. Cell Biol.* 156: 227–232, 2002.

[36501] 10053. Li, X.; Yin, W.; Perez-Jurado, L.; Bonadio, J.; Francke, U.: Mapping of human and murine genes for latent TGF- β binding protein-2 (LTBP2). *Mammalian Genome* 6: 42–45, 1995.

[36502] 10054. Sawicki, M.; Arnold, E.; Ebrahimi, S.; Duell, T.; Jin, S.; Wood, T.; Chakrabarti, R.; Peters, J.; Wan, Y.; Samara, G.; Weier, H.-U. G.; Udar, N.; Passaro, E., Jr.; Srivatsan, E. S.: A transcript map encompassing the multiple endocrine neoplasia type-1 (MEN1) locus on chromosome 11q13. *Genomics* 42: 405–412, 1997.

[36503] 10055. Yin, W.; Smiley, E.; Germiller, J.; Mechan, R. P.; Florer, J. B.; Wenstrup, R. J.; Bonadio, J.: Isolation of a novel latent transforming growth factor- β binding protein gene (LTBP-3). *J. Biol. Chem.* 270:10147–10160, 1995.

[36504] 10056. Moren, A.; Olofsson, A.; Stenman, G.; Sahlin, P.; Kanzaki, T.; Claesson-Welsh, L.; ten Dijke, P.; Miyazono,

K.; Heldin, C.-H.: Identification and characterization of LTBP-2, a novel latent transforming growth factor-beta-binding protein. *J. Biol. Chem.* 269: 32469-32478, 1994.

- [36505] 10057. Scherer, S. W.; Duvoisin, R. M.; Kuhn, R.; Heng, H. H. Q.; Belloni, E.; Tsui, L.-C.: Localization of two metabotropic glutamate receptor genes, GRM3 and GRM8, to human chromosome 7q. *Genomics* 31: 230-233, 1996.
- [36506] 10058. Scherer, S. W.; Soder, S.; Duvoisin, R. M.; Huizenga, J. J.; Tsui, L.-C.: The human metabotropic glutamate receptor 8 (GRM8) gene: a disproportionately large gene located at 7q31.3-q32.1. *Genomics* 44: 232-236, 1997.
- [36507] 10059. Wu, S.; Wright, R. A.; Rockey, P. K.; Burgett, S. G.; Arnold, J. S.; Rosteck, P. R., Jr.; Johnson, B. G.; Schoepp, D. D.; Belagaje, R. M.: Group III human metabotropic glutamate receptors 4, 7 and 8: molecular cloning, functional expression, and comparison of pharmacological properties in RGT cells. *Molec. Brain Res.* 53: 88-97, 1998.
- [36508] 10060. Meckelein, B.; Rohan de Silva, H. A.; Roses, A. D.; Rao, P. N.; Pettenati, M. J.; Xu, P.-T.; Hodge, R.; Gluckman, M. J.; Abraham, C. R.: Human endopeptidase (THOP1) is localized on chromosome 19 within the linkage region for the late-onset Alzheimer disease AD2 locus. *Genomics*

31: 246–249, 1996.

- [36509] 10061.Papastoitsis, G.; Siman, R.; Scott, R.; Abraham, C. R.: Identification of a metalloprotease from Alzheimer's disease brain able to degrade the beta-amyloid precursor protein and generate amyloidogenic fragments. *Biochemistry* 33:192–199, 1994.
- [36510] 10062.Torres, M. P.; Prange, C.; Lennon, G.: Human endopeptidase 24.15 (THOP1) is localized on chromosome 19p13.3 and is excluded from the linkage region for late-onset Alzheimer disease. *Genomics* 53: 239–240, 1998.
- [36511] 10063.Bram, R. J.; Crabtree, G. R.: Calcium signalling in T cells stimulated by a cyclophilin B-binding protein. *Nature* 371: 355–358, 1994.
- [36512] 10064.Bram, R. J.; Valentine, V.; Shapiro, D. N.; Jenkins, N. A.; Gilbert, D. J.; Copeland, N. G.: The gene for calcium-modulating cyclophilin ligand (CAMLG) is located on human chromosome 5q23 and a syntenic region of mouse chromosome 13. *Genomics* 31: 257–260, 1996.
- [36513] 10065.Bussemakers, M. J. G.; van Bokhoven, A.; Voller, M.; Smit, F. P.; Schalken, J. A.: The genes for the calcium-dependent cell adhesion molecules P- and E-cadherin are tandemly arranged in the human genome. *Biochem. Biophys. Res. Commun.* 203: 1291–1294, 1994.

- [36514] 10066.Carmeliet, P.; Lampugnani, M.-G.; Moons, L.; Breviario, F.; Compernelle,V.; Bono, F.; Balconi, G.; Spagnuolo, R.; Oosthuyse, B.; Dewerchin,M.; Zanetti, A.; Angelillo, A.; and 11 others: Targeted deficiencyof cytosolic truncation of the VE-cadherin gene in mice impairs VEGF-mediatedendothelial survival and angiogenesis. *Cell* 98: 147–157, 1999.
- [36515] 10067.Huber, P.; Dalmon, J.; Engiles, J.; Breviario, F.; Gory, S.; Siracusa,L. D.; Buchberg, A. M.; Dejana, E.: Genomic structure and chromosomal mapping of the mouse VE-cadherin gene (*Cdh5*). *Genomics* 32: 21–28,1996.
- [36516] 10068.Salomon, D.; Ayalon, O.; Patel-King, R.; Hynes, R. O.; Geiger,B.: Extrajunctional distribution of N-cadherin in cultured humanendothelial cells. *J. Cell Sci.* 102: 7–17, 1992.
- [36517] 10069.Carmeliet, P.; Moons, L.; Luttun, A.; Vincenti, V.; Compernelle,V.; De Mol, M.; Wu, Y.; Bono, F.; Devy, L.; Beck, H.; Scholz, D.;Acker, T.; and 17 others: Synergism between vascular endothelialgrowth factor and placental growth factor contributes to angiogenesisand plasma extravasation in pathological conditions. *Nature Med.* 7:575–583, 2001.
- [36518] 10070.Luttun, A.; Tjwa, M.; Moons, L.; Wu, Y.; Angelillo-

Scherrer, A.; Liao, F.; Nagy, J. A.; Hooper, A.; Priller, J.; De Klerck, B.; Compernelle, V.; Daci, E.; and 10 others: Revascularization of ischemic tissues by PlGF treatment, and inhibition of tumor angiogenesis, arthritis and atherosclerosis by anti-Flt1. *Nature Med.* 8: 831–840, 2002.

[36519] 10071. Maglione, D.; Guerriero, V.; Viglietto, G.; Delli-Bovi, P.; Persico, M. G.: Isolation of a human placenta cDNA coding for a protein related to the vascular permeability factor. *Proc. Nat. Acad. Sci.* 88: 9267–9271, 1991.

[36520] 10072. Mattei, M.-G.; Borg, J.-P.; Rosnet, O.; Marme, D.; Birnbaum, D.: Assignment of vascular endothelial growth factor (VEGF) and placenta growth factor (PLGF) genes to human chromosome 6p12–p21 and 14q24–q31 regions, respectively. *Genomics* 32: 168–169, 1996.

[36521] 10073. Le Coniat, M.; Choi, D.-S.; Maroteaux, L.; Launay, J.-M.; Berger, R.: The 5-HT_{2B} receptor gene maps to 2q36.3–2q37.1. *Genomics* 32: 172–173, 1996.

[36522] 10074. Nebigil, C. G.; Choi, D.-S.; Dierich, A.; Hickel, P.; Le Meur, M.; Messaddeq, N.; Launay, J.-M.; Maroteaux, L.: Serotonin 2B receptor is required for heart development. *Proc. Nat. Acad. Sci.* 97: 9508–9513, 2000.

[36523] 10075. Goshima, Y.; Nakamura, F.; Strittmatter, P.; Strittmatter, S. M.: Collapsin-induced growth cone col-

lapse mediated by an intracellular protein related to UNC-33. *Nature* 376: 509–514, 1995.

- [36524] 10076. Charrin, S.; Le Naour, F.; Oualid, M.; Billard, M.; Faure, G.; Hanash, S. M.; Boucheix, C.; Rubinstein, E.: The major CD9 and CD81 molecular partner: identification and characterization of the complexes. *J. Biol. Chem.* 276: 14329–14337, 2001.
- [36525] 10077. Nagase, T.; Kikuno, R.; Ishikawa, K.; Hirosawa, M.; Ohara, O.: Prediction of the coding sequences of unidentified human genes. XVI. The complete sequences of 150 new cDNA clones from brain which code for large proteins in vitro. *DNA Res.* 7: 65–73, 2000.
- [36526] 10078. Orlicky, D. J.; Berry, R.; Sikela, J. M.: Human chromosome 1 localization of the gene for a prostaglandin F₂-alpha receptor negative regulatory protein. *Hum. Genet.* 97: 655–658, 1996.
- [36527] 10079. Stipp, C. S.; Orlicky, D.; Hemler, M. E.: FPRP, a major, highly stoichiometric, highly specific CD81- and CD9-associated protein. *J. Biol. Chem.* 276: 4853–4862, 2001.
- [36528] 10080. Oliver, G.; Wehr, R.; Jenkins, N. A.; Copeland, N. G.; Cheyette, B. N. R.; Hartenstein, V.; Zipursky, S. L.; Gruss, P.: Homeobox genes and connective tissue patterning. De-

velopment 121: 693–705, 1995.

- [36529] 10081.Casellas, R.; Jankovic, M.; Meyer, G.; Gazumyan, A.; Luo, Y.; Roeder, R. G.; Nussenzweig, M. C.: OcaB is required for normal transcription and V(D)J recombination of a subset of immunoglobulin kappa genes. *Cell* 110:575–585, 2002.
- [36530] 10082.Gstaiger, M.; Knoepfel, L.; Georgiev, O.; Schaffner, W.; Hovens, C. M.: A B-cell coactivator of octamer-binding transcription factors. *Nature* 373:360–362, 1995.
- [36531] 10083.Junker, S.; Brondum-Nielsen, K.; Newell, J. W.; Matthias, P.; Tommerup, N.: Assignment of the human gene for Oct-binding factor-1 (OBF1), a B-cell-specific coactivator of octamer-binding transcription factors 1 and 2, to 11q23.1 by somatic cell hybridization and in situ hybridization. *Genomics* 33:143–145, 1996.
- [36532] 10084.Urness, L. D.; Sorensen, L. K.; Li, D. Y.: Arteriovenous malformations in mice lacking activin receptor-like kinase-1. *Nature Genet.* 26:328–331, 2000.
- [36533] 10085.Jantti, J.; Lahdenranta, J.; Olkkonen, V. M.; Soderlund, H.; Keranen, S.: SEM1, a homologue of the split hand/split foot malformation candidate gene Dss1, regulates exocytosis and pseudohyphal differentiation in yeast. *Proc. Nat. Acad. Sci.* 96: 909–914, 1999.

- [36534] 10086.Han, L. Wong, D.; Dhaka, A.; Afar, D.; White, M.; Xie, W.; Herschman,H.; Witte, O.; Colicelli, J.: Protein binding and signaling properties of RIN1 suggest a unique effector function. *Proc. Nat. Acad. Sci.* 94:4954–4959, 1997.
- [36535] 10087.Leffers, H.; Madsen, P.; Rasmussen, H. H.; Honore, B.; Andersen,A. H.; Walbum, E.; Vandekerckhove, J.; Celis, J. E.: Molecular cloning and expression of the transformation sensitive epithelial marker stratifin:a member of a protein family that has been involved in the protein kinase C signalling pathway. *J. Molec. Biol.* 231: 982–998, 1993.
- [36536] 10088.Jaspers, M.; Marynen, P.; Aly, M. S.; Cuppens, H.; Hilliker, C.;Cassiman, J.–J.: Localization of the gene encoding the alpha–2 subunit of the human VLA–2 receptor to chromosome 5q23–31. *Somat. Cell Molec.Genet.* 17: 505–511, 1991.
- [36537] 10089.Hammond, J. W.; Potter, M.; Wilcken, B.; Truscott, R.: Siblings with gamma–glutamyltransferase deficiency. *J. Inherit. Metab. Dis.* 18:82–83, 1995.
- [36538] 10090.Heisterkamp, N.; Groffen, J.: Duplication of the bcr and gamma–glutamyltranspeptidase genes. *Nucleic Acids Res.* 16: 8045–8056, 1988.
- [36539] 10091.Laperche, Y.; Bulle, F.; Aissani, T.; Chobert, M. N.; Aggerbeck,M.; Hanoune, J.; Guellaen, G.: Molecular

cloning and nucleotide sequence of rat kidney gamma-glutamyl transpeptidase cDNA. *Proc. Nat. Acad. Sci.* 83: 937–941, 1986.

[36540] 10092. O'Daley, S.: An abnormal sulphhydryl compound in urine. (Abstract) *Irish J. Med. Sci.* 7: 578–579, 1968.

[36541] 10093. Pawlak, A.; Lahuna, O.; Bulle, F.; Suzuki, A.; Ferry, N.; Siegrist, S.; Chikhi, N.; Chobert, M. N.; Guellaen, G.; Laperche, Y.: Gamma-glutamyl transpeptidase: a single copy gene in the rat and a multigene family in the human genome. *J. Biol. Chem.* 263: 9913–9916, 1988.

[36542] 10094. Rouleau, G. A.; Bazanowski, A.; Cohen, E. H.; Guellaen, G.; Gusella, J. F.: Gamma-glutamyl transferase locus (GGT) displays a PvuII polymorphism. *Nucleic Acids Res.* 16: 11848 only, 1988.

[36543] 10095. Sakamuro, D.; Yamazoe, M.; Matsuda, Y.; Kangawa, K.; Taniguchi, N.; Matsuo, H.; Yoshikawa, H.; Ogasawara, N.: The primary structure of human gamma-glutamyl transpeptidase. *Gene* 73: 1–9, 1988.

[36544] 10096. Schulman, J. D.; Goodman, S. I.; Mace, J. W.; Patrick, A. D.; Tietze, F.; Butler, E. J.: Glutathionuria: inborn error of metabolism due to tissue deficiency of gamma-glutamyl transpeptidase. *Biochem. Biophys. Res. Commun.* 65: 68–74, 1975.

- [36545] 10097. Annabi, B.; Hiraiwa, H.; Mansfield, B. C.; Lei, K.-J.; Ubagai, T.; Polymeropoulos, M. H.; Moses, S. W.; Parvari, R.; HersHKovitz, E.; Mandel, H.; Fryman, M.; Chou, J. Y.: The gene for glycogen-storage disease type 1b maps to chromosome 11q23. *Am. J. Hum. Genet.* 62:400–405, 1998.
- [36546] 10098. Jaeken, J.; Goemans, N.; Fryns, J.-P.; Francois, I.; de Zegher, F.: Association of hyperprolinaemia type I and heparin cofactor II deficiency with CATCH 22 syndrome: evidence for a contiguous gene syndrome locating the proline oxidase gene. *J. Inherit. Metab. Dis.* 19:275–277, 1996.
- [36547] 10099. Harvey, C. B.; Hollox, E. J.; Poulter, M.; Wang, Y.; Rossi, M.; Auricchio, S.; Iqbal, T. H.; Cooper, B. T.; Barton, R.; Sarner, M.; Korpela, R.; Swallow, D. M.: Lactase haplotype frequencies in Caucasians: association with the lactase persistence/non-persistence polymorphism. *Ann. Hum. Genet.* 62: 215–223, 1998.
- [36548] 10100. Harvey, C. B.; Pratt, W. S.; Islam, I.; Whitehouse, D. B.; Swallow, D. M.: DNA polymorphisms in the lactase gene: linkage disequilibrium across the 70-kb region. *Europ. J. Hum. Genet.* 3: 27–41, 1995.
- [36549] 10101. Hollox, E. J.; Poulter, M.; Zvarik, M.; Ferak, V.; Krause, A.; Jenkins, T.; Saha, N.; Kozlov, A. I.; Swallow, D.

M.: Lactase haplotypediversity in the Old World. *Am. J. Hum. Genet.* 68: 160–172, 2001.

- [36550] 10102.Murano, S.; Thweatt, R.; Reis, R. J. S.; Jones, R. A.; Moerman,E. J.; Goldstein, S.: Diverse gene sequences are overexpressed inWerner syndrome fibroblasts undergoing premature replicative senescence. *Molec.Cell. Biol.* 11: 3905–3914, 1991.
- [36551] 10103.Li, S.; Strelow, A.; Fontana, E. J.; Wesche, H.: IRAK–4: a novelmember of the IRAK family with the properties of an IRAK–kinase. *Proc.Nat. Acad. Sci.* 99: 5567–5572, 2002.
- [36552] 10104.Scanlan, M. J.; Gordon, J. D.; Williamson, B.; Stock–ert, E.; Bander,N. H.; Jongeneel, V.; Gure, A. O.; Jager, D.; Jager, E.; Knuth, A.;Chen, Y.–T.; Old, L. J.: Antigens recog–nized by autologous antibodyin patients with renal–cell carcinoma. *Int. J. Cancer* 83: 456–464,1999.
- [36553] 10105.Scott, A. F.: Personal Communication. Baltimore, Md. 4/25/2002.
- [36554] 10106.Suzuki, N.; Suzuki, S.; Duncan, G. S.; Millar, D. G.; Wada, T.;Mirtsos, C.; Takada, H.; Wakeham, A.; Itie, A.; Li, S.; Penninger,J. M.; Wesche, H.; Ohashi, P. S.; Mak, T. W.; Yeh, W.–C.: Severeimpairment of interleukin–1 and Toll–like receptor signalling in micelacking IRAK–4. *Nature*

416: 750–754, 2002.

[36555] 10107. Pennarun, G.; Chapelin, C.; Escudier, E.; Bridoux, A.-M.; Dastot, F.; Cacheux, V.; Goossens, M.; Amselem, S.; Duriez, B.: The human dynein intermediate chain 2 gene (DNAI2): cloning, mapping, expression pattern, and evaluation as a candidate for primary ciliary dyskinesia.

Hum. Genet. 107: 642–649, 2000.

[36556] 10108. Amerik, A. Y.; Petukhova, G. V.; Grigorenko, V. G.; Lykov, I. P.; Yarovoi, S. V.; Lipkin, V. M.; Gorbalenya, A. E.: Cloning and sequence analysis of cDNA for a human homolog of eubacterial ATP-dependent Lon proteases. FEBS Lett. 340: 25–28, 1994.

[36557] 10109. Wang, N.; Gottesman, S.; Willingham, M. C.; Gottesman, M. M.; Maurizi, M. R.: A human mitochondrial ATP-dependent protease that is highly homologous to bacterial Lon protease. Proc. Nat. Acad. Sci. 90: 11247–11251, 1993.

[36558] 10110. Popovici, C.; Mattei, M.-G.; Rattner, J. B.; Birnbaum, D.; Pebusque, M.-J.: Assignment of the centrosomal protein 110 gene (Cep110) to mouse chromosome bands 2B–C1 by in situ hybridization. Cytogenet. Cell Genet. 89: 216–217, 2000.

[36559] 10111. Castagnola, P.; Gennari, M.; Morello, R.; Tonachini,

L.; Marin, O.; Gaggero, A.; Cancedda, R.: Cartilage associated protein (CASP) is a novel developmentally regulated chick embryo protein. *J. Cell Sci.* 110: 1351–1359, 1997.

[36560] 10112. Morello, R.; Tonachini, L.; Monticone, M.; Viggiano, L.; Rocchi, M.; Cancedda, R.; Castagnola, P.: cDNA cloning, characterization and chromosome mapping of Crtp encoding the mouse cartilage associated protein. *Matrix Biol.* 18: 319–324, 1999.

[36561] 10113. Tonachini, L.; Morello, R.; Monticone, M.; Skaug, J.; Scherer, S. W.; Cancedda, R.; Castagnola, P.: cDNA cloning, characterization and chromosome mapping of the gene encoding human cartilage associated protein (CRTAP). *Cytogenet. Cell Genet.* 87: 191–194, 1999.

[36562] 10114. Shibamura, M.; Mashimo, J.; Mita, A.; Kuroki, T.; Nose, K.: Cloning from a mouse osteoblastic cell line of a set of transforming-growth-factor-beta-1-regulated genes, one of which seems to encode a follistatin-related polypeptide. *Europ. J. Biochem.* 217: 13–19, 1993.

[36563] 10115. Tanaka, M.; Ozaki, S.; Osakada, F.; Mori, K.; Okubo, M.; Nakao, K.: Cloning of follistatin-related protein as a novel autoantigen in systemic rheumatic diseases. *Int. Immun.* 10: 1305–1314, 1998.

[36564] 10116. Zwijsen, A.; Blockx, H.; van Arnhem, W.; Willems, J.;

Fransen,L.; Devos, K.; Raymackers, J.; van de Voorde, A.; Slegers, H.: Characterization of a rat C6 glioma–secreted follistatin–related protein (FRP): cloning and sequence of the human homologue. *Europ. J. Biochem.* 225: 937–946,1994.

[36565] 10117.Fang, M.; Jaffrey, S. R.; Sawa, A.; Ye, K.; Luo, X.; Snyder, S.H.: Dexas1: a G protein specifically coupled to neuronal nitric oxide synthase via CAPON. *Neuron* 28: 183–193, 2000.

[36566] 10118.Kemppainen, R. J.; Behrend, E. N.: Dexamethasone rapidly induces a novel Ras superfamily member–related gene in AtT–20 cells. *J. Biol.Chem.* 273: 3129–3131, 1998.

[36567] 10119.Tu, Y.; Wu, C.: Cloning, expression and characterization of a novel human Ras–related protein that is regulated by glucocorticoid hormone. *Biochim. Biophys. Acta* 1489: 452–456, 1999.

[36568] 10120.Jaffrey, S. R.; Snowman, A. M.; Eliasson, M. J. L.; Cohen, N. A.; Snyder, S. H.: CAPON: a protein associated with neuronal nitric oxide synthase that regulates its interactions with PSD95. *Neuron* 115–124,1998.

[36569] 10121.Thuresson, A.–C.; Astrom, J.; Astrom, A.; Gronvik, K.–O.; Virtanen,A.: Multiple forms of poly(A) polymerases

in human cells. *Proc.Nat. Acad. Sci.* 91: 979–983, 1994.

- [36570] 10122.Yamauchi, T.; Sugimoto, J.; Hatakeyama, T.; Asakawa, S.; Shimizu,N.; Isobe, M.: Assignment of the human poly(A) polymerase (PAP) gene to chromosome 14q32.1–q32.2 and isolation of a polymorphic CA repeat-sequence. *J. Hum. Genet.* 44: 253–255, 1999.
- [36571] 10123.Boles, K. S.; Nakajima, H.; Colonna, M.; Chuang, S. S.; Stepp,S. E.; Bennett, M.; Kumar, V.; Mathew, P. A.: Molecular characterization of a novel human natural killer cell receptor homologous to mouse 2B4. *Tissue Antigens* 54: 27–34, 1999.
- [36572] 10124.Fort, M. M.; Cheung, J.; Yen, D.; Li, J.; Zurawski, S. M.; Lo,S.; Menon, S.; Clifford, T.; Hunte, B.; Lesley, R.; Muchamuel, T.;Hurst, S. D.; Zurawski, G.; Leach, M. W.; Gorman, D. M.; Rennick,D. M.: IL-25 induces IL-4, IL-5, and IL-13 and Th2-associated pathologies in vivo. *Immunity* 15: 985–995, 2001.
- [36573] 10125.Scott, A. F.: Personal Communication. Baltimore, Md. 2/16/2001.
- [36574] 10126.Mori, T.; Fukuda, Y.; Kuroda, H.; Matsumura, T.; Ota, S.; Sugimoto,T.; Nakamura, Y.; Inazawa, J.: Cloning and characterization of a novel Rab-family gene, Rab36, within the region at 22q11.2 that is homozygously deleted

in malignant rhabdoid tumors. Biochem. Biophys. Res. Commun. 254: 594–600, 1999.

[36575] 10127. Zhou, J.-Y.; Fogelgren, B.; Wang, Z.; Roe, B. A.; Biegel, J. A.: Isolation of genes from the rhabdoid tumor deletion region in chromosome band 22q11.2. Gene 241: 133–141, 2000.

[36576] 10128. Sood, R.; Makalowska, I.; Carpten, J. D.; Robbins, C. M.; Stephan, D. A.; Connors, T. D.; Morgenbesser, S. D.; Su, K.; Pinkett, H. W.; Graham, C. L.; Quesenberry, M. I.; Baxevanis, A. D.; Klinger, K. W.; Trent, J. M.; Bonner, T. I.: The human RGL (RalGDS-like) gene: cloning, expression analysis and genomic organization. Biochim. Biophys. Acta 1491: 285–288, 2000.

[36577] 10129. Acquati, F.; Accarino, M.; Nucci, C.; Fumagalli, P.; Jovine, L.; Ottolenghi, S.; Taramelli, R.: The gene encoding DRAP (BACE2), a glycosylated transmembrane protein of the aspartic protease family, maps to the Down syndrome critical region. FEBS Lett. 468: 59–64, 2000.

[36578] 10130. Bennett, B. D.; Babu-Khan, S.; Loeloff, R.; Louis, J.-C.; Curran, E.; Citron, M.; Vassar, R.: Expression analysis of BACE2 in brain and peripheral tissues. J. Biol. Chem. 275: 20647–20651, 2000.

[36579] 10131. Saunders, A. J.; Kim, T.-W.; Tanzi, R. E.: BACE maps

to chromosome 11 and a BACE homolog, BACE2, reside in the obligate Down syndrome region of chromosome 21. Science 286: 1255A only, 1999.

- [36580] 10132. Solans, A.; Estivill, X.; de la Luna, S.: A new aspartyl protease on 21q22.3, BACE2, is highly similar to Alzheimer's amyloid precursor protein beta-secretase. Cytogenet. Cell Genet. 89: 177–184, 2000.
- [36581] 10133. Xin, H.; Stephans, J. C.; Duan, X.; Harrowe, G.; Kim, E.; Grieshammer, U.; Kingsley, C.; Giese, K.: Identification of a novel aspartic-like protease differentially expressed in human breast cancer cell lines. Biochim. Biophys. Acta 1501: 125–137, 2000.
- [36582] 10134. Kuang, W. W.; Thompson, D. A.; Hoch, R. V.; Weigel, R. J.: Differential screening and suppression subtractive hybridization identified genes differentially expressed in an estrogen receptor-positive breast carcinoma cell line. Nucleic Acids Res. 26: 1116–1123, 1998.
- [36583] 10135. Adamec, J.; Rusnak, F.; Owen, W. G.; Naylor, S.; Benson, L. M.; Gacy, A. M.; Isaya, G.: Iron-dependent self-assembly of recombinant yeast frataxin: implications for Friedreich ataxia. Am. J. Hum. Genet. 67: 549–562, 2000.
- [36584] 10136. Campuzano, V.; Montermini, L.; Lutz, Y.; Cova, L.; Hindelang, C.; Jiralerspong, S.; Trottier, Y.; Kish, S. J.;

Faucheux, B.; Trouillas, P.; Authier, F. J.; Durr, A.; Mandel, J.-L.; Vescovi, A.; Pandolfo, M.; Koenig, M.: Frataxin is reduced in Friedreich ataxia patients and is associated with mitochondrial membranes. *Hum. Molec. Genet.* 6:1771–1780, 1997.

[36585] 10137. Cavadini, P.; O'Neill, H. A.; Benada, O.; Isaya, G.: Assembly and iron-binding properties of human frataxin, the protein deficient in Friedreich ataxia. *Hum. Molec. Genet.* 11: 217–227, 2002.

[36586] 10138. Chamberlain, S.; Pook, M.; Carvajal, J.; Doudney, K.; Hillermann, R.: Frataxin fracas. (Letter) *Nature Genet.* 15: 337–338, 1997.

[36587] 10139. Cossee, M.; Campuzano, V.; Koutnikova, H.; Fischbeck, K.; Mandel, J.-L.; Koenig, M.; Bidichandani, S. I.; Patel, P. I.; Molte, M. D.; Canizares, J.; De Frutos, R.; Piane, L.; Cavalcanti, F.; Monticelli, A.; Coccozza, S.; Monteneri, L.; Pandolfo, M.: Frataxin fracas. (Letter) *Nature Genet.* 15: 337–338, 1997.

[36588] 10140. Bartles, J. R.; Wierda, A.; Zheng, L.: Identification and characterization of espin, an actin-binding protein localized to the F-actin-rich junctional plaques of Sertoli cell ectoplasmic specializations. *J. Cell Sci.* 109:1229–1239, 1996.

- [36589] 10141. Bartles, J. R.; Zheng, L.; Li, A.; Wierda, A.; Chen, B.: Smallespin: a third actin-bundling protein and potential forked protein ortholog in brush border microvilli. *J. Cell Biol.* 143: 107–119, 1998.
- [36590] 10142. Chen, B.; Li, A.; Wang, D.; Wang, M.; Zheng, L.; Bartles, J. R.: Espin contains an additional actin-binding site in its N terminus and is a major actin-bundling protein of the Sertoli cell-spermatid ectoplasmic specialization junctional plaque. *Molec. Biol. Cell* 10:4327–4339, 1999.
- [36591] 10143. Zheng, L.; Sekerkova, G.; Vranich, K.; Tilney, L. G.; Mugnaini, E.; Bartles, J. R.: The deaf jerker mouse has a mutation in the gene encoding the espin actin-bundling proteins of hair cell stereocilia and lacks espins. *Cell* 102: 377–385, 2000.
- [36592] 10144. Illarioshkin, S. N.; Ivanova-Smolenskaya, I. A.; Tanaka, H.; Poleshchuk, V. V.; Markova, E. D.; Tsuji, S.: Refined genetic location of the chromosome 2p-linked progressive muscular dystrophy gene. *Genomics* 42:345–348, 1997.
- [36593] 10145. Illarioshkin, S. N.; Ivanova-Smolenskaya, I. A.; Tanaka, H.; Vereshchagin, N. V.; Markova, E. D.; Poleshchuk, V. V.; Lozhnikova, S. M.; Sukhorukov, V. S.; Limborska, S. A.; Slominsky, P. A.; Bulayeva, K. B.; Tsuji, S.:

Clinical and molecular analysis of a large family with three distinct phenotypes of progressive muscular dystrophy. *Brain* 119:1895–1909, 1996.

[36594] 10146. McNally, E. M.; Ly, C. T.; Rosenmann, H.; Rosenbaum, S. M.; Jiang, W.; Anderson, L. V. B.; Soffer, D.; Argov, Z.: Splicing mutation in dysferlin produces limb-girdle muscular dystrophy with inflammation. *Am. J. Med. Genet.* 91: 305–312, 2000.

[36595] 10147. Weiler, T.; Greenberg, C. R.; Nylen, E.; Halliday, W.; Morgan, K.; Eggertson, D.; Wrogemann, K.: Limb-girdle muscular dystrophy and Miyoshi myopathy in an aboriginal Canadian kindred map to LGMD2B and segregate with the same haplotype. *Am. J. Hum. Genet.* 59: 872–878, 1996.

[36596] 10148. Crosbie, R. H.; Lim, L. E.; Moore, S. A.; Hirano, M.; Hays, A. P.; Maybaum, S. W.; Collin, H.; Dovico, S. A.; Stolle, C. A.; Fardeau, M.; Tome, F. M. S.; Campbell, K. P.: Molecular and genetic characterization of sarcospan: insights into sarcoglycan-sarcospan interactions. *Hum. Molec. Genet.* 9: 2019–2027, 2000.

[36597] 10149. Augustin, I.; Betz, A.; Herrmann, C.; Jo, T.; Brose, N.: Differential expression of two novel Munc13 proteins in rat brain. *Biochem. J.* 337:363–371, 1999.

[36598] 10150. Augustin, I.; Rosenmund, C.; Sudhof, T. C.; Brose,

N.: Munc13-1 is essential for fusion competence of glutamatergic synaptic vesicles. *Nature* 400:457–461, 1999.

- [36599] 10151. Rhee, J.-S.; Betz, A.; Pyott, S.; Reim, K.; Varoqueaux, F.; Augustin, I.; Hesse, D.; Sudhof, T. C.; Takahashi, M.; Rosenmund, C.; Brose, N.: Beta phorbol ester- and diacylglycerol-induced augmentation of transmitter release is mediated by Munc13s and not by PKCs. *Cell* 108:121–133, 2002.
- [36600] 10152. Rosenmund, C.; Sigler, A.; Augustin, I.; Reim, K.; Brose, N.; Rhee, J.-S.: Differential control of vesicle priming and short-term plasticity by Munc13 isoforms. *Neuron* 33: 411–424, 2002.
- [36601] 10153. Song, Y.; Ailenberg, M.; Silverman, M.: Cloning of a novel gene in the human kidney homologous to rat munc13s: its potential role in diabetic nephropathy. *Kidney Int.* 53: 1689–1695, 1998.
- [36602] 10154. Ji, Y.; Walkowicz, M. J.; Buiting, K.; Johnson, D. K.; Tarvin, R. E.; Rinchik, E. M.; Horsthemke, B.; Stubbs, L.; Nicholls, R. D.: The ancestral gene for transcribed, low-copy repeats in the Prader-Willi/Angelman region encodes a large protein implicated in protein trafficking, which is deficient in mice with neuromuscular and spermiogenic abnormalities. *Hum. Molec. Genet.* 8: 533–542, 1999.

- [36603] 10155.Manda, R.; Kohno, T.; Matsuno, Y.; Takenoshita, S.; Kuwano, H.;Yokota, J.: Identification of genes (SPON2 and C20orf2) differentiallyexpressed between cancerous and noncancerous lung cells by mRNA differentialdisplay. Genomics 61: 5–14, 1999.
- [36604] 10156.Zhang, Y.; Heidebrecht, H.–J.; Rott, A.; Schlegelberger, B.; Parwaresch,R.: Assignment of human proliferation associated p100 gene (C20orf1)to human chromosome band 20q11.2 by in situ hybridization. Cytogenet.Cell Genet. 84: 182–183, 1999.
- [36605] 10157.Parker, N. J.; Begley, C. G.; Smith, P. J.; Fox, R. M.: Molecularcloning of a novel human gene (D11S4896E) at chromosomal region 11p15.5. Genomics 37:253–256, 1996.
- [36606] 10158.Sabbioni, S.; Veronese, A.; Trubia, M.; Taramelli, R.; Barbanti–Brodano,G.; Croce, C. M.; Negrini, M.: Exon structure and promoter identificationof STIM1 (alias GOK), a human gene causing growth arrest of the humantumor cell lines G401 and RD. Cytogenet. Cell. Genet. 86: 214–218,1999.
- [36607] 10159.Boudin, H.; Doan, A.; Xia, J.; Shigemoto, R.; Huganir, R. L.; Worley,P.; Craig, A. M.: Presynaptic clustering of mGluR7a requires thePICK1 PDZ domain binding

site. *Neuron* 28: 485–497, 2000.

[36608] 10160.Dev, K. K.; Nishimune, A.; Henley, J. M.; Nakanishi, S.: The proteinkinase C- α binding protein PICK1 interacts with short but not long form alternative splice variants of AMPA receptor subunits. *Neuropharmacology* 38:635–644, 1999.

[36609] 10161.Staudinger, J.; Zhou, J.; Burgess, R.; Elledge, S. J.; Olson, E.N.: PICK1: a perinuclear binding protein and substrate for proteinkinase C isolated by the yeast two-hybrid system. *J. Cell Biol.* 128:263–271, 1995.

[36610] 10162.Takeya, R.; Takeshige, K.; Sumimoto, H.: Interaction of the PDZ domain of human PICK1 with class I ADP-ribosylation factors. *Biochem.Biophys. Res. Commun.* 267: 149–155, 2000.

[36611] 10163.Xia, J.; Zhang, X.; Staudinger, J.; Huganir, R. L.: Clustering of AMPA receptors by the synaptic PDZ domain-containing protein PICK1. *Neuron* 22:179–187, 1999.

[36612] 10164.Blencowe, B. J.; Bauren, G.; Eldridge, A. G.; Issner, R.; Nickerson, J. A.; Rosonina, E.; Sharp, P. A.: The SRm160/300 splicing coactivator subunits. *RNA* 6: 111–120, 2000.

[36613] 10165.Sawada, Y.; Miura, Y.; Umeki, K.; Tamaoki, T.; Fujinaga, K.; Ohtaki, S.: Cloning and characterization of a

novel RNA-binding protein SRL300 with RS domains.

Biochim. Biophys. Acta 1492: 191–195, 2000.

- [36614] 10166. Hirose, K.; Morita, M.; Ema, M.; Mimura, J.; Hamada, H.; Fujii, H.; Saijo, Y.; Gotoh, O.; Sogawa, K.; Fujii-Kuriyama, Y.: cDNA cloning and tissue-specific expression of a novel basic helix-loop-helix/PAS factor (Arnt2) with close sequence similarity to the aryl hydrocarbon receptor nuclear translocator (Arnt). *Molec. Cell. Biol.* 16: 1706–1713, 1996.
- [36615] 10167. Keith, B.; Adelman, D. M.; Simon, M. C.: Targeted mutation of the murine aryl hydrocarbon receptor nuclear translocator 2 (Arnt2) gene reveals partial redundancy with Arnt. *Proc. Nat. Acad. Sci.* 98:6692–6697, 2001.
- [36616] 10168. Michaud, J. L.; DeRossi, C.; May, N. R.; Holdener, B. C.; Fan, C.-M.: ARNT2 acts as the dimerization partner of SIM1 for the development of the hypothalamus. *Mech. Dev.* 90: 253–261, 2000.
- [36617] 10169. Wende, H.; Volz, A.; Ziegler, A.: Extensive gene duplications and a large inversion characterize the human leukocyte receptor cluster. *Immunogenetics* 51:703–713, 2000.
- [36618] 10170. Fukunaga-Johnson, N.; Lee, S. W.; Liebert, M.; Grossman, H. B.: Molecular analysis of a gene, BB1, over-

expressed in bladder and breast carcinoma. *Anticancer Res.* 16: 1085–1090, 1996.

- [36619] 10171. Saito, H.; Papaconstantinou, J.; Sato, H.; Goldstein, S.: Regulation of a novel gene encoding a lysyl oxidase-related protein in cellular adhesion and senescence. *J. Biol. Chem.* 272: 8157–8160, 1997.
- [36620] 10172. Hsu, S. Y.; Liang, S.-G.; Hsueh, A. J. W.: Characterization of two LGR genes homologous to gonadotropin and thyrotropin receptors with extracellular leucine-rich repeats and a G protein-coupled, seven-transmembrane region. *Molec. Endocr.* 12: 1830–1845, 1998.
- [36621] 10173. Loh, E. D.; Broussard, S. R.; Kolakowski, L. F.: Molecular characterization of a novel glycoprotein hormone G-protein-coupled receptor. *Biochem. Biophys. Res. Commun.* 282: 757–764, 2001.
- [36622] 10174. Loh, E. D.; Broussard, S. R.; Liu, Q.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Kolakowski, L. F., Jr. Chromosomal localization of GPR48, a novel glycoprotein hormone receptor like GPCR, in human and mouse with radiation hybrid and interspecific backcross mapping. *Cytogenet. Cell Genet.* 89: 2–5, 2000.
- [36623] 10175. Giffon, T.; Lepourcelet, M.; Pichon, L.; Jezequel, P.;

Bouric,P.; Carn, G.; Pontarotti, P.; Le Gall, J.-Y.; David, V.: Cloning of a human homologue of the mouse Tctex-5 gene within the MHC class I region. *Immunogenetics* 44: 331-339, 1996.

[36624] 10176. Lepourcelet, M.; Andrieux, N.; Giffon, T.; Pichon, L.; Hampe, A.; Galibert, F.; Mosser, J.: Systematic sequencing of the human HLA-A/HLA-F region: establishment of a cosmid contig and identification of a new gene cluster within 37 kb of sequence. *Genomics* 37: 316-326, 1996.

[36625] 10177. Zhang, J.; Zhang, L.; Zhao, S.; Lee, E. Y. C.: Identification and characterization of the human HCG V gene product as a novel inhibitor of protein phosphatase-1. *Biochemistry* 37: 16728-16734, 1998.

[36626] 10178. Hayakawa, A.; Matsuda, Y.; Daibata, M.; Nakamura, H.; Sano, K.: Genomic organization, tissue expression, and cellular localization of AF3p21, a fusion partner of MLL in therapy-related leukemia. *Genes Chromosomes Cancer* 30: 364-374, 2001.

[36627] 10179. Sano, K.; Hayakawa, A.; Piao, J.-H.; Kosaka, Y.; Nakamura, H.: Novel SH3 protein encoded by the AF3p21 gene is fused to the mixed lineage leukemia protein in a therapy-related leukemia with t(3;11)(p21;q23). *Blood* 95:1066-1068, 2000.

- [36628] 10180. Alagramam, K. N.; Murcia, C. L.; Kwon, H. Y.; Pawlowski, K. S.; Wright, C. G.; Woychik, R. P.: The mouse Ames waltzer hearing-loss mutant is caused by mutation of Pcdh15, a novel protocadherin gene. *Nature Genet.* 27: 99–102, 2001.
- [36629] 10181. Alagramam, K. N.; Yuan, H.; Kuehn, M. H.; Murcia, C. L.; Wayne, S.; Srisailpathy, C. R. S.; Lowry, R. B.; Knaus, R.; Van Laer, L.; Bernier, F. P.; Schwartz, S.; Lee, C.; Morton, C. C.; Mullins, R. F.; Ramesh, A.; Van Camp, G.; Hagemen, G. S.; Woychik, R. P.; Smith, R. J. H.: Mutations in the novel protocadherin PCDH15 cause Usher syndrome type 1F. *Hum. Molec. Genet.* 10: 1709–1718, 2001.
- [36630] 10182. Cao, H.; Hegele, R. A.: Identification of single-nucleotide polymorphisms in the human LPIN1 gene. *J. Hum. Genet.* 47: 370–372, 2002.
- [36631] 10183. Peterfy, M.; Phan, J.; Xu, P.; Reue, K.: Lipodystrophy in the fld mouse results from mutation of a new gene encoding a nuclear protein, lipin. *Nature Genet.* 27: 121–124, 2001.
- [36632] 10184. Reue, K.; Xu, P.; Wang, X.-P.; Slavin, B. G.: Adipose tissue deficiency, glucose intolerance, and increased atherosclerosis result from mutation in the mouse fatty liver dystrophy (fld) gene. *J. Lipid Res.* 41: 1067–1076,

2000.

- [36633] 10185.Shakhov, A. N.; Rubtsov, A. V.; Lyakhov, I. G.; Tumanov, A. V.;Nedospasov, S. A.: SPLASH (PLA(2)IID), a novel member of phospholipaseA2 family, is associated with lymphotoxin-deficiency. *Genes Immun.* 1:191–199, 2000.
- [36634] 10186.Bodine, S. C.; Latres, E.; Baumhueter, S.; Lai, V. K.–M.; Nunez,L.; Clarke, B. A.; Poueymirou, W. T.; Panaro, F. J.; Na, E.; Dharmarajan,K.; Pan, Z.–Q.; Valenzuela, D. M.; DeChiara, T. M.; Stitt, T. N.;Yancopoulos, G. D.; Glass, D. J.: Identification of ubiquitin ligasesrequired for skeletal muscle atrophy. *Science* 294: 1704–1708, 2001.
- [36635] 10187.Domanski, T. L.; Finta, C.; Halpert, J. R.; Zaphiropoulos, P. G.: cDNA cloning and initial characterization of CYP3A43, a novel humancytochrome P450. *Molec. Pharm.* 59: 386–392, 2001.
- [36636] 10188.Gellner, K.; Eiselt, R.; Hustert, E.; Arnold, H.; Koch, I.; Haberl,M.; Deglmann, C. J.; Burk, O.; Buntefuss, D.; Escher, S.; Bishop,C.; Koebe, H.–G.; Brinkmann, U.; Klenk, H.–P.; Kleine, K.; Meyer,U. A.; Wojnowski, L.: Genomic organization of the human CYP3A locus:identification of a new, inducible CYP3A gene. *Pharmacogenetics* 11:111–121, 2001.

- [36637] 10189. Westlind, A.; Malmebo, S.; Johansson, I.; Otter, C.; Andersson, T. B.; Ingelman-Sundberg, M.; Oscarson, M.: Cloning and tissue distribution of a novel human cytochrome P450 of the CYP3A subfamily, CYP3A43. *Biochem. Biophys. Res. Commun.* 281: 1349–1355, 2001.
- [36638] 10190. Holt, J. R.; Gillespie, S. K. H.; Provance, D. W., Jr.; Shah, K.; Shokat, K. M.; Corey, D. P.; Mercer, J. A.; Gillespie, P. G.: A chemical-genetic strategy implicates myosin-1c in adaptation by hair cells. *Cell* 108:371–381, 2002.
- [36639] 10191. Nakayama, T.; Yaoi, T.; Kuwajima, G.: Localization and subcellular distribution of N-copine in mouse brain. *J. Neurochem.* 72: 373–379, 1999.
- [36640] 10192. Nakayama, T.; Yaoi, Y.; Yasui, M.; Kuwajima, G.: N-copine: a novel two C2-domain-containing protein with neuronal activity-regulated expression. *FEBS Lett.* 428: 80–84, 1998.
- [36641] 10193. Runnels, L. W.; Yue, L.; Clapham, D. E.: The TRPM7 channel is inactivated by PIP(2) hydrolysis. *Nature Cell Biol.* 4: 329–336, 2002.
- [36642] 10194. Scott, A. F.: Personal Communication. Baltimore, Md. 3/8/2001.
- [36643] 10195. Chen, D.; Guo, J.; Miki, T.; Tachibana, M.; Gahl, W. A.: Molecular cloning of two novel rab genes from human

melanocytes. *Gene* 174:129–134, 1996.

- [36644] 10196. Centner, T.; Yano, J.; Kimura, E.; McElhinny, A. S.; Pelin, K.; Witt, C. C.; Bang, M.-L.; Trombitas, K.; Granzier, H.; Gregorio, C.C.; Sorimachi, H.; Labeit, S.: Identification of muscle specific ring finger proteins as potential regulators of the titin kinase domain. *J. Molec. Biol.* 306: 717–726, 2001.
- [36645] 10197. Scott, A. F.: Personal Communication. Baltimore, Md. 7/20/2001.
- [36646] 10198. Kirikoshi, H.; Koike, J.; Sagara, N.; Saitoh, T.; Tokuhara, M.; Tanaka, K.; Sekihara, H.; Hirai, M.; Katoh, M.: Molecular cloning and genomic structure of human Frizzled-3 at chromosome 8p21. *Biochem. Biophys. Res. Commun.* 271: 8–14, 2000.
- [36647] 10199. Sala, C. F.; Formenti, E.; Terstappen, G. C.; Caricasole, A.: Identification, gene structure, and expression of human frizzled-3 (FZD3). *Biochem. Biophys. Res. Commun.* 273: 27–34, 2000.
- [36648] 10200. Eggenschwiler, J. T.; Espinoza, E.; Anderson, K. V.: Rab23 is an essential negative regulator of the mouse Sonic hedgehog signalling pathway. *Nature* 412: 194–198, 2001.
- [36649] 10201. Zhang, Q.-H.; Ye, M.; Wu, X.-Y.; Ren, S.-X.; Zhao,

M.; Zhao, C.-J.; Fu, G.; Shen, Y.; Fan, H.-Y.; Lu, G.; Zhong, M.; Xu, X.-R.; and 9 others: Cloning and functional analysis of cDNAs with open reading frames for 300 previously undefined genes expressed in CD34+ hematopoietic stem/progenitor cells. *Genome Res.* 10: 1546–1560, 2000.

[36650] 10202. Saitoh, T.; Hirai, M.; Katoh, M.: Molecular cloning and characterization of human Frizzled-8 gene on chromosome 10p11.2. *Int. Oncol.* 18:991–996, 2001.

[36651] 10203. Wittenberger, T.; Schaller, H. C.; Hellebrand, S.: An expressed sequence tag (EST) data mining strategy succeeding in the discovery of new G-protein coupled receptors. *J. Molec. Biol.* 307: 799–813, 2001.

[36652] 10204. Communi, D.; Gonzalez, N. S.; Detheux, M.; Brezillon, S.; Lannoy, V.; Parmentier, M.; Boeynaems, J.-M.: Identification of a novel human ADP receptor coupled to G(i). *J. Biol. Chem.* 276: 41479–41485, 2001.

[36653] 10205. Chen, H.; Ross, C. A.; Wang, N.; Huo, Y.; MacKinnon, D. F.; Potash, J. B.; Simpson, S. G.; McMahon, F. J.; DePaulo, J. R., Jr.; McInnis, M. G.: NEDD4L on human chromosome 18q21 has multiple forms of transcripts and is a homologue of the mouse Nedd4-2 gene. *Europ. J. Hum. Genet.* 9:922–930, 2001.

[36654] 10206. Erdeniz, N.; Rothstein, R.: Rsp5, a ubiquitin-pro-

tein ligase, is involved in degradation of the single-stranded-DNA binding protein Rfa1 in *Saccharomyces cerevisiae*. *Molec. Cell. Biol.* 20: 224–232, 2000.

[36655] 10207. Lu, Q. R.; Park, J. K.; Noll, E.; Chan, J. A.; Alberta, J.; Yuk, D.; Alzamora, M. G.; Louis, D. N.; Stiles, C. D.; Rowitch, D. H.; Black, P. M.: Oligodendrocyte lineage genes (OLIG) as molecular markers for human glial brain tumors. *Proc. Nat. Acad. Sci.* 98: 10851–10856, 2001.

[36656] 10208. Riggins, G. J.; Thiagalingam, S.; Rozenblum, E.; Weinstein, C. L.; Kern, S. E.; Hamilton, S. R.; Willson, J. K. V.; Markowitz, S. D.; Kinzler, K. W.; Vogelstein, B.: Mad-related genes in the human. *Nature Genet.* 13: 347–349, 1996.

[36657] 10209. Harteneck, C.; Wedel, B.; Koesling, D.; Malkewitz, J.; Bohme, E.; Schultz, G.: Molecular cloning and expression of a new alpha-subunit of soluble guanylyl cyclase. Interchangeability of the alpha-subunit of the enzyme. *FEBS Lett.* 292: 217–222, 1991.

[36658] 10210. Yu, F.; Warburton, D.; Wellington, S.; Danziger, R. S.: Assignment of GUCIA2, the gene coding for the alpha-2 subunit of soluble guanylyl cyclase, to position 11q21–q22 on human chromosome 11. *Genomics* 33: 334–336, 1996.

[36659] 10211. Abe, K.; Yamamura, K.; Suzuki, M.: Molecular and

embryological characterization of a new transgene-induced null allele of mouse Brachyury locus. *Mammalian Genome* 11: 238–240, 2000.

[36660] 10212. European Consortium on MEN1: Mapping of the gene encoding the β 56-subunit of protein phosphatase 2A (PPP2R5B) to a 0.5-Mb region of chromosome 11q13 and its exclusion as a candidate gene for multiple endocrine neoplasia type 1 (MEN1). *Hum. Genet.* 100: 481–485, 1997.

[36661] 10213. Arsenijevic, D.; Onuma, H.; Pecqueur, C.; Raimbault, S.; Manning, B. S.; Miroux, B.; Couplan, E.; Alves-Guerra, M.-C.; Goubert, M.; Surwit, R.; Bouillard, F.; Richard, D.; Collins, S.; Ricquier, D.: Disruption of the uncoupling protein-2 gene in mice reveals a role in immunity and reactive oxygen species production. *Nature Genet.* 26:435–439, 2000.

[36662] 10214. Bouchard, C.; Perusse, L.; Chagnon, Y. C.; Warden, C.; Ricquier, D.: Linkage between markers in the vicinity of the uncoupling protein 2 gene and resting metabolic rate in humans. *Hum. Molec. Genet.* 6:1887–1889, 1997.

[36663] 10215. Brauner, P.; Nibbelink, M.; Flachs, P.; Vitkova, I.; Kopecky, P.; Mertelíková, I.; Janderová, L.; Penicaud, L.; Casteilla, L.; Plavka, R.; Kopecky, J.: Fast decline of

hematopoiesis and uncoupling protein2 content in human liver after birth: location of the protein in Kupffercells. *Pediat. Res.* 49: 440–447, 2001.

[36664] 10216.Esterbauer, H.; Schneitler, C.; Oberkofler, H.; Ebenbichler, C.; Paulweber, B.; Sandhofer, F.; Ladurner, G.; Hell, E.; Strosberg, A.D.; Patsch, J. R.; Krempler, F.; Patsch, W.: A common polymorphism in the promoter of UCP2 is associated with decreased risk of obesity in middle-aged humans. *Nature Genet.* 28: 178–183, 2001.

[36665] 10217.Fleury, C.; Neverova, M.; Collins, S.; Raimbault, S.; Champigny, O.; Levi-Meyrueis, C.; Bouillaud, F.; Seldin, M. F.; Surwit, R. S.; Ricquier, D.; Warden, C. H.: Uncoupling protein-2: a novel gene linked to obesity and hyperinsulinemia. *Nature Genet.* 15: 269–272, 1997.

[36666] 10218.Flier, J. S.; Lowell, B. B.: Obesity research springs a proton leak. *Nature Genet.* 15: 223–224, 1997.

[36667] 10219.Millet, L.; Vidal, H.; Andreelli, F.; Larrouy, D.; Riou, J.-P.; Ricquier, D.; Laville, M.; Langin, D.: Increased uncoupling protein-2 and -3 mRNA expression during fasting in obese and lean humans. *J.Clin. Invest.* 100: 2665–2670, 1997.

[36668] 10220.Edwards, Y. H.; Putt, W.; Lekoape, K. M.; Stott, D.; Fox, M.; Hopkinson, D. A.; Sowden, J.: The human homolog

T of the mouse T (Brachyury) gene: gene structure, cDNA sequence, and assignment to chromosome 6q27. *Genome Res.* 6: 226–233, 1996.

- [36669] 10221. Morrison, K.; Papapetrou, C.; Attwood, J.; Hol, F.; Lynch, S. A.; Sampath, A.; Hamel, B.; Burn, J.; Sowden, J.; Stott, D.; Mariman, E.; Edwards, Y. H.: Genetic mapping of the human homologue (T) of mouse T (Brachyury) and a search for allele association between human T and spina bifida. *Hum. Molec. Genet.* 5: 669–674, 1996.
- [36670] 10222. Papapetrou, C.; Drummond, F.; Reardon, W.; Winter, R.; Spitz, L.; Edwards, Y. H.: A genetic study of the human T gene and its exclusion as a major candidate gene for sacral agenesis with anorectal atresia. *J. Med. Genet.* 36: 208–213, 1999.
- [36671] 10223. Shields, D. C.; Ramsbottom, D.; Donoghue, C.; Pinjon, E.; Kirke, P. N.; Molloy, A. M.; Edwards, Y. H.; Mills, J. L.; Mynett-Johnson, L.; Weir, D. G.; Scott, J. M.; Whitehead, A. S.: Association between historically high frequencies of neural tube defects and the human T homologue of mouse T (Brachyury). *Am. J. Med. Genet.* 92: 206–211, 2000.
- [36672] 10224. Speer, M. C.; Melvin, E. C.; Viles, K. D.; Bauer, K. A.; Rampersaud, E.; Drake, C.; George, T. M.; Enterline, D. S.; Mackey, J. F.; Worley, G.; Gilbert, J. R.; Nye, J. S.; NTD Col-

laborative Group: T locus shows no evidence for linkage disequilibrium or mutation in American Caucasian neural tube defect families. *Am. J. Med. Genet.* 110: 215–218, 2002.

[36673] 10225. Trembath, D.; Sherbondy, A. L.; Vandyke, D. C.; Shaw, G. M.; Todoroff, K.; Lammer, E. J.; Finnell, R. H.; Marker, S.; Lerner, G.; Murray, J. C.: Analysis of select folate pathway genes, PAX3, and human Tin in a midwestern neural tube defect population. *Teratology* 59: 331–341, 1999.

[36674] 10226. Bellomo, D.; Headrick, J. P.; Silins, G. U.; Paterson, C. A.; Thomas, P. S.; Gartside, M.; Mould, A.; Cahill, M. M.; Tonks, I. D.; Grimmond, S. M.; Townson, S.; Wells, C.; Little, M.; Cummings, M. C.; Hayward, N. K.; Kay, G. F.: Mice lacking the vascular endothelial growth factor-B gene (Vegfb) have smaller hearts, dysfunctional coronary vasculature, and impaired recovery from cardiac ischemia. *Circ. Res.* 86: e29–e35, 2000.

[36675] 10227. Gerace, L.; Cirenei, N.; Cappelletti, M.; Petraroli, R.; Sebastiani, F.; Marziliano, N.: Assignment of the mouse Vegfb gene to mouse chromosome 19B by in situ hybridization. *Cytogenet. Cell Genet.* 95: 242–243, 2001.

[36676] 10228. Grimmond, S.; Lagercrantz, J.; Drinkwater, C.;

Silins, G.; Townson, S.; Pollock, P.; Gotley, D.; Carson, E.; Rakar, S.; Nordenskjold, M.; Ward, L.; Hayward, N.; Weber, G.: Cloning and characterization of a novel human gene related to vascular endothelial growth factor. *Genome Res.* 6: 124–131, 1996.

[36677] 10229. Olofsson, B.; Pajusola, K.; Kaipainen, A.; von Euler, G.; Joukov, V.; Saksela, O.; Orpana, A.; Pettersson, R. F.; Alitalo, K.; Eriksson, U.: Vascular endothelial growth factor B, a novel growth factor for endothelial cells. *Proc. Nat. Acad. Sci.* 93: 2576–2581, 1996.

[36678] 10230. Olofsson, B.; Pajusola, K.; von Euler, G.; Chilov, D.; Alitalo, K.; Eriksson, U.: Genomic organization of the mouse and human genes for vascular endothelial growth factor B (VEGF-B) and characterization of a second splice isoform. *J. Biol. Chem.* 271: 19310–19317, 1996.

[36679] 10231. Gunduz, M.; Ouchida, M.; Fukushima, K.; Hanafusa, H.; Etani, T.; Nishioka, S.; Nishizaki, K.; Shimizu, K.: Genomic structure of the human ING1 gene and tumor-specific mutations detected in head and neck squamous cell carcinomas. *Cancer Res.* 60: 3143–3146, 2000.

[36680] 10232. Hasegawa, H.; Kiyokawa, E.; Tanaka, S.; Nagashima, K.; Gotoh, N.; Shibuya, M.; Kurata, T.; Matsuda, M.: DOCK180, a major CRK-binding protein, alters cell

morphology upon translocation to the membrane.

Molec.Cell Biol. 16: 1770–176, 1996.

[36681] 10233.Savill, J.: Phagocytic docking without shocking. Nature 442–443,1998.

[36682] 10234.Takai, S.; Hasegawa, H.; Kiyokawa, E.; Yamada, K.; Kurata, T.;Matsuda, M.: Chromosomal mapping of the gene encoding DOCK180, amajor Crk–binding protein, to 10q26.13–q26.3 by fluorescence in situhybridization. Genomics 35: 403–404, 1996.

[36683] 10235.Wu, Y.–C.; Horvitz, H. R.: C. elegans phagocytosis and cell–migrationprotein CED–5 is similar to human DOCK180. Nature 392: 501–504,1998.

[36684] 10236.Alexander, J. M.; Bikkal, H. A.; Zervas, N. T.; Laws, E. R., Jr.;Klibanski, A.: Tumor–specific expression and alternate splicing ofmessenger ribonucleic acid encoding activin/transforming growth factor–betareceptors in human pituitary adenomas. J. Clin. Endocr. Metab. 81:783–790, 1996.

[36685] 10237.Su, G. H.; Bansal, R.; Murphy, K. M.; Montgomery, E.; Yeo, C. J.;Hruban, R. H.; Kern, S. E.: ACVR1B (ALK4, activin receptor type 1B)gene mutations in pancreatic carcinoma. Proc. Nat. Acad. Sci. 98:3254–3257, 2001.

[36686] 10238.Xu, J.; Matsuzaki, K.; McKeehan, K.; Wang, F.; Kan,

M.; McKeehan, W. L.: Genomic structure and cloned cDNAs predict that four variants in the kinase domain of serine/threonine kinase receptors arise by alternative splicing and poly(A) addition. *Proc. Nat. Acad. Sci.* 91:7957–7961, 1994.

[36687] 10239. Zhou, Y.; Sun, H.; Danila, D. C.; Johnson, S. R.; Sigai, D. P.; Zhang, X.; Klibanski, A.: Truncated activin type I receptor *Alk4* isoforms are dominant negative receptors inhibiting activin signaling. *Molec. Endocr.* 14: 2066–2075, 2000.

[36688] 10240. Mann, S. S.; Pettenati, M. J.; von Karp-herr, C.; Hart, T. C.: Reassignment of peptidyl prolyl isomerase-like 1 gene (*PPIL1*) to human chromosome region 6p21.1 by radiation hybrid mapping and fluorescence in situ hybridization. *Cytogenet. Cell Genet.* 83: 228–229, 1998.

[36689] 10241. Ozaki, K.; Fujiwara, T.; Kawai, A.; Shimizu, F.; Takami, S.; Okuno, S.; Takeda, S.; Shimada, Y.; Nagata, M.; Watanabe, T.; Takaichi, A.; Takahashi, E.; Nakamura, Y.; Shin, S.: Cloning, expression and chromosomal mapping of a novel cyclophilin-related gene (*PPIL1*) from human fetal-brain. *Cytogenet. Cell Genet.* 72: 242–245, 1996.

[36690] 10242. Graef, I. A.; Chen, F.; Chen, L.; Kuo, A.; Crabtree, G. R.: Signal transduced by Ca^{2+} /calcineurin and

NFATc3/c4 pattern the developing vasculature. *Cell* 105: 863–875, 2001.

- [36691] 10243. Wang, M. G.; Yi, H.; Guerini, D.; Klee, C. B.; McBride, O. W.: Calcineurin A alpha (PPP3CA), calcineurin A beta (PPP3CB) and calcineurin B (PPP3R1) are located on human chromosomes 4, 10q21–q22 and 2p16–p15, respectively. *Cytogenet. Cell Genet.* 72: 236–241, 1996.
- [36692] 10244. Zeng, H.; Chattarji, S.; Barbarosie, M.; Rondi-Reig, L.; Philpot, B. D.; Miyakawa, T.; Bear, M. F.; Tonegawa, S.: Forebrain-specific calcineurin knockout selectively impairs bidirectional synaptic plasticity and working/episodic-like memory. *Cell* 107: 617–629, 2001.
- [36693] 10245. Kolodrubetz, D.; Burgum, A.: Sequence and genetic analysis of NHP2: a moderately abundant high mobility group-like nuclear protein with an essential function in *Saccharomyces cerevisiae*. *Yeast* 7: 79–90, 1991.
- [36694] 10246. Saito, H.; Fujiwara, T.; Shin, S.; Okui, K.; Nakamura, Y.: Cloning and mapping of a human novel cDNA (NHP2L1) that encodes a protein highly homologous to yeast nuclear protein NHP2. *Cytogenet. Cell Genet.* 72: 191–193, 1996.
- [36695] 10247. Mooseker, M. S.; Cheney, R. E.: Unconventional myosins. *Annu. Rev. Cell Dev. Biol.* 11: 633–675, 1995.

- [36696] 10248.Bement, W. M.; Wirth, J. A.; Mooseker, M. S.: Cloning and mRNA expression of human unconventional myosin-IC: a homologue of amoeboid myosins-I with a single IQ motif and an SH3 domain. *J. Molec. Biol.* 243:356–363, 1994.
- [36697] 10249.Bement, W. M.; Hasson, T.; Wirth, J. A.; Cheney, R. E.; Mooseker, M. S.: Identification and overlapping expression of multiple unconventional myosin genes in vertebrate cell types. *Proc. Nat. Acad. Sci.* 91:6549–6553, 1994. Erratum: *Proc. Nat. Acad. Sci.* 91: 11767, 1994.
- [36698] 10250.Crozet, F.; Amraoui, A. E.; Blanchard, S.; Lenoir, M.; Ripoll, C.; Vago, P.; Hamel, C.; Fizames, C.; Levi-Acobas, F.; Depetris, D.; Mattei, M.-G.; Weil, D.; Pujol, R.; Petit, C.: Cloning of the genes encoding two murine and human cochlear unconventional type I myosins. *Genomics* 40:332–341, 1997.
- [36699] 10251.Goppelt, A.; Stelzer, G.; Lottspeich, F.; Meisterernst, M.: A mechanism for repression of class II gene transcription through specific binding of NC2 to TBP-promoter complexes via heterodimeric histone fold domains. *EMBO J.* 15: 3105–3116, 1996.
- [36700] 10252.Inostroza, J. A.; Mermeistein, F. H.; Ha, I.; Lane, W. S.; Reinberg, D.: Dr1, a TATA-binding protein-associated

phosphoprotein and inhibitor of class II gene transcription. Cell 70: 477–489, 1992.

[36701] 10253. Kamada, K.; Shu, F.; Chen, H.; Malik, S.; Stelzer, G.; Roeder, R. G.; Meisterernst, M.; Burley, S. K.: Crystal structure of negative cofactor 2 recognizing the TBP–DNA transcription complex. Cell 106:71–81, 2001.

[36702] 10254. Mermelstein, F.; Yeung, K.; Cao, J.; Inostroza, J. A.; Erdjument-Bromage, H.; Egelson, K.; Landsman, D.; Levitt, P.; Tempst, P.; Reinberg, D.: Requirement of a corepressor for Dr1-mediated repression of transcription. Genes Dev. 10: 1033–1048, 1996.

[36703] 10255. Purello, M.; Di Pietro, C.; Rapisarda, A.; Viola, A.; Corsaro, C.; Motta, S.; Grzeschik, K.–H.; Sichel, G.: Genomic localization of the human gene encoding Dr1, a negative modulator of transcription of class II and class III genes. Cytogenet. Cell Genet. 75: 186–189, 1996.

[36704] 10256. Rozet, J.–M.; Gerber, S.; Perrault, I.; Camuzat, A.; Calvas, P.; Viegas-Pequignot, E.; Molina-Gomes, D.; Le Paslier, D.; Chumakov, I.; Munnich, A.; Kaplan, J.: Structure and physical mapping of DR1, a TATA-binding protein-associated phosphoprotein gene, to chromosome 1p22.1 and its exclusion in Stargardt disease (STGD). Genomics 36:554–556, 1996.

- [36705] 10257.Willy, P. J.; Kobayashi, R.; Kadonaga, J. T.: A basal transcriptionfactor that activates or represses transcription. *Science* 290: 982–984,2000.
- [36706] 10258.Yeung, K.; Kim, S.; Reinberg, D.: Functional dissection of a humanDr1–DRAP1 repressor complex. *Molec. Cell. Biol.* 17: 36–45, 1997.
- [36707] 10259.Dong, L. Q.; Du, H.; Porter, S. G.; Kolakowski, L. F., Jr.; Lee,A. V.; Mandarino, J.; Fan, J.; Yee, D.; Liu, F.: Cloning, chromosomelocalization, expression, and characterization of an Src homology2 and pleckstrin homology domain–containing insulin receptor bindingprotein hGrb10–gamma. *J. Biol. Chem.* 272: 29104–29112, 1997.
- [36708] 10260.Margolis, B.; Silvennoinen, O.; Comoglio, F.; Roonprapunt, C.;Skolnik, E.; Ullrich, A.; Schlessinger, J.: High-efficiency expression/cloningof epidermal growth factor–receptor–binding proteins with Src homology2 domains. *Proc. Nat. Acad. Sci.* 89: 8894–8898, 1992.
- [36709] 10261.Skolnik, E. Y.; Margolis, B.; Mohammadi, M.; Lowenstein, E.; Fischer,R.; Drepps, A.; Ullrich, A.; Schlessinger, J.: Cloning of PI3 kinase–associated p85 utilizing a novel method for expression/cloning oftarget proteins for receptor tyrosine kinases. *Cell* 65: 83–90, 1991.
- [36710] 10262.Tanaka, S.; Mori, M.; Akiyoshi, T.; Tanaka, Y.; Ma–

funo, K.; Wands, J. R.; Sugimachi, K.: A novel variant of human Grb7 is associated with invasive esophageal carcinoma. J. Clin. Invest. 102: 821–827, 1998.

- [36711] 10263. Blagitko, N.; Mergenthaler, S.; Schulz, U.; Wollmann, H. A.; Craigen, W.; Eggermann, T.; Ropers, H.-H.; Kalscheuer, V. M.: Human GRB10 is imprinted and expressed from the paternal and maternal allele in a highly tissue- and isoform-specific fashion. *Hum. Molec. Genet.* 9:1587-1595, 2000.
- [36712] 10264. Frantz, J. D.; Giorgetti-Peraldi, S.; Ottinger, E. A.; Shoelson, S. E.: Human GRB-IR-beta/GRB10: splice variants of an insulin and growth factor receptor-binding protein with PH and SH2 domains. *J. Biol. Chem.* 272: 2659-2667, 1997.
- [36713] 10265. Hannula, K.; Lipsanen-Nyman, M.; Kontiokari, T.; Kere, J.: A narrow segment of maternal uniparental disomy of chromosome 7q31-qter in Silver-Russell syndrome delimits a candidate gene region. *Am. J. Hum. Genet.* 68: 247-253, 2001.
- [36714] 10266. Jerome, C. A.; Scherer, S. W.; Tsui, L.-C.; Gietz, R. D.; Triggs-Raine, B.: Assignment of growth factor receptor-bound protein 10 (GRB10) to human chromosome 7p11.2-p12. *Genomics* 40: 215-216, 1997.
- [36715] 10267. Liu, F.; Roth, R. A.: Grb-IR: a SH2-domain-containing protein that binds to the insulin receptor and inhibits its function. *Proc. Nat. Acad. Sci.* 92:

10287–10291, 1995.

[36716] 10268.Ooi, J.; Yajnik, V.; Immanuel, D.; Gordon, M.; Moskow, J. J.;Buchberg, A. M.; Margolis, B.: The cloning of Grb10 reveals a newfamily of SH2 domain proteins. *Oncogene* 10: 1621–1630, 1995.

[36717] 10269.Daly, R. J.; Sanderson, G. M.; Janes, P. W.; Sutherland, R. L.: Cloning and characterization of GRB14, a novel member of the GRB7gene family. *J. Biol. Chem.* 271: 12502–12510, 1996.

[36718] 10270.Hakala, B. E.; White, C.; Recklies, A. D.: Human cartilage gp–39,a major secretory product of articular chondrocytes and synovial cells,is a mammalian member of a chitinase protein family. *J. Biol. Chem.* 268:25803–25810, 1993.

[36719] 10271.Rehli, M.; Krause, S. W.; Andreesen, R.: Molecular characterizationof the gene for human cartilage gp–39 (CHI3L1), a member of the chitinaseprotein family and marker for late stages of macrophage differentiation. *Genomics* 43:221–225, 1997.

[36720] 10272.Dry, K.; Kenwrick, S.; Rosenthal, A.; Platzer, M.: The complete sequence of the human locus for NgCAM–related cell adhesion moleculereveals a novel alternative exon in chick and man and conserved genomicorganization for

the L1 subfamily. *Gene* 273: 115–122, 2001.

[36721] 10273.Grumet, M.; Mauro, V.; Burgoon, M. P.; Edelman, G. M.; Cunningham, B. A.: Structure of a new nervous system glycoprotein, Nr–CAM, and its relationship to subgroups of neural cell adhesion molecules. *J. Cell. Biol.* 113: 1399–1412, 1991.

[36722] 10274.Kayem, J. F.; Roman, J. M.; de la Rosa, E. J.; Schwarz, U.; Dreyer, W. J.: Bravo/Nr–CAM is closely related to the cell adhesion molecules L1 and Ng–CAM and has a similar heterodimer structure. *J. Cell. Biol.* 118:1259–1270, 1992.

[36723] 10275.Lane, R. P.; Chen, X.–N.; Yamakawa, K.; Vielmetter, J.; Korenberg, J. R.; Dreyer, W. J.: Characterization of a highly conserved human homolog to the chicken neural cell surface protein Bravo/Nr–CAM that maps to chromosome band 7q31. *Genomics* 35: 456–465, 1996.

[36724] 10276.Wang, B.; Williams, H.; Du, J.–S.; Terrett, J.; Kenwick, S.: Alternative splicing of human NrCAM in neural and nonneural tissues. *Molec. Cell. Neurosci.* 10: 287–295, 1998.

[36725] 10277.Damen, J. E.; Liu, L.; Rosten, P.; Humphries, R. K.; Jefferson, A. B.; Majerus, P. W.; Krystal, G.: The 145–kDa protein induced to associate with Shc by multiple cytokines

is an inositol tetrakisphosphate and phosphatidylinositol 3,4,5-trisphosphate 5-phosphatase. *Proc. Nat. Acad. Sci.* 93: 1689–1693, 1996.

[36726] 10278. Drayer, A. L.; Pesesse, X.; De Smedt, F.; Woscholski, R.; Parker, P.; Erneux, C.: Cloning and expression of a human placenta inositol 1,3,4,5-tetrakisphosphate and phosphatidylinositol 3,4,5-trisphosphate 5-phosphatase. *Biochem. Biophys. Res. Commun.* 225: 243–249, 1996.

[36727] 10279. Helgason, C. D.; Damen, J. E.; Rosten, P.; Grewal, R.; Sorensen, P.; Chappel, S. M.; Borowski, A.; Jirik, F.; Krystal, G.; Humphries, R. K.: Targeted disruption of SHIP leads to hemopoietic perturbations, lung pathology, and a shortened life span. *Genes Dev.* 12: 1610–1620, 1998.

[36728] 10280. Huber, M.; Helgason, C. D.; Damen, J. E.; Liu, L.; Humphries, R. K.; Krystal, G.: The src homology 2-containing inositol phosphatase (SHIP) is the gatekeeper of mast cell degranulation. *Proc. Nat. Acad. Sci.* 95: 11330–11335, 1998.

[36729] 10281. Kavanaugh, W. M.; Pot, D. A.; Chin, S. M.; Deuter-Reinhard, M.; Jefferson, A. B.; Norris, F. A.; Masiarz, F. R.; Cousens, L. S.; Majerus, P. W.; Williams, L. T.: Multiple forms of an inositol polyphosphate 5-phosphatase form signaling complexes with Shc and Grb2. *Curr. Biol.* 6:

438–445, 1996.

- [36730] 10282.Lioubin, M. N.; Algate, P. A.; Tsai, S.; Carlberg, K.; Aebersold,R.; Rohrschneider, L. R.: p150(Ship), a signal transduction moleculewith inositol polyphosphate–5–phosphatase activity. *Genes Dev.* 10:1084–1095, 1996.
- [36731] 10283.Liu, Q.; Amgen EST Program; Dumont, D. J.: Molecular cloningand chromosomal localization in human and mouse of the SH2–containinginositol phosphatase, INPP5D (SHIP). *Genomics* 39: 109–112, 1997.
- [36732] 10284.Liu, Q.; Shalaby, F.; Jones, J.; Bouchard, D.; Dumont, D. J.:The SH2–containing inositol polyphosphate 5–phosphatase, Ship, isexpressed during hematopoiesis and spermatogenesis. *Blood* 91: 2753–2759,1998.
- [36733] 10285.Takeshita, S.; Namba, N.; Zhao, J. J.; Jiang, Y.; Genant, H. K.;Silva, M. J.; Brodt, M. D.; Helgason, C. D.; Kalesnikoff, J.; Rauh,M. J.; Humphries, R. K.; Krystal, G.; Teitelbaum, S. L.; Ross, F.P.: SHIP–deficient mice are severely osteoporotic due to increasednumbers of hyper–resorptive osteoclasts. *Nature Med.* 8: 943–949,2002.
- [36734] 10286.Wang, J.–W.; Howson, J. M.; Ghansah, T.; Desponts, C.; Ninos,J. M.; May, S. L.; Nguyen, K. H. T.; Toyama–Sorimachi, N.; Kerr, W.G.: Influence of SHIP on the NK repertoire and allogeneic bone marrowtransplantation.

Science 295: 2094–2097, 2002.

- [36735] 10287. Ware, M. D.; Rosten, P.; Damen, J. E.; Liu, L.; Humphries, R.K.; Krystal, G.: Cloning and characterization of human SHIP, the 145-kD inositol 5-phosphatase that associates with SHC after cytokine stimulation. *Blood* 88: 2833–2840, 1996.
- [36736] 10288. Cates, C. A.; Michael, R. L.; Staybrook, K. R.; Harvey, K. A.; Burke, Y. D.; Randall, S. K.; Crowell, P. L.; Crowell, D. N.: Prenylation of oncogenic human PTP(CAAX) protein tyrosine phosphatases. *Cancer Lett.* 110: 49–55, 1996.
- [36737] 10289. Montagna, M.; Serova, O.; Sylla, B. S.; Feunteun, J.; Lenoir, G.M.: A 100-kb physical and transcriptional map around the EDH17B2 gene: identification of three novel genes and a pseudogene of a human homologue of the rat PRL-1 tyrosine phosphatase. *Hum. Genet.* 96: 532–538, 1995.
- [36738] 10290. Zeng, Q.; Hong, W.; Tan, Y. H.: Mouse PRL-2 and PRL-3, two potentially prenylated protein tyrosine phosphatases homologous to PRL-1. *Biochem. Biophys. Res. Commun.* 244: 421–427, 1998.
- [36739] 10291. Muneer, S.; Ramalingam, V.; Wyatt, R.; Schultz, R. A.; Minna, J.D.; Kamibayashi, C.: Genomic organization and mapping of the gene encoding the PP2A B56-gamma

regulatory subunit. *Genomics* 79: 344–348, 2002.

- [36740] 10292. Deveraux, Q.; Jensen, C.; Rechsteiner, M.: Molecular cloning and expression of a 26 S protease subunit enriched in dileucine repeats. *J. Biol. Chem.* 270: 23726–23729, 1995.
- [36741] 10293. Deveraux, Q.; Ustrell, V.; Pickart, C.; Rechsteiner, M.: A 26S protease subunit that binds ubiquitin conjugates. *J. Biol. Chem.* 269:7059–7061, 1994.
- [36742] 10294. Hu, R.-J.; Lee, M. P.; Johnson, L. A.; Feinberg, A. P.: A novel human homologue of yeast nucleosome assembly protein, 65 kb centromeric to the p57(KIP2) gene, is biallelically expressed in fetal and adult tissues. *Hum. Molec. Genet.* 5: 1743–1748, 1996.
- [36743] 10295. Abderrahim, H.; Jaramillo-Babb, V. L.; Zhou, Z.; Vollrath, D.: Characterization of the murine TIGR/myocilin gene. *Mammalian Genome* 9:673–675, 1998.
- [36744] 10296. Adam, M. F.; Belmouden, A.; Binisti, P.; Brezin, A. P.; Valtot, F.; Bechetoille, A.; Dascotte, J.-C.; Copin, B.; Gomez, L.; Chaventre, A.; Bach, J.-F.; Garchon, H.-J.: Recurrent mutations in a single exon encoding the evolutionarily conserved olfactomedin-homology domain of TIGR in familial open-angle glaucoma. *Hum. Molec. Genet.* 6: 2091–2097, 1997.

- [36745] 10297. Baird, P. N.; Dickinson, J.; Craig, J. E.; Mackey, D. A.: The Taa1 restriction enzyme provides a simple means to identify the Q368STOP mutation of the myocilin gene in primary open angle glaucoma. *Am.J. Ophthalmol.* 131: 510–511, 2001.
- [36746] 10298. Rodriguez, P.; Munroe, D.; Prawitt, D.; Chu, L. L.; Bric, E.; Kim, J.; Reid, L. H.; Davies, C.; Nakagama, H.; Loebbert, R.; Winterpacht, A.; Petruzzi, M.-J.; Higgins, M. J.; Nowak, N.; Evans, G.; Shows, T.; Weissman, B. E.; Zabel, B.; Housman, D. E.; Pelletier, J.: Functional characterization of human nucleosome assembly protein-2 (NAP1L4) suggests a role as a histone chaperone. *Genomics* 44: 253–265, 1997.
- [36747] 10299. Stegmaier, K.; Pendse, S.; Barker, G. F.; Bray-Ward, P.; Ward, D. C.; Montgomery, K. T.; Krauter, K. S.; Reynolds, C.; Sklar, J.; Donnelly, M.; Bohlander, S. K.; Rowley, J. D.; Sallan, S. E.; Gilliland, D. G.; Golub, T. R.: Frequent loss of heterozygosity at the TEL gene locus in acute lymphoblastic leukemia of childhood. *Blood* 86: 38–44, 1995.
- [36748] 10300. Arakawa, H.; Nagase, H.; Hayashi, N.; Fujiwara, T.; Ogawa, M.; Shin, S.; Nakamura, Y.: Molecular cloning and expression of a novel human gene that is highly homolo-

gous to human FK506-binding protein12kDa (hFKBP-12) and characterization of two alternatively spliced transcripts. *Biochem. Biophys. Res. Commun.* 200: 836–843, 1994.

[36749] 10301.Xin, H.-B.; Senbonmatsu, T.; Cheng, D.-S.; Wang, Y.-X. Copello, J. A.; Ji, G.-J.; Collier, M. L.; Deng, K.-Y.; Jeyakumar, L. H.; Magnuson, M. A.; Inagami, T.; Kotlikoff, M. I.; Fleischer, S.: Oestrogen protects FKBP12.6 null mice from cardiac hypertrophy. *Nature* 416: 334–337, 2002.

[36750] 10302.Stein, R.; Mori, N.; Matthews, K.; Lo, L.-C.; Anderson, D. J.:The NGF-inducible SCG10 mRNA encodes a novel membrane-bound protein present in growth cones and abundant in developing neurons. *Neuron* 1:463–476, 1988.

[36751] 10303.Dong, J.-T.; Isaacs, W. B.; Barrett, J. C.; Isaacs, J. T.: Genomic organization of the human KAI1 metastasis-suppressor gene. *Genomics* 41:25–32, 1997.

[36752] 10304.Dong, J.-T.; Lamb, P. W.; Rinker-Schaeffer, C. W.; Vukanovic, J.; Ichikawa, T.; Isaacs, J. T.; Barrett, J. C.: KAI1, a metastasis suppressor gene for prostate cancer on human chromosome 11p11.2. *Science* 268:884–886, 1995.

[36753] 10305.Guo, X.-Z.; Friess, H.; Di Mola, F. F.; Heinicke, J.-M.; Abou-Shady, M.; Graber, H. U.; Baer, H. U.; Zimmer-

mann, A.; Korc, M.; Buchler, M. W.: KAI1, a new metastasis suppressor gene, is reduced in metastatic hepatocellular carcinoma. *Hepatology* 28: 1481–1488, 1998.

[36754] 10306. Mashimo, T.; Watabe, M.; Hirota, S.; Hosobe, S.; Miura, K.; Tegtmeyer, P. J.; Rinker-Shaeffer, C. W.; Watabe, K.: The expression of the KAI1 gene, a tumor metastasis suppressor, is directly activated by p53. *Proc. Nat. Acad. Sci.* 95: 11307–11311, 1998.

[36755] 10307. Miyazaki, T.; Kato, H.; Shitara, Y.; Yoshikawa, M.; Tajima, K.; Masuda, N.; Shouji, H.; Tsukada, K.; Nakajima, T.; Kuwano, H.: Mutation and expression of the metastasis suppressor gene KAI1 in esophageal squamous cell carcinoma. *Cancer* 89: 955–962, 2000.

[36756] 10308. Gupta, P.; Soyombo, A. A.; Atashband, A.; Wisniewski, K. E.; Shelton, J. M.; Richardson, J. A.; Hammer, R. E.; Hofmann, S. L.: Disruption of PPT1 or PPT2 causes neuronal ceroid lipofuscinosis in knockout mice. *Proc. Nat. Acad. Sci.* 98: 13566–13571, 2001.

[36757] 10309. Ardell, M. D.; Aragon, I.; Oliveira, L.; Porche, G. E.; Burke, E.; Pittler, S. J.: The beta subunit of human rod photoreceptor cGMP-gated cation channel is generated from a complex transcription unit. *FEBS Lett.* 389: 213–218, 1996.

- [36758] 10310.Ardell, M. D.; Makhija, A. K.; Oliveira, L.; Miniou, P.; Viegas-Pequignot, E.; Pittler, S. J.: cDNA, gene structure, and chromosomal localization of human GAR1 (CNCG3L), a homolog of the third subunit of bovine photoreceptor-cGMP-gated channel. *Genomics* 28: 32–38, 1995.
- [36759] 10311.Bareil, C.; Hamel, C. P.; Delague, V.; Arnaud, B.; Demaille, J.; Claustres, M.: Segregation of a mutation in CNGB1 encoding the beta-subunit of the rod cGMP-gated channel in a family with autosomal recessive retinitis pigmentosa. *Hum. Genet.* 108: 328–334, 2001.
- [36760] 10312.Bird, A. C.: Retinal photoreceptor dystrophies. LI. Edward Jackson Memorial Lecture. *Am. J. Ophthalm.* 119: 543–562, 1995.
- [36761] 10313.Chen, T.-Y.; Illing, M.; Molday, L. L.; Hsu, Y.-T.; Yau, K.-W.; Molday, R. S.: Subunit 2 (or beta) of retinal rod cGMP-gated cation channel is a component of the 240-kDa channel-associated protein and mediates Ca^{2+} -calmodulin modulation. *Proc. Nat. Acad. Sci.* 91:11757–11761, 1994.
- [36762] 10314.Chen, T. Y.; Peng, Y. W.; Dhallan, R. S.; Ahamed, B.; Reed, R.R.; Yau, K. W.: A new subunit of the cyclic nucleotide-gated cation channel in retinal rods. *Nature* 362: 764–767, 1993.

- [36763] 10315.Korschen, H. G.; Beyermann, M.; Muller, F.; Heck, M.; Vantler, M.; Koch, K.-W.; Kellner, R.; Wolfrum, U.; Bode, C.; Hofmann, K. P.; Kaupp, U. B.: Interaction of glutamic-acid-rich proteins with the cGMP signalling pathway in rod photoreceptors. *Nature* 400: 761-766, 1999.
- [36764] 10316.Neufeld, E. F.: Personal Communication. Los Angeles, Calif. 7/24/1987.
- [36765] 10317.Blair, H. J.; Ho, M.; Monaco, A. P.; Fisher, S.; Craig, I. W.; Boyd, Y.: High-resolution comparative mapping of the proximal region of the mouse X chromosome. *Genomics* 28: 305-310, 1995.
- [36766] 10318.Cox, J. P. D.; Yamamoto, K.; Christie, P. T.; Wooding, C.; Feest, T.; Flinter, F. A.; Goodyer, P. R.; Leumann, E.; Neuhaus, T.; Reid, C.; Williams, P. F.; Wrong, O.; Thakker, R. V.: Renal chloride channel, CLCN5, mutations in Dent's disease. *J. Bone Min. Res.* 14: 1536-1542, 1999.
- [36767] 10319.Craig, I.: Personal Communication. Oxford, England 1/31/1995.
- [36768] 10320.Devuyst, O.; Christie, P. T.; Courtoy, P. J.; Beauwens, R.; Thakker, R. V.: Intra-renal and subcellular distribution of the human chloride channel, CLC-5, reveals a pathophysiological basis for Dent's disease. *Hum. Molec. Genet.* 8: 247-257, 1999.

- [36769] 10321.Dutzler, R.; Campbell, E. B.; Cadene, M.; Chait, B. T.; MacKinnon, R.: X-ray structure of a ClC chloride channel at 3.0 angstrom reveals the molecular basis of anion selectivity. *Nature* 415: 287–294, 2002.
- [36770] 10322.Fisher, S. E.; Black, G. C. M.; Lloyd, S. E.; Hatchwell, E.; Wrong, O.; Thakker, R. V.; Craig, I. W.: Isolation and partial characterization of a chloride channel gene which is expressed in kidney and is a candidate for Dent's disease (an X-linked hereditary nephrolithiasis). *Hum.Molec. Genet.* 3: 2053–2059, 1994.
- [36771] 10323.Fisher, S. E.; Van Bakel, I.; Lloyd, S. E.; Pearce, S. H. S.; Thakker, R. V.; Craig, I. W.: Cloning and characterization of CLCN5, the human kidney chloride channel gene implicated in Dent disease (an X-linked hereditary nephrolithiasis). *Genomics* 29: 598–606, 1995.
- [36772] 10324.Gunther, W.; Luchow, A.; Cluzeaud, F.; Vandewalle, A.; Jentsch, T. J.: ClC-5, the chloride channel mutated in Dent's disease, colocalizes with the proton pump in endocytotically active kidney cells. *Proc.Nat. Acad. Sci.* 95: 8075–8080, 1998.
- [36773] 10325.Igarashi, T.; Hayakawa, H.; Shiraga, H.; Kawato, H.; Yan, K.; Kawaguchi, H.; Yamanaka, T.; Tsuchida, S.; Akagi, K.: Hypercalciuria and nephrocalcinosis in patients with id-

idiopathic low molecular weight proteinuria in Japan: is the disease identical to Dent's disease in the United Kingdom? *Nephron*. 69:242–247, 1995.

[36774] 10326. Lloyd, S. E.; Gunther, W.; Pearce, S. H. S.; Thomson, A.; Bianchi, M. L.; Bosio, M.; Craig, I. W.; Fisher, S. E.; Scheinman, S. J.; Wrong, O.; Jentsch, T. J.; Thakker, R. V.: Characterisation of renal chloride channel, CLCN5, mutations in hypercalciuric nephrolithiasis (kidney stones) disorders. *Hum. Molec. Genet.* 6: 1233–1239, 1997.

[36775] 10327. Lloyd, S. E.; Pearce, S. H. S.; Fisher, S. E.; Steinmeyer, K.; Schwappach, B.; Scheinman, S. J.; Harding, B.; Bolino, A.; Devoto, M.; Goodyer, P.; Rigden, S. P. A.; Wrong, O.; Jentsch, T. J.; Craig, I. W.; Thakker, R. V.: A common molecular basis for three inherited kidney stone diseases. *Nature* 370: 445–449, 1996.

[36776] 10328. Lloyd, S. E.; Pearce, S. H. S.; Gunther, W.; Kawaguchi, H.; Igarashi, T.; Jentsch, T. J.; Thakker, R. V.: Idiopathic low molecular weight proteinuria associated with hypercalciuric nephrocalcinosis in Japanese children is due to mutations of the renal chloride channel (CLCN5). *J. Clin. Invest.* 99: 967–974, 1997.

[36777] 10329. Oudet, C.; Martin-Coignard, D.; Pannetier, S.; Praud, E.; Champion, G.; Hanauer, A.: A second family with

XLRH displays the mutation S244L in the CLCN5 gene.

Hum. Genet. 99: 781–784, 1997.

- [36778] 10330. Piwon, N.; Gunther, W.; Schwake, M.; Bosl, M. R.; Jentsch, T.J.: CIC-5 Cl(–)-channel disruption impairs endocytosis in a mouse model for Dent's disease. Nature 408: 369–373, 2000.
- [36779] 10331. Schurman, S. J.; Norden, A. G. W.; Scheinman, S. J.: X-linked recessive nephrolithiasis: presentation and diagnosis in children. J. Pediatr. 132: 859–862, 1998.
- [36780] 10332. Cabeza-Arvelaiz, Y.; Shih, L.-C. N.; Hardman, N.; Asselbergs, F.; Bilbe, G.; Schmitz, A.; White, B.; Siciliano, M. J.; Lachman, L. B.: Cloning and genetic organization of the human kinesin light-chain (KLC) gene. DNA Cell Biol. 12: 881–892, 1993.
- [36781] 10333. Pearce, S. H. S.; Cheetham, T.; Imrie, H.; Vaidya, B.; Barnes, N. D.; Bilous, R. W.; Carr, D.; Meeran, K.; Shaw, N. J.; Smith, C. S.; Toft, A. D.; Williams, G.; Kendall-Taylor, P.: A common and recurrent 13-bp deletion in the autoimmune regulator gene in British kindreds with autoimmune polyendocrinopathy type 1. Am. J. Hum. Genet. 63: 1675–1684, 1998.
- [36782] 10334. Mitnick, M.; Reichlin, S.: Enzymatic synthesis of thyrotropin-releasing hormone (TRH) by hypothalamic

'TRH synthetase.'. *Endocrinology* 91:1145–1153, 1972.

[36783] 10335. Battini, R.; Ferrari, S.; Kaczmarek, L.; Calabretta, B.; Chen, S.-T.; Baserga, R.: Molecular cloning of a cDNA for a human ADP/ATP carrier which is growth-regulated. *J. Biol. Chem.* 262: 4355–4359, 1987.

[36784] 10336. Chen, S.-T.; Chang, C.-D.; Huebner, K.; Ku, D.-H.; McFarland, M.; DeRiel, J. K.; Baserga, R.; Wurzel, J.: A human ADP/ATP translocase gene has seven pseudogenes and localizes to chromosome X. *Somat. Cell Molec. Genet.* 16: 143–149, 1990.

[36785] 10337. Ku, D.-H.; Kagan, J.; Chen, S.-T.; Chang, C.-D.; Baserga, R.; Wurzel, J.: The human fibroblast adenine nucleotide translocator gene: molecular cloning and sequence. *J. Biol. Chem.* 265: 16060–16063, 1990.

[36786] 10338. Schiebel, K.; Mertz, A.; Winkelmann, M.; Nagaraja, R.; Rappold, G.: Localization of the adenine nucleotide translocase gene ANT2 to chromosome Xq24–q25 with tight linkage to DXS425. *Genomics* 24:605–606, 1994.

[36787] 10339. Lu, Q. R.; Sun, T.; Zhu, Z.; Ma, N.; Garcia, M.; Stiles, C. D.; Rowitch, D. H.: Common developmental requirement for Olig function indicates a motor neuron/oligodendrocyte connection. *Cell* 109: 75–86, 2002.

[36788] 10340. Lu, Q. R.; Yuk, D.; Alberta, J. A.; Zhu, Z.; Pawlitzky,

I.; Chan,J.; McMahon, A. P.; Stiles, C. D.; Rowitch, D. H.: Sonic hedgehog-regulated oligodendrocyte lineage genes encoding bHLH proteins in the mammalian central nervous system. *Neuron* 25: 317–329, 2000.

[36789] 10341. Raff, M. C.; Miller, R. H.; Noble, M.: A glial progenitor cell that develops in vitro into an astrocyte or an oligodendrocyte depending on culture medium. *Nature* 303: 390–396, 1983.

[36790] 10342. Zhou, Q.; Anderson, D. J.: The bHLH transcription factors OLIG2 and OLIG1 couple neuronal and glial subtype specification. *Cell* 109:61–73, 2002.

[36791] 10343. Zhou, Q.; Wang, S.; Anderson, D. J.: Identification of a novel family of oligodendrocyte lineage-specific basic helix–loop–helix transcription factors. *Neuron* 25: 331–343, 2000.

[36792] 10344. Guimaraes, M. J.; Peterson, D.; Vicari, A.; Cocks, B. G.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Ferrick, D. A.; Kastelein, R. A.; Bazan, J. F.; Zlotnik, A.: Identification of a novel selenocysteine homologue from eukaryotes, bacteria, and archaea: is there an autoregulatory mechanism in selenocysteine metabolism? *Proc. Nat. Acad. Sci.* 93:15086–15091, 1996.

[36793] 10345. Koesters, R.; Adams, V.; Betts, D.; Moos, R.;

Schmid, M.; Siemann, A.; Hassam, S.; Weitz, S.; Lichter, P.; Heitz, P. U.; von Knebel Doeberitz, M.; Briner, J.: Human eukaryotic initiation factor EIF2C1 gene: cDNA sequence, genomic organization, localization to chromosomal bands 1q34–p35, and expression. *Genomics* 61: 210–218, 1999.

[36794] 10346. Martinez, J.; Patkaniowska, A.; Urlaub, H.; Luhrmann, R.; Tusch, T.: Single-stranded antisense siRNAs guide target RNA cleavage in RNAi. *Cell* 110: 563–574, 2002.

[36795] 10347. Suetsugu, S.; Miki, H.; Takenawa, T.: Identification of two human WAVE/SCAR homologues as general actin regulatory molecules which associate with the Arp2/3 complex. *Biochem. Biophys. Res. Commun.* 260: 296–302, 1999.

[36796] 10348. Myers, L. C.; Gustafsson, C. M.; Bushnell, D. A.; Lui, M.; Erdjument-Bromage, H.; Tempst, P.; Kornberg, R. D.: The Med proteins of yeast and their function through the RNA polymerase II carboxy-terminal domain. *Genes Dev.* 12: 45–54, 1998.

[36797] 10349. Hanaoka, E.; Ozaki, T.; Ohira, M.; Nakamura, Y.; Suzuki, M.; Takahashi, E.; Moriya, H.; Nakagawara, A.; Sakiyama, S.: Molecular cloning and expression analysis of

the human DA41 gene and its mapping to chromosome 9q21.2–q21.3. *J. Hum. Genet.* 45: 188–191, 2000.

[36798] 10350. Ozaki, T.; Hishiki, T.; Toyama, Y.; Yuasa, S.; Nakagawara, A.; Sakiyama, S.: Identification of a new cellular protein that can interact specifically with DAN. *DNA Cell Biol.* 16: 985–991, 1997.

[36799] 10351. Venkateswarlu, K.; Gunn-Moore, F.; Oatey, P. B.; Tavaré, J. M.; Cullen, P. J.: Nerve growth factor- and epidermal growth factor-stimulated translocation of the ADP-ribosylation factor-exchange factor GRP1 to the plasma membrane of PC12 cells requires activation of phosphatidylinositol 3-kinase and the GRP1 pleckstrin homology domain. *Biochem. J.* 335: 139–146, 1998.

[36800] 10352. Dammann, R.; Li, C.; Yoon, J.-H.; Chin, P. L.; Bates, S.; Pfeifer, G. P.: Epigenetic inactivation of a RAS association domain family protein from the lung tumour suppressor locus 3p21.3. *Nature Genet.* 25: 315–319, 2000.

[36801] 10353. Dreijerink, K.; Braga, E.; Kuzmin, I.; Geil, L.; Duh, F.-M.; Angeloni, D.; Zbar, B.; Lerman, M. I.; Stanbridge, E. J.; Minna, J. D.; Protopopov, A.; Li, J.; Kashuba, V.; Klein, G.; Zbarovsky, E. R.: The candidate tumor suppressor gene, RASSF1A, from human chromosome 3p21.3 is involved in kidney tumorigenesis. *Proc. Nat. Acad. Sci.* 98:

7504–7509, 2001.

- [36802] 10354. Harada, K.; Toyooka, S.; Maitra, A.; Maruyama, R.; Toyooka, K.O.; Timmons, C. F.; Tomlinson, G. E.; Mastrangelo, D.; Hay, R. J.; Minna, J. D.; Gazdar, A. F.: Aberrant promoter methylation and silencing of the RASSF1A gene in pediatric tumors and cell lines. *Oncogene* 21:4345–4349, 2002.
- [36803] 10355. Sekido, Y.; Ahmadian, M.; Wistuba, I. I.; Latif, F.; Bader, S.; Wei, M.-H.; Duh, F.-M.; Gazdar, A. F.; Lerman, M. I.; Minna, J. D.: Cloning of a breast cancer homozygous deletion junction narrows the region of search for a 3p21.3 tumor suppressor gene. *Oncogene* 16:3151–3157, 1998.
- [36804] 10356. Dann, C. E.; Hsieh, J.-C.; Rattner, A.; Sharma, D.; Nathans, J.; Leahy, D. J.: Insights into Wnt binding and signalling from the structures of two Frizzled cysteine-rich domains. *Nature* 412: 86–90, 2001.
- [36805] 10357. Hoang, B.; Moos, M., Jr.; Vukicevic, S.; Luyten, F. P.: Primary structure and tissue distribution of FRZB, a novel protein related to *Drosophila* frizzled, suggest a role in skeletal morphogenesis. *J. Biol. Chem.* 271: 26131–26137, 1996.
- [36806] 10358. Leyns, L.; Bouwmeester, T.; Kim, S.-H.; Piccolo, S.; De Robertis, E. M.: Frzb-1 is a secreted antagonist of Wnt

signaling expressed in the Spemann organizer. *Cell* 88: 747–756, 1997.

[36807] 10359. Schumann, H.; Holtz, J.; Zerkowski, H.-R.; Hatzfeld, M.: Expression of secreted frizzled related proteins 3 and 4 in human ventricular myocardium correlates with apoptosis related gene expression. *Cardiovasc. Res.* 45: 720–728, 2000.

[36808] 10360. Bouchon, A.; Dietrich, J.; Colonna, M.: Cutting edge: inflammatory responses can be triggered by TREM-1, a novel receptor expressed on neutrophils and monocytes. *J. Immun.* 164: 4991–4995, 2000.

[36809] 10361. Bouchon, A.; Facchetti, F.; Weigand, M. A.; Colonna, M.: TREM-1 amplifies inflammation and is a crucial mediator of septic shock. *Nature* 410: 1103–1107, 2001.

[36810] 10362. Yu, Y.; Xu, F.; Peng, H.; Fang, X.; Zhao, S.; Li, Y.; Cuevas, B.; Kuo, W.-L.; Gray, J. W.; Siciliano, M.; Mills, G. B.; Bast, R. C., Jr.: NOEY2 (ARHI), an imprinted putative tumor suppressor gene in ovarian and breast carcinomas. *Proc. Nat. Acad. Sci.* 96: 214–219, 1999.

[36811] 10363. Walowsky, C.; Fitzhugh, D. J.; Castano, I. B.; Ju, J. Y.; Levin, N. A.; Christman, M. F.: The topoisomerase-related function gene TRF4 affects cellular sensitivity to the

antitumor agent camptothecin. J.Biol. Chem. 274:
7302–7308, 1999.

[36812] 10364.Wang, Z.; Castano, I. B.; De Las Penas, A.; Adams, C.; Christman,M. F.: Pol kappa: a DNA polymerase re-
quired for sister chromatidcohesion. Science 289:
774–779, 2000.

[36813] 10365.Clark, J.; Lu, Y.-J.; Sidhar, S. K.; Parker, C.; Gill, S.; Smedley,D.; Hamoudi, R.; Linehan, W. M.; Shipley, J.; Cooper, C. S.: Fusionof splicing factor genes PSF and NonO (p54–nrb) to the TFE3 gene inpapillary renal cell carcinoma. Oncogene 15: 2233–2239, 1997.

[36814] 10366.Patton, J. G.; Porro, E. B.; Galceran, J.; Tempst, P.; Nadal–Ginard,B.: Cloning and characterization of PSF, a novel pre–mRNA splicingfactor. Genes Dev. 7: 393–406, 1993.

[36815] 10367.Piquemal, D.; Joulia, D.; Balaguer, P.; Basset, A.; Marti, J.;Commes, T.: Differential expression of the RTP/ Drg1/Ndr1 gene productin proliferating and growth ar-
rested cells. Biochim. Biophys. Acta 1450:364–373, 1999.

[36816] 10368.LeClerc, S.; Palaniswami, R.; Xie, B.; Govdan, M. V.: Molecularcloning and characterization of a factor that binds the human glucocorticoidreceptor gene and re-
presses its expression. J. Biol. Chem. 266:17333–17340,

1991.

- [36817] 10369.Tikoo, A.; Czekay, S.; Viars, C.; White, S.; Heath, J. K.; Arden, K.; Maruta, H.: p190-A, a human tumor suppressor gene, maps to the chromosomal region 19q13.3 that is reportedly deleted in some gliomas. *Gene* 257:23–31, 2000.
- [36818] 10370.Boccaccio, I.; Glatt-Deeley, H.; Watrin, F.; Roeckel, N.; Lalande, M.; Muscatelli, F.: The human MAGEL2 gene and its mouse homologue are paternally expressed and mapped to the Prader-Willi region. *Hum. Molec. Genet.* 8: 2497–2505, 1999.
- [36819] 10371.Lee, S.; Kozlov, S.; Hernandez, L.; Chamberlain, S. J.; Brannan, C. I.; Stewart, C. L.; Wevrick, R.: Expression and imprinting of MAGEL2 suggest a role in Prader-Willi syndrome and the homologous murine imprinting phenotype. *Hum. Molec. Genet.* 9: 1813–1819, 2000.
- [36820] 10372.Wang, A. H.; Bertos, N. R.; Vezmar, M.; Pelletier, N.; Crosato, M.; Heng, H. H.; Th'ng, J.; Han, J.; Yang, X.-J.: HDAC4, a human histone deacetylase related to yeast HDA1, is a transcriptional corepressor. *Molec. Cell. Biol.* 19: 7816–7827, 1999.
- [36821] 10373.Nishimura, Y.; Hayashi, M.; Inada, H.; Tanaka, T.: Molecular cloning and characterization of mammalian ho-

mologues of vesicle-associated membrane protein-associated (VAMP-associated) proteins. *Biochem. Biophys. Res. Commun.* 254: 21–26, 1999.

[36822] 10374. Weir, M. L.; Klip, A.; Trimble, W. S.: Identification of a human homologue of the vesicle-associated membrane protein (VAMP)-associated protein of 33 kDa (VAP-33): a broadly expressed protein that binds to VAMP. *Biochem. J.* 333: 247–251, 1998.

[36823] 10375. Perez Jurado, L. A.; Wang, Y.-K.; Francke, U.; Cruces, J.: TBL2, a novel transducin family member in the WBS deletion: characterization of the complete sequence, genomic structure, transcriptional variants and the mouse ortholog. *Cytogenet. Cell Genet.* 86: 277–284, 1999.

[36824] 10376. Akhmanova, A.; Hoogenraad, C. C.; Drabek, K.; Stepanova, T.; Dortland, B.; Verkerk, T.; Vermeulen, W.; Burgering, B. M.; De Zeeuw, C. I.; Grosveld, F.; Galjart, N.: CLASPs are CLIP-115 and -170 associating proteins involved in the regional regulation of microtubule dynamics in motile fibroblasts. *Cell* 104: 923–935, 2001.

[36825] 10377. Cattanach, B. M.; Barr, J. A.; Beechey, C. V.; Martin, J.; Noebels, J.; Jones, J.: A candidate model for Angelman syndrome in the mouse. *Mammalian Genome* 8: 472–478, 1997.

- [36826] 10378.Dhar, M.; Webb, L. S.; Smith, L.; Hauser, L.; Johnson, D.; West,D. B.: A novel ATPase on mouse chromosome 7 is a candidate gene forincreased body fat. *Physiol. Genomics* 4: 93–100, 2000.
- [36827] 10379.Halleck, M. S.; Lawler, J. F., Jr.; Blackshaw, S.; Gao, L.; Nagarajan,P.; Hacker, C.; Pyle, S.; Newman, J. T.; Nakanishi, Y.; Ando, H.;Weinstock, D.; Williamson, P.; Schlegel, R. A.: Differential expressionof putative transbi-layer amhipath transporters. *Physiol. Genomics* 1:139–150, 1999.
- [36828] 10380.Fitzgibbon, J.; Neat, M. J.; Foot, N.; Hill, A. S.; Lister, T.A.; Gupta, R. K.: Assignment of brain acid-soluble protein 1 (BASP1)to human chromosome 5p15.1–p14, differential expression in human cancer cell lines as a result of alterations in gene dosage. *Cytogenet.Cell Genet.* 89: 147–149, 2000.
- [36829] 10381.Mosevitsky, M. I.; Capony, J. P.; Skladchikova, G. Y.; Novitskaya,V. A.; Plekhanov, A. Y.; Zakharov, V. V.: The BASP1 family of myristoylatedproteins abundant in axonal termini: primary structure analysis andphysico-chemical properties. *Biochimie* 79: 373–384, 1997.
- [36830] 10382.Park, S.; Kim, Y.–I.; Kim, B.; Seong, C.; Oh, Y.; Baek, K.; Yoon,J.: Characterization of bovine and human cDNAs

encoding NAP-22 (22kDa neuronal tissue-enriched acidic protein) homologs. *Molec. Cells* 8:471-477, 1998.

- [36831] 10383. Shichijo, S.; Nakao, M.; Imai, Y.; Takasu, H.; Kawamoto, M.; Niiya, F.; Yang, D.; Toh, Y.; Yamana, H.; Itoh, K.: A gene encoding antigenic peptides of human squamous cell carcinoma recognized by cytotoxic T lymphocytes. *J. Exp. Med.* 187: 277-288, 1998.
- [36832] 10384. Valenta, R.; Natter, S.; Seiberler, S.; Wichlas, S.; Maurer, D.; Hess, M.; Pavelka, M.; Grote, M.; Ferreira, F.; Szepfalusi, Z.; Valent, P.; Stingl, G.: Molecular characterization of an autoallergen, Homs 1, identified by serum IgE from atopic dermatitis patients. *J. Invest. Derm.* 111: 1178-1183, 1998.
- [36833] 10385. Loftus, B. J.; Kim, U.-J.; Sneddon, V. P.; Kalush, F.; Brandon, R.; Fuhrmann, J.; Mason, T.; Crosby, M. L.; Barnstead, M.; Cronin, L.; Mays, A. D.; Cao, Y.; Xu, R. X.; Kang, H.-L.; Mitchell, S.; Eichler, E. E.; Harris, P. C.; Venter, J. C.; Adams, M. D.: Genome duplications and other features in 12 Mb of DNA sequence from human chromosome 16p and 16q. *Genomics* 60: 295-308, 1999.
- [36834] 10386. Brauner-Osborne, H.; Krogsgaard-Larsen, P.: Sequence and expression pattern of a novel human orphan G-protein-coupled receptor, GPRC5B, a family C receptor

with a short amino-terminal domain. *Genomics* 65:121–128, 2000.

[36835] 10387. Robbins, M. J.; Michalovich, D.; Hill, J.; Calver, A. R.; Medhurst, A. D.; Gloger, I.; Sims, M.; Middlemiss, D. N.; Pangalos, M. N.: Molecular cloning and characterization of two novel retinoic acid-inducible orphan G-protein-coupled receptors (GPRC5B and GPRC5C). *Genomics* 67:8–18, 2000.

[36836] 10388. Autieri, M. V.; Feuerstein, G. Z.; Yue, T.-L.; Ohlstein, E. H.; Douglas, S. A.: Use of differential display to identify differentially expressed mRNAs induced by rat carotid artery balloon angioplasty. *Lab. Invest.* 72: 656–661, 1995.

[36837] 10389. Autieri, M. V.; Haines, D. S.; Romanic, A. M.; Ohlstein, E. H.: Expression of 14-3-3-gamma in injured arteries and growth factor- and cytokine-stimulated human vascular smooth muscle cells. *Cell Growth Diff.* 7: 1453–1460, 1996.

[36838] 10390. Horie, M.; Suzuki, M.; Takahashi, E.; Tanigami, A.: Cloning, expression, and chromosomal mapping of the human 14-3-3-gamma gene (YWHAG) to 7q11.23. *Genomics* 60:241–243, 1999.

[36839] 10391. Morrison, D.: 14-3-3: modulators of signaling

proteins? Science 266:56–57, 1994.

[36840] 10392.Brazas, R.; Ganem, D.: A cellular homolog of hepatitis delta antigen:implications for viral replication and evolution. Science 274: 90–94,1996.

[36841] 10393.Long, M.; de Souza, S. J.; Gilbert, W.: Delta-interacting proteinA and the origin of hepatitis delta antigen. Science 276: 824–825,1997.

[36842] 10394.Robertson, H. D.: How did replicating and coding RNAs first gettogether? Science 274: 66–67, 1996.

[36843] 10395.Asada, H.; Kawamura, Y.; Maruyama, K.; Kume, H.; Ding, R.–G.; Kanbara,N.; Kuzume, H.; Sanbo, M.; Yagi, T.; Obata, K.: Cleft palate anddecreased brain gamma-aminobutyric acid in mice lacking the 67–kDaisoform of glutamic acid decarboxylase. Proc. Nat. Acad. Sci. 94:6496–6499, 1997.

[36844] 10396.Brilliant, M. H.; Szabo, G.; Katarova, Z.; Kozak, C. A.; Glaser,T. M.; Greenspan, R. J.; Housman, D. E.: Sequences homologous toglutamic acid decarboxylase cDNA are present on mouse chromosomes2 and 10. Genomics 6: 115–122, 1990.

[36845] 10397.Bu, D.–F.; Tobin, A. J.: The exon–intron organization of the genes(GAD1 and GAD2) encoding two human glutamate decarboxylases (GAD–67and GAD–65) suggests

that they derive from a common ancestral GAD. *Genomics* 21:222–228, 1994.

[36846] 10398. Condie, B. G.; Bain, G.; Gottlieb, D. I.; Capecchi, M. R.: Cleftpalate in mice with a targeted mutation in the gamma-aminobutyricacid-producing enzyme glutamic acid decarboxylase 67. *Proc. Nat.Acad. Sci.* 94: 11451–11455, 1997.

[36847] 10399. Erlander, M. G.; Tillakaratne, N. J. K.; Feldblum, S.; Patel, N.; Tobin, A. J.: Two genes encode distinct glutamate decarboxylases. *Neuron* 7:91–100, 1991.

[36848] 10400. Kelly, C. D.; Edwards, Y.; Johnstone, A. P.; Harfst, E.; Nogradi, A.; Nussey, S. S.; Povey, S.; Carter, N. D.: Nucleotide sequence and chromosomal assignment of a cDNA encoding the large isoform of human glutamate decarboxylase. *Ann. Hum. Genet.* 56: 255–265, 1992.

[36849] 10401. Krishnamoorthy, K. S.: Pyridoxine-dependency seizure: report of a rare presentation. *Ann. Neurol.* 13: 103–104, 1983.

[36850] 10402. Sparkes, R. S.; Kaufman, D. L.; Heinzmann, C.; Tobin, A. J.; Mohandas, T.: Brain glutamate decarboxylase (GAD) gene assigned to human chromosome 2 by somatic cell hybrid analysis. (Abstract) *Cytogenet. Cell Genet.* 46:696 only, 1987.

- [36851] 10403.Kedra, D.; Seroussi, E.; Fransson, I.; Trifunovic, J.; Clark, M.; Lagercrantz, J.; Blennow, E.; Mehlin, H.; Duman-ski, J.: The germinalcenter kinase gene and a novel CDC25-like gene are located in the vicinity of the PYGM gene on 11q13. *Hum. Genet.* 100: 611–619, 1997.
- [36852] 10404.Hirsch, D. S.; Pirone, D. M.; Burbelo, P. D.: A new family of Cdc42 effector proteins, CEPs, function in fibroblast and epithelial cell shape changes. *J. Biol. Chem.* 276: 875–883, 2001.
- [36853] 10405.Joberty, G.; Perlungher, R. R.; Macara, I. G.: The Borgs, a new family of Cdc42 and TC10 GTPase-interacting proteins. *Molec. Cell Biol.* 19: 6585–6597, 1999.
- [36854] 10406.Yang, Z.-Q.; Imoto, I.; Fukuda, Y.; Pimkhaokham, A.; Shimada, Y.; Imamura, M.; Sugano, S.; Nakamura, Y.; Inazawa, J.: Identification of a novel gene, GASC1, within an amplicon at 9p23–24 frequently detected in esophageal cancer cell lines. *Cancer Res.* 60: 4735–4739, 2000.
- [36855] 10407.LeCouter, J.; Kowalski, J.; Foster, J.; Hass, P.; Zhang, Z.; Dillard-Telm, L.; Frantz, G.; Rangell, L.; DeGuzman, L.; Keller, G.-A.; Peale, F.; Gurney, P.; Hillan, K. J.; Ferrara, N.: Identification of an angiogenic mitogen selective for endocrine gland endothelium. *Nature* 412: 877–884, 2001.

- [36856] 10408.Li, M.; Bullock, C. M.; Knauer, D. J.; Ehlert, F. J.; Zhou, Q.Y.: Identification of two prokineticin cDNAs: recombinant proteinspotently contract gastrointestinal smooth muscle. *Molec. Pharm.* 59:692–698, 2001.
- [36857] 10409.Sun, Q.–A.; Wu, Y.; Zappacosta, F.; Jeang, K.–T.; Lee, B. J.; Hatfield,D. L.; Gladyshev, V. N.: Redox regulation of cell signaling by selenocysteinein mammalian thioredoxin reductases. *J. Biol. Chem.* 274: 24522–24530,1999.
- [36858] 10410.Nagase, T.; Nakayama, M.; Nakajima, D.; Kikuno, R.; Ohara, O.:Prediction of the coding sequences of unidentified human genes. XX.The complete sequences of 100 new cDNA clones from brain which codefor large proteins in vitro. *DNA Res.* 8: 85–95, 2001.
- [36859] 10411.Telford, E. A. R.; Moynihan, L. M.; Markham, A. F.; Lench, N. J.: Isolation and characterisation of a cDNA encoding the precursorfor a novel member of the acyl–CoA dehydrogenase gene family. *Biochim.Biophys. Acta* 1446: 371–376, 1999.
- [36860] 10412.Jaquemar, D.; Schenker, T.; Trueb, B.: An ankyrin-like proteinwith transmembrane domains is specifically lost after oncogenic transformationof human fibroblasts. *J. Biol. Chem.* 274: 7325–7333, 1999.

- [36861] 10413.Schenker, T.; Trueb, B.: Down-regulated proteins of mesenchymaltumor cells. *Exp. Cell Res.* 239: 161–168, 1998.
- [36862] 10414.Cerretti, D. P.; DuBose, R. F.; Black, R. A.; Nelson, N.: Isolationof two novel metalloproteinase–disintegrin (ADAM) cDNAs that showtestis–specific gene expression. *Biochem. Biophys. Res. Commun.* 263:810–815, 1999.
- [36863] 10415.Xu, R.; Cai, J.; Xu, T.; Zhou, W.; Ying, B.; Deng, K.; Zhao, S.;Li, C.: Molecular cloning and mapping of a novel ADAM gene (ADAM29)to human chromosome 4. *Genomics* 62: 537–539, 1999.
- [36864] 10416.Jacobs, S.; Schilf, C.; Fliegert, F.; Koling, S.; Weber, Y.; Schurmann,A.; Joost, H.–G.: ADP–ribosylation factor (ARF)–like 4, 6, and 7represent a subgroup of the ARF family characterized by rapid nucleotideexchange and a nuclear localization signal. *FEBS Lett.* 456: 384–388,1999.
- [36865] 10417.Lako, M.; Lindsay, S.; Bullen, P.; Wilson, D. I.; Robson, S. C.;Strachan, T.: A novel mammalian Wnt gene, WNT8B, shows brain–restrictedexpression in early development, with sharply delimited expressionboundaries in the developing forebrain. *Hum. Molec. Genet.* 7: 813–822,1998.
- [36866] 10418.Lako, M.; Strachan, T.; Curtis, A. R. J.; Lindsay, S.:

Isolation and characterization of WNT8B, a novel human Wnt gene that maps to 10q24. *Genomics* 35: 386–388, 1996.

[36867] 10419. Toda, T.; Iida, A.; Miwa, T.; Nakamura, Y.; Imai, T.: Isolation and characterization of a novel gene encoding nuclear protein at a locus (D11S636) tightly linked to multiple endocrine neoplasia type 1 (MEN1). *Hum. Molec. Genet.* 3: 465–470, 1994.

[36868] 10420. Nusse, R.; Varmus, H. E.: Many tumors induced by the mouse mammary tumor virus contain a provirus integrated in the same region of the host genome. *Cell* 31: 99–109, 1982.

[36869] 10421. Scholler, J. K.; Kanner, S. B.: The human p167 gene encodes a unique structural protein that contains centrosomin A homology and associates with a multicomponent complex. *DNA Cell Biol.* 16: 515–531, 1997.

[36870] 10422. Abdulkadir, S. A.; Magee, J. A.; Peters, T. J.; Kaleem, Z.; Naughton, C. K.; Humphrey, P. A.; Milbrandt, J.: Conditional loss of Nkx3.1 in adult mice induces prostatic intraepithelial neoplasia. *Molec. Cell. Biol.* 22: 1495–1503, 2002.

[36871] 10423. Bieberich, C. J.; Fujita, K.; He, W. W.; Jay, G.: Prostate-specific and androgen-dependent expression of a

novel homeobox gene. J. Biol.Chem. 271: 31779–31782, 1996.

- [36872] 10424.Korkmaz, K. S.; Korkmaz, C. G; Ragnhildstveit, E.; Kizildag, S.;Pretlow, T. G.; Saatcioglu, F.: Full-length cDNA sequence and genomicorganization of human NKX3A--alternative forms and regulation by bothandro-gens and estrogens. Gene 260: 25–36, 2000.
- [36873] 10425.He, W. W.; Sciavolino, P. J.; Wing, J.; Augustus, M.; Hudson, P.;Meissner, P. S.; Curtis, R. T.; Shell, B. K.; Bost-wick, D. G.; Tindall,D. J.; Gelmann, E. P.; Abate-Shen, C.; Carter, K. C.: A novel humanprostate-specific, androgen-regulated homeobox gene (NKX3.1) thatmaps to 8p21, a region frequently deleted in prostate cancer. Genomics 43:69–77, 1997.
- [36874] 10426.Argyropoulos, G.; Brown, A. M.; Willi, S. M.; Zhu, J.; He, Y.;Reitman, M.; Gevao, S. M.; Spruill, I.; Garvey, W. T.: Effects ofmutations in the human uncoupling protein 3 gene on the respiratoryquotient and fat oxidation in se-vere obesity and type 2 diabetes. J.Clin. Invest. 102: 1345–1351, 1998.
- [36875] 10427.Boss, O.; Giacobino, J.-P.; Muzzin, P.: Genomic structure of uncouplingprotein-3 (UCP3) and its assign-ment to chromosome 11q13. Genomics 47:425–426,

1998.

- [36876] 10428.Boss, O.; Samec, S.; Paoloni-Giacobino, A.; Rossier, C.; Dulloo, A.; Seydoux, J.; Muzzin, P.; Giacobino, J.-P.: Uncoupling protein-3: a new member of the mitochondrial carrier family with tissue-specific expression. *FEBS Lett.* 408: 39–42, 1997.
- [36877] 10429.Brown, A. M.; Dolan, J. W.; Willi, S. M.; Garvey, W. T.; Argyropoulos, G.: Endogenous mutations in human uncoupling protein 3 alter its functional properties. *FEBS Lett.* 464: 189–193, 1999.
- [36878] 10430.Bilaud, T.; Brun, C.; Ancelin, K.; Koering, C. E.; Laroche, T.; Gilson, E.: Telomeric localization of TRF2, a novel human telobox protein. *Nature Genet.* 17: 236–239, 1997.
- [36879] 10431.Broccoli, D.; Smogorzewska, A.; Chong, L.; de Lange, T.: Human telomeres contain two distinct Myb-related proteins, TRF1 and TRF2. *Nature Genet.* 17: 231–235, 1997.
- [36880] 10432.Karlseder, J.; Smogorzewska, A.; de Lange, T.: Senescence induced by altered telomere state, not telomere loss. *Science* 295: 2446–2449, 2002.
- [36881] 10433.Sakaguchi, A. Y.; Padalecki, S. S.; Mattern, V.; Rodriguez, A.; Leach, R. J.; McGill, J. R.; Chavez, M.; Gi-

ambernardi, T. A.: Chromosomal sublocalization of the transcribed human telomere repeat binding factor 2 gene and comparative mapping in the mouse. *Somat. Cell Molec. Genet.* 24:157–163, 1998.

[36882] 10434. van Steensel, B.; Smogorzewska, A.; de Lange, T.: TRF2 protects human telomeres from end-to-end fusions. *Cell* 92: 401–413, 1998.

[36883] 10435. Parr, B. A.; McMahon, A. P.: Sexually dimorphic development of the mammalian reproductive tract requires Wnt-7a. *Nature* 395: 707–710, 1998.

[36884] 10436. Barron-Casella, E. A.; Torres, M. A.; Scherer, S. W.; Heng, H. H. Q.; Tsui, L.-C.; Casella, J. F.: Sequence analysis and chromosomal localization of human Cap Z: conserved residues within the actin-binding domain may link Cap Z to gelsolin/severin and profilin protein families. *J. Biol. Chem.* 270: 21472–21479, 1995.

[36885] 10437. Hart, M. C.; Korshunova, Y. O.; Cooper, J. A.: Mapping of the mouse actin capping protein alpha subunit genes and pseudogenes. *Genomics* 39:264–270, 1997.

[36886] 10438. Cooper, J. A.: Personal Communication. St. Louis, Mo. 3/8/1999.

[36887] 10439. Cardoso, C.; Mignon, C.; Hetet, G.; Grandchamps, B.; Fontes, M.; Colleaux, L.: The human EZH2 gene: ge-

nomie organisation and revised mapping in 7q35 within the critical region for malignant myeloid disorders. *Eur. J. Hum. Genet.* 8: 174–180, 2000.

- [36888] 10440. Chen, H.; Rossier, C.; Antonarakis, S. E.: Cloning of a human homolog of the *Drosophila* enhancer of zeste gene (EZH2) that maps to chromosome 21q22.2. *Genomics* 38: 30–37, 1996.
- [36889] 10441. Dohner, K.; Brown, J.; Hehmann, U.; Hetzel, C.; Stewart, J.; Lowther, G.; Scholl, C.; Frohling, S.; Cuneo, A.; Tsui, L. C.; Lichter, P.; Scherer, S. W.; Dohner, H.: Molecular cytogenetic characterization of a critical region in bands 7q35–q36 commonly deleted in malignant myeloid disorders. *Blood* 92: 4031–4035, 1998.
- [36890] 10442. Laible, G.; Haynes, A. R.; Lebersorger, A.; O'Carroll, D.; Mattei, M.-G.; Denny, P.; Brown, S. D. M.; Jenuwein, T.: The murine polycomb-group genes *Ezh1* and *Ezh2* map close to Hox gene clusters on mouse chromosomes 11 and 6. *Mammalian Genome* 10: 311–314, 1999.
- [36891] 10443. Varambally, S.; Dhanasekaran, S. M.; Zhou, M.; Barrette, T. R.; Kumar-Sinha, C.; Sanda, M. G.; Ghosh, D.; Pienta, K. J.; Sewalt, R. G. A. B.; Otte, A. P.; Rubin, M. A.; Chinnaiyan, A. M.: The polycomb group protein EZH2 is involved in progression of prostate cancer. *Nature*

419:624–629, 2002.

[36892] 10444.Morohoshi, F.; Arai, K.; Takahashi, E.; Tanigami, A.; Ohki, M.: Cloning and mapping of a human RBP56 gene encoding a putative RNA binding protein similar to FUS/TLS and EWS proteins. *Genomics* 38:51–57, 1996.

[36893] 10445.Batt, J.; Asa, S.; Fladd, C.; Rotin, D.: Pituitary, pancreatic and gut neuroendocrine defects in protein tyrosine phosphatase-sigma-deficient mice. *Molec. Endocr.* 16: 155–169, 2002.

[36894] 10446.Elchebly, M.; Wagner, J.; Kennedy, T. E.; Lanctot, C.; Michaliszyn, E.; Itie, A.; Drouin, J.; Tremblay, M. L.: Neuroendocrine dysplasia in mice lacking protein tyrosine phosphatase sigma. *Nature Genet.* 21:330–333, 1999.

[36895] 10447.Pulido, R.; Serra-Pages, C.; Tang, M.; Streuli, M.: The LAR/PTPdelta/PTP sigma subfamily of transmembrane protein-tyrosine-phosphatases: multiple human LAR, PTP delta, and PTP sigma isoforms are expressed in a tissue-specific manner and associate with the LAR-interacting protein LIP.1. *Proc. Nat. Acad. Sci.* 92: 11686–11690, 1995.

[36896] 10448.Wagner, J.; Gordon, L. A.; Heng, H. H. Q.; Tremblay, M. L.; Olsen, A. S.: Physical mapping of receptor type protein tyrosine phosphatase sigma (PTPRS) to human chro-

mosome 19p13.3. Genomics 38: 76–78, 1996.

- [36897] 10449.Wallace, M. J.; Batt, J.; Fladd, C. A.; Henderson, J. T.; Skarnes,W.; Rotin, D.: Neuronal defects and posterior pituitary hypoplasia in mice lacking the receptor tyrosine phosphatase PTP-sigma. Nature Genet. 21: 334–338, 1999.
- [36898] 10450.Bruyns, E.; Mincheva, A.; Bruyns, R. M.; Kirchgessner, H.; Weitz,S.; Lichter, P.; Meuer, S.; Schraven, B.: Sequence, genomic organization, and chromosomal localization of the human LPAP (PTPRCAP) and mouse CD45-AP/LSM-1 genes. Genomics 38: 79–83, 1996.
- [36899] 10451.Schraven, B.; Schoenhaut, D.; Bruyns, E.; Koretzky, G.; Eckerskorn,C.; Wallich, R.; Kirchgessner, H.; Sakorafas, P.; Labkovsky, B.; Ratnofsky,S.; Meuer, S.: LPAP, a novel 32-kDa phosphoprotein that interacts with CD45 in human lymphocytes. J. Biol. Chem. 269: 29102–29111, 1994.
- [36900] 10452.Spicer, A. P.; Augustine, M. L.; McDonald, J. A.: Molecular cloning and characterization of a putative mouse hyaluronan synthase. J. Biol. Chem. 271: 23400–23406, 1996.
- [36901] 10453.Skalhegg, B. S.; Huang, Y.; Su, T.; Idzerda, R. L.; McKnight, G.S.; Burton, K. A.: Mutation of the C-alpha

subunit of PKA leads to growth retardation and sperm dysfunction. *Molec. Endocr.* 16: 630–639, 2002.

- [36902] 10454.Tasken, K.; Solberg, R.; Zhao, Y.; Hansson, V.; Jahnsen, T.; Siciliano, M. J.: The gene encoding the catalytic subunit C- α of cAMP-dependent protein kinase (locus PRKACA) localizes to human chromosome region 19p13.1. *Genomics* 36: 535–538, 1996.
- [36903] 10455.Kim, J.; Uyemura, K.; Van Dyke, M. K.; Legaspi, A. J.; Rea, T.H.; Shuai, K.; Modlin, R. L.: A role for IL-12 receptor expression and signal transduction in host defense in leprosy. *J. Immun.* 167:779–786, 2001.
- [36904] 10456.Morton, S. M.; Bocaccio, I.; Depetris, D.; Mattei, M.; Dessein, A.: Assignment of IL12RB2 to human chromosome 1p31.3–p31.2 between D1S230 and D1S198. *Cytogenet. Cell Genet.* 79: 282–283, 1997.
- [36905] 10457.McCright, B.; Brothman, A. R.; Virshup, D. M.: Assignment of human protein phosphatase 2A regulatory subunit genes B56- α , B56- β , B56- γ , B56- δ , and B56- ϵ (PPP2R5A–PPP2R5E), highly expressed in muscle and brain, to chromosome regions 1q41, 11q12, 3p21, 6p21.1, and 7p11.2–to–p12. *Genomics* 36: 168–170, 1996.
- [36906] 10458.McCright, B.; Virshup, D. M.: Identification of a new

family of protein phosphatase 2A regulatory subunits. J. Biol. Chem. 270:26123–26128, 1995.

- [36907] 10459. Horikawa, S.; Tsukada, K.: Molecular cloning and developmental expression of a human kidney S-adenosylmethionine synthetase. FEBS Lett. 312: 37–41, 1992.
- [36908] 10460. Mao, Z.; Liu, S.; Cai, J.; Huang, Z.-Z.; Lu, S. C.: Cloning and functional characterization of the 5-prime-flanking region of human methionine adenosyltransferase 2A gene. Biochem. Biophys. Res. Commun. 248:479–484, 1998.
- [36909] 10461. Clark, J. A.; Bonner, T. I.; Kim, A. S.; Usdin, T. B.: Multiple regions of ligand discrimination revealed by analysis of chimeric parathyroid hormone 2 (PTH2) and PTH/PTH-related peptide (PTHrP) receptors. Molec. Endocr. 12: 193–206, 1998.
- [36910] 10462. Usdin, T. B.; Gruber, C.; Bonner, T. I.: Identification and functional expression of a receptor selectively recognizing parathyroid hormone, the PTH2 receptor. J. Biol. Chem. 270: 15455–15458, 1995.
- [36911] 10463. Usdin, T. B.; Modi, W.; Bonner, T. I.: Assignment of the human PTH2 receptor gene (PTHR2) to chromosome 2q33 by fluorescence in situ hybridization. Genomics 37:

140–141, 1996.

- [36912] 10464. Faure, S.; Meyer, L.; Costagliola, D.; Vaneensberghe, C.; Genin, E.; Autran, B.; French ALT and IMMUNOCO Study Groups; Delfraissy, J.-F.; SEROCO Study Group; McDermott, D. H.; Murphy, P. M.; Debre, P.; Theodorou, I.; Cambadiere, C.: Rapid progression to AIDS in HIV+ individuals with a structural variant of the chemokine receptor CX(3)CR1. *Science* 287:2274–2277, 2000.
- [36913] 10465. Imai, T.; Hieshima, K.; Haskell, C.; Baba, M.; Nagira, M.; Nishimura, M.; Kakizaki, M.; Takagi, S.; Nomiyama, H.; Schall, T. J.; Yoshie, O.: Identification and molecular characterization of fractalkine receptor CX3CR1, which mediates both leukocyte migration and adhesion. *Cell* 91:521–530, 1997.
- [36914] 10466. Moatti, D.; Faure, S.; Fumeron, F.; Amara, M. E. W.; Seknadji, P.; McDermott, D. H.; Debre, P.; Aumont, M. C.; Murphy, P. M.; de Prost, D.; Combadiere, C.: Polymorphism in the fractalkine receptor CX3CR1 as a genetic risk factor for coronary artery disease. *Blood* 97:1925–1928, 2001.
- [36915] 10467. Raport, C. J.; Schweickart, V. L.; Eddy, R. L., Jr.; Shows, T. B.; Gray, P. W.: The orphan G-protein-coupled receptor-encoding gene V28 is closely related to genes for

chemokine receptors and is expressed in lymphoid and neural tissues. *Gene* 163: 295–299, 1995.

[36916] 10468. Tripp, R. A.; Jones, L. P.; Haynes, L. M.; Zheng, H.; Murphy, P. M.; Anderson, L. J.: CX3C chemokine mimicry by respiratory syncytial virus G glycoprotein. *Nature Immun.* 2: 732–738, 2001.

[36917] 10469. Lafreniere, R. G.; Rochefort, D. L.; Chretien, N.; Neville, C. E.; Korneluk, R. G.; Zuo, L.; Wei, Y.; Lichter, J.; Rouleau, G. A.: Isolation and genomic structure of a human homolog of the yeast periodic tryptophan protein 2 (PWP2) gene mapping to 21q22.3. *Genome Res.* 6: 1216–1226, 1996.

[36918] 10470. Lalioti, M. D.; Chen, H.; Rossier, C.; Shafaatian, R.; Reid, J. D.; Antonarakis, S. E.: Cloning the cDNA of human PWP2, which encodes a protein with WD repeats and maps to 21q22.3. *Genomics* 35: 321–327, 1996.

[36919] 10471. Yamakawa, K.; Gao, D.-Q.; Korenberg, J. R.: A periodic tryptophan protein 2 gene homologue (PWP2H) in the candidate region of progressive myoclonus epilepsy on 21q22.3. *Cytogenet. Cell Genet.* 74: 140–145, 1996.

[36920] 10472. Hanes, J.; von der Kammer, H.; Klaudiny, J.; Scheit, K. H.: Characterization by cDNA cloning of two new human protein kinases: evidence by sequence comparison of a

new family of mammalian protein kinases. *J. Molec.Biol.* 244: 665–672, 1994.

- [36921] 10473.Talmadge, C. B.; Finkernagel, S.; Sumegi, J.; Sciorra, L.; Rabinow,L.: Chromosomal mapping of three human LAMMER protein–kinase–encodinggenes. *Hum. Genet.* 103: 523–524, 1998.
- [36922] 10474.Andersen, G.; Poterszman, A.; Egly, J. M.; Moras, D.; Thierry,J.–C: The crystal structure of human cyclin H. *FEBS Lett.* 397:65–69, 1996.
- [36923] 10475.Eki, T.; Okumura, K.; Abe, M.; Kagotani, K.; Taguchi, H.; Murakami,Y.; Pan, Z.–Q.; Hanaoka, F.: Mapping of the human genes encodingcyclin H (CCNH) and the CDK–activating kinase (CAK) assembly factorMAT1 (MNAT1) to chromosome bands 5q13.3–q14 and 14q23, respectively. *Genomics* 47:115–120, 1998.
- [36924] 10476.Fisher, R. P.; Morgan, D. O.: A novel cyclin associates with MO15/CDK7to form the CDK–activating kinase. *Cell* 78: 713–724, 1994.
- [36925] 10477.Shiekhattar, R.; Mermelstein, F.; Fisher, R. P.; Drapkin, R.; Dynlacht,B.; Wessling, H. C.; Morgan, D. O.; Reinberg: Cdk–activating kinasecomplex is a component of human transcription factor TFIIH. *Nature* 374:283–287, 1995.

- [36926] 10478.Alders, M.; Hodges, M.; Hadjantonakis, A.-K.; Postmus, J.; vanWijk, I.; Blik, J.; de Meulemeester, M.; Westerveld, A.; Guillemot, F.; Oudejans, C.; Little, P.; Mannens, M.: The human Achaete-Scutehomologue 2 (ASCL2, HASH2) maps to chromosome 11p15.5, close to IGF2 and is expressed in extravillous trophoblasts. *Hum. Molec. Genet.* 6:859-867, 1997.
- [36927] 10479.Guillemot, F.; Nagy, A.; Auerbach, A.; Rossant, J.; Joyner, A.L.: Essential role of Mash-2 in extraembryonic development. *Nature* 371:333-336, 1994.
- [36928] 10480.Banga, S. S.; Ozer, H. L.; Park, S.-K.; Lee, S.-T.: Assignment of PTK7 encoding a receptor protein tyrosine kinase-like molecule to human chromosome 6p21.1-p12.2 by fluorescence in situ hybridization. *Cytogenet. Cell Genet.* 76: 43-44, 1997.
- [36929] 10481.Lee, S.-T.; Strunk, K. M.; Spritz, R. A.: A survey of protein tyrosine kinase mRNAs expressed in normal human melanocytes. *Oncogene* 8:3403-3410, 1993.
- [36930] 10482.Mossie, K.; Jallal, B.; Alves, F.; Sures, I.; Plowman, G. D.; Ullrich, A.: Colon carcinoma kinase-4 defines a new subclass of the receptor tyrosine kinase family. *Oncogene* 11: 2179-2184, 1995.
- [36931] 10483.Park, S.-K.; Lee, H.-S.; Lee, S.-T.: Characterization

of the human full-length PTK7 cDNA encoding a receptor protein tyrosine kinase-like molecule closely related to chick KLG. *J. Biochem.* 119: 235–239, 1996.

[36932] 10484. Sotiropoulou, G.; Anisowicz, A.; Sager, R.: Identification, cloning, and characterization of cystatin M, a novel cysteine proteinase inhibitor, down-regulated in breast cancer. *J. Biol. Chem.* 272: 903–910, 1997.

[36933] 10485. Stenman, G.; Astrom, A.-K.; Roijer, E.; Sotiropoulou, G.; Zhang, M.; Sager, R.: Assignment of a novel cysteine proteinase inhibitor (CST6) to 11q13 by fluorescence in situ hybridization. *Cytogenet. Cell Genet.* 76: 45–46, 1997.

[36934] 10486. Kohler, M.; Ansieau, S.; Prehn, S.; Leutz, A.; Haller, H.; Hartmann, E.: Cloning of two novel human importin- α subunits and analysis of the expression pattern of the importin- α protein family. *FEBS Lett.* 417: 104–108, 1997.

[36935] 10487. Moore, M. S.; Blobel, G.: The two steps of nuclear import, targeting to the nuclear envelope and translocation through the nuclear pore, require different cytosolic factors. *Cell* 69: 939–950, 1992.

[36936] 10488. Takeda, S.; Fujiwara, T.; Shimizu, F.; Kawai, A.; Shinomiya, K.; Okuno, S.; Ozaki, K.; Katagiri, T.; Shimada, Y.;

Nagata, M.; Watanabe, T.; Takaichi, A.; Kuga, Y.; Suzuki, M.; Hishigaki, H.; Takahashi, E.; Shin, S.; Nakamura, Y.; Hirai, Y.: Isolation and mapping of karyopherin α -3 (KPNA3), a human gene that is highly homologous to genes encoding *Xenopus* importin, yeast SRP1 and human RCH1. *Cytogenet. Cell Genet.* 76:87–93, 1997.

[36937] 10489. Zhang, C.-Y.; Baffy, G.; Perret, P.; Krauss, S.; Peroni, O.; Grujic, D.; Hagen, T.; Vidal-Puig, A.; Boss, O.; Kim, Y.-B.; Zheng, X. X.; Wheeler, M. B.; Shulman, G. I.; Chan, C. B.; Lowell, B. B.: Uncoupling protein-2 negatively regulates insulin secretion and is a major link between obesity, beta cell dysfunction, and type 2 diabetes. *Cell* 105:745–755, 2001.

[36938] 10490. Edlund, A.; Johansson, T.; Leidvik, B.; Hansson, L.: Structure of the human kappa-casein gene. *Gene* 174: 65–69, 1996.

[36939] 10491. Sprecher, C. A.; Morgenstern, K. A.; Mathewes, S.; Dahlen, J. R.; Schrader, S. K.; Foster, D. C.; Kisiel, W.: Molecular cloning, expression, and partial characterization of two novel members of the ovalbumin family of serine proteinase inhibitors. *J. Biol. Chem.* 270: 29854–29861, 1995.

[36940] 10492. Chevalier, D.; Cauffiez, C.; Bernard, C.; Lo-Guidice,

J.-M.; Allorge,D.; Fazio, F.; Ferrari, N.; Libersa, C.; Lhermitte, M.; D'Halluin,J.-C.; Broly, F.: Characterization of new mutations in the codingsequence and 5-prime-untranslated region of the human prostacyclin-synthase gene (CYP8A1). Hum. Genet. 108: 148-155, 2001.

[36941] 10493.Barrett, A. J.; Rawlings, N. D.: Evolutionary lines of cysteinepeptidases. Biol. Chem. 382: 727-733, 2001.

[36942] 10494.Fernandes-Alnemri, T.; Takahashi, A.; Armstrong, R.; Krebs, J.;Fritz, L.; Tomaselli, K. J.; Wang, L.; Yu, Z.; Croce, C. M.; Salveson,G.; Earnshaw, W. C.; Litwack, G.; Alnemri, E. S.: Mch3, a novel humanapoptotic cysteine protease highly related to CPP32. Cancer Res. 55:6045-6052, 1995.

[36943] 10495.Riedl, S. J.; Fuentes-Prior, P.; Renatus, M.; Kairies, N.; Krapp,S.; Huber, R.; Salvesen, G. S.; Bode, W.: Structural basis for theactivation of human procaspase-7. Proc. Nat. Acad. Sci. 98: 14790-14795,2001.

[36944] 10496.Park, W. S.; Lee, J. H.; Shin, M. S.; Park, J. Y.; Kim, H. S.;Lee, J. H.; Kim, Y. S.; Lee, S. N.; Xiao, W.; Park, C. H.; Lee, S.H.; Yoo, N. J.; Lee, J. Y.: Inactivating mutations of the caspase-10gene in gastric cancer. Oncogene 21: 2919-2925, 2002.

- [36945] 10497. Shin, M. S.; Kim, H. S.; Kang, C. S.; Park, W. S.; Kim, S. Y.; Lee, S. N.; Lee, J. H.; Park, J. Y.; Jang, J. J.; Kim, C. W.; Kim, S. H.; Lee, J. Y.; Yoo, N. J.; Lee, S. H.: Inactivating mutations of CASP10 gene in non-Hodgkin lymphomas. *Blood* 99: 4094–4099, 2002.
- [36946] 10498. Vincenz, C.; Dixit, V. M.: Fas-associated death domain protein interleukin-1-beta-converting enzyme 2 (FLICE2), an ICE/Ced-3 homologue, is proximally involved in CD95- and p55-mediated death signaling. *J. Biol. Chem.* 272: 6578–6583, 1997.
- [36947] 10499. Wang, J.; Chun, H. J.; Wong, W.; Spencer, D. M.; Lenardo, M. J.: Caspase-10 is an initiator caspase in death receptor signaling. *Proc. Nat. Acad. Sci.* 98: 13884–13888, 2001.
- [36948] 10500. Wang, J.; Zheng, L.; Lobito, A.; Chan, F. K.; Dale, J.; Sneller, M.; Yao, X.; Puck, J. M.; Straus, S. E.; Lenardo, M. J.: Inherited human caspase 10 mutations underlie defective lymphocyte and dendritic cell apoptosis in autoimmune lymphoproliferative syndrome type II. *Cell* 98: 47–58, 1999.
- [36949] 10501. Chun, H. J.; Zheng, L.; Ahmad, M.; Wang, J.; Speirs, C. K.; Siegel, R. M.; Dale, J. K.; Puck, J.; Davis, J.; Hall, C. G.; Skoda-Smith, S.; Atkinson, T. P.; Straus, S. E.; Lenardo, M.

J.: Pleiotropic defects in lymphocyte activation caused by caspase-8 mutations lead to human immunodeficiency. *Nature* 419: 395–399, 2002.

[36950] 10502. Grenet, J.; Teitz, T.; Wei, T.; Valentine, V.; Kidd, V. J.: Structure and chromosome localization of the human CASP8 gene. *Gene* 226: 225–232, 1999.

[36951] 10503. Kischkel, F. C.; Kioschis, P.; Weitz, S.; Poustka, A.; Lichter, P.; Krammer, P. H.: Assignment of CASP8 to human chromosome band 2q33–q34 and Casp8 to the murine syntenic region on chromosome 1B–proximal C by in situ hybridization. *Cytogenet. Cell Genet.* 82: 95–96, 1998.

[36952] 10504. Sanchez, I.; Xu, C.-J.; Juo, P.; Kakizaka, A.; Blenis, J.; Yuan, J.: Caspase-8 is required for cell death induced by expanded polyglutamine repeats. *Neuron* 22: 623–633, 1999.

[36953] 10505. Berkovic, S. F.; Kennerson, M. L.; Howell, R. A.; Scheffer, I. E.; Hwang, P. A.; Nicholson, G. A.: Phenotypic expression of benign familial neonatal convulsions linked to chromosome 20. *Arch. Neurol.* 51: 1125–1128, 1994.

[36954] 10506. Varfolomeev, E. E.; Schuchmann, M.; Luria, V.; Chinnikulkulchai, N.; Beckmann, J. S.; Mett, I. L.; Rebrikov, D.; Brodianski, V. M.; Kemper, O. C.; Kollet, O.; Lapidot, T.; Soffer, D.; Sobe, T.; Avraham, K. B.; Goncharov, T.; Holt-

mann, H.; Lonai, P.; Wallach, D.: Targeted disruption of the mouse Caspase 8 gene ablates cell death induction by the TNF receptors, Fas/Apo1, and DR3 and is lethal prenatally. *Immunity* 9:267–276, 1998.

[36955] 10507. Kalchman, M. A.; Koide, H. B.; McCutcheon, K.; Graham, R. K.; Nichol, K.; Nishiyama, K.; Kazemi-Esfarjani, P.; Lynn, F. C.; Wellington, C.; Metzler, M.; Goldberg, Y. P.; Kanazawa, I.; Gietz, R. D.; Hayden, M. R.: HIP1, a human homologue of *S. cerevisiae* Slp2, interacts with membrane-associated huntingtin in the brain. *Nature Genet.* 16:44–53, 1997.

[36956] 10508. Wanker, E. E.; Rovira, C.; Scherzinger, E.; Hasenbank, R.; Walter, S.; Tait, D.; Colicelli, J.; Lehrach, H.: HIP-1: a huntingtin interacting protein isolated by the yeast two-hybrid system. *Hum. Molec. Genet.* 6:487–495, 1997.

[36957] 10509. Eki, T.; Okumura, K.; Shiratori, A.; Abe, M.; Nogami, M.; Taguchi, H.; Shibata, T.; Murakami, Y.; Hanaoka, F.: Assignment of the closest human homologue (DNA2L; KIAA0083) of the yeast Dna2 helicase gene to chromosome band 10q21.3–q22.1 *Genomics* 37: 408–410, 1996.

[36958] 10510. Nagase, T.; Miyajima, N.; Tanaka, A.; Sazuka, T.; Seki, N.; Sato, S.; Tabata, S.; Ishikawa, K.; Kawarabayashi, Y.; Kotani, H.; Nomura, N.: Prediction of the coding se-

quences of unidentified human genes.III. The coding sequences of 40 new genes (KIAA0081–KIAA0120) deduced by analysis of cDNA clones from human cell line KG–1. DNA Res. 2:37–43, 1995.

- [36959] 10511.Kim, J. W.; Lee, Y.; Lee, I. A.; Kang, H. B.; Choe, Y. K.; Choe, I. S.: Cloning and expression of human cDNA encoding Na(+),K(+)-ATPase gamma-subunit. Biochim. Biophys. Acta 1350: 133–135, 1997.
- [36960] 10512.Langkopf, A.; Hammarback, J. A.; Muller, R.; Vallee, R. B.; Garner, C. C.: Microtubule-associated proteins 1A and LC2: Two proteins encoded in one messenger RNA. J. Biol. Chem. 267: 16561–16566, 1992.
- [36961] 10513.Sun, X.–H.; Copeland, N. G.; Jenkins, N. A.; Baltimore, D.: Id proteins Id1 and Id2 selectively inhibit DNA binding by one class of helix–loop–helix proteins. Molec. Cell. Biol. 11: 5603–5611, 1991.
- [36962] 10514.Kordeli, E.; Bennett, V.: Distinct ankyrin isoforms at neuron cell bodies and nodes of Ranvier resolved using erythrocyte ankyrin-deficient mice. J. Cell Biol. 114: 1243–1259, 1991.
- [36963] 10515.Reche, P. A.; Soumelis, V.; Gorman, D. M.; Clifford, T.; Liu, M.; Travis, M.; Zurawski, S. M.; Johnston, J.; Liu, Y.–J.; Spits, H.; de Waal Malefyt, R.; Kastelein, R. A.; Bazan,

J. F.: Human thymicstromal lymphopoietin preferentially stimulates myeloid cells. *J.Immun.* 167: 336–343, 2001.

[36964] 10516.Nagase, T.; Kikuno, R.; Nakayama, M.; Hirosawa, M.; Ohara, O.:Prediction of the coding sequences of unidentified human genes. XVIII.The complete sequences of 100 new cDNA clones from brain which codefor large proteins in vitro. *DNA Res.* 7: 273–281, 2000.

[36965] 10517.Mukai, J.; Hachiya, T.; Shoji–Hoshino, S.; Kimura, M. T.; Nadano,D.; Suvanto, P.; Hanaoka, T.; Li, Y.; Irie, S.; Greene, L. A.; Sato,T.–A.: NADE, a p75NTR–associated cell death executor, is involvedin signal transduction mediated by the common neurotrophin receptorp75NTR. *J. Biol. Chem.* 275: 17566–17570, 2000.

[36966] 10518.Rapp, G.; Freudenstein, J.; Klaudiny, J.; Mucha, J.; Wempe, F.;Zimmer, M.; Scheit, K. H.: Characterization of three abundant mRNAsfrom human ovarian granulosa cells. *DNA Cell Biol.* 9: 479–485, 1990.

[36967] 10519.Scott, A. F.: Personal Communication. Baltimore, Md. 10/8/2001.

[36968] 10520.Kurochkin, I. V.; Yonemitsu, N.; Funahashi, S.; No–mura, H.: ALEX1,a novel human armadillo repeat protein that is expressed differentiallyin normal tissues and carci–nomas. *Biochem. Biophys. Res. Commun.* 280:340–347,

2001.

- [36969] 10521. Nagase, T.; Ishikawa, K.; Miyajima, N.; Tanaka, A.; Kotani, H.; Nomura, N.; Ohara, O.: Prediction of the coding sequences of unidentified human genes. IX. The complete sequences of 100 new cDNA clones from brain which can code for large proteins in vitro. *DNA Res.* 5: 31–39, 1998.
- [36970] 10522. Chuang, T.–H.; Ulevitch, R. J.: Cloning and characterization of a sub-family of human Toll-like receptors: hTLR7, hTLR8 and hTLR9. *Europ. Cytokine Netw.* 11: 372–378, 2000.
- [36971] 10523. Du, X.; Poltorak, A.; Wei, Y.; Beutler, B.: Three novel mammalian toll-like receptors: gene structure, expression, and evolution. *Europ. Cytokine Netw.* 11: 362–371, 2000.
- [36972] 10524. Kadowaki, N.; Ho, S.; Antonenko, S.; de Waal Malefyt, R.; Kastelein, R. A.; Bazan, F.; Liu, Y.–J.: Subsets of human dendritic cell precursors express different Toll-like receptors and respond to different microbial antigens. *J. Exp. Med.* 194: 863–869, 2001.
- [36973] 10525. Wirth, B.; Schmidt, T.; Hahnen, E.; Rudnik-Schoneborn, S.; Krawczak, M.; Muller-Myhsok, B.; Schoning, J.; Zerres, K.: De novo rearrangements found in 2% of index patients with spinal muscular atrophy: mutational mechanisms, parental origin, mutation rate, and im-

plications for genetic counseling. Am. J. Hum. Genet. 61: 1102–1111, 1997.

[36974] 10526. Frattini, A.; Orchard, P. J.; Sobacchi, C.; Giliani, S.; Abinun, M.; Mattsson, J. P.; Keeling, D. J.; Andersson, A.-K.; Wallbrandt, P.; Zecca, L.; Notarangelo, L. D.; Vezzoni, P.; Villa, A.: Defects in TCIRG1 subunit of the vacuolar proton pump are responsible for a subset of human autosomal recessive osteopetrosis. Nature Genet. 25:343–346, 2000.

[36975] 10527. Afzal, A. R.; Rajab, A.; Fenske, C. D.; Oldridge, M.; Elanko, N.; Ternes-Pereira, E.; Tuysuz, B.; Murday, V. A.; Patton, M. A.; Wilkie, A. O. M.; Jeffery, S.: Recessive Robinow syndrome, allelic to dominant brachydactyly type B, is caused by mutation of ROR2. Nature Genet. 25:419–422, 2000.

[36976] 10528. van Bokhoven, H.; Celli, J.; Kayserili, H.; van Beusekom, E.; Balci, S.; Brussel, W.; Skovby, F.; Kerr, B.; Percin, E. F.; Akarsu, N.; Brunner, H. G.: Mutation of the gene encoding the ROR2 tyrosine kinase causes autosomal recessive Robinow syndrome. Nature Genet. 25:423–426, 2000.

[36977] 10529. den Dunnen, J. T.; Grootsholten, P. M.; Bakker, E.; Blonden, L. A. J.; Ginjaar, H. B.; Wapenaar, M. C.; van

Paassen, H. M. B.; van Broeckhoven, C.; Pearson, P. L.; van Ommen, G. J. B.: Topography of the Duchenne muscular dystrophy (DMD) gene: FIGE and cDNA analysis of 194 cases reveals 115 deletions and 13 duplications. *Am. J. Hum. Genet.* 45: 835–847, 1989.

[36978] 10530. Claudio, J. O.; Liew, C.-C.; Dempsey, A. A.; Cukerman, E.; Stewart, A. K.; Na, E.; Atkins, H. L.; Iscove, N. N.; Hawley, R. G.: Identification of sequence-tagged transcripts differentially expressed within the human hematopoietic hierarchy. *Genomics* 50: 44–52, 1998.

[36979] 10531. Claudio, J. O.; Liew, C.-C.; Ma, J.; Heng, H. H. Q.; Stewart, A. K.; Hawley, R. G.: Cloning and expression analysis of a novel WD repeat gene, WDR3, mapping to 1p12–p13. *Genomics* 59: 85–89, 1999.

[36980] 10532. Eng, B. H.; Guerinot, M. L.; Eide, D.; Saier, M. H., Jr.: Sequence analyses and phylogenetic characterization of the ZIP family of metal ion transport proteins. *J. Membr. Biol.* 166: 1–7, 1998.

[36981] 10533. Lioumi, M.; Ferguson, C. A.; Sharpe, P. T.; Freeman, T.; Marenholz, I.; Mischke, D.; Heizmann, C.; Ragoussis, J.: Isolation and characterization of human and mouse ZIRT, a member of the IRT1 family of transporters, mapping within the epidermal differentiation complex. *Genomics*

62:272–280, 1999.

- [36982] 10534.Lioumi, M.; Olavesen, M. G.; Nizetic, D.; Ragoussis, J.: High-resolutionYAC fragmentation map of 1q21. *Genomics* 49: 200–208, 1998.
- [36983] 10535.Leiper, J. M.; Santa Maria, J.; Chubb, A.; MacAllister, R. J.;Charles, I. G.; Whitley, G. S.; Vallance, P.: Identification of twohuman dimethylarginine dimethylaminohydrolases with distinct tissuedistributions and homology with microbial arginine deiminases. *Biochem.J.* 343: 209–214, 1999.
- [36984] 10536.Tran, C. T. L.; Fox, M. F.; Vallance, P.; Leiper, J. M.: Chromosomallocalization, gene structure, and expression pattern of DDAH1: comparisonwith DDAH2 and implications for evolutionary origins. *Genomics* 68:101–105, 2000.
- [36985] 10537.Vanhalst, K.; Kools, P.; Eynde, E. V.; van Roy, F.: The humanand murine protocadherin-beta one-exon gene families show high evolutionaryconservation, despite the difference in gene number. *FEBS Lett.* 495:120–125, 2001.
- [36986] 10538.Wu, Q.; Maniatis, T.: A striking organization of a large familyof human neural cadherin like cell adhesion genes. *Cell* 97: 779–790,1999.

- [36987] 10539. Katayama, H.; Ota, T.; Morita, K.; Terada, Y.; Suzuki, F.; Katoh, O.; Tatsuka, M.: Human AIM-1: cDNA cloning and reduced expression during endomitosis in megakaryocyte-lineage cells. *Gene* 224: 1-7, 1998.
- [36988] 10540. Kimura, M.; Matsuda, Y.; Yoshioka, T.; Sumi, N.; Okano, Y.: Identification and characterization of STK12/Aik2: a human gene related to aurora of *Drosophila* and yeast IPL1. *Cytogenet. Cell Genet.* 82: 147-152, 1998.
- [36989] 10541. Tatsuka, M.; Katayama, H.; Ota, T.; Tanaka, T.; Odashima, S.; Suzuki, F.; Terada, Y.: Multinuclearity and increased ploidy caused by overexpression of the aurora- and Ipl1-like midbody-associated protein mitotic kinase in human cancer cells. *Cancer Res.* 58: 4811-4816, 1998.
- [36990] 10542. Chen, C. M.; Kraut, N.; Groudine, M.; Weintraub, H.: I-mf, a novel myogenic repressor, interacts with members of the MyoD family. *Cell* 86: 731-741, 1996.
- [36991] 10543. Kraut, N.: The gene encoding I-mf (Mdfi) maps to human chromosome 6p21 and mouse chromosome 17. *Mammalian Genome* 8: 618-619, 1997.
- [36992] 10544. Kraut, N.; Snider, L.; Chen, C.-M. A.; Tapscott, S. J.; Groudine, M.: Requirement of the mouse I-mfa gene for placental development and skeletal patterning. *EMBO J.* 17: 6276-6288, 1998.

- [36993] 10545.Boynton, S.; Tully, T.: Latheo, a new gene involved in associative learning and memory in *Drosophila melanogaster*, identified from P-element mutagenesis. *Genetics* 131: 655–672, 1992.
- [36994] 10546.Pinto, S.; Quintana, D. G.; Smith, P.; Mihalek, R. M.; Hou, Z.-H.; Boynton, S.; Jones, C. J.; Hendricks, M.; Velinzon, K.; Wohlschlegel, J. A.; Austin, R. J.; Lane, W. S.; Tully, T.; Dutta, A.: Latheo encodes a subunit of the origin recognition complex and disrupts neuronal proliferation and adult olfactory memory when mutant. *Neuron* 23:45–54, 1999.
- [36995] 10547.Springer, J.; Nanda, I.; Hoehn, K.; Schmid, M.; Grummt, F.: Identification and chromosomal localization of murine ORC3, a new member of the mouse origin recognition complex. *Cytogenet. Cell Genet.* 87: 245–251, 1999.
- [36996] 10548.Tugal, T.; Zou-Yang, X. H.; Gavin, K.; Pappin, D.; Canas, B.; Kobayashi, R.; Hunt, T.; Stillman, B.: The Orc4p and Orc5p subunits of the *Xenopus* and human origin recognition complex are related to Orc1p and Cdc6p. *J. Biol. Chem.* 273: 32421–32429, 1998.
- [36997] 10549.Butcher, S.; Arney, K. L.; Cook, G. P.: MAFA-L, an ITIM-containing receptor encoded by the human NK cell gene complex and expressed by basophils and NK cells.

Europ. J. Immun. 28: 3755–3762, 1998.

[36998] 10550.Hanke, T.; Corral, L.; Vance, R. E.; Raulet, D. H.: 2F1 antigen,the mouse homolog of the rat 'mast cell function-associated antigen',is a lectin-like type II transmembrane receptor expressed by naturalkiller cells. Europ. J. Immun. 28: 4409–4417, 1998.

[36999] 10551.Lamers, M. B. A. C.; Lamont, A. G.; Williams, D. H.: Human MAFAhas alternatively spliced variants. Biochim. Biophys. Acta 1399:209–212, 1998.

[37000] 10552.Voehringer, D.; Kaufmann, M.; Pircher, H.: Genomic structure,alternative splicing, and physical mapping of the killer cell lectin-like receptor G1 gene (KLRG1), the mouse homologue of MAFA. Immunogenetics 52:206–211, 2001.

[37001] 10553.Bahler, M.: Myosins on the move to signal transduction. Curr.Opin. Cell Biol. 8: 18–22, 1996.

[37002] 10554.Gorman, S. W.; Haider, N. B.; Grieshammer, U.; Swiderski, R. E.;Kim E; Welch, J. W.; Searby, C.; Leng, S.; Carmi, R.; Sheffield, V.C.; Duhl, D. M.: The cloning and developmental expression of unconventionalmyosin IXA (MYO9A) a gene in the Bardet–Biedl syndrome (BBS4) regionat chromosome 15q22–q23. Genomics 59: 150–160, 1999.

[37003] 10555.Dunham, I.; Shimizu, N.; Roe, B. A.; Chissoe, S.;

Hunt, A. R.; Collins, J. E.; Bruskiewich, R.; Beare, D. M.; Clamp, M.; Smink, L.J.; Ainscough, R.; Almeida, J. P.; and 205 others: The DNA sequence of human chromosome 22. *Nature* 402: 489–495, 1999.

[37004] 10556. Kimura, T.; Ivell, R.; Rust, W.; Mizumoto, Y.; Ogita, K.; Kusui, C.; Matsumura, Y.; Azuma, C.; Murata, Y.: Molecular cloning of a human MafF homologue, which specifically binds to the oxytocin receptor gene in term myometrium. *Biochem. Biophys. Res. Commun.* 264: 86–92, 1999.

[37005] 10557. Hiki, K.; D'Andrea, R. J.; Furze, J.; Crawford, J.; Woollatt, E.; Sutherland, G. R.; Vadas, M. A.; Gamble, J. R.: Cloning, characterization, and chromosomal location of a novel human K(+)-Cl(-) cotransporter. *J. Biol. Chem.* 274: 10661–10667, 1999.

[37006] 10558. Mount, D. B.: Personal Communication. Nashville, Tenn. 3/28/2000.

[37007] 10559. Mount, D. B.; Mercado, A.; Song, L.; Xu, J.; George, A. L., Jr.; Delpire, E.; Gamba, G.: Cloning and characterization of KCC3 and KCC4, new members of the cation-chloride cotransporter gene family. *J. Biol. Chem.* 274: 16355–16362, 1999.

[37008] 10560. Herzing, L. B. K.; Kim, S.-J.; Cook, E. H., Jr.; Led-

better, D.H.: The human aminophospholipid-transporting ATPase gene ATP10C maps adjacent to UBE3A and exhibits similar imprinted expression. *Am.J. Hum. Genet.* 68: 1501-1505, 2001.

[37009] 10561.Dode, C.; Le Du, N.; Cuisset, L.; Letourneur, F.; Berthelot, J.-M.; Vaudour, G.; Meyrier, A.; Watts, R. A.; Scott, D. G. I.; Nicholls, A.; Granel, B.; Frances, C.; Garcier, F.; Edery, P.; Boulinguez, S.; Domergues, J.-P.; Delpech, M.; Grateau, G.: New mutations of CIAS1 that are responsible for Muckle-Wells syndrome and familial cold urticaria: a novel mutation underlies both syndromes. *Am. J. Hum. Genet.* 70:1498-1506, 2002.

[37010] 10562.Feldmann, J.; Prieur, A.-M.; Quartier, P.; Berquin, P.; Certain, S.; Cortis, E.; Teillac-Hamel, D.; Fischer, A.; de Saint Basile, G.: Chronic infantile neurological cutaneous and articular syndrome is caused by mutations in CIAS1, a gene highly expressed in polymorphonuclear cells and chondrocytes. *Am. J. Hum. Genet.* 71: 198-203, 2002.

[37011] 10563.Greeve, I.; Hermans-Borgmeyer, I.; Brellinger, C.; Kasper, D.; Gomez-Isla, T.; Behl, C.; Levkau, B.; Nitsch, R. M.: The human DIMINUTO/DWARF1 homolog seladin-1 confers resistance to Alzheimer's disease-associated neurodegeneration and oxidative stress. *J. Neu-*

rosci. 20: 7345–7352,2000.

- [37012] 10564.Sarkar, D.; Imai, T.; Kambe, F.; Shibata, A.; Ohmori, S.; Siddiq,A.; Hayasaka, S.; Funahashi, H.; Seo, H.: The human homolog of Diminuto/Dwarf1gene (hDiminuto): a novel ACTH-responsive gene overexpressed in benign cortisol-producing adrenocortical adenomas. J. Clin. Endocr. Metab. 86:5130–5137, 2001.
- [37013] 10565.Weidenhammer, E. M.; Singh, M.; Ruiz–Noriega, M.; Woolford, J.L., Jr.: The PRP31 gene encodes a novel protein required for pre-mRNA splicing in *Saccharomyces cerevisiae*. Nucleic Acids Res. 24: 1164–1170,1996.
- [37014] 10566.Hattori, M.; Fujiyama, A.; Taylor, T. D.; Watanabe, H.; Yada, T.;Park, H.–S.; Toyoda, A.; Ishii, K.; Totoki, Y.; Choi, D.–K.; Groner,Y.; Soeda, E.; and 52 others: The DNA sequence of human chromosome21. Nature 405: 311–319, 2000. Note: Erratum: Nature: 407: 110 only,2000.
- [37015] 10567.Wilcox, E. R.; Burton, Q. L.; Naz, S.; Riazuddin, S.; Smith, T.N.; Ploplis, B.; Belyatseva, I.; Ben–Yosef, T.; Liburd, N. A.; Morell,R. J.; Kachar, B.; Wu, D. K.; Griffith, A. J.; Riazuddin, S.; Friedman,T. B.: Mutations in the gene encoding tight junction claudin–14 cause recessive deafness DFNB29. Cell 104: 165–172, 2001.

- [37016] 10568.Dunlevy, J. R.; Berryhill, B. L.; Vergnes, J.-P.; SundarRaj, N.;Hassell, J. R.: Cloning, chromosomal localization, and characterization of cDNA from a novel gene, SH3BP4, expressed by human corneal fibroblasts. *Genomics* 62:519–524, 1999.
- [37017] 10569.Wong, W. T.; Schumacher, C.; Salcini, A. E.; Romano, A.; Castagnino,P.; Pelicci, P. G.; DiFiore, P. P.: A protein-binding domain, EH,identified in the receptor tyrosine kinase substrate Eps15 and conserved in evolution. *Proc. Nat. Acad. Sci.* 92: 9530–9534, 1995.
- [37018] 10570.Yamadori, T.; Baba, Y.; Matsushita, M.; Hashimoto, S.; Kurosaki,M.; Kurosaki, T.; Kishimoto, T.; Tsukada, S.: Bruton's tyrosine kinase activity is negatively regulated by Sab, the Btk-SH3 domain-binding protein. *Proc. Nat. Acad. Sci.* 96: 6341–6346, 1999.
- [37019] 10571.Engqvist-Goldstein, A. E. Y.; Kessels, M. M.; Chopra, V. S.; Hayden,M. R.; Drubin, D. G.: An actin-binding protein of the Sla2/Huntingtin interacting protein 1 family is a novel component of clathrin-coated pits and vesicles. *J. Cell Biol.* 147: 1503–1518, 1999.
- [37020] 10572.Baba, Y.; Matsushita, M.; Matsuda, Y.; Inazawa, J.; Yamadori, T.;Hashimoto, S.; Kishimoto, T.; Tsukada, S.: Assignment of SH3BP5/Sh3bp5 encoding Sab, an SH3 do-

main-binding protein which preferentially associates with Bruton's tyrosine kinase, to human chromosome 1q43 and mouse chromosome 14B by in situ hybridization. Cytogenet. Cell Genet. 87:221–222, 1999.

[37021] 10573. Matsushita, M.; Yamadori, T.; Kato, S.; Takemoto, Y.; Inazawa, J.; Baba, Y.; Hashimoto, S.; Sekine, S.; Arai, S.; Kunikata, T.; Kurimoto, M.; Kishimoto, T.; Tsukada, S.: Identification and characterization of a novel SH3-domain binding protein, Sab, which preferentially associates with Bruton's tyrosine kinase (Btk). Biochem. Biophys. Res. Commun. 245:337–343, 1998.

[37022] 10574. Itoh, T.; Koshiba, S.; Kigawa, T.; Kikuchi, A.; Yokoyama, S.; Takenawa, T.: Role of the ENTH domain in phosphatidylinositol-4,5-bisphosphate binding and endocytosis. Science 291: 1047–1051, 2001.

[37023] 10575. Seki, N.; Muramatsu, M.; Sugano, S.; Suzuki, Y.; Nakagawara, A.; Ohhira, M.; Hayashi, A.; Hori, T.; Saito, T.: Cloning, expression analysis, and chromosomal localization of HIP1R, an isolog of huntingtin interacting protein (HIP1). J. Hum. Genet. 43: 268–271, 1998.

[37024] 10576. Marton, M. J.; Vazquez de Aldana, C. R.; Qiu, H.; Chakraburttty, K.; Hinnebusch, A. G.: Evidence that GCN1 and GCN20, translational regulators of GCN4, function on

elongating ribosomes in activation of eIF2- α kinase GCN2. *Molec. Cell. Biol.* 17: 4474–4489, 1997.

- [37025] 10577. Nash, S. R.; Giros, B.; Kingsmore, S. F.; Kim, K. M.; El-Mestikawy, S.; Dong, Q.; Fumagalli, F.; Seldin, M. F.; Caron, M. G.: Cloning, gene structure and genomic localization of an orphan transporter from mouse kidney with six alternatively-spliced isoforms. *Receptors Channels* 6:113–128, 1998.
- [37026] 10578. Scott, A. F.: Personal Communication. Baltimore, Md. 2/5/2001.
- [37027] 10579. Meguro, M.; Kashiwagi, A.; Mitsuya, K.; Nakao, M.; Kondo, I.; Saitoh, S.; Oshimura, M.: A novel maternally expressed gene, ATP10C, encodes a putative aminophospholipid translocase associated with Angelman syndrome. *Nature Genet.* 28: 19–20, 2001.
- [37028] 10580. Nakakura, E. K.; Watkins, D. N.; Schuebel, K. E.; Sriuranpong, V.; Borges, M. W.; Nelkin, B. D.; Ball, D. W.: Mammalian Scratch: a neural-specific Snail family transcriptional repressor. *Proc. Nat. Acad. Sci.* 98: 4010–4015, 2001.
- [37029] 10581. Scott, A. F.: Personal Communication. Baltimore, Md. 6/21/2001.
- [37030] 10582. Hoatlin, M. E.; Zhi, Y.; Ball, H.; Silvery, K.; Melnick,

A.; Stone, S.; Arai, S.; Hawe, N.; Owen, G.; Zelent, A.; Licht, J. D.: A novel BTB/POZ transcriptional repressor protein interacts with the Fanconianemia group C protein and PLZF. *Blood* 94: 3737–3747, 1999.

[37031] 10583. Lin, W.; Lai, C.-H.; Tang, C.-J. C.; Huang, C.-J.; Tang, T. K.: Identification and gene structure of a novel human PLZF-related transcription factor gene, TZFP. *Biochem. Biophys. Res. Commun.* 264:789–795, 1999.

[37032] 10584. Yokoyama-Kobayashi, M.; Sugano, S.; Kato, T.; Kato, S.: A signal sequence detection system using secreted protease activity as an indicator. *Gene* 163:193–196, 1995.

[37033] 10585. Yokoyama-Kobayashi, M.; Yamaguchi, T.; Sekine, S.; Kato, S.: Selection of cDNAs encoding putative type II membrane proteins on the cell surface from a human full-length cDNA bank. *Gene* 228: 161–167, 1999.

[37034] 10586. Olivier, E.; Soury, E.; Risler, J. L.; Smith, F.; Schneider, K.; Lochner, K.; Jouzeau, J. Y.; Fey, G. H.; Salier, J. P.: A novel set of hepatic mRNAs preferentially expressed during an acute inflammation in rat represents mostly intracellular proteins. *Genomics* 57: 352–364, 1999.

[37035] 10587. Olivier, E.; Soury, E.; Ruminy, P.; Husson, A.; Parmentier, F.; Daveau, M.; Salier, J.-P.: Fetuin-B, a second

member of the fetuin family in mammals. *Biochem. J.* 350: 589–597, 2000.

- [37036] 10588. Hampe, J.; Grebe, J.; Nikolaus, S.; Solberg, C.; Croucher, P. J. P.; Mascheretti, S.; Jahnsen, J.; Moum, B.; Klump, B.; Krawczak, M.; Mirza, M. M.; Foelsch, U. R.; Vatn, M.; Schreiber, S.: Association of NOD2 (CARD 15) genotype with clinical course of Crohn's disease: a cohort study. *Lancet* 359: 1661–1665, 2002.
- [37037] 10589. Hugot, J.-P.; Chamaillard, M.; Zouali, H.; Lesage, S.; Cezard, J.-P.; Belaiche, J.; Almer, S.; Tysk, C.; O'Morain, C. A.; Gassull, M.; Binder, V.; Finkel, Y.; and 8 others: Association of NOD2 leucine-rich repeat variants with susceptibility to Crohn's disease. *Nature* 411: 599–603, 2001.
- [37038] 10590. Murillo, L.; Crusius, J. B. A.; van Bodegraven, A. A.; Alizadeh, B. Z.; Pena, A. S.: CARD15 gene and the classification of Crohn's disease. *Immunogenetics* 54: 59–61, 2002.
- [37039] 10591. Ogura, Y.; Bonen, D. K.; Inohara, N.; Nicolae, D. L.; Chen, F. F.; Ramos, R.; Britton, H.; Moran, T.; Karaliuskas, R.; Duerr, R. H.; Achkar, J.-P.; Brant, S. R.; Bayless, T. M.; Kirschner, B. S.; Hanauer, S. B.; Nunez, G.; Cho, J. H.: A frameshift mutation in Nod2 associated with susceptibility to Crohn's disease. *Nature* 411: 603–606, 2001.

- [37040] 10592.Ogura, Y.; Inohara, N.; Benito, A.; Chen, F. F.; Yamaoka, S.; Nunez, G.: Nod2, a Nod1/Apaf-1 family member that is restricted to monocytes and activates NF-kappa-B. *J. Biol. Chem.* 276: 4812-4818, 2001.
- [37041] 10593.Lesage, S.; Zouali, H.; Cezard, J.-P.; EPWG-IBD Group; Colombel, J.-F.; EPIMAD Group; Belaiche, J.; GETAID Group; Almer, S.; Tysk, C.; O'Morain, C.; Gassull, M.; Binder, V.; Finkel, Y.; Modigliani, R.; Gower-Rousseau, C.; Macry, J.; Merlin, F.; Chamaillard, M.; Jannot, A.-S.; Thomas, G.; Hugot, J.-P.: CARD15/NOD2 mutational analysis and genotype-phenotype correlation in 612 patients with inflammatory bowel disease. *Am. J. Hum. Genet.* 70: 845-857, 2002.
- [37042] 10594.van Heel, D. A.; McGovern, D. P. B.; Cardon, L. R.; Dechairo, B.M.; Lench, N. J.; Carey, A. H.; Jewell, D. P.: Fine mapping of the IBD1 locus did not identify Crohn disease-associated NOD2 variants: implications for complex disease genetics. *Am. J. Med. Genet.* 111:253-259, 2002.
- [37043] 10595.Vermeire, S.; Wild, G.; Kocher, K.; Cousineau, J.; Dufresne, L.; Bitton, A.; Langelier, D.; Pare, P.; Lapointe, G.; Cohen, A.; Daly, M. J.; Rioux, J. D.: CARD15 genetic variation in a Quebec population: prevalence, genotype-phenotype relationship, and haplotype structure. *Am. J. Hum.*

Genet. 71: 74–83, 2002.

[37044] 10596.Yamazaki, K.; Takazoe, M.; Tanaka, T.; Kazumori, T.; Nakamura,Y.: Absence of mutation in the NOD2/CARD15 gene among 483 Japanese patients with Crohn's disease. J. Hum. Genet. 47: 469–472, 2002.

[37045] 10597.Fliss, M. S.; Hinkle, P. M.; Bancroft, C.: Expression cloning and characterization of PREB (prolactin regulatory element binding), a novel WD motif DNA-binding protein with a capacity to regulate prolactin promoter activity. Molec. Endocr. 13: 644–657, 1999.

[37046] 10598.Taylor Clelland, C. L.; Levy, B.; McKie, J. M.; Duncan, A. M. V.; Hirschhorn, K.; Bancroft, C.: Cloning and characterization of human PREB; a gene that maps to a genomic region associated with trisomy 2p syndrome. Mammalian Genome 11: 675–681, 2000.

[37047] 10599.Li, B.; Oestreich, S.; de Lange, T.: Identification of human Rap1: implications for telomere evolution. Cell 101: 471–483, 2000.

[37048] 10600.Lieb, J. D.; Liu, X.; Botstein, D.; Brown, P. O.: Promoter-specific binding of Rap1 revealed by genome-wide maps of protein–DNA association. Nature Genet. 28: 327–334, 2001.

[37049] 10601.Honore, B.; Leffers, H.; Madsen, P.; Rasmussen, H.

H.; Vandekerckhove, J.; Celis, J. E.: Molecular cloning and expression of a transformation-sensitive human protein containing the TPR motif and sharing identity to the stress-inducible yeast protein STI1. *J. Biol. Chem.* 267: 8485–8491, 1992.

- [37050] 10602. Scheufler, C.; Brinker, A.; Bourenkov, G.; Pegoraro, S.; Moroder, L.; Bartunik, H.; Hartl, F. U.; Moarefi, I.: Structure of TPR domain-peptide complexes: critical elements in the assembly of the Hsp70–Hsp90 multichaperon machine. *Cell* 101: 199–210, 2000.
- [37051] 10603. Chong, S. S.; Tanigami, A.; Roschke, A. V.; Ledbetter, D. H.: 14–3–3-epsilon has no homology to LIS1 and lies telomeric to it on chromosome 17p13.3 outside the Miller–Dieker syndrome chromosome region. *Genome Res.* 6: 735–741, 1996.
- [37052] 10604. Conklin, D. S.; Galaktionov, K.; Beach, D.: 14–3–3 proteins associate with cdc25 phosphatases. *Proc. Nat. Acad. Sci.* 92: 7892–7896, 1995.
- [37053] 10605. Jin, D.-Y.; Lyu, M. S.; Kozak, C. A.; Jeang, K.-T.: Function of 14–3–3 proteins. *Nature* 382: 308 only, 1996.
- [37054] 10606. Luk, S. C. W.; Garcia-Barcelo, M.; Tsui, S. K. W.; Fung, K. P.; Lee, C. Y.; Waye, M. M. Y.: Assignment of the human 14–3–3 epsilon isoform (YWHA E) to human chro-

mosome 17p13 by in situ hybridization. *Cytogenet. Cell Genet.* 78: 105–106, 1997.

[37055] 10607. Slentz–Kesler, K.; Moore, J. T.; Lombard, M.; Zhang, J.; Hollingsworth, R.; Weiner, M. P.: Identification of the human Mnk2 gene (MKNK2) through protein interaction with estrogen receptor beta. *Genomics* 69:63–71, 2000.

[37056] 10608. Li, Y.; He, X.; Schembri–King, J.; Jakes, S.; Hayashi, J.: Cloning and characterization of human Lnk, an adaptor protein with pleckstrin homology and Src homology 2 domains that can inhibit T cell activation. *J. Immunol.* 164: 5199–5206, 2000.

[37057] 10609. Takaki, S.; Sauer, K.; Iritani, B. M.; Chien, S.; Ebihara, Y.; Tsuji, K.; Takatsu, K.; Perlmutter, R. M.: Control of B cell production by the adaptor protein Lnk: definition of a conserved family of signal–modulating proteins. *Immunity* 13: 599–609, 2000.

[37058] 10610. Fiscella, M.; Zhang, H.; Fan, S.; Sakaguchi, K.; Shen, S.; Mercer, W. E.; Vande Woude, G. F.; O'Connor, P. M.; Appella, E.: Wip1, a novel human protein phosphatase that is induced in response to ionizing radiation in a p53–dependent manner. *Proc. Nat. Acad. Sci.* 94: 6048–6053, 1997.

[37059] 10611. Li, J.; Yang, Y.; Peng, Y.; Austin, R. J.; van Eynd–

hoven, W. G.; Nguyen, K. C. Q.; Gabriele, T.; McCurrach, M. E.; Marks, J. R.; Hoey, T.; Lowe, S. W.; Powers, S.: Oncogenic properties of PPM1D located within a breast cancer amplification epicenter at 17q23. *Nature Genet.* 31:133–134, 2002.

[37060] 10612. Nagase, T.; Seki, N.; Ishikawa, K.; Ohira, M.; Kawarabayashi, Y.; Fiscella, M.; Zhang, H.; Fan, S.; Sakaguchi, K.; Shen, S.; Mercer, W. E.; Vande Woude, G. F.; O'Connor, P. M.; Appella, E.: Wip1, a novel human protein phosphatase that is induced in response to ionizing radiation in a p53-dependent manner. *Proc. Nat. Acad. Sci.* 94: 6048–6053, 1997.

[37061] 10613. Stover, C. M.; Schwaebler, W. J.; Lynch, N. J.; Thiel, S.; Speicher, M. R.: Assignment of the gene encoding mannan-binding lectin-associated serine protease 2 (MASP2) to human chromosome 1p36.3–p36.2 by in situ hybridization and somatic cell hybrid analysis. *Cytogenet. Cell Genet.* 84:148–149, 1999.

[37062] 10614. Stover, C. M.; Thiel, S.; Thelen, M.; Lynch, N. J.; Vorup-Jensen, T.; Jensenius, J. C.; Schwaebler, W. J.: Two constituents of the initiation complex of the mannan-binding lectin activation pathway of complement are encoded by a single structural gene. *J. Immun.* 162:

3481–3490,1999.

- [37063] 10615.Takahashi, M.; Endo, Y.; Fujita, T.; Matsushita, M.: A truncated form of mannose-binding lectin-associated serine protease (MASP)-2 expressed by alternative polyadenylation is a component of the lectin complement pathway. *Int. Immun.* 11: 859–863, 1999.
- [37064] 10616.Thiel, S.; Vorup-Jensen, T.; Stover, C. M.; Schwaeble, W.; Laursen, S. B.; Poulsen, K.; Willis, A. C.; Eggleton, P.; Hansen, S.; Holmskov, U.; Reid, K. B. M.; Jensenius, J. C.: A second serine protease associated with mannan-binding lectin that activates complement. *Nature* 386:506–510, 1997.
- [37065] 10617.Ferguson, K. M.; Kavran, J. M.; Sankaran, V. G.; Fournier, E.; Isakoff, S. J.; Skolnik, E. Y.; Lemmon, M. A.: Structural basis for discrimination of 3-phosphoinositides by pleckstrin homology domains. *Molec.Cell* 6: 373–384, 2000.
- [37066] 10618.Franco, M.; Boretto, J.; Robineau, S.; Monier, S.; Goud, B.; Chardin, P.; Chavrier, P.: ARNO3, a Sec7-domain guanine nucleotide exchange factor for ADP ribosylation factor 1, is involved in the control of Golgi structure and function. *Proc. Nat. Acad. Sci.* 95: 9926–9931, 1998.
- [37067] 10619.Klarlund, J. K.; Guilherme, A.; Holik, J. J.; Virbasius,

J. V.;Chawla, A.; Czech, M. P.: Signaling by phosphoinositide-3,4,5-triphosphate through proteins containing pleckstrin and Sec7 homology domains. *Science* 275:1927-1930, 1997.

[37068] 10620.Drewes, T.; Senkel, S.; Holewa, B.; Ryffel, G. U.: Human hepatocytenuclear factor 4 isoforms are encoded by distinct and differentially expressed genes. *Molec. Cell. Biol.* 16: 925-931, 1996.

[37069] 10621.Taraviras, S.; Mantamadiotis, T.; Dong-Si, T.; Mincheva, A.; Lichter,P.; Drewes, T.; Ryffel, G. U.; Monaghan, A. P.; Schutz, G.: Primary structure, chromosomal mapping, expression and transcriptional activity of murine hepatocyte nuclear factor 4-gamma. *Biochim. Biophys. Acta* 1490:21-32, 2000.

[37070] 10622.Gauss, R.; Seifert, R.; Kaupp, U. B.: Molecular identification of a hyperpolarization-activated channel in sea urchin sperm. *Nature* 393:583-587, 1998.

[37071] 10623.Seifert, R.; Scholten, A.; Gauss, R.; Mincheva, A.; Lichter, P.;Kaupp, U. B.: Molecular characterization of a slowly gating human hyperpolarization-activated channel predominantly expressed in thalamus, heart, and testis. *Proc. Nat. Acad. Sci.* 96: 9391-9396, 1999.

[37072] 10624.Aihara, Y.; Mashima, H.; Onda, H.; Hisano, S.; Ka-

suya, H.; Hori,T.; Yamada, S.; Tomura, H.; Yamada, Y.; Inoue, I.; Kojima, I.; Takeda,J.: Molecular cloning of a novel brain-type Na(+)-dependent inorganicphosphate cotransporter. J. Neurochem. 74: 2622-2625, 2000.

[37073] 10625.Bellocchio, E. E.; Reimer, R. J.; Fremeau, R. T., Jr.; Edwards,R. H.: Uptake of glutamate into synaptic vesicles by an inorganicphosphate transporter. Science 289: 957-960, 2000.

[37074] 10626.Ni, B.; Du, Y.; Wu, X.; DeHoff, B. S.; Rosteck, P. R., Jr.; Paul,S. M.: Molecular cloning, expression, and chromosomal localizationof a human brain-specific Na(+)-dependent inorganic phosphate cotransporter. J.Neurochem. 66: 2227-2238, 1996.

[37075] 10627.Takamori, S.; Rhee, J. S.; Rosenmund, C.; Jahn, R.: Identificationof a vesicular glutamate transporter that defines a glutamatergicphenotype in neurons. Nature 407: 189-194, 2000.

[37076] 10628.Blackwood, D.: P300, a state and a trait marker in schizophrenia. Lancet 355:771-772, 2000.

[37077] 10629.Blackwood, D. H. R.; Fordyce, A.; Walker, M. T.; St. Clair, D.M.; Porteous, D. J.; Muir, W. J.: Schizophrenia and affective disorders--cosegregationwith a translocation at chromosome 1q42 that directly disrupts brain-

expressed genes: clinical and P300 findings in a family.

Am. J. Hum. Genet. 69:428–433, 2001.

- [37078] 10630. Ekelund, J.; Hovatta, I.; Parker, A.; Paunio, T.; Varilo, T.; Martin, R.; Suhonen, J.; Ellonen, P.; Chan, G.; Sinheimer, J. S.; Sobel, E.; Juvonen, H.; Arajärvi, R.; Partonen, T.; Suvisaari, J.; Lonnqvist, J.; Meyer, J.; Peltonen, L.: Chromosome 1 loci in Finnish schizophrenia families. Hum. Molec. Genet. 10: 1611–1617, 2001.
- [37079] 10631. Millar, J. K.; Wilson–Annan, J. C.; Anderson, S.; Christie, S.; Taylor, M. S.; Semple, C. A. M.; Devon, R. S.; St. Clair, D. M.; Muir, W. J.; Blackwood, D. H. R.; Porteous, D. J.: Disruption of two novel genes by a translocation cosegregating with schizophrenia. Hum. Molec. Genet. 9: 1415–1423, 2000.
- [37080] 10632. St. Clair, D.; Blackwood, D.; Muir, W.; Carothers, A.; Walker, M.; Spowart, G.; Gosden, C.; Evans, H. J.: Association within a family of a balanced autosomal translocation with major mental illness. Lancet 336:13–16, 1990.
- [37081] 10633. Bulfone, A.; Menguzzato, E.; Broccoli, V.; Marchiello, A.; Gattuso, C.; Mariani, M.; Consalez, G. G.; Martinez, S.; Ballabio, A.; Banfi, S.: Barhl1, a gene belonging to a new subfamily of mammalian homeobox genes, is expressed in migrating neurons of the CNS. Hum.

Molec.Genet. 9: 1443–1452, 2000.

- [37082] 10634. Alessi, D. R.; Deak, M.; Casamayor, A.; Caudwell, F. B.; Morrice, N.; Norman, D. G.; Gaffney, P.; Reese, C. B.; MacDougall, C. N.; Harbison, D.; Ashworth, A.; Bownes, M.: 3-phosphoinositide-dependent protein kinase-1 (PDK1): structural and functional homology with the *Drosophila* D-STPK61 kinase. *Curr. Biol.* 7: 776–789, 1997.
- [37083] 10635. Bertin, J.; Nir, W.-J.; Fischer, C. M.; Tayber, O. V.; Errada, P. R.; Grant, J. R.; Keilty, J. J.; Gosselin, M. L.; Robinson, K. E.; Wong, G. H. W.; Glucksmann, M. A.; DiStefano, P. S.: Human CARD4 protein is a novel CED-4/Apaf-1 cell death family member that activates NF- κ B. *J. Biol. Chem.* 274: 12955–12958, 1999.
- [37084] 10636. Cohen, D. E.; Green, R. M.; Wu, M. K.; Beier, D. R.: Cloning, tissue-specific expression, gene structure and chromosomal localization of human phosphatidylcholine transfer protein. *Biochim. Biophys. Acta* 1447: 265–270, 1999.
- [37085] 10637. van Helvoort, A.; de Brouwer, A.; Ottenhoff, R.; Brouwers, J. F. H. M.; Wijnholds, J.; Beijnen, J. H.; Rijneveld, A.; van der Valk, M. A.; Majoor, D.; Voorhout, W.; Wirtz, K. W. A.; Elferink, R. P. J. O.; Borst, P.: Mice without phosphatidylcholine transfer protein have no defects in the se-

cretion of phosphatidylcholine into bile or into lung
airspaces. *Proc. Nat. Acad. Sci.* 96: 11501–11506, 1999.

[37086] 10638. de Rooij, J.; Zwartkruis, F. J. T.; Verheijen, M. H. G.;
Cool, R. H.; Nijman, S. M. B.; Wittinghofer, A.; Bos, J. L.:
Epac is a Rap1 guanine–nucleotide–exchange factor di-
rectly activated by cyclic AMP. *Nature* 396: 474–477, 1998.

[37087] 10639. Kawasaki, H.; Springett, G. M.; Mochizuki, N.; Toki,
S.; Nakaya, M.; Matsuda, M.; Housman, D. E.; Graybiel, A.
M.: A family of cAMP–binding proteins that directly activate
Rap1. *Science* 282: 2275–2279, 1998.

[37088] 10640. Hahn, Y.; Lee, J.; Seong, C.; Yoon, J.; Chung, J. H.:
Structural analysis of phylogenetically conserved J domain
protein gene. *Biochim. Biophys. Acta* 1447: 325–333,
1999.

[37089] 10641. Hollopeter, G.; Jantzen, H.–M.; Vincent, D.; Li, G.;
England, L.; Ramakrishnan, V.; Yang, R.–B.; Nurden, P.;
Nurden, A.; Julius, D.; Conley, P. B.: Identification of the
platelet ADP receptor targeted by antithrombotic drugs.
Nature 409: 202–206, 2001.

[37090] 10642. Nurden, P.; Savi, P.; Heilmann, E.; Bihour, C.; Her-
bert, J.–M.; Maffrand, J.–P.; Nurden, A.: An inherited bleed-
ing disorder linked to a defective interaction between ADP
and its receptor on platelets: its influence on glycoprotein

IIb–IIIa complex function. *J. Clin. Invest.* 95: 1612–1622, 1995.

[37091] 10643. Hill, K. E.; Dasouki, M.; Phillips, J. A., III; Burk, R. F.: Human selenoprotein P gene maps to 5q31. *Genomics* 36: 550–551, 1996.

[37092] 10644. Hill, K. E.; Lloyd, R. S.; Burk, R. F.: Conserved nucleotide sequences in the open reading frame and 3-prime untranslated region of selenoprotein P mRNA. *Proc. Nat. Acad. Sci.* 90: 537–541, 1993.

[37093] 10645. Keshan Disease Research Group of the Chinese Academy of Medical Sciences: Observations on effect of sodium selenite in prevention of Keshan disease. *Chinese Med. J.* 92: 471–476, 1979.

[37094] 10646. Eberhart, C. G.; Maines, J. Z.; Wasserman, S. A.: Meiotic cell cycle requirement for a fly homologue of human Deleted in Azoospermia. *Nature* 381: 783–785, 1996.

[37095] 10647. Rugglu, M.; Speed, R.; Taggart, M.; McKay, S. J.; Kilanowski, F.; Saunders, P.; Derin, J.; Cooke, H. J.: The mouse Dazla gene encodes a cytoplasmic protein essential for gametogenesis. *Nature* 389: 73–77, 1997.

[37096] 10648. Shan, Z.; Hirschmann, P.; Seebacher, T.; Edelmann, A.; Jauch, A.; Morell, J.; Urbitsch, P.; Vogt, P. H.: A SPGY copy homologous to the mouse gene Dazla and the

Drosophila gene boule is autosomal and expressed only in the human male gonad. Hum. Molec. Genet. 5: 2005–2011, 1996.

- [37097] 10649. Slee, R.; Grimes, B.; Speed, R. M.; Taggart, M.; Maguire, S. M.; Ross, A.; McGill, N. I.; Saunders, P. T. K.; Cooke, H. J.: A human DAZ transgene confers partial rescue of the mouse Daz1 null phenotype. Proc. Nat. Acad. Sci. 96: 8040–8045, 1999.
- [37098] 10650. Yen, P. H.; Chai, N. N.; Salido, E. C.: The human autosomal gene DAZLA: testis specificity and a candidate for male infertility. Hum. Molec. Genet. 5: 2013–2017, 1996.
- [37099] 10651. Akiyama, T. E.; Sakai, S.; Lambert, G.; Nicol, C. J.; Matsusue, K.; Pimprale, S.; Lee, Y.-H.; Ricote, M.; Glass, C. K.; Brewer, H. B., Jr.; Gonzalez, F. J.: Conditional disruption of the peroxisome proliferator-activated receptor gamma gene in mice results in lowered expression of ABCA1, ABCG1, and apoE in macrophages and reduced cholesterol efflux. Molec. Cell. Biol. 22: 2607–2619, 2002.
- [37100] 10652. Ardlie, K. G.; Lunetta, K. L.; Seielstad, M.: Testing for population subdivision and association in four case-control studies. Am. J. Hum. Genet. 71: 304–311, 2002.
- [37101] 10653. Barak, Y.; Nelson, M. C.; Ong, E. S.; Jones, Y. Z.; Ruiz-Lozano, P.; Chien, K. R.; Koder, A.; Evans, R. M.:

PPAR-gamma is required for placental, cardiac, and adipose tissue development. *Molec. Cell* 4:585-595, 1999.

[37102] 10654. Barroso, I.; Gurnell, M.; Crowley, V. E. F.; Agostini, M.; Schwabe, J. W.; Soos, M. A.; Masien, G. L.; Williams, T. D. M.; Lewis, H.; Schafer, A. J.; Chatterjee, V. K. K.; O'Rahilly, S.: Dominant negative mutations in human PPAR-gamma associated with severe insulin resistance, diabetes mellitus and hypertension. *Nature* 402: 880-883, 1999.

[37103] 10655. Beamer, B. A.; Yen, C.-J.; Andersen, R. E.; Muller, D.; Elahi, D.; Cheskin, L. J.; Andres, R.; Roth, J.; Shuldiner, A. R.: Association of the pro12ala variant in the peroxisome proliferator-activated receptor-gamma-2 gene with obesity in two Caucasian populations. *Diabetes* 47: 1806-1808, 1998.

[37104] 10656. Beamer, B. A.; Negri, C.; Yen, C.-J.; Gavrilova, O.; Rumberger, J. M.; Durcan, M. J.; Yarnall, D. P.; Hawkins, A. L.; Griffin, C. A.; Burns, D. K.; Roth, J.; Reitman, M.; Shuldiner, A. R.: Chromosomal localization and partial genomic structure of the human peroxisome proliferator activated receptor-gamma (hPPAR-gamma) gene. *Biochem. Biophys. Res. Commun.* 233: 756-759, 1997.

[37105] 10657. Chawla, A.; Boisvert, W. A.; Lee, C.-H.; Laffitte, B.

A.; Barak,Y.; Joseph, S. B.; Liao, D.; Nagy, L.; Edwards, P. A.; Curtiss, L.K.; Evans, R. M.; Tontonoz, P.: A PPAR–gamma–LXR–ABCA1 pathway in macrophages is involved in cholesterol efflux and atherogenesis. *Molec.Cell* 7: 161–171, 2001.

[37106] 10658. Deeb, S. S.; Fajas, L.; Nemoto, M.; Pihlajamäki, J.; Mykkanen, L.; Kuusisto, J.; Laakso, M.; Fujimoto, W.; Auwerx, J.: A pro12alasubstitution in PPAR–gamma–2 associated with decreased receptor activity, lower body mass index and improved insulin sensitivity. *Nature Genet.* 20:284–287, 1998.

[37107] 10659. Elbrecht, A.; Chen, Y.; Cullinan, C. A.; Hayes, N.; Leibowitz, M. D.; Moller, D. E.; Berger, J.: Molecular cloning, expression and characterization of human peroxisome proliferator activated receptors gamma–1 and gamma–2. *Biochem. Biophys. Res. Commun.* 224: 431–437, 1996.

[37108] 10660. Fajas, L.; Auboeuf, D.; Raspe, E.; Schoonjans, K.; Lefebvre, A.M.; Saladin, R.; Najib, J.; Laville, M.; Fruchart, J.–C.; Deeb, S.; Vidal–Puig, A.; Flier, J.; Briggs, M. R.; Staels, B.; Vidal, H.; Auwerx, J.: The organization, promoter analysis, and expression of the human PPAR–gamma gene. *J. Biol. Chem.* 272: 18779–18789, 1997.

- [37109] 10661. Gampe, R. T., Jr.; Montana, V. G.; Lambert, M. H.; Miller, A. B.; Bledsoe, R. K.; Milburn, M. V.; Kliewer, S. A.; Willson, T. M.; Xu, H. E.: Asymmetry in the PPAR- γ /RXR- α crystal structure reveals the molecular basis of heterodimerization among nuclear receptors. *Molec. Cell* 5: 545–555, 2000.
- [37110] 10662. Chakraborti, A.; Lippman, D. L.; Loh, H. H.; Kozak, C. A.; Lee, N. M.: Genetic mapping of opioid binding protein gene(s) to mouse chromosome 9. *Mammalian Genome* 4: 179–182, 1993.
- [37111] 10663. Cho, T. M.; Hasegawa, J.-I.; Ge, B.-I.; Loh, H. H.: Purification to apparent homogeneity of a mu-type opioid receptor from rat brain. *Proc. Nat. Acad. Sci.* 83: 4138–4142, 1986.
- [37112] 10664. Shark, K. B.; Lee, N. M.: Cloning, sequencing and localization to chromosome 11 of a cDNA encoding a human opioid-binding cell adhesion molecule (OBCAM). *Gene* 155: 213–217, 1995.
- [37113] 10665. Burmeister, M.; Meyer, G. E.: The trefoil gene maps to mouse chromosome 17. *Mammalian Genome* 8: 223–224, 1997.
- [37114] 10666. Chinery, R.; Williamson, J.; Poulson, R.: The gene encoding human intestinal trefoil factor (TFF3) is located

on chromosome 21q22.3 clustered with other members of the trefoil peptide family. *Genomics* 32:281–284, 1996.

[37115] 10667. Mashimo, H.; Wu, D.-C.; Podolsky, D. K.; Fishman, M. C.: Impaired defense of intestinal mucosa in mice lacking intestinal trefoil factor. *Science* 274:262–265, 1996.

[37116] 10668. Podolsky, D. K.; Lynch-Devaney, K.; Stow, J. L.; Oates, P.; Murgue, B.; DeBeaumont, M.; Sands, B. E.; Mahida, Y. R.: Identification of human intestinal trefoil factor: goblet cell-specific expression of a peptide targeted for apical secretion. *J. Biol. Chem.* 268: 6694–6702, 1993.

[37117] 10669. Probst, J. C.; Zetzsche, T.; Weber, M.; Theilemann, P.; Skutella, T.; Landgraf, R.; Jirikowski, G. F.: Human intestinal trefoil factor is expressed in human hypothalamus and pituitary: evidence for a novel neuropeptide. *FASEB J.* 10: 1518–1523, 1996.

[37118] 10670. Schmitt, H.; Wundrack, I.; Beck, S.; Gott, P.; Welter, C.; Shizuya, H.; Simon, M. I.; Blin, N.: A third P-domain peptide gene (TFF3), human intestinal trefoil factor, maps to 21q22.3. *Cytogenet. Cell Genet.* 72: 299–302, 1996.

[37119] 10671. Taupin, D.; Wu, D.-C.; Jeon, W.-K.; Devaney, K.; Wang, T. C.; Podolsky, D. K.: The trefoil gene family are coordinately expressed immediate-early genes: EGF receptor- and MAP kinase-dependent interregulation. *J. Clin. In-*

vest. 103: R31–R38, 1999.

- [37120] 10672.Thim, L.: A new family of growth factor–like peptides: 'trefoil'disulphide loop structures as a common feature in breast cancer associatedpeptide (pS2), pancreatic spasmolytic polypeptide (PSP), and frogskin peptides (spasmolysins). FEBS Lett. 250: 85–90, 1989.
- [37121] 10673.Sugimoto, Y.; Yatsunami, K.; Tsujimoto, M.; Khorana, H. G.; Ichikawa,A.: The amino acid sequence of a glutamic acid–rich protein frombovine retina as deduced from the cDNA sequence. Proc. Nat. Acad.Sci. 88: 3116–3119, 1991.
- [37122] 10674.Marigo, V.; Roberts, D. J.; Lee, S. M. K.; Tsukurov, O.; Levi,T.; Gastier, J. M.; Epstein, D. J.; Gilbert, D. J.; Copeland, N. G.;Seidman, C. E.; Jenkins, N. A.; Seidman, J. G.; McMahon, A. P.; Tabin,C.: Cloning, expression, and chromosomal location of SHH and IHH:two human homologues of the Drosophila segment polarity gene hedgehog. Genomics 28:44–51, 1995.
- [37123] 10675.McIntire, J. J.; Umetsu, S. E.; Akbari, O.; Potter, M.; Kuchroo,V. K.; Barsh, G. S.; Freeman, G. J.; Umetsu, D. T.; DeKruyff, R. H.: Identification of Tapr (an airway hyperre–activity regulatory locus)and the linked Tim gene family. Nature Immun. 2: 1109–1116, 2001.

- [37124] 10676.Hwang, B. J.; Ford, J. M.; Hanawalt, P. C.; Chu, G.: Expression of the p48 xeroderma pigmentosum gene is p53-dependent and is involved in global genomic repair. Proc. Nat. Acad. Sci. 96: 424-428, 1999.
- [37125] 10677.Tahir, S. A.; Yang, G.; Ebara, S.; Timme, T. L.; Satoh, T.; Li, L.; Goltsov, A.; Ittmann, M.; Morrisett, J. D.; Thompson, T. C.: Secreted caveolin-1 stimulates cell survival/clonal growth and contributes to metastasis in androgen-insensitive prostate cancer. Cancer Res. 61:3882-3885, 2001.
- [37126] 10678.England, S. K.; Uebele, V. N.; Kodali, J.; Bennett, P. B.; Tamkun, M. M.: A novel K⁺ channel beta-subunit (hKv-beta-1.3) is produced via alternative mRNA splicing. J. Biol. Chem. 270: 28531-28534, 1995.
- [37127] 10679.Kratz, C. P.; Emerling, B. M.; Bonifas, J.; Wang, W.; Green, E.D.; Le Beau, M. M.; Shannon, K. M.: Genomic structure of the PIK3CG gene on chromosome band 7q22 and evaluation as a candidate myeloid tumor suppressor. Blood 99: 372-374, 2002.
- [37128] 10680.Temple, I. K.; Gardner, R. J.; Robinson, D. O.; Kibirige, M. S.; Ferguson, A. W.; Baum, J. D.; Barber, J. C. K.; James, R. S.; Shield, J. P. H.: Further evidence for an imprinted gene for neonatal diabetes localised to chromo-

some 6q22–q23. *Hum. Molec. Genet.* 5: 1117–1124, 1996.

- [37129] 10681. Ahmad, W.; Li, S.; Chen, H.; Tuck–Muller, C. M.; Pittler, S. J.; Aronson, N. N., Jr.: Lysosomal chitobiase (CTB) and the G–protein gamma–5 subunit (GNG5) genes co-localize to human chromosome 1p22. *Cytogenet. Cell Genet.* 71: 44–46, 1995.
- [37130] 10682. Fisher, K. J.; Aronson, N. N.: Characterization of the cDNA and genomic sequence of a G protein gamma subunit (gamma–5). *Molec. Cell Biol.* 12: 1585–1591, 1992.
- [37131] 10683. Fiaschi, T.; Marzella, R.; Veggi, D.; Marzocchini, R.; Raugei, G.; Rocchi, M.; Ramponi, G.: Assignment of the human erythrocyte acylphosphatase gene (ACYP1) to chromosome band 14q24.3. *Cytogenet. Cell Genet.* 81: 235–236, 1998.
- [37132] 10684. Fiaschi, T.; Raugei, G.; Marzocchini, R.; Chiarugi, P.; Cirri, P.; Ramponi, G.: Cloning and expression of the cDNA coding for the erythrocyte isoenzyme of human acylphosphatase. *FEBS Lett.* 367: 145–148, 1995.
- [37133] 10685. Goldowitz, D.; Smeyne, R. J.: Tune into the weaver channel. *Nature Genet.* 11: 107–109, 1995.
- [37134] 10686. Hess, E. J.: Identification of the weaver mouse mutation: the end of the beginning. *Neuron* 16: 1073–1076, 1996.

- [37135] 10687.Lane, P. W.: New mutation: Weaver, wv. Mouse News Letter 32–33,1964.
- [37136] 10688.Lesage, F.; Duprat, F.; Fink, M.; Guillemare, E.; Coppola, T.;Lazdunski, M.; Hugnot, J.–P.: Cloning provides evidence for a familyof inward rectifier and G–protein coupled K(+) channels in the brain. FEBSLett. 353: 37–42, 1994.
- [37137] 10689.Rakic, P.; Sidman, R. L.: Sequence of developmental abnormalitiesleading to granule cell deficit in cerebellar cortex of weaver mutantmice. J. Comp. Neurol. 152: 103–132, 1973.
- [37138] 10690.Sakura, H.; Bond, C.; Warren–Perry, M.; Horsley, S.; Kearney, L.;Tucker, S.; Adelman, J.; Turner, R.; Ashcroft, F. M.: Characterizationand variation of a human inwardly–rectifying K–channel gene (KCNJ6):a putative ATP–sensitive K–channel subunit. FEBS Lett. 367: 193–197,1995.
- [37139] 10691.Tsaur, M.–L.; Menzel, S.; Lai, F.–P.; Espinosa, R., III; Concannon,P.; Spielman, R. S.; Hanis, C. L.; Cox, N. J.; Le Beau, M. M.; German,M. S.; Jan, L. Y.; Bell, G. I.; Stoffel, M.: Isolation of a cDNAclone encoding a K(ATP) channel–like protein expressed in insulin–secretingcells, localization of the human gene to chromosome band 21q22.1 andlink–

age studies with NIDDM. Diabetes 44: 592–596, 1995.

- [37140] 10692.Yasuda, K.; Sakura, H.; Mori, Y.; Iwamoto, K.; Shimokawa, K.;Kadowaki, H.; Hagura, R.; Akanuma, Y.; Adelman, J. P.; Yazaki, Y.;Ashcroft, F. M.; Kadowaki, T.: No evidence for mutations in a putativesubunit of the beta-cell ATP-sensitive potassium channel (K-ATP channel)in Japanese NIDDM patients. Biochem. Biophys. Res. Commun. 211:1036–1040, 1995.
- [37141] 10693.Efiok, B. J. S.; Chiorini, J. A.; Safer, B.: A key transcriptionfactor for eukaryotic initiation factor-2-alpha is strongly homologousto developmental transcription factors and may link metabolic genesto cellular growth and development. J. Biol. Chem. 269: 18921–18930,1994.
- [37142] 10694.Gopalakrishnan, L.; Scarpulla, R. C.: Structure, expression, andchromosomal assignment of the human gene encoding nuclear respiratoryfactor 1. J. Biol. Chem. 270: 18019–18025, 1995.
- [37143] 10695.Spelbrink, J. N.; Van den Bogert, C.: The pre-mRNA of nuclearrespiratory factor 1, a regulator of mitochondrial biogenesis, isalternatively spliced in human tissues and cell lines. Hum. Molec.Genet. 4: 1591–1596, 1995.
- [37144] 10696.Tiranti, V.; Rossi, E.; Rocchi, M.; DiDonato, S.; Zufardi, O.;Zeviani, M.: The gene (NFE2L1) for human NRF-1,

an activator involved in nuclear-mitochondrial interactions, maps to 7q32. *Genomics* 27:555-557, 1995.

[37145] 10697. Ohshiro, T.; Yagami, T.; Zhang, C.; Matsuzaki, F.: Role of cortical tumour-suppressor proteins in asymmetric division of *Drosophila* neuroblast. *Nature* 408:593-596, 2000.

[37146] 10698. Peng, C.-Y.; Manning, L.; Albertson, R.; Doe, C. Q.: The tumour-suppressor genes *lgl* and *dlg* regulate basal protein targeting in *Drosophila* neuroblasts. *Nature* 408:596-600, 2000.

[37147] 10699. Strand, D.; Unger, S.; Corvi, R.; Hartenstein, K.; Schenkel, H.; Kalms, A.; Merdes, G.; Neumann, B.; Krieg-Schneider, F.; Coy, J.F.; Poustka, A.; Schwab, M.; Mechler B. M.: A human homologue of the *Drosophila* tumour suppressor gene *l(2)gl* maps to 17p11.2-12 and codes for a cytoskeletal protein that associates with nonmuscle myosin II heavy chain. *Oncogene* 11: 291-301, 1995.

[37148] 10700. Timchenko, L. T.; Miller, J. W.; Timchenko, N. A.; DeVore, D. R.; Datar, K. V.; Lin, L.; Roberts, R.; Caskey, C. T.; Swanson, M. S.: Identification of a (CUG)_n triplet repeat RNA-binding protein and its expression in myotonic dystrophy. *Nucleic Acids Res.* 24: 4407-4414, 1996.

[37149] 10701. Timchenko, L. T.; Timchenko, N. A.; Caskey, C. T.;

Roberts, R.: Novel proteins with binding specificity for DNA CTG repeats and RNA CUG repeats: implications for myotonic dystrophy. *Hum. Molec. Genet.* 5: 115–121, 1996.

[37150] 10702. Wary, K. K.; Mariotti, A.; Zurzolo, C.; Giancotti, F. G.: A requirement for caveolin-1 and associated kinase Fyn in integrin signaling and anchorage-dependent cell growth. *Cell* 94: 625–634, 1998.

[37151] 10703. Yang, G.; Truong, L. D.; Timme, T. L.; Ren, C.; Wheeler, T. M.; Park, S. H.; Nasu, Y.; Bangma, C. H.; Kattan, M. W.; Scardino, P. T.; Thompson, T. C.: Elevated expression of caveolin is associated with prostate and breast cancer. *Clin. Cancer Res.* 4: 1873–1880, 1998.

[37152] 10704. Meij, I. C.; Koenderink, J. B.; van Bokhoven, H.; Assink, K. F. H.; Groenestege, W. T.; de Pont, J. J. H. H. M.; Bindels, R. J. M.; Monnens, L. A. H.; van den Heuvel, L. P. W. J.; Knoers, N. V. A. M.: Dominant isolated renal magnesium loss is caused by misrouting of the Na⁺, K⁺-ATPase gamma-subunit. *Nature Genet.* 26: 265–266, 2000. Note: Erratum. *Nature Genet.* 27: 125 only, 2001.

[37153] 10705. Sweadner, K. J.; Rael, E.: The FXYD gene family of small ion transport regulators or channels: cDNA sequence, protein signature sequence, and expression. *Ge-*

nomics 68: 41–56, 2000.

- [37154] 10706.Sweadner, K. J.; Wetzel, R. K.; Arystarkhova, E.: Genomic organization of the human FXYD2 gene encoding the gamma subunit of the Na,K-ATPase. *Biochem.Biophys. Res. Commun.* 279: 196–201, 2000.
- [37155] 10707.Blair, E.; Redwood, C.; Ashrafian, H.; Oliveira, M.; Broxholme,J.; Kerr, B.; Salmon, A.; Ostman-Smith, I.; Watkins, H.: Mutations in the gamma-2 subunit of AMP-activated protein kinase cause familialhypertrophic cardiomyopathy: evidence for the central role of energy-compromise in disease pathogenesis. *Hum. Molec. Genet.* 10: 1215–1220,2001.
- [37156] 10708.Siderovski, D. P.; Heximer, S. P.; Forsdyke, D. R.: A human geneencoding a putative basic helix-loop-helix phosphoprotein whose mRNAincreases rapidly in cycloheximide-treated blood mononuclear cells. *DNACell Biol.* 13: 125–147, 1994.
- [37157] 10709.Hirai, H.; Roussel, M. F.; Kato, J.-Y.; Ashmun, R. A.; Sherr, C.J.: Novel INK4 proteins, p19 and p18, are specific inhibitors ofcyclin D-dependent kinases CDK4 and CDK6. *Molec. Cell. Biol.* 15:2672–2681, 1995.
- [37158] 10710.Okuda, T.; Hirai, H.; Valentine, V. A.; Shurtleff, S. A.; Kidd,V. J.; Lahti, J. M.; Sherr, C. J.; Downing, J. R.:

Molecular cloning, expression pattern, and chromosomal localization of human CDKN2D/INK4d, an inhibitor of cyclin D-dependent kinases. *Genomics* 29: 623–630, 1995.

[37159] 10711. Sago, H.; Kitagawa, M.; Obata, S.; Mori, N.; Taketani, S.; Rochelle, J. M.; Seldin, M. F.; Davidson, M.; St. John, T.; Suzuki, S. T.: Cloning, expression, and chromosomal localization of a novel cadherin-related protein, protocadherin-3. *Genomics* 29: 631–640, 1995.

[37160] 10712. Fiorucci, S.; Mencarelli, A.; Palazzetti, B.; Distrutti, E.; Vergnolle, N.; Hollenberg, M. D.; Wallace, J. L.; Morelli, A.; Cirino, G.: Proteinase-activated receptor 2 is an anti-inflammatory signal for colonic lamina propria lymphocytes in a mouse model of colitis. *Proc. Nat. Acad. Sci.* 98:13936–13941, 2001.

[37161] 10713. Nystedt, S.; Emilsson, K.; Larsson, A.-K.; Strombeck, B.; Sundelin, J.: Molecular cloning and functional expression of the gene encoding the human proteinase activated receptor 2. *Europ. J. Biochem.* 232:84–89, 1995.

[37162] 10714. Nystedt, S.; Emilsson, K.; Wahlestedt, C.; Sundelin, J.: Molecular cloning of a potential proteinase-activated receptor. *Proc. Nat. Acad. Sci.* 91: 9208–9212, 1994.

[37163] 10715. Devlin, A. M.; Ling, E.; Peerson, J. M.; Fernando, S.;

Clarke,R.; Smith, A. D.; Halsted, C. H.: Glutamate carboxypeptidase II:a polymorphism associated with lower levels of serum folate and hyperhomocysteinemia.

Hum.Molec. Genet. 9: 2837–2844, 2000.

[37164] 10716.Israeli, R. S.; Powell, C. T.; Fair, W. R.; Heston, W. D. W.:Molecular cloning of a complementary DNA encoding a prostate-specificmembrane antigen. Cancer Res. 53: 227–230, 1993.

[37165] 10717.Leek, J.; Lench, N.; Maraj, B.; Bailey, A.; Carr, I. M.; Andersen,S.; Cross, J.; Whelan, P.; MacLennan, K. A.; Meredith, D. M.; Markham,A. F.: Prostate-specific membrane antigen: evidence for the existenceof a second related human gene. Brit. J. Cancer 72: 583–588, 1995.

[37166] 10718.Maraj, B. H.; Leek, J. P.; Karayi, M.; Ali, M.; Lench, N. J.; Markham,A. F.: Detailed genetic mapping around a putative prostate-specificmembrane antigen locus on human chromosome 11p11.2. Cytogenet. CellGenet. 81: 3–9, 1998.

[37167] 10719.Ikemoto, M.; Arai, H.; Feng, D.; Tanaka, K.; Aoki, J.; Dohmae,N.; Takio, K.; Adachi, H.; Tsujimoto, M.; Inoue, K.: Identificationof a PDZ-domain-containing protein that interacts with the scavengerreceptor class B type I. Proc. Nat. Acad. Sci. 97: 6538–6543, 2000.

- [37168] 10720.O'Keefe, D. S.; Su, S. L.; Bacich, D. J.; Horiguchi, Y.; Luo, Y.;Powell, C. T.; Zandvliet, D.; Russell, P. J.; Molloy, P. L.; Nowak,N. J.; Shows, T. B.; Mullins, C.; Vonder Haar, R. A.; Fair, W. R.;Heston, W. D. W.: Mapping, genomic organization and promoter analysisof the human prostate-specific membrane antigen gene. *Biochim. Biophys.Acta* 1443: 113–127, 1998.
- [37169] 10721.Rinker-Schaeffer, C. W.; Hawkins, A. L.; Su, S. L.; Israeli, R.S.; Griffin, C. A.; Isaacs, J. T.; Heston, W. D. W.: Localizationand physical mapping of the prostate-specific membrane antigen (PSM)gene to human chromosome 11. *Genomics* 30: 105–108, 1995.
- [37170] 10722.Beltrame, J. F.; Sasayama, S.; Maseri, A.: Racial heterogeneityin coronary artery vasomotor reactivity: differences between Japaneseand Caucasian patients. *J. Am. Coll. Cardiol.* 33: 1442–1452, 1999.
- [37171] 10723.Inagaki, N.; Inazawa, J.; Seino, S.: cDNA sequence, gene structure,and chromosomal localization of the human ATP-sensitive potassiumchannel, uK(ATP)–1, gene (KCNJ8). *Genomics* 30: 102–104, 1995.
- [37172] 10724.Inagaki, N.; Tsuura, Y.; Namba, N.; Masuda, K.; Gono, T.; Horie,M.; Seino, Y.; Mizuta, M.; Seino, S.: Cloning and functional characterizationof a novel ATP–

sensitive potassium channel ubiquitously expressed in rat tissues, including pancreatic islets, pituitary, skeletal muscle, and heart. *J. Biol. Chem.* 270: 5691–5694, 1995.

[37173] 10725. MacAlpin, R. N.: Cardiac arrest and sudden unexpected death in variant angina: complications of coronary spasm that can occur in the absence of severe organic coronary stenosis. *Am. Heart J.* 125: 1011–1017, 1993.

[37174] 10726. Bartsch, J. W.; Mukai, H.; Takahashi, N.; Ronsiek, M.; Fuchs, S.; Jockusch, H.; Ono, Y.: The protein kinase N (PKN) gene PRKCL1/Prkcl1 maps to human chromosome 19p12–p13.1 and mouse chromosome 8 with close linkage to the myodystrophy (myd) mutation. *Genomics* 49: 129–132, 1998.

[37175] 10727. Mukai, H.; Ono, Y.: A novel protein kinase with leucine zipper-like sequences: its catalytic domain is highly homologous to that of protein kinase C. *Biochem. Biophys. Res. Commun.* 199: 897–904, 1994.

[37176] 10728. Palmer, R. H.; Ridden, J.; Parker, P. J.: Identification of multiple, novel, protein kinase C-related gene products. *FEBS Lett.* 356: 5–8, 1994.

[37177] 10729. Palmer, R. H.; Ridden, J.; Parker, P. J.: Cloning and expression patterns of two members of a novel protein-kinase-C-related kinase family. *Europ. J. Biochem.* 227:

344–351, 1995.

- [37178] 10730. Yu, W.; Liu, J.; Morrice, N. A.; Wettenhall, R. E. H.: Isolation and characterization of a structural homologue of human PRK2 from rat liver. *J. Biol. Chem.* 272: 10030–10034, 1997.
- [37179] 10731. Durkin, M. E.; Loechel, F.; Mattei, M.-G.; Gilpin, B. J.; Albrechtsen, R.; Wewer, U. M.: Tissue-specific expression of the human laminin α -5-chain, and mapping of the gene to human chromosome 20q13.2–13.3 and to distal mouse chromosome 2 near the locus for the ragged (Ra) mutation. *FEBS Lett.* 411: 296–300, 1997.
- [37180] 10732. Miner, J. H.; Lewis, R. M.; Sanes, J. R.: Molecular cloning of a novel laminin chain, α -5, and widespread expression in adult mouse tissues. *J. Biol. Chem.* 270: 28523–28526, 1995.
- [37181] 10733. Hernandez, A.; Park, J. P.; Lyon, G. J.; Mohandas, T. K.; St. Germain, D. L.: Localization of the type 3 iodothyronine deiodinase (DIO3) gene to human chromosome 14q32 and mouse chromosome 12F1. *Genomics* 53: 119–121, 1998.
- [37182] 10734. Huang, S. A.; Tu, H. M.; Harney, J. W.; Venihaki, M.; Butte, A. J.; Kozakewich, H. P. W.; Fishman, S. J.; Larsen, P. R.: Severe hypothyroidism caused by type 3 iodothyronine

deiodinase in infantile hemangiomas. *NewEng. J. Med.* 343: 185–189, 2000.

- [37183] 10735.Salvatore, D.; Low, S. C.; Berry, M.; Maia, A. L.; Harney, J. W.;Croteau, W.; St. German, D. L.; Larsen, P. R.: Type 3 iodothyroninedeiodinase: cloning, in vitro expression, and functional analysisof the placental selenoenzyme. *J. Clin. Invest.* 96: 2421–2430, 1995.
- [37184] 10736.Kolodkin, A. L.; Matthes, D. J.; Goodman, C. S.: The semaphoringenes encode a family of transmembrane and secreted growth cone guidancemolecules. *Cell* 75: 1389–1399, 1993.
- [37185] 10737.Luo, Y.; Raible, D.; Raper, J. A.: Collapsin: a protein in brainthat induces the collapse and paralysis of neuronal growth cones. *Cell* 75:217–227, 1993.
- [37186] 10738.Marin, O.; Yaron, A.; Bagri, A.; Tessier–Lavigne, M.; Rubenstein,J. L. R.: Sorting of striatal and cortical interneurons regulatedby semaphorin–neuropilin interactions. *Science* 293: 872–875, 2001.
- [37187] 10739.Sekido, Y.; Bader, S.; Latif, F.; Chen, J.–Y.; Duh, F.–M.; Wei,M.–H.; Albanesi, J. P.; Lee, C.–C.; Lerman, M. I.; Minna, J. D.:Human semaphorins A(V) and IV reside in the 3p21.3 small cell lungcancer deletion region and demonstrate distinct expression patterns. *Proc.Nat. Acad. Sci.*

93: 4120–4125, 1996.

- [37188] 10740.Xiang, R.-H.; Hensel, C. H.; Garcia, D. K.; Carlson, H. C.; Kok,K.; Daly, M. C.; Kerbacher, K.; van den Berg, A.; Veldhuis, P.; Buys,C. H. C. M.; Naylor S. L.: Isolation of the human semaphorin III/Fgene (SEMA3F) at chromosome 3p21, a region deleted in lung cancer. *Genomics* 32:39–48, 1996.
- [37189] 10741.Ardehali, H.; Tiller, G. E.; Printz, R. L.; Mochizuki, H.; Prochazka,M.; Granner, D. K.: A novel (TA)_n polymorphism in the hexokinase II gene: application to noninsulin-dependent diabetes mellitus in the Pima Indians. *Hum. Genet.* 97: 482–485, 1996.
- [37190] 10742.Echwald, S. M.; Bjorbaek, C.; Hansen, T.; Clausen, J. O.; Vestergaard,H.; Zierarth, J. R.; Printz, R. L.; Granner, D. K.; Pedersen, O.:Identification of four amino acid substitutions in hexokinase II and studies of relationships to NIDDM, glucose effectiveness, and insulin sensitivity. *Diabetes* 44: 347–353, 1995.
- [37191] 10743.Heikkinen, S.; Suppola, S.; Malkki, M.; Deeb, S. S.; Janne, J.;Laakso, M.: Mouse hexokinase II gene: structure, cDNA, promoter analysis,and expression pattern. *Mammalian Genome* 11: 91–96, 2000.
- [37192] 10744.Laakso, M.; Malkki, M.; Deeb, S. S.: Amino acid

substitutions in hexokinase II among patients with NIDDM. Diabetes 44: 330–334, 1995.

- [37193] 10745. Lehto, M.; Xiang, K.; Stoffel, M.; Espinosa, R., III; Groop, L.C.; Le Beau, M. M.; Bell, G. I.: Human hexokinase II: localization of the polymorphic gene to chromosome 2. Diabetologia 36: 1299–1302, 1993.
- [37194] 10746. Mathupala, S. P.; Heese, C.; Pedersen, P. L.: Glucose catabolism in cancer cells: the type II hexokinase promoter contains functionally active response elements for the tumor suppressor p53. J. Biol. Chem. 272: 22776–22780, 1997.
- [37195] 10747. Mathupala, S. P.; Rempel, A.; Pedersen, P. L.: Glucose catabolism in cancer cells: isolation, sequence, and activity of the promoter for type II hexokinase. J. Biol. Chem. 270: 16918–16925, 1995.
- [37196] 10748. Vidal-Puig, A.; Printz, R. L.; Stratton, I. M.; Granner, D. K.; Moller, D. E.: Analysis of the hexokinase II gene in subjects with insulin resistance and NIDDM and detection of a gln142-to-his substitution. Diabetes 44: 340–346, 1995.
- [37197] 10749. Luo, Y.; Roeder, R. G.: Cloning, functional characterization, and mechanism of action of the B-cell-specific transcriptional activator OCA-B. Molec. Cell. Biol. 15:

4115–4124, 1995.

- [37198] 10750. Staudt, L. M.; Lenardo, M. J.: Immunoglobulin gene transcription. *Ann. Rev. Immun.* 9: 373–398, 1991.
- [37199] 10751. Strubin, M.; Newell, J. W.; Matthias, P.: OBF-1, a novel B cell-specific coactivator that stimulates immunoglobulin promoter activity through association with octamer-binding proteins. *Cell* 80: 497–506, 1995.
- [37200] 10752. Hahm, K.; Kim, G.; Turck, C. W.; Smale, S. T.: Isolation of a murine gene encoding a nucleic acid-binding protein with homology to hnRNP K. *Nucleic Acids Res.* 21: 3894 only, 1993.
- [37201] 10753. Leffers, H.; Dejgaard, K.; Celis, J. E.: Characterisation of two major cellular poly(rC)-binding human proteins, each containing three K-homologous (KH) domains. *Europ. J. Biochem.* 230: 447–453, 1995.
- [37202] 10754. Bouchard, M. J.; Wang, L.-H.; Schneider, R. J.: Calcium signaling by HBx protein in hepatitis B virus DNA replication. *Science* 294: 2376–2378, 2001.
- [37203] 10755. Calalb, M. B.; Polte, T. R.; Hanks, S. K.: Tyrosine phosphorylation of focal adhesion kinase at sites in the catalytic domain regulates kinase activity: a role for Src family kinases. *Molec. Cell. Biol.* 15: 954–963, 1995.
- [37204] 10756. Ganem, D.: The X files— one step closer to closure.

Science 294:2299–2300, 2001.

- [37205] 10757. Herzog, H.; Nicholl, J.; Hort, Y. J.; Sutherland, G. R.; Shine, J.: Molecular cloning and assignment of FAK2, a novel human focal adhesion kinase, to 8p11.2–p22 by nonisotopic in situ hybridization. *Genomics* 32:484–486, 1996.
- [37206] 10758. Lev, S.; Moreno, H.; Martinez, R.; Canoll, P.; Peles, E.; Musacchio, J. M.; Plowman, G. D.; Rudy, B.; Schlessinger, J.: Protein tyrosine kinase PYK2 involved in Ca^{2+} –induced regulation of ion channel and MAP kinase functions. *Nature* 376: 737–745, 1995.
- [37207] 10759. Manser, E.; Leung, T.; Salihuddin, H.; Tan, L.; Lim, L.: A non-receptor tyrosine kinase that inhibits the GTPase activity of p21(cdc42). *Nature* 363:364–367, 1993.
- [37208] 10760. Matsuya, M.; Sasaki, H.; Aoto, H.; Mitaka, T.; Nagura, K.; Ohba, T.; Ishino, M.; Takahashi, S.; Suzuki, R.; Sasaki, T.: Cell adhesion kinase beta forms a complex with a new member, Hic-5, of proteins localized at focal adhesions. *J. Biol. Chem.* 273: 1003–1014, 1998.
- [37209] 10761. Aubry, F.; Mattei, M.-G.; Barque, J.-P.; Galibert, F.: Chromosomal localization and expression pattern of the RNase L inhibitor gene. *FEBS Lett.* 381: 135–139, 1996.
- [37210] 10762. Diriong, S.; Salehzada, T.; Bisbal, C.; Martinand, C.;

Taviaux,S.: Localization of the ribonuclease L inhibitor gene (RNS4I), anew member of the interferon-regulated 2-5A pathway, to 4q31 by fluorescencein situ hybridization. Genomics 32: 488-490, 1996.

[37211] 10763.Meyer, J.; Wirth, J.; Held, M.; Schempp, W.; Scherer, G.: SOX20,a new member of the SOX gene family, is located on chromosome 17p13. Cytogenet.Cell Genet. 72: 246-249, 1996.

[37212] 10764.Vujic, M.; Rajic, T.; Goodfellow, P. N.; Stevanovic, M.: cDNAcharacterization and high resolution mapping of the human SOX20 gene. MammalianGenome 9: 1059-1061, 1998.

[37213] 10765.Ide, H.; Saito-Ohara, F.; Ohnami, S.; Osada, Y.; Ikeuchi, T.; Yoshida,T.; Terada, M.: Assignment of the BMPR1A and BMPR1B genes to humanchromosome 10q22.3 and 4q23-q24 by in situ hybridization and radiationhybrid mapping. Cytogenet. Cell Genet. 81: 285-286, 1998.

[37214] 10766.Schuffenhauer, S.; Lichtner, P.; Peykar-Derakhshandeh, P.; Murken,J.; Haas, O. A.; Back, E.; Wolff, G.; Zabel, B.; Barisic, I.; Rauch,A.; Borochowitz, Z.; Dal-lapiccola, B.; Ross, M.; Meitinger, T.: Deletionmapping on chromosome 10p and definition of a critical region forthe

second DiGeorge syndrome locus (DGS2). *Europ. J. Hum. Genet.* 6:213–225, 1998.

- [37215] 10767. Pizzuti, A.; Amati, F.; Calabrese, G.; Mari, A.; Colosimo, A.; Silani, V.; Giardino, L.; Ratti, A.; Penso, D.; Calza, L.; Palka, G.; Scarlato, G.; Novelli, G.; Dallapiccola, B.: cDNA characterization and chromosomal mapping of two human homologs of the *Drosophila* dishevelled polarity-gene. *Hum. Molec. Genet.* 5: 953–958, 1996.
- [37216] 10768. Semenov, M. V.; Snyder, M.: Human dishevelled genes constitute a DHR-containing multigene family. *Genomics* 42: 302–310, 1997.
- [37217] 10769. Lee, J.; Hahn, Y.; Yun, J. H.; Mita, K.; Chung, J. H.: Characterization of JDP genes, an evolutionarily conserved J domain-only protein family, from human and moths. *Biochim. Biophys. Acta* 1491: 355–363, 2000.
- [37218] 10770. Qiu, J.; Qian, Y.; Chen, V.; Guan, M.-X.; Shen, B.: Human exonuclease 1 functionally complements its yeast homologues in DNA recombination, RNA primer removal, and mutation avoidance. *J. Biol. Chem.* 274: 17893–17900, 1999.
- [37219] 10771. Schmutte, C.; Marinescu, R. C.; Sadoff, M. M.; Guerrette, S.; Overhauser, J.; Fishel, R.: Human exonuclease I interacts with the mismatch repair protein hMSH2.

Cancer Res. 58: 4537–4542, 1998.

[37220] 10772. Tishkoff, D. X.; Amin, N. S.; Viars, C. S.; Arden, K. C.; Kolodner, R. D.: Identification of a human gene encoding a homologue of *Saccharomyces cerevisiae* EXO1, an exonuclease implicated in mismatch repair and recombination. Cancer Res. 58: 5027–5031, 1998.

[37221] 10773. Wilson, D. M., III; Carney, J. P.; Coleman, M. A.; Adamson, A. W.; Christensen, M.; Lamerdin, J. E.: Hex1: a new human Rad2 nuclease family member with homology to yeast exonuclease 1. Nucleic Acids Res. 26: 3762–3768, 1998.

[37222] 10774. Carson-Walter, E. B.; Watkins, D. N.; Nanda, A.; Vogelstein, B.; Kinzler, K. W.; St. Croix, B.: Cell surface tumor endothelial markers are conserved in mice and humans. Cancer Res. 61: 6649–6655, 2001.

[37223] 10775. Christian, S.; Ahorn, H.; Koehler, A.; Eisenhaber, F.; Rodi, H.-P.; Garin-Chesa, P.; Park, J. E.; Rettig, W. J.; Lenter, M. C.: Molecular cloning and characterization of endosialin, a C-type lectin-like cell surface receptor of tumor endothelium. J. Biol. Chem. 276: 7408–7414, 2001.

[37224] 10776. St. Croix, B.; Rago, C.; Velculescu, V.; Traverso, G.; Romans, K. E.; Montgomery, E.; Lal, A.; Riggins, G. J.; Lengauer, C.; Vogelstein, B.; Kinzler, K. W.: Genes ex-

pressed in human tumor endothelium. *Science* 289:1197–1202, 2000.

- [37225] 10777.Gosling, J.; Dairaghi, D. J.; Wang, Y.; Hanley, M.; Talbot, D.; Miao, Z.; Schall, T. J.: Cutting edge: identification of a novel chemokine receptor that binds dendritic cell- and T cell-active chemokines including ELC, SLC, and TECK. *J. Immun.* 164: 2851–2856, 2000.
- [37226] 10778.Cohn, R. D.; Campbell, K. P.: Molecular basis of muscular dystrophies. *Muscle Nerve* 23: 1456–1471, 2000.
- [37227] 10779.Gillespie, C. S.; Lee, M.; Fantes, J. F.; Brophy, P. J.: The gene encoding the Schwann cell protein periaxin localizes on mouse chromosome 7 (Prx). *Genomics* 41: 297–298, 1997.
- [37228] 10780.Gillespie, C. S.; Sherman, D. L.; Fleetwood-Walker, S. M.; Cottrell, D. F.; Tait, S.; Garry, E. M.; Wallace, V. C. J.; Ure, J.; Griffiths, I. R.; Smith, A. Brophy, P. J.: Peripheral demyelination and neuropathic pain behavior in periaxin-deficient mice. *Neuron* 26: 523–531, 2000.
- [37229] 10781.Schuler, G. D.: Sequence mapping by electronic PCR. *Genome Res.* 7:541–550, 1997.
- [37230] 10782.Takashima, H.; Boerkoel, C. F.; De Jonghe, P.; Ceuterick, C.; Martin, J.-J.; Voit, T.; Schroder, J.-M.; Williams, A.; Brophy, P. J.; Timmerman, V.; Lupski, J. R.: Periaxin mu-

tations cause a broad spectrum of demyelinating neuropathies. *Ann. Neurol.* 51: 709–715, 2002.

- [37231] 10783.Liu, Q.-Y.; Wang, L. F.; Miao, S. Y.; Catterall, J. F.: Expression and characterization of a novel human sperm membrane protein. *Biol.Reprod.* 54: 323–330, 1996.
- [37232] 10784.Miao, S.; Yan, Y.; Li, Y.; Bai, Y.; Wei, S.; Zong, C.; Zhao, M.;Zong, S.; Wang, L.: cDNA encoding a human sperm membrane protein BS-84. *Prog. Natural Sci.* 5: 119–122, 1995.
- [37233] 10785.Wang, H.; Miao, S.; Chen, D.; Wang, L.; Koide, S. S.: Assignment of chromosomal locus and evidence for alternatively spliced mRNAs of a human sperm membrane protein (hSMP-1). *Biochim. Biophys. Acta* 1447:119–124, 1999.
- [37234] 10786.Horie, M.; Mitsumoto, Y.; Kyushiki, H.; Kanemoto, N.; Watanabe,A.; Taniguchi, Y.; Nishino, N.; Okamoto, T.; Kondo, M.; Mori, T.;Noguchi, K.; Nakamura, Y.; Takahashi, E.; Tanigami, A.: Identification and characterization of TM-EFF2, a novel survival factor for hippocampal and mesencephalic neurons. *Genomics* 67: 146–152, 2000.
- [37235] 10787.Uchida, T.; Wada, K.; Akamatsu, T.; Yonezawa, M.; Noguchi, H.;Mizoguchi, A.; Kasuga, M.; Sakamoto, C.: A novel epidermal growth factor-like molecule containing

two follistatin modules stimulate tyrosine phosphorylation of erbB-4 in MKN28 gastric cancer cells.

Biochem. Biophys. Res. Commun. 266: 593–602, 1999.

- [37236] 10788. Young, J.; Biden, K. G.; Simms, L. A.; Huggard, P.; Karamatic, R.; Eyre, H. J.; Sutherland, G. R.; Herath, N.; Barker, M.; Anderson, G. J.; Fitzpatrick, D. R.; Ramm, G. A.; Jass, J. R.; Leggett, B. A.: HPP1: a transmembrane protein-encoding gene commonly methylated in colorectal polyps and cancers. *Proc. Nat. Acad. Sci.* 98: 265–270, 2001.
- [37237] 10789. Blasina, A.; de Weyer, I. V.; Laus, M. C.; Luyten, W. H.; Parker, A. E.; McGowan, C. H.: A human homologue of the checkpoint kinase Cds1 directly inhibits Cdc25 phosphatase. *Curr. Biol.* 14: 1–10, 1999.
- [37238] 10790. Brown, A. L.; Lee, C.-H.; Schwarz, J. K.; Mitiku, N.; Piwnicka-Worms, H.; Chung, J. H.: A human Cds1-related kinase that functions downstream of ATM protein in the cellular response to DNA damage. *Proc. Nat. Acad. Sci.* 96: 3745–3750, 1999.
- [37239] 10791. Chaturvedi, P.; Eng, W. K.; Zhu, Y.; Mattern, M. R.; Mishra, R.; Hurle, M. R.; Zhang, X.; Annan, R. S.; Lu, Q.; Faucette, L. F.; Scott, G. F.; Li, X.; Carr, S. A.; Johnson, R. K.; Winkler, J. D.; Zhou, B. B.: Mammalian Chk2 is a downstream effector of the ATM-dependent DNA damage

checkpoint pathway. *Oncogene* 18: 4047–4054, 1999.

- [37240] 10792. Chehab, N. H.; Malikzay, A.; Appel, M.; Hala-zonitis, T. D.: Chk2/hCds1 functions as a DNA damage checkpoint in G-1 by stabilizing p53. *Genes Dev.* 14: 278–288, 2000.
- [37241] 10793. Ino, Y.; Wahrer, D. C. R.; Bell, D. W.; Haber, D. A.; Louis, D. N.: Mutation analysis of the hCHK2 gene in primary human malignant gliomas. (Letter) *Neurogenetics* 3: 45–46, 2000.
- [37242] 10794. Lee, S. B.; Kim, S. H.; Bell, D. W.; Wahrer, D. C. R.; Schiripo, T. A.; Jorczak, M. M.; Sgroi, D. C.; Garber, J. E.; Li, F. P.; Nichols, K. E.; Varley, J. M.; Godwin, A. K.; Shannon, K. M.; Harlow, E.; Haber, D. A.: Destabilization of CHK2 by a missense mutation associated with Li-Fraumeni syndrome. *Cancer Res.* 61: 8062–8067, 2001.
- [37243] 10795. Lopes, M.; Cotta-Ramusino, C.; Pelliccioli, A.; Liberi, G.; Plevani, P.; Muzi-Falconi, M. Newlon, C. S.; Foiani, M.: The DNA replication checkpoint response stabilizes stalled replication forks. *Nature* 412: 557–561, 2001.
- [37244] 10796. Matsuoka, S.; Huang, M.; Elledge, S. J.: Linkage of ATM to cell cycle regulation by the Chk2 protein kinase. *Science* 282: 1893–1897, 1998.
- [37245] 10797. Meijers-Heijboer, H.; van den Ouweland, A.; Klijn,

J.; Wasielewski, M.; de Snoo, A.; Oldenburg, R.; Hollestelle, A.; Houben, M.; Crepin, E.; van Veghel-Plandsoen, M.; Elstrodt, F.; van Duijn, C.; and 29 others: Low-penetrance susceptibility to breast cancer due to CHEK2*1100delC in noncarriers of BRCA1 or BRCA2 mutations. *Nature Genet.* 31: 55–59, 2002.

[37246] 10798. Miller, C. W.; Ikezoe, T.; Krug, U.; Hofmann, W.-K.; Tavor, S.; Vegesna, V.; Tsukasaki, K.; Takeuchi, S.; Koeffler, H. P.: Mutations of the CHK2 gene are found in some osteosarcomas, but are rare in breast, lung, and ovarian tumors. *Genes Chromosomes Cancer* 33: 17–21, 2002.

[37247] 10799. Sogo, J. M.; Lopes, M.; Foiani, M.: Fork reversal and ssDNA accumulation at stalled replication forks owing to checkpoint defects. *Science* 297: 599–602, 2002.

[37248] 10800. Vahteristo, P.; Bartkova, J.; Eerola, H.; Syrjäkoski, K.; Ojala, S.; Kilpivaara, O.; Tamminen, A.; Kononen, J.; Aittomäki, K.; Heikkilä, P.; Holli, K.; Blomqvist, C.; Bartek, J.; Kallioniemi, O.-P.; Nevanlinna, H.: A CHEK2 genetic variant contributing to a substantial fraction of familial breast cancer. *Am. J. Hum. Genet.* 71: 432–438, 2002.

[37249] 10801. Vahteristo, P.; Tamminen, A.; Karvinen, P.; Eerola, H.; Eklund, C.; Aaltonen, L. A.; Blomqvist, C.; Aittomäki, K.; Nevanlinna, H.: p53, CHK2, and CHK1 genes in Finnish

families with Li-Fraumeni syndrome: further evidence of
CHK2 in inherited cancer predisposition. *Cancer Res.* 61:
5718–5722, 2001.

[37250] 10802. De, S. K.; Enders, G. C.; Andrews, G. K.: High levels
of metallothionein messenger RNAs in male germ cells of
the adult mouse. *Molec. Endocr.* 5:628–636, 1991.

[37251] 10803. Salehi-Ashtiani, K.; Widrow, R. J.; Markert, C. L.;
Goldberg, E.: Testis-specific expression of a metalloth-
ionein I-driven transgene correlates with undermethylation
of the locus in testicular DNA. *Proc. Nat. Acad. Sci.* 90:
8886–8890, 1993.

[37252] 10804. Sugihara, T.; Wadhwa, R.; Kaul, S. C.; Mitsui, Y.: A
novel testis-specific metallothionein-like protein, tesmin,
is an early marker of male germ cell differentiation. *Ge-
nomics* 57: 130–136, 1999.

[37253] 10805. Larrouy, D.; Vidal, H.; Andreelli, F.; Laville, M.; Lan-
gin, D.: Cloning and mRNA tissue distribution of human
PPAR-gamma coactivator-1. *Int. J. Obesity* 23: 1327–1332,
1999.

[37254] 10806. Lin, J.; Wu, H.; Tarr, P. T.; Zhang, C.-Y.; Wu, Z.;
Boss, O.; Michael, L. F.; Puigserver, P.; Isotani, E.; Olson, E.
N.; Lowell, B. B.; Bassel-Duby, R.; Spiegelman, B. M.: Tran-
scriptional co-activator PGC-1-alpha drives the formation

of slow-twitch muscle fibres. *Nature* 418: 797–801,2002.

[37255] 10807.Monsalve, M.; Wu, Z.; Adelmant, G.; Puigserver, P.; Fan, M.; Spiegelman,B. M.: Direct coupling of transcription and mRNA processing throughthe thermogenic coactivator PGC-1. *Molec. Cell* 6: 307–316, 2000.

[37256] 10808.Puigserver, P.; Adelmant, G.; Wu, Z.; Fan, M.; Xu, J.; O'Malley,B.; Spiegelman, B. M.: Activation of PPAR-gamma coactivator-1 throughtranscription factor docking. *Science* 286: 1368–1371, 1999.

[37257] 10809.Puigserver, P.; Rhee, J.; Lin, J.; Wu, Z.; Yoon, J. C.; Zhang,C.-Y.; Krauss, S.; Mootha, V. K.; Lowell, B. B.; Spiegelman, B. M.: Cytokine stimulation of energy expenditure through p38 MAP kinaseactivation of PPAR-gamma coactivator-1. *Molec. Cell* 8: 971–982,2001.

[37258] 10810.Puigserver, P.; Wu, Z.; Park, C. W.; Graves, R.; Wright, M.; Spiegelman,B. M.: A cold-inducible coactivator of nuclear receptors linked toadaptive thermogenesis. *Cell* 92: 829–839, 1998.

[37259] 10811.Waite, L. L.; Person, E. C.; Zhou, Y.; Lim, K.-H.; Scanlan, T.S.; Taylor, R. N.: Placental peroxisome proliferator-activated receptor-gammais up-regulated by pregnancy serum. *J. Clin. Endocr. Metab.* 85:3808–3814, 2000.

[37260] 10812.Wu, Z.; Puigserver, P.; Andersson, U.; Zhang, C.;

Adelmant, G.; Mootha, V.; Troy, A.; Cinti, S.; Lowell, B.; Scarpulla, R. C.; Spiegelman, B. M.: Mechanisms controlling mitochondrial biogenesis and respiration through the thermogenic coactivator PGC-1. *Cell* 98: 115–124, 1999.

[37261] 10813. Yoon, J. C.; Puigserver, P.; Chen, G.; Donovan, J.; Wu, Z.; Rhee, J.; Adelmant, G.; Stafford, J.; Kahn, C. R.; Granner, D. K.; Newgard, C. B.; Spiegelman, B. M.: Control of hepatic gluconeogenesis through the transcriptional coactivator PGC-1. *Nature* 413: 131–138, 2001.

[37262] 10814. Ellis, J. H.; Ashman, C.; Burden, M. N.; Kilpatrick, K. E.; Morse, M. A.; Hamblin, P. A.: GRID: a novel Grb-2-related adapter protein that interacts with the activated T cell costimulatory receptor CD28. *J. Immun.* 164: 5805–5814, 2000.

[37263] 10815. Qiu, M.; Hua, S.; Agrawal, M.; Li, G.; Cai, J.; Chan, E.; Zhou, H.; Luo, Y.; Liu, M.: Molecular cloning and expression of human Grap-2, a novel leukocyte-specific SH2- and SH3-containing adaptor-like protein that binds to Gab-1. *Biochem. Biophys. Res. Commun.* 253: 443–447, 1998.

[37264] 10816. Yoder, J.; Pham, C.; Iizuka, Y.-M.; Kanagawa, O.; Liu, S. K.; McGlade, J.; Cheng, A. M.: Requirement for the SLP-76 adaptor GADS in T cell development. *Science* 291:

1987–1991, 2001.

- [37265] 10817. Granger, S. W.; Butrovich, K. D.; Houshmand, P.; Edwards, W. R.; Ware, C. F.: Genomic characterization of LIGHT reveals linkage to an immune response locus on chromosome 19p13.3 and distinct isoforms generated by alternate splicing or proteolysis. *J. Immun.* 167: 5122–5128, 2001.
- [37266] 10818. Mauri, D. N.; Ebner, R.; Montgomery, R. I.; Kochel, K. D.; Cheung, T. C.; Yu, G. L.; Ruben, S.; Murphy, M.; Eisenberg, R. J.; Cohen, G. H.; Spear, P. G.; Ware, C. F.: LIGHT, a new member of the TNF superfamily, and lymphotoxin alpha are ligands for herpesvirus entry mediator. *Immunity* 8: 21–30, 1998.
- [37267] 10819. Harrop, J. A.; McDonnell, P. C.; Brigham–Burke, M.; Lyn, S. D.; Minton, J.; Tan, K. B.; Dede, K.; Spanpanato, J.; Silverman, C.; Hensley, P.; DiPrinzio, R.; Emery, J. G.; Deen, K.; Eichman, C.; Chabot–Fletcher, M.; Truneh, A.; Young, P. R.: Herpesvirus entry mediator ligand (HVEM–L), a novel ligand for HVEM/TR2, stimulates proliferation of T cells and inhibits HT29 cell growth. *J. Biol. Chem.* 273: 27548–27556, 1998.
- [37268] 10820. Hadjantonakis, A.–K.; Formstone, C. J.; Little, P. F. R.: mCelsr1 is an evolutionarily conserved seven–pass

transmembrane receptor and is expressed during mouse embryonic development. *Mech. Dev.* 78:91–95, 1998.

[37269] 10821. Hadjantonakis, A.-K.; Sheward, W. J.; Harmar, A. J.; de Galan, L.; Hoovers, J. M. N.; Little, P. F. R.: Celsr1, a neural-specific gene encoding an unusual seven-pass transmembrane receptor, maps to mouse chromosome 15 and human chromosome 22qter. *Genomics* 45: 97–104, 1997.

[37270] 10822. Hewett-Emmett, D.; Tashian, R. E.: Functional diversity, conservation, and convergence in the evolution of the alpha-, beta-, and gamma-carbonic anhydrase gene families. *Molec. Phylogenet. Evol.* 5: 50–77, 1996.

[37271] 10823. Hewett-Emmett, D.; Wiebauer, K.: Personal Communication. Houston, Texas 9/1999.

[37272] 10824. Kleiderlein, J. J.; Nisson, P. E.; Jessee, J.; Li, W.-B.; Becker, K. G.; Derby, M. L.; Ross, C. A.; Margolis, R. L.: CCG repeats in cDNAs from human brain. *Hum. Genet.* 103: 666–673, 1998.

[37273] 10825. Lovejoy, D. A.; Hewett-Emmett, D.; Porter, C. A.; Cepoi, D.; Sheffield, A.; Vale, W. W.; Tashian, R. E.: Evolutionarily conserved, 'acatalytic' carbonic anhydrase-related protein XI contains a sequence motif present in the neuropeptide sauvagine: the human CA-RP XI gene (CA11) is

embedded between the secretor gene cluster and the DBP gene at 19q13.3. *Genomics* 54:484–493, 1998.

- [37274] 10826. Bellingham, J.; Gregory-Evans, K.; Gregory-Evans, C. Y.: Sequence and tissue expression of a human novel carbonic anhydrase-related protein, CARP-2, mapping to chromosome 19q13.3. *Biochem. Biophys. Res. Comm.* 253: 364–367, 1998.
- [37275] 10827. Fujikawa-Adachi, K.; Nishimori, I.; Taguchi, T.; Yuri, K.; Onishi, S.: cDNA sequence, mRNA expression, and chromosomal localization of human carbonic anhydrase-related protein, CA-RP XI. *Biochim. Biophys. Acta* 1431: 518–524, 1999.
- [37276] 10828. Lindsay, M. E.; Plafker, K.; Smith, A. E.; Clurman, B. E.; Macara, I. G.: Np60/Nup50 is a tri-stable switch that stimulates importin- α : β -mediated nuclear protein import. *Cell* 110: 349–360, 2002.
- [37277] 10829. Trichet, V.; Shkolny, D.; Dunham, I.; Beare, D.; McDermid, H. E.: Mapping and complex expression pattern of the human NPAP60L nucleoporin gene. *Cytogenet. Cell Genet.* 85: 221–226, 1999.
- [37278] 10830. de Veer, M. J.; Sim, H.; Whisstock, J. C.; Devenish, R. J.; Ralph, S. J.: IFI60/ISG60/IFIT4, a new member of the human IFI54/IFIT2 family of interferon-stimulated genes.

Genomics 54: 267–277, 1998.

[37279] 10831. Yu, M.; Tong, J.-H.; Mao, M.; Kan, L.-X.; Liu, M.-M.; Sun, Y.-W.; Fu, G.; Jing, Y.-K.; Yu, L.; Lepaslier, D.; Lanotte, M.; Wang, Z.-Y.; Chen, Z.; Waxman, S.; Wang, Y.-X.; Tan, J.-Z.; Chen, S.-J.: Cloning of a gene (RIG-G) associated with retinoic acid-induced differentiation of acute promyelocytic leukemia cells and representing a new member of a family of interferon-stimulated genes. *Proc. Nat. Acad. Sci.* 94: 7406–7411, 1997.

[37280] 10832. Zhu, H.; Cong, J.-P.; Shenk, T.: Use of differential display analysis to assess the effect of human cytomegalovirus infection on the accumulation of cellular RNAs: induction of interferon-responsive RNAs. *Proc. Nat. Acad. Sci.* 94: 13985–13990, 1997.

[37281] 10833. Hirai, H.; Tanaka, K.; Takano, S.; Ichimasa, M.; Nakamura, M.; Nagata, K.: Cutting edge: agonistic effect of indomethacin on a prostaglandin D₂ receptor, CRTH2. *J. Immun.* 168: 981–985, 2002.

[37282] 10834. Nagata, K.; Tanaka, K.; Ogawa, K.; Kemmotsu, K.; Imai, Yoshie, O.; Abe, H.; Tada, K.; Nakamura, M.; Sugamura, K.; Takano, S.: Selective expression of a novel surface molecule by human Th2 cells in vivo. *J. Immun.* 162: 1278–1286, 1999.

- [37283] 10835.Grundemann, D.; Schechinger, B.; Rappold, G. A.; Schomig, E.:Molecular identification of the corticosterone-sensitive extraneuronalcatecholamine transporter. *Nature Neurosci.* 1: 349–351, 1998.
- [37284] 10836.Boettger, T.; Hubner, C. A.; Maler, H.; Rust, M. B.; Beck, F. X.;Jentsch, T. J.: Deafness and renal tubular acidosis in mice lackingthe K–Cl co–transporter Kcc4. *Nature* 416: 874–878, 2002.
- [37285] 10837.Kotake, K.; Ozaki, N.; Mizuta, M.; Sekiya, S.; Inagaki, N.; Seino,S.: Noc2, a putative zinc finger protein involved in exocytosis inendocrine cells. *J. Biol. Chem.* 272: 29407–29410, 1997.
- [37286] 10838.Smith, J. S.; Tachibana, I.; Allen, C.; Chiappa, S. A.; Lee, H.K.; Mclver, B.; Jenkins, R. B.; Raffel, C.: Cloning of a human ortholog(RPH3AL) of (RNO)Rph3al from a candidate 17p13.3 medulloblastoma tumorsuppressor locus. *Genomics* 59: 97–101, 1999.
- [37287] 10839.Nomura, N.; Miyajima, N.; Sazuka, T.; Tanaka, A.; Kwarabayasi,Y.; Sato, S.; Nagase, T.; Seki, N.; Ishikawa, K.; Tabata, S.: Predictionof the coding sequences of unidentified human genes. I. The codingsequences of 40 new genes (KIAA0001–KIAA0040) deduced by analysisof randomly sampled cDNA clones from human immature

myeloid cell line KG-1. DNA Res. 1: 27-35, 1994.

- [37288] 10840. Kimura, M.; Okano, Y.: Identification and assignment of the human NIMA-related protein kinase 7 gene (NEK7) to human chromosome 1q31.3. Cytogenet. Cell Genet. 94: 33-38, 2001.
- [37289] 10841. Li, M. Z.; Yu, L.; Liu, Q.; Chu, J. Y.; Zhao, S. Y.: Assignment of NEK6, a NIMA-related gene, to human chromosome 9q33.3-q34.11 by radiation hybrid mapping. Cytogenet. Cell Genet. 87: 271-272, 1999.
- [37290] 10842. Favier, D.; Gonda, T. J.: Detection of proteins that bind to the leucine zipper motif of c-Myb. Oncogene 9: 305-311, 1994.
- [37291] 10843. Keough, R.; Woollatt, E.; Crawford, J.; Sutherland, G. R.; Plummer, S.; Casey, G.; Gonda, T. J.: Molecular cloning and chromosomal mapping of the human homologue of MYB binding protein (P160) 1A (MYBBP1A) to 17p13.3. Genomics 62: 483-489, 1999.
- [37292] 10844. Tavner, F. J.; Simpson, R.; Tashiro, S.; Favier, D.; Jenkins, N. A.; Gilbert, D. J.; Copeland, N. G.; Macmillan, E. M.; Lutwyche, J.; Keough, R. A.; Ishii, S.; Gonda, T. J.: Molecular cloning reveals that the p160 Myb-binding protein is a novel, predominantly nucleolar protein which may play a role in transactivation by Myb. Molec. Cell Biol. 18:

989–1002, 1998.

- [37293] 10845. Gilbert, D. J.; Engel, H.; Wang, X.; Grzeschik, K.-H.; Copeland, N. G.; Jenkins, N. A.; Kilimann, M. W.: The neurobeachin gene (Nbea) identifies a new region of homology between mouse central chromosome 3 and human chromosome 13q13. *Mamm. Genome* 10: 1030–1031, 1999.
- [37294] 10846. Nagle, D. L.; Karim, M. A.; Woolf, E. A.; Holmgren, L.; Bork, P.; Misumi, D. J.; McGrail, S. H.; Dussault, B. J., Jr.; Perou, C. M.; Boissy, R. E.; Duyk, G. M.; Spritz, R. A.; Moore, K. J.: Identification and mutation analysis of the complete gene for Chediak–Higashi syndrome. *Nature–Genet.* 14: 307–311, 1996.
- [37295] 10847. Wang, X.; Herberg, F. W.; Laue, M. M.; Wullner, C.; Hu, B.; Petrasch–Parwez, E.; Kilimann, M. W.: Neurobeachin: a protein kinase A–anchoring, beige/Chediak–Higashi protein homolog implicated in neuronal membrane traffic. *J. Neurosci.* 20: 8551–8565, 2000.
- [37296] 10848. Bunn, R. C.; Jensen, M. A.; Reed, B. C.: Protein interactions with the glucose transporter binding protein GLUT1CBP that provide a link between GLUT1 and the cytoskeleton. *Molec. Biol. Cell* 10: 819–832, 1999.
- [37297] 10849. De Vries, L.; Lou, X.; Zhao, G.; Zheng, B.; Farquhar,

M. G.: GIPC, a PDZ domain containing protein, interacts specifically with the Cterminus of RGS-GAIP. Proc. Nat. Acad. Sci. 95: 12340–12345, 1998.

[37298] 10850. Von Kap-Herr, C.; Kandala, G.; Mann, S. S.; Hart, T. C.; Pettenati, M. J.; Setaluri, V.: Assignment of PDZ domain-containing protein GIPC gene (C19orf3) to human chromosome band 19p13.1 by in situ hybridization and radiation hybrid mapping. Cytogenet. Cell Genet. 89: 234–235, 2000.

[37299] 10851. Alessi, D. R.; James, S. R.; Downes, C. P.; Holmes, A. B.; Gaffney, P. R. J.; Reese, C. B.; Cohen, P.: Characterization of a 3-phosphoinositide-dependent protein kinase which phosphorylates and activates protein kinase B-alpha. Curr. Biol. 7: 261–269, 1997.

[37300] 10852. Sandrini, F.; Farmakidis, C.; Kirschner, L. S.; Wu, S.-M.; Tullio-Pelet, A.; Lyonnet, S.; Metzger, D. L.; Bourdony, C. J.; Tiosano, D.; Chan, W.-Y.; Stratakis, C. A.: Spectrum of mutations of the AAAS gene in Allgrove syndrome: lack of mutations in six kindreds with isolated resistance to corticotropin. J. Clin. Endocr. Metab. 86: 5433–5437, 2001.

[37301] 10853. Ludwig, D.; Lorenz, J.; Dejana, E.; Bohlen, P.; Hicklin, D. J.; Witte, L.; Pytowski, B.: cDNA cloning, chromoso-

mal mapping, and expression analysis of human VE-cadherin-2. *Mammalian Genome* 11: 1030–1033, 2000.

[37302] 10854. Telo, P.; Breviario, F.; Huber, P.; Panzeri, C.; Dejana, E.: Identification of a novel cadherin (vascular endothelial cadherin-2) located at intercellular junctions in endothelial cells. *J. Biol. Chem.* 273: 17565–17572, 1998.

[37303] 10855. Adams, J. C.; Seed, B.; Lawler, J.: Muskulin, a novel intracellular mediator of cell adhesive and cytoskeletal responses to thrombospondin-1. *EMBO J.* 17: 4964–4974, 1998.

[37304] 10856. Adams, J. C.; Zhang, L.: cDNA cloning of human muskulin and localisation of the muskulin (MKLN1) gene to human chromosome 7q32 and mouse chromosome 6B1/B2 by physical mapping and FISH. *Cytogenet. Cell Genet.* 87:19–21, 1999.

[37305] 10857. Moynihan, T. P.; Ardley, H. C.; Nuber, U.; Rose, S. A.; Jones, P. F.; Markham, A. F.; Scheffner, M.; Robinson, P. A.: The ubiquitin-conjugating enzymes UbcH7 and UbcH8 interact with RING finger/IBR motif-containing domains of HHARI and H7-AP1. *J. Biol. Chem.* 274: 30963–30968, 1999.

[37306] 10858. Tan, N. G. S.; Ardley, H. C.; Rose, S. A.; Leek, J. P.; Markham, A. F.; Robinson, P. A.: Characterisation of the

human and mouse orthologues of the *Drosophila ariadne* gene. *Cytogenet. Cell Genet.* 90: 242–245, 2000.

- [37307] 10859. Janssen, J. W. G.; Imoto, I.; Inoue, J.; Shimada, Y.; Ueda, M.; Imamura, M.; Bartram, C. R.; Inazawa, J.: MYEOV, a gene at 11q13, is coamplified with CCND1, but epigenetically inactivated in a subset of esophageal squamous cell carcinomas. *J. Hum. Genet.* 47: 460–464, 2002.
- [37308] 10860. Janssen, J. W. G.; Vaandrager, J.-W.; Heuser, T.; Jauch, A.; Kluin, P. M.; Geelen, E.; Bergsagel, P. L.; Kuehl, W. M.; Drexler, H. G.; Otsuki, T.; Bartram, C. R.; Schuurin, E.: Concurrent activation of a novel putative transforming gene, *myeov*, and cyclin D1 in a subset of multiple myeloma cell lines with t(11;14)(q13;q32). *Blood* 95:2691–2698, 2000.
- [37309] 10861. Di Cunto, F.; Calautti, E.; Hsiao, J.; Ong, L.; Topley, G.; Turco, E.; Dotto, G. P.: Citron Rho-interacting kinase, a novel tissue-specific ser/thr kinase encompassing the Rho-Rac-binding protein citron. *J. Biol. Chem.* 273: 29706–29711, 1998.
- [37310] 10862. Di Cunto, F.; Imarisio, S.; Hirsch, E.; Broccoli, V.; Bulfone, A.; Migheli, A.; Atzori, C.; Turco, E.; Triolo, R.; Dotto, G. P.; Silengo, L.; Altruda, F.: Defective neurogenesis in citron kinase knockout mice by altered cytokinesis and

massive apoptosis. *Neuron* 28:115–127, 2000.

- [37311] 10863.Madaule, P.; Furuyashiki, T.; Reid, T.; Ishizaki, T.; Watanabe,G.; Morii, N.; Narumiya, S.: A novel partner for the GTP-bound formsof rho and rac. *FEBS Lett.* 377: 243–248, 1995.
- [37312] 10864.Ishizaki, J.; Suzuki, N.; Higashino, K.; Yokota, Y.; Ono, T.; Kawamoto,K.; Fujii, N.; Arita, H.; Hanasaki, K.: Cloning and characterizationof novel mouse and human secretory phospholipase A(2)s. *J. Biol.Chem.* 274: 24973–24979, 1999.
- [37313] 10865.Baudat, F.; Manova, K.; Yuen, J. P.; Jasin, M.; Keeney, S.: Chromosomesynapsis defects and sexually di-morphic meiotic progression in micelacking Spo11. *Molec. Cell* 6: 989–998, 2000.
- [37314] 10866.Romanienko, P. J.; Camerini-Otero, R. D.: The mouse Spo11 geneis required for meiotic chromosome synapsis. *Molec. Cell* 6: 975–987,2000.
- [37315] 10867.Romanienko, P. J.; Camerini-Otero, R. D.: Cloning, characterization,and localization of mouse and human SPO11. *Genomics* 61: 156–169,1999.
- [37316] 10868.Shannon, M.; Richardson, L.; Christian, A.; Handel, M. A.; Thelen,M. P.: Differential gene expression of mam-malian SPO11/TOP6A homologsduring meiosis. *FEBS Lett.*

462: 329–334, 1999.

- [37317] 10869.Lopez–Coviella, I.; Berse, B.; Krauss, R.; Thies, R. S.; Blusztajn, J. K.: Induction and maintenance of the neuronal cholinergic phenotype in the central nervous system by BMP–9. *Science* 289: 313–316, 2000.
- [37318] 10870.Burn, T. C.; Connors, T. D.; Van Raay, T. J.; Dackowski, W. R.; Millholland, J. M.; Klinger, K. W.; Landes, G. M.: Generation of a transcriptional map for a 700–kb region surrounding the polycystic kidney disease type 1 (PKD1) and tuberous sclerosis type 2 (TSC2) disease genes on human chromosome 16p13.3. *Genome Res.* 6: 525–537, 1996.
- [37319] 10871.Pullen, N.; Dennis, P. B.; Andjelkovic, M.; Dufner, A.; Kozma, S. C.; Hemmings, B. A.; Thomas, G.: Phosphorylation and activation of p70(s6k) by PDK1. *Science* 279: 707–710, 1998.
- [37320] 10872.Stephens, L.; Anderson, K.; Stokoe, D.; Erdjument–Bromage, H.; Painter, G. F.; Holmes, A. B.; Gaffney, P. R. J.; Reese, C. B.; McCormick, F.; Tempst, P.; Coadwell, J.; Hawkins, P. T.: Protein kinase B kinases that mediate phosphatidylinositol 3,4,5–triphosphate–dependent activation of protein kinase B. *Science* 279: 710–714, 1998.
- [37321] 10873.Uebele, V. N.; Lagrutta, A.; Wade, T.; Figueroa, D.

J.; Liu, Y.; McKenna, E.; Austin, C. P.; Bennett, P. B.; Swanson, R.: Cloning and functional expression of two families of beta-subunits of the large conductance calcium-activated K(+) channel. *J. Biol. Chem.* 275:23211–23218, 2000.

- [37322] 10874. Kuroku, Y.; Soyama, A.; Fujita, E.; Urase, K.; Tsukahara, T.; Momoi, T.: RA70 is a src kinase-associated protein expressed ubiquitously. *Biochem. Biophys. Res. Comm.* 252: 738–742, 1998.
- [37323] 10875. Liu, J.; Kang, H.; Raab, M.; da Silva, A. J.; Kraeft, S.-K.; Rudd, C. E.: FYB (FYN binding protein) serves as a binding partner for lymphoid protein and FYN kinase substrate SKAP55 and a SKAP55-related protein in T cells. *Proc. Nat. Acad. Sci.* 95: 8779–8784, 1998.
- [37324] 10876. Marie-Cardine, A.; Verhagen, A. M.; Eckerskorn, C.; Schraven, B.: SKAP-HOM, a novel adaptor protein homologous to the FYN-associated protein SKAP55. *FEBS Lett.* 435: 55–60, 1998.
- [37325] 10877. Chai, J.; Du, C.; Wu, J.-W.; Kyin, S.; Wang, X.; Shi, Y.: Structural and biochemical basis of apoptotic activation by Smac/DIABLO. *Nature* 406:855–862, 2000.
- [37326] 10878. Du, C.; Fang, M.; Li, Y.; Li, L.; Wang, X.: Smac, a mitochondrial protein that promotes cytochrome c-

dependent caspase activation by eliminating IAP inhibition.
Cell 102: 33–42, 2000.

- [37327] 10879.Okada, H.; Suh, W.-K.; Jin, J.; Woo, M.; Du, C.; Elia, A.; Duncan, G. S.; Wakeham, A.; Itie, A.; Lowe, S. W.; Wang, X.; Mak, T. W.: Generation and characterization of Smac/ DIABLO-deficient mice. Molec. Cell. Biol. 22: 3509–3517, 2002.
- [37328] 10880.Scott, A. F.: Personal Communication. Baltimore, Md. 8/18/2000.
- [37329] 10881.Verhagen, A. M.; Ekert, P. G.; Pakusch, M.; Silke, J.; Connolly, L. M.; Reid, G. E.; Moritz, R. L.; Simpson, R. J.; Vaux, D. L.: Identification of DIABLO, a mammalian protein that promotes apoptosis by binding to and antagonizing IAP proteins. Cell 102: 43–53, 2000.
- [37330] 10882.Behrens, R.; Nolting, A.; Reimann, F.; Schwarz, M.; Waldschutz, R.; Pongs, O.: hKCNMB3 and hKCNMB4, cloning and characterization of two members of the large-conductance calcium-activated potassium channel beta subunit family. FEBS Lett. 474: 99–106, 2000.
- [37331] 10883.Brenner, R.; Jegla, T. J.; Wickenden, A.; Liu, Y.; Aldrich, R.W.: Cloning and functional characterization of novel large conductance calcium-activated potassium channel beta subunits, hKCNMB3 and hKCNMB4. J. Biol.

Chem. 275: 6453–6461, 2000.

- [37332] 10884.Riazi, M. A.; Brinkman–Mills, P.; Johnson, A.; Naylor, S. L.; Minoshima,S.; Shimizu, N.; Baldini, A.; McDermid, H. E.: Identification of a putative regulatory subunit of a calcium–activated potassium channel in the dup(3q) syndrome region and a related sequence on 22q11.2. *Genomics* 62:90–94, 1999.
- [37333] 10885.Hemmi, H.; Takeuchi, O.; Kawai, T.; Kaisho, T.; Sato, S.; Sanjo,H.; Matsumoto, M.; Hoshino, K.; Wagner, H.; Takeda, K.; Akira, S.: A toll–like receptor recognizes bacterial DNA. *Nature* 408: 740–745,2000.
- [37334] 10886.Leadbetter, E. A.; Rifkin, I. R.; Hohlbaum, A. M.; Beaudette, B.C.; Shlomchik, M. J.; Marshak–Rothstein, A.: Chromatin–IgG complexes activate B cells by dual engagement of IgM and Toll–like receptors. *Nature* 416:603–607, 2002.
- [37335] 10887.Takeshita, F.; Leifer, C. A.; Gursel, I.; Ishii, K. J.; Takeshita,S.; Gursel, M.; Klinman, D. M.: Cutting edge: role of Toll–like receptor9 in CpG DNA–induced activation of human cells. *J. Immun.* 167: 3555–3558,2001.
- [37336] 10888.Verthelyi, D.; Ishii, K. J.; Gursel, M.; Takeshita, F.; Klinman,D. K.: Human peripheral blood cells differentially recognize and respond to two distinct CpG motifs. *J. Im–*

mun. 166: 2372–2377, 2001.

- [37337] 10889.Bagrodia, S.; Taylor, S. J.; Jordon, K. A.; Van Aelst, L.; Cerione, R. A.: A novel regulator of p21-activated kinases. *J. Biol. Chem.* 273:23633–23636, 1998.
- [37338] 10890.Oh, W. K.; Yoo, J. C.; Jo, D.; Song, Y. H.; Kim, M. G.; Park, D.: Cloning of a SH3 domain-containing proline-rich protein, p85SPR, and its localization in focal adhesion. *Biochem. Biophys. Res. Commun.* 235:794–798, 1997.
- [37339] 10891.Ehrmann, D. A.; Schwarz, P. E. H.; Hara, M.; Tang, X.; Horikawa, Y.; Imperial, J.; Bell, G. I.; Cox, N. J.: Relationship of calpain-10 genotype to phenotypic features of polycystic ovary syndrome. *J. Clin. Endocr. Metab.* 87: 1669–1673, 2002.
- [37340] 10892.Elbein, S. C.; Chu, W.; Ren, Q.; Hemphill, C.; Schay, J.; Cox, N. J.; Hanis, C. L.; Hasstedt, S. J.: Role of calpain-10 gene variants in familial type 2 diabetes in Caucasians. *J. Clin. Endocr. Metab.* 87:650–654, 2002.
- [37341] 10893.Fullerton, S. M.; Bartoszewicz, A.; Ybazeta, G.; Horikawa, Y.; Bell, G. I.; Kidd, K. K.; Cox, N. J.; Hudson, R. R.; Di Rienzo, A.: Geographic and haplotype structure of candidate type 2 diabetes-susceptibility variants at the calpain-10 locus. *Am. J. Hum. Genet.* 70: 1096–1106, 2002.

- [37342] 10894.Tsai, H.-J.; Sun, G.; Weeks, D. E.; Kaushal, R.; Wolujewicz, M.;McGarvey, S. T.; Tufa, J.; Viali, S.; Deka, R.: Type 2 diabetes andthree calpain-10 gene polymorphisms in Samoans: no evidence of association. *Am.J. Hum. Genet.* 69: 1236-1244, 2001.
- [37343] 10895.Weiss, K. M.; Terwilliger, J. D.: How many diseases does it taketo map a gene with SNPs? *Nature Genet.* 26: 151-157, 2000.
- [37344] 10896.Hanis, C. L.; Boerwinkle, E.; Chakraborty, R.; Ellsworth, D. L.;Concannon, P.; Stirling, B.; Morrison, V. A.; Wapelhorst, B.; Spielman,R. S.; Gogolin-Ewens, K. J.; Shephard, J. M.; Williams, S. R.; and21 others: A genome-wide search for human non-insulin-dependent(type 2) diabetes genes reveals a major susceptibility locus on chromosome2. *Nature Genet.* 13: 161-166, 1996.
- [37345] 10897.Weber, A.; Wienker, T. F.; Jung, M.; Easton, D.; Dean, H. J.; Heinrichs,C.; Reis, A.; Clark, A. J. L.: Linkage of the gene for the tripleA syndrome to chromosome 12q13 near the type II keratin gene cluster. *Hum.Molec. Genet.* 5: 2061-2066, 1996.
- [37346] 10898.Bousquet, O.; Basseville, M.; Vila-Porcile, E.; Billette de Villemeur,T.; Hauw, J.-J.; Landrieu, P.; Portier, M.-M.: Aggregation of a subpopulationof vimentin filaments in

cultured human skin fibroblasts derived from patients with giant axonal neuropathy. *Cell. Motil. Cytoskeleton* 33:115–129, 1996.

- [37347] 10899. Kuhlénbaumer, G.; Young, P.; Oberwittler, C.; Hunermund, G.; Schirmacher, A.; Domschke, K.; Ringelstein, B.; Stogbauer, F.: Giant axonal neuropathy (GAN): case report and two novel mutations in the *gigaxonin* gene. *Neurology* 58:1273–1276, 2002. Note: Erratum: *Neurology* 58: 1444, 2002.
- [37348] 10900. Pena, S. D.: Giant axonal neuropathy: an inborn error of organization of intermediate filaments. *Muscle Nerve* 5: 166–172, 1982.
- [37349] 10901. Prineas, J. W.; Ouvrier, R. A.; Wright, R. G.; Walsh, J. C.; McLeod, J. G.: Giant axonal neuropathy: a generalized disorder of cytoplasmic microfilament formation. *J. Neuropath. Exp. Neurol.* 35: 458–470, 1976.
- [37350] 10902. Bowe, A. E.; Finnegan, R.; Jan de Beur, S. M.; Cho, J.; Levine, M. A.; Kumar, R.; Schiavi, S. C.: FGF-23 inhibits renal tubular phosphate transport and is a PHEX substrate. *Biochem. Biophys. Res. Commun.* 284:977–981, 2001.
- [37351] 10903. Shimada, T.; Mizutani, S.; Muto, T.; Yoneya, T.; Hino, R.; Takeda, S.; Takeuchi, Y.; Fujita, T.; Fukumoto, S.; Yamashita, T.: Cloning and characterization of FGF23 as a

causative factor of tumor-induced osteomalacia. *Proc. Nat. Acad. Sci.* 98: 6500–6505, 2001.

[37352] 10904. Strewler, G. J.: FGF23, hypophosphatemia, and rickets: has phosphorylation been found? (Commentary) *Proc. Nat. Acad. Sci.* 98: 5945–5946, 2001.

[37353] 10905. White, K. E.; Jonsson, K. B.; Carn, G.; Hampson, G.; Spector, T. D.; Mannstadt, M.; Lorenz-Depiereux, B.; Miyauchi, A.; Yang, I. M.; Ljunggren, O.; Meitinger, T.; Strom, T. M.; Juppner, H.; Econs, M. J.: The autosomal dominant hypophosphatemic rickets (ADHR) gene is a secreted polypeptide overexpressed by tumors that cause phosphate wasting. *J. Clin. Endocr. Metab.* 86: 497–500, 2001.

[37354] 10906. Yamashita, T.; Yoshioka, M.; Itoh, N.: Identification of a novel fibroblast growth factor, FGF-23, preferentially expressed in the ventrolateral thalamic nucleus of the brain. *Biochem. Biophys. Res. Commun.* 277: 494–498, 2000.

[37355] 10907. Barker, R. L.; Gleich, G. J.; Pease, L. R.: Acidic precursor revealed in human eosinophil granule major basic protein cDNA. *J. Exp. Med.* 168: 1493–1498, 1988.

[37356] 10908. Li, M.-S.; Sun, L.; Satoh, T.; Fisher, L. M.; Spry, C. J. F.: Human eosinophil major basic protein, a mediator of

allergic inflammation, is expressed by alternative splicing from two promoters. *Biochem.J.* 305: 921–927, 1995.

[37357] 10909. McGrogan, M.; Simonsen, C.; Scott, R.; Griffith, J.; Ellis, N.; Kennedy, J.; Campanelli, D.; Nathan, C.; Gabay, J.: Isolation of a complementary DNA clone encoding a precursor to human eosinophil major basic protein. *J. Exp. Med.* 168: 2295–2308, 1988.

[37358] 10910. Plager, D. A.; Weiler, D. A.; Loegering, D. A.; Johnson, W. B.; Haley, L.; Eddy, R. L.; Shows, T. B.; Gleich, G. J.: Comparative structure, proximal promoter elements, and chromosome location of the human eosinophil major basic protein genes. *Genomics* 71: 271–281, 2001.

[37359] 10911. Wasmoen, T. L.; Bell, M. P.; Loegering, D. A.; Gleich, G. J.; Prendergast, F. G.; McKean, D. J.: Biochemical and amino acid sequence analysis of human eosinophil granule major basic protein. *J. Biol. Chem.* 263: 12559–12563, 1988.

[37360] 10912. Weller, P. F.; Ackerman, S. J.; Smith, J. A.: Eosinophil granule proteins: major basic protein is distinct from the smaller subunit of eosinophil peroxidase. *J. Leukoc. Biol.* 43: 1–4, 1988.

[37361] 10913. Yoshimatsu, K.; Ohya, Y.; Shikata, Y.; Seto, T.; Hasegawa, Y.; Tanaka, I.; Kawamura, T.; Kitoh, K.;

Toyoshima, S.; Osawa, T.: Purification and cDNA cloning of a novel factor produced by a human T-cell hybridoma: sequence homology with animal lectins. *Molec. Immun.* 29: 537–546, 1992.

[37362] 10914. Frey, N.; Richardson, J. A.; Olson, E. N.: Calsarcins, a novel family of sarcomeric calcineurin-binding proteins. *Proc. Nat. Acad. Sci.* 97: 14632–14637, 2000.

[37363] 10915. Faulkner, G.; Pallavicini, A.; Comelli, A.; Salamon, M.; Bortoletto, G.; Ievolella, C.; Trevisan, S.; Kojic, S.; Dalla Vecchia, F.; Laveder, P.; Valle, G.; Lanfranchi, G.: FATZ, a filamin-, actinin-, and telethonin-binding protein of the Z-disc of skeletal muscle. *J. Biol. Chem.* 275: 41234–41242, 2000.

[37364] 10916. Takada, F.; Vander Woude, D. L.; Tong, H.-Q.; Thompson, T. G.; Watkins, S. C.; Kunkel, L. M.; Beggs, A. H.: Myozenin: an alpha-actinin- and gamma-filamin-binding protein of skeletal muscle Z lines. *Proc. Nat. Acad. Sci.* 98: 1595–1600, 2001.

[37365] 10917. Scott, A. F.: Personal Communication. Baltimore, Md. 3/13/2001.

[37366] 10918. Drane, P.; Barel, M.; Balbo, M.; Frade, R.: Identification of RB18A, a 205 kDa new p53 regulatory protein which shares antigenic and functional properties with p53.

Oncogene 15: 3013–3024, 1997.

- [37367] 10919.Mansharamani, M.; Hewetson, A.; Chilton, B. S.: Cloning and characterization of an atypical type IV P-type ATPase that binds to the RING motif of RUSH transcription factors. J. Biol. Chem. 276: 3641–3649, 2001.
- [37368] 10920.Nakayama, Y.; Weissman, S. M.; Bothwell, A. L. M.: BXMAS1 identifies a cluster of homologous genes differentially expressed in B cells. Biochem. Biophys. Res. Commun. 285: 830–837, 2001.
- [37369] 10921.Desai, R.; Peretz, A.; Idelson, H.; Lazarovici, P.; Attali, B.: Ca(2+)-activated K(+) channels in human leukemic Jurkat T cells: molecular cloning, biochemical and functional characterization. J. Biol. Chem. 275: 39954–39963, 2000.
- [37370] 10922.Maguchi, M.; Nishida, W.; Kohara, K.; Kuwano, A.; Kondo, I.; Hiwada, K.: Molecular cloning and gene mapping of human basic and acidic calponins. Biochem. Biophys. Res. Commun. 217: 238–244, 1995.
- [37371] 10923.Miano, J. M.; Krahe, R.; Garcia, E.; Elliott, J. M.; Olson, E.N.: Expression, genomic structure and high resolution mapping to 19p13.2 of the human smooth muscle cell calponin gene. Gene 197:215–224, 1997.
- [37372] 10924.Miano, J. M.; Thomas, S.; Disteché, C. M.: Expres-

sion and chromosomal mapping of the mouse smooth muscle calponin gene. *Mammalian Genome* 12:187–191, 2001.

- [37373] 10925. Strasser, P.; Gimona, M.; Moessler, H.; Herzog, M.; Small, J. V.: Mammalian calponin: identification and expression of genetic variants. *FEBS Lett.* 330: 13–18, 1993.
- [37374] 10926. Duetsch, G.; Illig, T.; Loesgen, S.; Rohde, K.; Kloop, N.; Herbon, N.; Cohlke, H.; Altmueller, J.; Wjst, M.: STAT6 as an asthma candidate gene: polymorphism–screening, association and haplotype analysis in a Caucasian sib–pair study. *Hum. Molec. Genet.* 11: 613–621, 2002.
- [37375] 10927. Wu, H.–K.; Heng, H. H. Q.; Shi, X.–M.; Forsdyke, D. R.; Tsui, L.–C.; Mak, T. W.; Minden, M. D.; Siderovski, D. P.: Differential expression of a basic helix–loop–helix phosphoprotein gene, G0S8, in acute leukemia and localization to human chromosome 1q31. *Leukemia* 9: 1291–1298, 1995.
- [37376] 10928. Bogerd, H. P.; Fridell, R. A.; Madore, S.; Cullen, B. R.: Identification of a novel cellular cofactor for the Rev/Rex class of retroviral regulatory proteins. *Cell* 82: 485–494, 1995.
- [37377] 10929. Fritz, C. C.; Zapp, M. L.; Green, M. R.: A human nucleoporin–like protein that specifically interacts with HIV

Rev. Nature 376: 530–533,1995.

- [37378] 10930.Jones, T.; Sheer, D.; Bevec, D.; Kappel, B.; Hauber, J.; Steinkasserer,A.: The human HIV–1 Rev binding–protein hRIP/Rab (HRB) maps to chromosome2q36. Genomics 40: 198–199, 1997.
- [37379] 10931.Kang–Decker, N.; Mantchev, G. T.; Juneja, S. C.; McNiven, M. A.;van Deursen, J. M. A.: Lack of acrosome formation in Hrb–deficientmice. Science 294: 1531–1533, 2001.
- [37380] 10932.Salcini, A. E.; Confalonieri, S.; Doria, M.; Santolini, E.; Tassi,E.; Minenkova, O.; Cesareni, G.; Pelicci, P. G.; Di Fiore, P. P.:Binding specificity and in vivo targets of the EH domain, a novelprotein–protein interaction module. Genes Dev. 11: 2239–2249, 1997.
- [37381] 10933.Fish, K. J.; Cegielska, A.; Getman, M. E.; Landes, G. M.; Virshup,D. M.: Isolation and characterization of human casein kinase I–epsilon(CKI), a novel member of the CKI gene family. J. Biol. Chem. 270:14875–14883, 1995.
- [37382] 10934.Kloss, B.; Price, J. L.; Saez, L.; Blau, J.; Rothenfluh, A.; Wesley,C. S.; Young, M. W.: The Drosophila clock gene double–time encodesa protein closely–related to human casein kinase I–epsilon. Cell 94:97–107, 1998.
- [37383] 10935.Lowrey, P. L.; Shimomura, K.; Antoch, M. P.; Ya–

mazaki, S.; Zamenides, P. D.; Ralph, M. R.; Menaker, M.; Takahashi, J. S.: Positional syntenic cloning and functional characterization of the mammalian circadian mutation tau. Science 288: 483–491, 2000.

[37384] 10936. Graves, P. R.; Haas, D. W.; Hagedorn, C. H.; De-Paoli-Roach, A.A.; Roach, P. J.: Molecular cloning, expression, and characterization of a 49-kilodalton casein kinase I isoform from rat testis. J. Biol. Chem. 268: 6394–6401, 1993.

[37385] 10937. Kusuda, J.; Hidari, N.; Hirai, M.; Hashimoto, K.: Sequence analysis of the cDNA for the human casein kinase 1-delta (CSNK1D) gene and its chromosomal localization. Genomics 32: 140–143, 1996.

[37386] 10938. Kools, P. F. J.; Roebroek, A. J. M.; van de Velde, H. J. K.; Marynen, P.; Bullerdiek, J.; Van de Ven, W. J. M.: Regional mapping of the human NSP gene to chromosome region 14q21–q22 by fluorescence in situ hybridization analysis. Cytogenet. Cell Genet. 66: 48–50, 1994.

[37387] 10939. Roebroek, A. J. M.; Ayoubi, T. A. Y.; van de Velde, H. J. K.; Schoenmakers, E. F. P. M.; Pauli, I. G. L.; Van de Ven, W. J. M.: Genomic organization of the human NSP gene, prototype of a novel gene family encoding reticulons. Genomics 32: 191–199, 1996.

- [37388] 10940. Roebroek, A. J. M.; van de Velde, H. J. K.; Van Bokhoven, A.; Broers, J. L. V.; Ramaekers, F. C. S.; Van de Ven, W. J. M.: Cloning and expression of alternative transcripts of a novel neuroendocrine-specific gene and identification of its 135-kDa translational product. *J. Biol. Chem.* 268: 13439–13447, 1993.
- [37389] 10941. Senden, N. H. M.; van de Velde, H. J. K.; Broers, J. L. V.; Timmer, E. D. J.; Roebroek, A. J. M.; van de Ven, W. J. M.; Ramaekers, F. C. S.: Cluster-10 lung-cancer antibodies recognize NSPs, novel neuro-endocrine proteins associated with membranes of the endoplasmic reticulum. *Int. J. Cancer (Suppl. 8)*: 84–88, 1994.
- [37390] 10942. van de Velde, H. J. K.; Roebroek, A. J. M.; van Leeuwen, F. W.; Van de Ven, W. J. M.: Molecular analysis of expression in rat brain of NSP-A, a novel neuroendocrine-specific protein of the endoplasmic reticulum. *Molec. Brain Res.* 23: 81–92, 1994.
- [37391] 10943. van de Velde, H. J. K.; Senden, N. H. M.; Roskams, T. A. D.; Broers, J. L. V.; Ramaekers, F. C. S.; Roebroek, A. J. M.; Van de Ven, W. J. M.: NSP-encoded reticulons are neuroendocrine markers of a novel category in human lung cancer diagnosis. *Cancer Res.* 54: 4769–4776, 1994.
- [37392] 10944. Baron, B. W.; Anastasi, J.; Thirman, M. J.; Furukawa,

Y.; Fears,S.; Kim, D. C.; Simone, F.; Birkenbach, M.; Montag, A.; Sadhu, A.;Zeleznik–Le, N.; McKeithan, T. W.: The human programmed cell death–2(PDCD2) gene is a target of BCL6 repression: implications for a roleof BCL6 in the down–regulation of apoptosis. *Proc. Nat. Acad. Sci.* 99:2860–2865, 2002.

[37393] 10945.Cohen, J. J.; Duke, R. C.: Glucocorticoid activation of a calcium–dependentendonuclease in thymocyte nuclei leads to cell death. *J. Immun.* 132:38–42, 1984.

[37394] 10946.Kawakami, T.; Furukawa, Y.; Sudo, K.; Saito, H.; Takami, S.; Takahashi,E.; Nakamura, Y.: Isolation and mapping of a human gene (PDCD2) thatis highly homologous to Rp8, a rat gene associated with programmedcell death. *Cytogenet. Cell Genet.* 71: 41–43, 1995.

[37395] 10947.Owens, G. P.; Cohen, J. J.: Identification of genes involved inprogrammed cell death. *Cancer Metastasis Rev.* 11: 149–156, 1992.

[37396] 10948.Sellins, K. S.; Cohen, J. J.: Gene induction by gamma–irradiationleads to DNA fragmentation in lymphocytes. *J. Immun.* 139: 3199–3206,1987.

[37397] 10949.Wyllie, A. H.: Glucocorticoid–induced thymocyte apoptosis is associatedwith endogenous endonuclease activation. *Nature* 284: 555–556, 1980.

- [37398] 10950. Bullrich, F.; Druck, T.; Kunapuli, P.; Gomez, J.; Gripp, K. W.; Schlegelberger, B.; Lasota, J.; Aronson, M.; Cannizzaro, L. A.; Huebner, K.; Benovic, J. L.: Chromosomal mapping of the genes GPRK5 and GPRK6 encoding G protein-coupled receptor kinases GRK5 and GRK6. *Cytogenet. Cell Genet.* 70: 250–254, 1995.
- [37399] 10951. Haribabu, B.; Snyderman, R.: Identification of additional members of human G-protein-coupled receptor kinase multigene family. *Proc. Nat. Acad. Sci.* 90: 9398–9402, 1993.
- [37400] 10952. Miki, T.; Suzuki, M.; Shibasaki, T.; Uemura, H.; Sato, T.; Yamaguchi, K.; Koseki, H.; Iwanaga, T.; Nakaya, H.; Seino, S.: Mouse model of Prinzmetal angina by disruption of the inward rectifier Kir6.1. *Nature Med.* 8: 466–472, 2002.
- [37401] 10953. Prinzmetal, M.; Kenamer, R.; Merliss, R.; Wada, T.; Bor, N.: Angina pectoris. 1. A variant form of angina pectoris: preliminary report. *Am. J. Med.* 27: 375–388, 1959.
- [37402] 10954. Pristipino, C.; Beltrame, J. F.; Finocchiaro, M. L.; Hattori, R.; Fujita, M.; Mongiardo, R.; Cianflone, D.; Sanna, T.; Sasayama, S.; Maseri, A.: Major racial differences in coronary constrictor response between Japanese and Caucasians with recent myocardial infarction. *Circulation*

101:1102–1108, 2000.

- [37403] 10955. Cherel, M.; Sorel, M.; Apiou, F.; Lebeau, B.; Dubois, S.; Jacques, Y.; Minvielle, S.: The human interleukin–11 receptor alpha gene (IL11RA): genomic organization and chromosome mapping. *Genomics* 32: 49–53, 1996.
- [37404] 10956. Cherel, M.; Sorel, M.; Lebeau, B.; Dubois, S.; Moreau, J.–F.; Bataille, R.; Minvielle, S.; Jacques, Y.: Molecular cloning to two isoforms of a receptor for the human hematopoietic cytokine interleukin–11. *Blood* 86: 2534–2540, 1995.
- [37405] 10957. Neuhaus, H.; Bettenhausen, B.; Bilinski, P.; Simon–Chazottes, D.; Guenet, J.–L.; Gossler, A.: Etl2, a novel putative type–I cytokine receptor expressed during mouse embryogenesis at high levels in skin and cells with skeletogenic potential. *Dev. Biol.* 166: 531–542, 1994.
- [37406] 10958. Van Leuven, F.; Stas, L.; Hilliker, C.; Miyake, Y.; Bilinski, P.; Gossler, A.: Molecular cloning and characterization of the human interleukin–11 receptor alpha–chain gene, IL11RA, located on chromosome 9p13. *Genomics* 31: 65–70, 1996.
- [37407] 10959. Magrangeas, F.; Pitiot, G.; Dubois, S.; Bragado–Nils–son, E.; Cherel, M.; Jobert, S.; Lebeau, B.; Boisteau, O.; Lethe, B.; Mallet, J.; Jacques, Y.; Minvielle, S.: Cotranscrip–

tion and intergenic splicing of humangalactose-1-phosphate uridylyltransferase and interleukin-11 receptoralpha-chain genes generate a fusion mRNA in normal cells: implicationfor the production of multidomain proteins during evolution. J. Biol.Chem. 273: 16005-16010, 1998.

- [37408] 10960.Avraham, K. B.; Hasson, T.; Sobe, T.; Balsara, B.; Testa, J. R.;Skvorak, A. B.; Morton, C. C.; Copeland, N. G.; Jenkins, N. A.: Characterizationof unconventional MYO6, the human homologue of the gene responsiblefor deafness in Snell's waltzer mice. Hum. Molec. Genet. 6: 1225-1231,1997.
- [37409] 10961.Avraham, K. B.; Hasson, T.; Steel, K. P.; Kingsley, D. M.; Russell,L. B.; Mooseker, M. S.; Copeland, N. G.; Jenkins, N. A.: The mouseSnell's waltzer deafness gene encodes an unconventional myosin requiredfor structural integrity of inner ear hair cells. Nature Genet. 11:369-375, 1995.
- [37410] 10962.Hasson, T.; Mooseker, M. S.: Porcine myosin-VI: characterizationof a new mammalian unconventional myosin. J. Cell. Biol. 127: 425-440,1994.
- [37411] 10963.Hasson, T.; Skowron, J. F.; Gilbert, D. J.; Avraham, K. B.; Perry,W. L.; Bement, W. M.; Anderson, B. L.; Sherr, E.

H.; Chen, Z.-Y.; Greene, L. A.; Ward, D. C.; Corey, D. P.; Mooseker, M. S.; Copeland, N. G.; Jenkins, N. A.: Mapping of unconventional myosins in mouse and human. *Genomics* 36: 431–439, 1996.

[37412] 10964. Rock, R. S.; Rice, S. E.; Wells, A. L.; Purcell, T. J.; Spudich, J. A.; Sweeney, H. L.: Myosin VI is a processive motor with a large step size. *Proc. Nat. Acad. Sci.* 98: 13655–13659, 2001.

[37413] 10965. Wells, A. L.; Lin, A. W.; Chen, L.-Q.; Safer, D.; Cain, S. M.; Hasson, T.; Carragher, B. O.; Milligan, R. A.; Sweeney, H. L.: Myosin VI is an actin-based motor that moves backwards. *Nature* 401: 505–508, 1999.

[37414] 10966. Melchionda, S.; Ahituv, N.; Bisceglia, L.; Sobe, T.; Glaser, F.; Rabionet, R.; Arbones, M. L.; Notarangelo, A.; Di Iorio, E.; Carella, M.; Zelante, L.; Estivill, X.; Avraham, K. B.; Gasparini, P.: MYO6, the human homologue of the gene responsible for deafness in Snell's Waltzer mice, is mutated in autosomal dominant nonsyndromic hearing loss. *Am. J. Hum. Genet.* 69: 635–640, 2001.

[37415] 10967. Chittum, H. S.; Himeno, S.; Hill, K. E.; Burk, R. F.: Multiple forms of selenoprotein P in rat plasma. *Arch. Biochem. Biophys.* 325: 124–128, 1996.

[37416] 10968. Bonne, S.; van Hengel, J.; van Roy, F.: Chromosomal

mapping of human armadillo genes belonging to the p120(ctn)/plakophilin subfamily. *Genomics* 51:452–454, 1998.

[37417] 10969. Drab, M.; Verkade, P.; Elger, M.; Kasper, M.; Lohn, M.; Lauterbach, B.; Menne, J.; Lindschau, C.; Mende, F.; Luft, F. C.; Schedl, A.; Haller, H.; Kurzchalia, T. V.: Loss of caveolae, vascular dysfunction, and pulmonary defects in caveolin-1 gene-disrupted mice. *Science* 293:2449–2452, 2001.

[37418] 10970. Engelman, J. A.; Zhang, X.; Galbiati, F.; Volonte, D.; Sotgia, F.; Pestell, R. G.; Minetti, C.; Scherer, P. E.; Okamoto, T.; Lisanti, M. P.: Molecular genetics of the caveolin gene family: implications for human cancers, diabetes, Alzheimer disease, and muscular dystrophy. *Am. J. Hum. Genet.* 63: 1578–1587, 1998.

[37419] 10971. Engelman, J. A.; Zhang, X. L.; Galbiati, F.; Lisanti, M. P.: Chromosomal localization, genomic organization, and developmental expression of the murine caveolin gene family (Cav-1, -2, and -3): Cav-1 and Cav-2 genes map to a known tumor suppressor locus (6-A2/7q31). *FEBS Lett.* 429:330–336, 1998.

[37420] 10972. Engelman, J. A.; Zhang, X. L.; Lisanti, M. P.: Genes encoding human caveolin-1 and -2 are co-localized to the

D7S522 locus (7q31.1), a known fragile site (FRA7G) that is frequently deleted in human cancers. FEBS Lett. 436: 403–410, 1998.

- [37421] 10973. Feron, O.; Dessy, C.; Moniotte, S.; Desager, J.-P.; Balligand, J.-L.: Hypercholesterolemia decreases nitric oxide production by promoting the interaction of caveolin and endothelial nitric oxide synthase. J. Clin. Invest. 103: 897–905, 1999.
- [37422] 10974. Giordano, S.; Ponzetto, C.; Di Renzo, M. F.; Cooper, C. S.; Comoglio, P. M.: Tyrosine kinase receptor indistinguishable from the c-met protein. Nature 339: 155–156, 1989.
- [37423] 10975. Glenney, J. R., Jr.: The sequence of human caveolin reveals identity with VIP21, a component of transport vesicles. FEBS Lett. 314: 45–48, 1992.
- [37424] 10976. Kurzchalia, T. V.; Dupree, P.; Parton, R. G.; Kellner, R.; Virta, H.; Lehnert, M.; Simons, K.: VIP21, a 21-kD membrane protein is an integral component of trans-Golgi-network-derived transport vesicles. J. Cell Biol. 118: 1003–1014, 1992.
- [37425] 10977. Scherer, P. E.; Okamoto, T.; Chun, M.; Nishimoto, I.; Lodish, H. F.; Lisanti, M. P.: Identification, sequence, and expression of caveolin-2 defines a caveolin gene family.

Proc. Nat. Acad. Sci. 93:131–135, 1996.

- [37426] 10978.Scherer, P. E.; Tang, Z.; Chun, M.; Sargiacomo, M.; Lodish, H.F.; Lisanti, M. P.: Caveolin isoforms differ in their N-terminalprotein sequence and subcellular distribution: identification andepitope mapping of an isoform-specific monoclonal antibody probe. J.Biol. Chem. 270 16395–16401, 1995.
- [37427] 10979.Razani, B.; Engelman, J. A.; Wang, X. B.; Schubert, W.; Zhang,X. L.; Marks, C. B.; Macaluso, F.; Russell, R. G.; Li, M.; Pestell,R. G.; Di Vizio, D.; Hou, H., Jr.; Kneitz, B.; Lagaud, G.; Christ,G. J.; Edelman, W.; Lisanti, M. P.: Caveolin-1 null mice are viablebut show evidence of hyperproliferative and vascular abnormalities. J.Biol. Chem. 276: 38121–38138, 2001.
- [37428] 10980.Bell, D. R.; Plant, N. J.; Rider, C. G.; Na, L.; Brown, S.; Ateitalla,I.; Acharya, S. K.; Davies, M. H.; Elias, E.; Jenkins, N. A.; Gilbert,D. J.; Copeland, N. G.; Elcombe, C. R.: Species-specific inductionof cytochrome P-450 4A RNAs: PCR cloning of partial guinea-pig, humanand mouse CYP4A cDNAs. Biochem. J. 294: 173–180, 1993.
- [37429] 10981.Imaoka, S.; Ogawa, H.; Kimura, S.; Gonzalez, F. J.: Complete cDNAsequence and cDNA-directed expression of CYP4A11, a fatty acid omega-hydroxylaseexpressed in

human kidney. DNA Cell Biol. 12: 893–899, 1993.

[37430] 10982.Kawashima, H.; Kusunose, E.; Kikuta, Y.; Kinoshita, H.; Tanaka,S.; Yamamoto, S.; Kishimoto, T.; Kusunose, M.: Purification and cDNAcloning of human liver CYP4A fatty acid omega–hydroxylase. J. Biochem. 116:74–80, 1994.

[37431] 10983.Palmer, C. N. A.; Richardson, T. H.; Griffin, K. J.; Hsu, M.–H.;Muerhoff, A. S.; Clark, J. E.; Johnson, E. F.: Characterization of a cDNA encoding a human kidney, cytochrome P–450 4A fatty acid omega–hydroxylase and the cognate enzyme expressed in Escherichia coli. Biochim. Biophys.Acta 1172: 161–166, 1993.

[37432] 10984.Muller, J.; Ory, S.; Copeland, T.; Piwnica–Worms, H.; Morrison,D. K.: C–TAK1 regulates Ras signaling by phosphorylating the MAPKscaffold, KSR1. Molec. Cell 8: 983–993, 2001.

[37433] 10985.Rosenthal, R. L.: Haemorrhage in PTA (factor XI) deficiency.(Abstract) Proc. 10th Int. Cong. Soc. Hemat., Stockholm , 1964.

[37434] 10986.Laporte, J.; Hu, L. J.; Kretz, C.; Mandel, J.–L.; Kioschis, P.;Coy, J. F.; Klauck, S. M.; Poustka, A.; Dahl, N.: A gene mutated in X–linked myotubular myopathy defines a new putative tyrosine phosphatase family conserved in yeast. Nature Genet. 13: 175–182, 1996.

- [37435] 10987.Rosenthal, R. L.; Dreskin, O. H.; Rosenthal, N.: Plasma thromboplastinantecedent (PTA) deficiency: clinical, coagulation, therapeutic and hereditary aspects of a new hemophilia-like disease. Blood 10: 120–131, 1955.
- [37436] 10988.Seligsohn, U.: Factor XI (PTA) deficiency. In: Goodman, R. E.; Motulsky, A. G.: Genetic Diseases Among Ashkenazi Jews. New York: Raven Press (pub.) 1979. Pp. 141–148.
- [37437] 10989.Vinazzer, H.: Partieller familiaerer Faktor–XI–Mangel. Blut 15:263–267, 1967.
- [37438] 10990.Wistinghausen, B.; Reischer, A.; Oddoux, C.; Ostrer, H.; Nardi, M.; Karpatkin, M.: Severe factor XI deficiency in an Arab family associated with a novel mutation in exon 11. Brit. J. Haemat. 99:575–577, 1997.
- [37439] 10991.Zivelin, A.; Bauduer, F.; Ducout, L.; Peretz, H.; Rosenberg, N.; Yativ, R.; Seligsohn, U.: Factor XI deficiency in French Basques caused predominantly by an ancestral cys38-to-arg mutation in the factor XI gene. Blood 99: 2448–2454, 2002.
- [37440] 10992.Hsueh, Y.-P.; Wang, T.-F.; Yang, F.-C.; Sheng, M.: Nuclear transcription and transcription regulation by the membrane-associated guanylate kinase CASK/LIN-2. Nature 404: 298–302, 2000.

- [37441] 10993.De Smet, C.; Lurquin, C.; van der Bruggen, P.; De Plaen, E.; Brasseur,F.; Boon, T.: Sequence and expression pattern of the human MAGE2gene. Immunogenetics 39: 121–129, 1994.
- [37442] 10994.Chambost, H.; Van Baren, N.; Brasseur, F.; Godelaine, D.; Xerri,L.; Landi, S. J.; Theate, I.; Plumas, J.; Spagnoli, G. C.; Michel,G.; Coulie, P. G.; Olive, D.: Expression of gene MAGE–A4 in Reed–Sternbergcells. Blood 95: 3530–3533, 2000.
- [37443] 10995.Nagase, T.; Ishikawa, K.; Suyama, M.; Kikuno, R.; Miyajima, N.;Tanaka, A.; Kotani, H.; Nomura, N.; Ohara, O.: Prediction of thecoding sequences of unidentified human genes. XI. The complete sequencesof 100 new cDNA clones from brain which code for large proteins invitro. DNA Res. 5: 277–286, 1998.
- [37444] 10996.Gospe, S. M., Jr.; Lazaro, R. P.; Lava, N. S.; Grootscholten,P. M.; Scott, M. O.; Fischbeck, K. H.: Familial X–linked myalgiaand cramps: a nonprogressive myopathy associated with a deletion inthe dystrophin gene. Neurology 39: 1277–1280, 1989.
- [37445] 10997.Kingston, H. M.; Sarfarazi, M.; Thomas, N. S. T.; Harper, P. S.: Localisation of the Becker muscular dystrophy gene on the shortarm of the X chromosome by link–

age to cloned DNA sequences. Hum.Genet. 67: 6–17, 1984.

[37446] 10998.Kingston, H. M.; Thomas, N. S. T.; Pearson, P. L.; Sarfarazi,M.; Harper, P. S.: Genetic linkage between Becker muscular dystrophyand a polymorphic DNA sequence on the short arm of the X chromosome. J.Med. Genet. 20: 255–258, 1983.

[37447] 10999.Reardon, W.; Middleton–Price, H. R.; Sandkuijl, L.; Phelps, P.;Bellman, S.; Luxon, L.; Pembrey, M. E.; Malcolm, S.: A multipedigreelinkage study of X–linked deafness: linkage to Xq13–q21 and evidencefor genetic heterogeneity. Genomics 11: 885–894, 1991.

[37448] 11000.Cardoso, C.; Timsit, S.; Villard, L.; Khrestchatisky, M.; Fontes,M.; Colleaux, L.: Specific interaction between the XNP/ATR–X geneproduct and the SET domain of the human EZH2 protein. Hum. Molec.Genet. 7: 679–684, 1998.

[37449] 11001.Bairoch, A.: Personal Communication. Geneva, Switzerland 5/13/1994.

[37450] 11002.Kamal, A.; Stokin, G. B.; Yang, Z.; Xia, C.–H.; Goldstein, L. S.: Axonal transport of amyloid precursor protein is mediated by directbinding to the kinesin light chain subunit of kinesin–I. Neuron 28:449–459, 2000.

- [37451] 11003.Luo, Y.; Bolon, B.; Kahn, S.; Bennett, B. D.; Babu-Khan, S.; Denis,P.; Fan, W.; Kha, H.; Zhang, J.; Gong, Y.; Martin, L.; Louis, J.-C.;Yan, Q.; Richards, W. G.; Citron, M.; Vassar, R.: Mice deficientin BACE1, the Alzheimer's beta-secretase, have normal phenotype andabolished beta-amyloid generation. *Nature Neurosci.* 4: 231–232,2001.
- [37452] 11004.Schiemann, B.; Gommerman, J. L.; Vora, K.; Cachero, T. G.; Shulga-Morskaya,S.; Dobles, M.; Frew, E.; Scott, M. L.: An essential role for BAFFin the normal development of B cells through a BCMA-independent pathway. *Science* 2111–2114,2001.
- [37453] 11005.Okamoto, N.; Hori, S.; Akazawa, C.; Hayashi, Y.; Shigemoto, R.;Mizuno, N.; Nakanishi, S.: Molecular characterization of a new metabotropicglutamate receptor mGluR7 coupled to inhibitory cyclic AMP signaltransduction. *J. Biol. Chem.* 269: 1231–1236, 1994.
- [37454] 11006.Ango, F.; Prezeau, L.; Muller, T.; Tu, J. C.; Xiao, B.; Worley,P. F.; Pin, J. P.; Bockaert, J.; Fagni, L.: Agonist-independent activationof metabotropic glutamate receptors by the intracellular protein Homer. *Nature* 411:962–965, 2001.
- [37455] 11007.Offenberg, H. H.; Schalk, J. A. C.; Meuwissen, R. L. J.; van Aalderen,M.; Kester, H. A.; Dietrich, A. J. J.; Heyting,

C.: SCP2: a major protein component of the axial elements of synaptonemal complexes of the rat. *Nucleic Acids Res.* 26: 2572–2579, 1998.

- [37456] 11008. Schalk, J. A. C.; Offenberg, H. H.; Peters, E.; Groot, N. P. B.; Hoovers, J. M. N.; Heyting, C.: Isolation and characterization of the human SCP2 cDNA and chromosomal localization of the gene. *Mammalian Genome* 10: 642–644, 1999.
- [37457] 11009. Komada, M.; McLean, D. J.; Griswold, M. D.; Russell, L. D.; Soriano, P.: E-MAP-115, encoding a microtubule-associated protein, is a retinoic acid-inducible gene required for spermatogenesis. *Genes Dev.* 14: 1332–1342, 2000.
- [37458] 11010. Masson, D.; Kreis, T. E.: Identification and molecular characterization of E-MAP-115, a novel microtubule-associated protein predominantly expressed in epithelial cells. *J. Cell Biol.* 123: 357–371, 1993.
- [37459] 11011. Kowalski, P. E.; Freeman, J. D.; Nelson, D. T.; Mager, D. L.: Genomic structure and evolution of a novel gene (PLA2L) with duplicated phospholipase A2-like domains. *Genomics* 39: 38–46, 1997.
- [37460] 11012. Liu, M.; Parker, R. M. C.; Darby, K.; Eyre, H. J.; Copeland, N. G.; Crawford, J.; Gilbert, D. J.; Sutherland, G.

R.; Jenkins, N. A.; Herzog, H.: GPR56, a novel secretin-like human G-protein-coupled receptor gene. *Genomics* 55: 296–305, 1999.

[37461] 11013. Zendman, A. J. W.; Cornelissen, I. M. H. A.; Weidle, U. H.; Ruiter, D. J.; van Muijen, G. N. P.: TM7XN1, a novel human EGF-TM7-like cDNA, detected with mRNA differential display using human melanoma cell lines with different metastatic potential. *FEBS Lett.* 446: 292–298, 1999.

[37462] 11014. Nicolaou, M.; Song, Y.-Q.; Sato, C. A.; Orlacchio, A.; Kawarai, T.; Medeiros, H.; Liang, Y.; Sorbi, S.; Richard, E.; Rogaev, E. I.; Moliaka, Y.; Bruni, A. C.; Jorge, R.; Percy, M.; Duara, R.; Farrer, L. A.; St George-Hyslop, P.; Rogaeva, E. A.: Mutations in the open reading frame of the beta-site APP cleaving enzyme (BACE) locus are not a common cause of Alzheimer's disease. *Neurogenetics* 3: 203–206, 2001.

[37463] 11015. Saunders, A. J.; Kim, T.-W.; Tanzi, R. E.: BACE maps to chromosome 11 and a BACE homolog, BACE2, reside in the obligate Down syndrome region of chromosome 21. *Science* 286: 1255a, 1999. Note: Electronic Publication.

[37464] 11016. Sinha, S.; Anderson, J. P.; Barbour, R.; Basi, G. S.; Caccavello, R.; Davis, D.; Doan, M.; Dovey, H. F.; Frigon, N.; Hong, J.; Jacobson-Croak, K.; Jewett, N.; and 15 others: Purification and cloning of amyloid precursor protein beta-

secretase from human brain. *Nature* 402: 537–540, 1999.

[37465] 11017. Vassar, R.; Bennett, B. D.; Babu-Khan, S.; Kahn, S.; Mendiaz, E. A.; Dents, P.; Taplow, D. B.; Ross, S.; Amaranta, P.; Loeloff, R.; Luo, Y.; Fisher, S.; and 12 others: Beta-secretase cleavage of Alzheimer's amyloid precursor protein by the transmembrane aspartic protease BACE. *Science* 286: 735–741, 1999.

[37466] 11018. Yan, R.; Bienkowski, M. J.; Shuck, M. E.; Miao, H.; Tory, M. C.; Pauley, A. M.; Brashler, J. R.; Stratman, N. C.; Mathews, W. R.; Buhl, A. E.; Carter, D. B.; Tomasselli, A. G.; Parodi, L. A.; Heinrikson, R. L.; Gurney, M. E.: Membrane-anchored aspartyl protease with Alzheimer's disease beta-secretase activity. *Nature* 402: 533–536, 1999.

[37467] 11019. Jagla, K.; Dolle, P.; Mattei, M.-G.; Jagla, T.; Schuhbauer, B.; Dretzen, G.; Bellard, F.; Bellard, M.: Mouse Lbx1 and human LBX1 define a novel mammalian homeobox gene family related to the Drosophila ladybird genes. *Mech. Dev.* 53: 345–356, 1995.

[37468] 11020. Mennerich, D.; Schafer, K.; Braun, T.: Pax-3 is necessary but not sufficient for lbx1 expression in myogenic precursor cells of the limb. *Mech. Dev.* 73: 147–158, 1998.

[37469] 11021. Schafer, K.; Braun, T.: Early specification of limb muscle precursor cells by the homeobox gene Lbx1h. *Na-*

ture Genet. 23: 213–216, 1999.

- [37470] 11022. Wilson, P. J.; McGlinn, E.; Marsh, A.; Evans, T.; Arnold, J.; Wright, K.; Biden, K.; Young, J.; Wainwright, B.; Wicking, C.; Chenevix-Trench, G.: Sequence variants of DLC1 in colorectal and ovarian tumours. Hum. Mutat. 15: 156–165, 2000.
- [37471] 11023. Yuan, B.-Z.; Miller, M. J.; Keck, C. L.; Zimonjic, D. B.; Thorgeirsson, S. S.; Popescu, N. C.: Cloning, characterization, and chromosomal localization of a gene frequently deleted in human liver cancer (DLC-1) homologous to rat RhoGAP. Cancer Res. 58: 2196–2199, 1998.
- [37472] 11024. Yuan, B.-Z.; Yang, Y.; Keck-Waggoner, C. L.; Zimonjic, D. B.; Thorgeirsson, S. S.; Popescu, N. C.: Assignment and cloning of mouse Arhgap7 to chromosome 8A4–B2, a conserved syntenic region of human chromosome 8p22–p21. Cytogenet. Cell Genet. 87: 189–190, 1999.
- [37473] 11025. Arnould, C.; Philippe, C.; Bourdon, V.; Gregoire, M. J.; Berger, R.; Jonveaux, P.: The signal transducer and activator of transcription STAT5b gene is a new partner of retinoic acid receptor alpha in acute promyelocytic-like leukaemia. Hum. Molec. Genet. 8: 1741–1749, 1999.
- [37474] 11026. Lin, J.-X.; Mietz, J.; Modi, W. S.; John, S.; Leonard,

W. J.:Cloning of human Stat5B: reconstitution of interleukin-2-induced Stat5A and Stat5B DNA binding activity in COS-7 cells. *J. Biol. Chem.* 271:10738–10744, 1996.

- [37475] 11027. Shinoura, N.; Shamraj, O. I.; Hugenholtz, H.; Zhu, J. G.; McBlack, P.; Warnick, R.; Tew, J. J.; Wani, M. A.; Menon, A. G.: Identification and partial sequence of a cDNA that is differentially expressed in human brain tumors. *Cancer Lett.* 89: 215–221, 1995.
- [37476] 11028. Rumsby, G.; Cregeen, D. P.: Identification and expression of a cDNA for human hydroxypyruvate/glyoxylate reductase. *Biochim. Biophys. Acta* 1446: 383–388, 1999.
- [37477] 11029. Webster, K. E.; Ferree, P. M.; Holmes, R. P.; Cramer, S. D.: Identification of missense, nonsense, and deletion mutations in the GRHPR gene in patients with primary hyperoxaluria type II (PH2). *Hum. Genet.* 107:176–185, 2000.
- [37478] 11030. Cremona, O.; Di Paolo, G.; Wenk, M. R.; Luthi, A.; Kim, W. T.; Takei, K.; Daniell, L.; Nemoto, Y.; Shears, S. B.; Flavell, R. A.; McCormick, D. A.; De Camilli, P.: Essential role of phosphoinositide metabolism in synaptic vesicle recycling. *Cell* 99: 179–188, 1999.
- [37479] 11031. Cremona, O.; Nimmakayalu, M.; Haffner, C.; Bray-Ward, P.; Ward, D. C.; De Camilli, P.: Assignment of SYNJ1

to human chromosome 21q22.2 and Synj12 (sic) to the murine homologous region on chromosome 16C3–4 by in situ hybridization. *Cytogenet. Cell Genet.* 88: 89–90, 2000.

[37480] 11032. Haffner, C.; Takei, K.; Chen, H.; Ringstad, N.; Hudson, A.; Butler, M. H.; Salcini, A. E.; Di Fiore, P. P.; De Camilli, P.: Synaptojanin1: localization on coated endocytic intermediates in nerve terminals and interaction of its 170 kDa isoform with Eps15. *FEBS Lett.* 419:175–180, 1997.

[37481] 11033. McPherson, P. S.; Garcia, E. P.; Slepnev, V. I.; David, C.; Zhang, X.; Grabs, D.; Sossin, W. S.; Bauerfeind, R.; Nemoto, Y.; De Camilli, P.: A presynaptic inositol-5-phosphatase. *Nature* 379: 353–357, 1996.

[37482] 11034. Saito, T.; Guan, F.; Papolos, D. F.; Lau, S.; Klein, M.; Fann, C. S. J.; Lachman, H. M.: Mutation analysis of SYNJ1: a possible candidate gene for chromosome 21q22-linked bipolar disorder. *Molec. Psychiat.* 6: 387–395, 2001.

[37483] 11035. Aita, V. M.; Liang, X. H.; Murty, V. V. V. S.; Pincus, D. L.; Yu, W.; Cayanis, E.; Kalachikov, S.; Gilliam, T. C.; Levine, B.: Cloning and genomic organization of beclin 1, a candidate tumor suppressor gene on chromosome 17q21. *Genomics* 59: 59–65, 1999.

- [37484] 11036.Liang, X. H.; Jackson, S.; Seaman, M.; Brown, K.; Kempkes, B.;Hibshoosh, H.; Levine, B.: Induction of autophagy and inhibitionof tumorigenesis by beclin 1. Nature 402: 672–676, 1999.
- [37485] 11037.Liang, X. H.; Kleeman, L. K.; Jiang, H. H.; Gordon, G.; Goldman,J. E.; Berry, G.; Herman, B.; Levine, B.: Protection against fatalSindbis virus encephalitis by beclin, a novel Bcl-2-interacting protein. J.Virol. 72: 8586–8596, 1998.
- [37486] 11038.Liang, X. H.; Yu, J.; Brown, K.; Levine, B.: Beclin 1 containsa leucine-rich nuclear export signal that is required for its autophagyand tumor suppressor function. Cancer Res. 61: 3443–3449, 2001.
- [37487] 11039.Hatamura, I.; Kanauchi, Y.; Takahara, M.; Fujiwara, M.; Muragaki,Y.; Ooshima, A.; Ogino, T.: A nonsense mutation in TRPS1 in a Japanesefamily with trichorhino-phalangeal syndrome type I. (Letter) Clin.Genet. 59: 366–367, 2001.
- [37488] 11040.Hilton, M. J.; Sawyer, J. M.; Gutierrez, L.; Hogart, A.; Kung,T. C.; Wells, D. E.: Analysis of novel and recurrent mutations responsiblefor the tricho-rhino-phalangeal syndromes. J. Hum. Genet. 47: 103–106,2002.
- [37489] 11041.Patel, N.; Brinkman–Van der Linden, E. C. M.; Alt–

mann, S. W.; Gish, K.; Balasubramanian, S.; Timans, J. C.; Peterson, D.; Bell, M. P.; Bazan, J. F.; Varki, A.; Kastelein, R. A.: OB-BP1/Siglec-6: a leptin- and sialic acid-binding protein of the immunoglobulin superfamily. *J. Biol. Chem.* 274: 22729-22738, 1999. Note: Erratum: *J. Biol. Chem.* 274: 28058 only, 1999.

[37490] 11042. Miller, A. F.; Harvey, S. A. K.; Thies, R. S.; Olson, M. S.: Bonemorphogenetic protein-9: an autocrine/paracrine cytokine in the liver. *J. Biol. Chem.* 275: 17937-17945, 2000.

[37491] 11043. Scott, A. F.: Personal Communication. Baltimore, Md. 7/13/2000.

[37492] 11044. Lin, C.-Y.; Anders, J.; Johnson, M.; Sang, Q. A.; Dickson, R. B.: Molecular cloning of cDNA for matriptase, a matrix-degrading serine protease with trypsin-like activity. *J. Biol. Chem.* 274: 18231-18236, 1999.

[37493] 11045. Oberst, M.; Anders, J.; Xie, B.; Singh, B.; Ossandon, M.; Johnson, M.; Dickson, R. B.; Lin, C.-Y.: Matriptase and HAI-1 are expressed by normal and malignant epithelial cells in vitro and in vivo. *Am. J. Path.* 158: 1301-1311, 2001.

[37494] 11046. Kandil, E.; Kohda, K.; Ishibashi, T.; Tanaka, K.; Kasahara, M.: PA28 subunits of the mouse proteasome:

primary structures and chromosomal localization of the genes. *Immunogenetics* 46: 337–344, 1997.

[37495] 11047. Nikaido, T.; Shimada, K.; Shibata, M.; Hata, M.; Sakamoto, M.; Takasaki, Y.; Sato, C.; Takahashi, T.; Nishida, Y.: Cloning and nucleotide sequence of cDNA for Ki antigen, a highly conserved nuclear protein detected with sera from patients with systemic lupus erythematosus. *Clin. Exp. Immun.* 79: 209–214, 1990.

[37496] 11048. Bednarek, A. K.; Keck–Waggoner, C. L.; Daniel, R. L.; Laflin, K. J.; Bergsagel, P. L.; Kiguchi, K.; Brenner, A. J.; Aldaz, C. M.: WWOX, the FRA16D gene, behaves as a suppressor of tumor growth. *Cancer Res.* 61: 8068–8073, 2001.

[37497] 11049. Bednarek, A. K.; Laflin, K. J.; Daniel, R. L.; Liao, Q.; Hawkins, K. A.; Aldaz, C. M.: WWOX, a novel WW domain-containing protein mapping to human chromosome 16q23.3–24.1, a region frequently affected in breast cancer. *Cancer Res.* 60: 2140–2145, 2000.

[37498] 11050. Chang, N.–S.; Pratt, N.; Heath, J.; Schultz, L.; Sleve, D.; Carey, G. B.; Zevotek, N.: Hyaluronidase induction of a WW domain-containing oxidoreductase that enhances tumor necrosis factor cytotoxicity. *J. Biol. Chem.* 276: 3361–3370, 2001.

- [37499] 11051.Krummel, K. A.; Denison, S. R.; Calhoun, E.; Phillips, L. A.; Smith,D. I.: The common fragile site FRA16D and its associated gene WWOXare highly conserved in the mouse at Fra8E1. *Genes Chromosomes Cancer* 34:154–167, 2002.
- [37500] 11052.Miyake, A.; Mochizuki, S.; Yokoi, H.; Kohda, M.; Furuchi, K.:New ether-a-go-go K(+) channel family members localized in human telencephalon. *J.Biol. Chem.* 274: 25018–25025, 1999.
- [37501] 11053.Acampora, D.; Postiglione, M. P.; Avantaggiato, V.; Di Bonito,M.; Vaccarino, F. M.; Michaud, J.; Simeone, A.: Progressive impairmentof developing neuroendocrine cell lineages in the hypothalamus ofmice lacking the Orthopedia gene. *Genes Dev.* 13: 2787–2800, 1999.
- [37502] 11054.Lin, X.; State, M. W.; Vaccarino, F. M.; Greally, J.; Hass, M.;Leckman, J. F.: Identification, chromosomal assignment, and expressionanalysis of the human homeodomain-containing gene Orthopedia (OTP). *Genomics* 60:96–104, 1999.
- [37503] 11055.Gongora, C.; David, G.; Pintard, L.; Tissot, C.; Hua, T. D.; Dejean,A.; Mechti, N.: Molecular cloning of a new interferon-induced PMLnuclear body-associated protein. *J. Biol. Chem.* 272: 19457–19463,1997.

- [37504] 11056. Mattei, M. G.; Tissot, C.; Gongora, C.; Mechti, N.: Assignment of ISG20 encoding a new interferon-induced PML nuclear body-associated protein, to chromosome 15q26 by in situ hybridization. *Cytogenet. Cell Genet.* 79: 286–287, 1997.
- [37505] 11057. Pentecost, B. T.: Expression and estrogen regulation of the HEM45 mRNA in human tumor lines and in the rat uterus. *J. Steroid Biochem. Molec. Biol.* 64: 25–33, 1998.
- [37506] 11058. Berchtold, S.; Muhl-Zurbes, P.; Heufler, C.; Winkelhner, P.; Schuler, G.; Steinkasserer, A.: Cloning, recombinant expression and biochemical characterization of the murine CD83 molecule which is specifically upregulated during dendritic cell maturation. *FEBS Lett.* 461: 211–216, 1999.
- [37507] 11059. Fujimoto, Y.; Tu, L.; Miller, A. S.; Bock, C.; Fujimoto, M.; Doyle, C.; Steeber, D. A.; Tedder, T. F.: CD83 expression influences CD4+ T cell development in the thymus. *Cell* 108: 755–767, 2002.
- [37508] 11060. Kozlow, E. J.; Wilson, G. L.; Fox, C. H.; Kehrl, J. H.: Subtractive cDNA cloning of a novel member of the Ig gene superfamily expressed at high levels in activated B lymphocytes. *Blood* 81: 454–461, 1993.

- [37509] 11061.Olavesen, M.G.; Bentley, E.; Mason, R. V.; Stephens, R. J.; Ragoussis,J.: Fine mapping of 39 ESTs on human chromosome 6p23–p25. *Genomics* 46:303–306, 1997.
- [37510] 11062.Scholler, N.; Hayden–Ledbetter, M.; Hellstrom, K.–E.; Hellstrom,I.; Ledbetter, J. A.: CD83 is a sialic acid–binding Ig–like lectin(Siglec) adhesion receptor that binds monocytes and a subset of activatedCD8(+) T cells. *J. Immun.* 166: 3865–3872, 2001.
- [37511] 11063.Schinkmann, K.; Blenis, J.: Cloning and characterization of a human STE20–like protein kinase with unusual cofactor requirements. *J.Biol. Chem.* 272: 28695–28703, 1997.
- [37512] 11064.Zhou, T.–H.; Ling, K.; Guo, J.; Zhou, H.; Wu, Y.–L.; Jing, Q.;Ma, L.; Pei, G.: Identification of a human brain–specific isoformof mammalian STE20–like kinase 3 that is regulated by cAMP–dependentprotein kinase. *J. Biol. Chem.* 275: 2513–2519, 2000.
- [37513] 11065.St–Pierre, M. V.; Hagenbuch, B.; Ugele, B.; Meier, P. J.; Stallmach,T.: Characterization of an organic anion–transporting polypeptide(OATP–B) in human placenta. *J. Clin. Endocr. Metab.* 87: 1856–1863,2002.
- [37514] 11066.Murthy, A.; Gonzalez–Agosti, C.; Cordero, E.; Pinney, D.; Candia,C.; Solomon, F.; Gusella, J.; Ramesh, V.:

NHE-RF, a regulatory cofactor for Na⁽⁺⁾-H⁽⁺⁾ exchange, is a common interactor for merlin and ERM(MERM) proteins. J. Biol. Chem. 273: 1273–1276, 1998.

[37515] 11067. Reczek, D.; Berryman, M.; Bretscher, A.: Identification of EBP50: a PDZ-containing phosphoprotein that associates with members of the ezrin-radixin-moesin family. J. Cell Biol. 139: 169–179, 1997.

[37516] 11068. Shenolikar, S.; Voltz, J. W.; Minkoff, C. M.; Wade, J. B.; Weinman, E. J.: Targeted disruption of the mouse NHERF-1 gene promotes internalization of proximal tubule sodium-phosphate cotransporter type IIa and renal phosphate wasting. Proc. Nat. Acad. Sci. 99: 11470–11475, 2002.

[37517] 11069. Boucher, C. A.; Winchester, C. L.; Hamilton, G. M.; Winter, A. D.; Johnson, K. J.; Bailey, M. E. S.: Structure, mapping and expression of the human gene encoding the homeodomain protein, SIX2. Gene 247: 145–151, 2000.

[37518] 11070. Hasan, S.; Stucki, M.; Hassa, P. O.; Imhof, R.; Gehrig, P.; Hunziker, P.; Hubscher, U.; Hottiger, M. O.: Regulation of human flap endonuclease-1 activity by acetylation through the transcriptional coactivator p300. Molec. Cell 7: 1221–1231, 2001.

[37519] 11071. Hiraoka, L. R.; Harrington, J. J.; Gerhard, D. S.;

Lieber, M. R.;Hsieh, C.-L.: Sequence of human FEN-1, a structure-specific endonuclease,and chromosomal localization of the gene (FEN1) in mouse and human. *Genomics* 25:220-225, 1995.

[37520] 11072.Hosfield, D. J.; Mol, C. D.; Shen, B.; Tainer, J. A.: Structureof the DNA repair and replication endonuclease and exonuclease FEN-1:coupling DNA and PCNA binding to FEN-1 activity. *Cell* 95: 135-146,1998.

[37521] 11073.Kucherlapati, M.; Yang, K.; Kuraguchi, M.; Zhao, J.; Lia, M.; Heyer,J.; Kane, M. F.; Fan, K.; Russell, R.; Brown, A. M. C.; Kneitz, B.;Edelmann, W.; Kolodner, R. D.; Lipkin, M.; Kucherlapati, R.: Haploinsufficiencyof Flap endonuclease (Fen1) leads to rapid tumor progression. *Proc.Nat. Acad. Sci.* 99: 9924-9929, 2002.

[37522] 11074.Otto, C. J.; Almqvist, E.; Hayden, M. R.; Andrew, S. E.: The 'flap'endonuclease gene FEN1 is excluded as a candidate gene implicatedin the CAG repeat expansion underlying Huntington disease. *Clin.Genet.* 59: 122-127, 2001.

[37523] 11075.Spiro, C.; Pelletier, R.; Rolfsmeier, M. L.; Dixon, M. J.; Lahue,R. S.; Gupta, G.; Park, M. S.; Chen, X.; Mariappan, S. V. S.; McMurray,C. T.: Inhibition of FEN-1 processing by DNA secondary structureat trinucleotide repeats. *Molec.*

Cell 4: 1079–1085, 1999.

- [37524] 11076.Tishkoff, D. X.; Filosi, N.; Gaida, G. M.; Kolodner, R. D.: Anovel mutation avoidance mechanism dependent on *S. cerevisiae* RAD27is distinct from DNA mismatch repair. Cell 88: 253–263, 1997.
- [37525] 11077.Vermeesch, J. R.; Mertens, G.; David, G.; Marynen, P.: Assignmentof the human glypican gene (GPC1) to 2q35–q37 by fluorescence in situhybridization. Genomics 25: 327–329, 1995.
- [37526] 11078.Ono, Y.; Ohno, M.; Shimura, Y.: Identification of a putative RNAhelicase (HRH1), a human homolog of yeast Prp22. Molec. Cell. Biol. 14:7611–7620, 1994.
- [37527] 11079.Fulop, V.; Bocskei, Z.; Polgar, L.: Prolyl oligopepti- dase: anunusual beta–propeller domain regulates proteol- ysis. Cell 94: 161–170,1998.
- [37528] 11080.Goossens, F. J.; Wauters, J. G.; Vanhoof, G. C.; Bossuyt, P. J.;Schatterman, K. A.; Loens, K.; Scharpe, S. L.: Subregional mappingof the human lymphocyte prolyl oligopeptidase gene (PREP) to humanchromosome 6q22. Cytogenet. Cell Genet. 74: 99–101, 1996.
- [37529] 11081.Vanhoof, G.; Goossens, F.; Hendriks, L.; De Meester, I.; Hendriks,D.; Vriend, G.; Van Broeckhoven, C.; Scharpe, S.: Cloning and sequenceanalysis of the gene en–

coding human lymphocyte prolyl endopeptidase. *Gene* 149:363–366, 1994.

[37530] 11082. Goldstein, L. A.; Gherzi, G.; Pineiro-Sanchez, M. L.; Salamone, M.; Yeh, Y.; Flessate, D.; Chen, W.-T.: Molecular cloning of seprase: a serine integral membrane protease from human melanoma. *Biochim. Biophys. Acta* 1361: 11–19, 1997.

[37531] 11083. Mathew, S.; Scanlan, M. J.; Mohan Raj, B. K.; Murty, V. V. V. S.; Garin-Chesa, P.; Old, L. J.; Rettig, W. J.; Chaganti, R. S. K.: The gene for fibroblast activation protein alpha (FAP), a putative cell surface-bound serine protease expressed in cancer stroma and wound healing, maps to chromosome band 2q23. *Genomics* 25: 335–337, 1995.

[37532] 11084. Pineiro-Sanchez, M. L.; Goldstein, L. A.; Dodt, J.; Howard, L.; Yeh, Y.; Tran, H.; Argraves, W. S.; Chen, W.-T.: Identification of the 170-kDa melanoma membrane-bound gelatinase (seprase) as a serine integral membrane protease. *J. Biol. Chem.* 272: 7595–7601, 1997.

[37533] 11085. Scanlan, M. J.; Raj, B. K. M.; Calvo, B.; Garin-Chesa, P.; Sanz-Moncasi, M. P.; Healey, J. H.; Old, L. J.; Rettig, W. J.: Molecular cloning of fibroblast activation protein alpha, a member of the serine protease family selectively expressed in stromal fibroblasts of epithelial cancers. *Proc.*

Nat. Acad. Sci. 91: 5657–5661, 1994.

[37534] 11086.Okumura, K.; Nogami, M.; Taguchi, H.; Dean, F. B.; Chen, M.; Pan,Z.–Q.; Hurwitz, J.; Shiratori, A.; Murakami, Y.; Ozawa, K.; Eki, T.: Assignment of the 36.5–kDa (RFC5), 37–kDa (RFC4), 38–kDa (RFC3),and 40–kDa (RFC2) sub–unit genes of human replication factor C to chromosome–bands 12q24.2–q24.3, 3q27, 13q12.3–q13, and 7q11.23. Genomics 25:274–278, 1995.

[37535] 11087.Peoples, R.; Perez–Jurado, L.; Wang, Y.–K.; Kaplan, P.; Francke,U.: The gene for replication factor C subunit 2 (RFC2) is withinthe 7q11.23 Williams syndrome deletion. (Letter) Am. J. Hum. Genet. 58:1370–1373, 1996.

[37536] 11088.Wang, Y.; Cortez, D.; Yazdi, P.; Neff, N.; Elledge, S. J.; Qin,J.: BASC, a super complex of BRCA1–associated proteins involved inthe recognition and repair of aberrant DNA structures. Genes Dev. 14:927–939, 2000.

[37537] 11089.Thim, L.; Woldike, H. F.; Nielsen, P. F.; Christensen, M.; Lynch–Devaney,K.; Podolsky, D. K.: Characterization of human and rat intestinaltrefoil factor produced in yeast. Biochemistry 34: 4757–4764, 1995.

[37538] 11090.Itoh, T.; Mori, T.; Ohkubo, H.; Yamaizumi, M.: A newly identifiedpatient with clinical xeroderma pigmento–sum phenotype has a non–sensemutation in the DDB2

gene and incomplete repair in (6–4) photoproducts.

J. Invest. Derm. 113: 251–257, 1999.

- [37539] 11091. Acebron, A.; Aza-Blanc, P.; Rossi, D. L.; Lamas, L.; Santisteban, P.: Congenital human thyroglobulin defect due to low expression of the thyroid-specific transcription factor TTF-1. J. Clin. Invest. 96:781–785, 1995.
- [37540] 11092. Guazzi, S.; Price, M.; De Felice, M.; Damante, G.; Mattei, M.-G.; Di Lauro, R.: Thyroid nuclear factor 1 (TTF-1) contains a homeodomain and displays a novel DNA binding specificity. EMBO J. 9: 3631–3639, 1990.
- [37541] 11093. Ikeda, K.; Clark, J. C.; Shaw-White, J. R.; Stahlman, M. T.; Boutell, C. J.; Whitsett, J. A.: Gene structure and expression of human thyroid transcription factor-1 in respiratory epithelial cells. J. Biol. Chem. 270: 8108–8114, 1995.
- [37542] 11094. Kimura, S.; Hara, Y.; Pineau, T.; Fernandez-Salguero, P.; Fox, C. H.; Ward, J. M.; Gonzalez, F. J.: The T/ebp null mouse: thyroid-specific enhancer-binding protein is essential for the organogenesis of the thyroid, lung, ventral forebrain, and pituitary. Genes Dev. 10:60–69, 1996.
- [37543] 11095. Fernandes-Alnemri, T.; Armstrong, R. C.; Krebs, J.; Srinivasula, S. M.; Wang, L.; Bullrich, F.; Fritz, L. C.; Tra-

pani, J. A.; Tomaselli, K. J.; Litwack, G.; Alnemri, E. S.: In vitro activation of CPP32 and Mch3 by Mch4, a novel human apoptotic cysteine protease containing two FADD-like domains. *Proc. Nat. Acad. Sci.* 93: 7464–7469, 1996.

[37544] 11096. Fernandes-Alnemri, T.; Litwack, G.; Alnemri, E. S.: CPP32, a novel human apoptotic protein with homology to *Caenorhabditis elegans* cell death protein Ced-3 and mammalian interleukin-1 beta-converting enzyme. *J. Biol. Chem.* 269: 30761–30764, 1994.

[37545] 11097. Fernando, P.; Kelly, J. F.; Balazsi, K.; Slack, R. S.; Megeney, L. A.: Caspase 3 activity is required for skeletal muscle differentiation. *Proc. Nat. Acad. Sci.* 99: 11025–11030, 2002.

[37546] 11098. Huang, Y.; Shin, N.-H.; Sun, Y.; Wang, K. K. W.: Molecular cloning and characterization of a novel caspase-3 variant that attenuates apoptosis induced by proteasome inhibition. *Biochem. Biophys. Res. Commun.* 283: 762–769, 2001.

[37547] 11099. Kuida, K.; Zheng, T. S.; Na, S.; Kuan, C.; Yang, D.; Karasuyama, H.; Rakio, P.; Flavell, R. A.: Decreased apoptosis in the brain and premature lethality in CPP32-deficient mice. *Nature* 384: 368–372, 1996.

[37548] 11100. Levkau, B.; Koyama, H.; Raines, E. W.; Clurman, B.

E.; Herren,B.; Orth, K.; Roberts, J. M.; Ross, R.: Cleavage of p21(Cip1/Waf1)and p27(Kip1) mediates apoptosis in endothelial cells through activationof Cdk2: role of a caspase cascade. *Molec. Cell* 1: 553–563, 1998.

[37549] 11101.Nasir, J.; Theilmann, J. L.; Chopra, V.; Jones, A. M.; Walker,D.; Rasper, D. M.; Vaillancourt, J. P.; Hewitt, J. E.; Nicholson,D. W.; Hayden, M. R.: Localization of the cell death genes CPP32and Mch–2 to human chromosome 4q. *Mammalian Genome* 8: 56–59, 1997.

[37550] 11102.Tiso, N.; Pallavicini, A.; Muraro, T.; Zimbello, R.; Apolloni,E.; Valle, G.; Lanfranchi, G.; Danieli, G. A.: Chromosomal localizationof the human genes, CPP32, Mch2, Mch3, and Ich–1, involved in cellularapoptosis. *Biochem. Biophys. Res. Commun.* 225: 983–989, 1996.

[37551] 11103.Woo, M.; Hakem, R.; Soengas, M. S.; Duncan, G. S.; Shahinian,A.; Kagi, K.; Hakem, A.; McCurrach, M.; Khoo, W.; Kaufman, S. A.;Senaldi, G.; Howard, T.; Lowe, S. W.; Mak, T. W.: Essential contributionof caspase 3/CPP32 to apoptosis and its associated nuclear changes. *GenesDev.* 12: 806–819, 1998.

[37552] 11104.Nicholson, D. W.; Ali, A.; Thornberry, N. A.; Vaillancourt, J.P.; Ding, C. K.; Gallant, M.; Gareau, Y.; Griffin, P. R.; Labelle,M.; Lazebnik, Y. A.; Munday, N. A.; Raju, S. M.;

Smulson, M. E.; Yamin, T.-T.; Yu, V. L.; Miller, D. K.: Identification and inhibition of the ICE/CED-3 protease necessary for mammalian apoptosis. *Nature* 376:37-43, 1995.
MEDLINE UID: 95319529

[37553] 11105. Bergeron, L.; Perez, G. I.; Macdonald, G.; Shi, L.; Sun, Y.; Jurisicova, A.; Varmuza, S.; Latham, K. E.; Flaws, J. A.; Salter, J. C. M.; Hara, H.; Moskowitz, M. A.; Li, E.; Greenberg, A.; Tilly, J. L.; Yuan, J.: Defects in regulation of apoptosis in caspase-2-deficient mice. *Genes Dev.* 12: 1304-1314, 1998.

[37554] 11106. Garcia, C. K.; Goldstein, J. L.; Pathak, R. K.; Anderson, R. G. W.; Brown, M. S.: Molecular characterization of a membrane transporter for lactate, pyruvate, and other monocarboxylates: implications for the Cori cycle. *Cell* 76: 865-873, 1994.

[37555] 11107. Garcia, C. K.; Li, X.; Luna, J.; Francke, U.: cDNA cloning of the human monocarboxylate transporter 1 and chromosomal localization of the SLC16A1 locus to 1p13.2-p12. *Genomics* 23: 500-503, 1994.

[37556] 11108. Kim, C. M.; Goldstein, J. L.; Brown, M. S.: cDNA cloning of mev, a mutant protein that facilitates cellular uptake of mevalonate, and identification of the point mutation responsible for its gain of function. *J. Biol. Chem.*

267: 23113–23121, 1992.

- [37557] 11109.Kingsmore, S. F.; Souryal, C. A.; Watson, M. L.; Patel, D. D.;Seldin, M. F.: Physical and genetic linkage of the genes encodingLy-9 and CD48 on mouse and human chromosomes 1. Immunogenetics 42:59–62, 1995.
- [37558] 11110.Kozak, C. A.; Davidson, W. F.; Morse, H. C., III: Genetic andfunctional relationships of the retroviral and lymphocyte alloantigenloci on mouse chromosome 1. Immunogenetics 19: 163–168, 1984.
- [37559] 11111.Sandrin, M. S.; Gumley, T. P.; Henning, M. M.; Vaughan, H. A.;Gonez, L. J.; Trapani, J. A.; McKenzie, I. F. C.: Isolation and characterizationof cDNA clones for mouse Ly-9. J. Immun. 149: 1636–1641, 1992.
- [37560] 11112.Leek, J. P.; Moynihan, T. P.; Anwar, R.; Bonthron, D. T.; Markham,A. F.; Lench, N. J.: Assignment of Indian hedgehog (IHH) to humanchromosome bands 2q33–q35 by in situ hybridization. Cytogenet. CellGenet. 76: 187–188, 1997.
- [37561] 11113.Porter, J. A.; Young, K. E.; Beachy, P. A.: Cholesterol modificationof hedgehog signaling proteins in animal development. Science 274:255–259, 1996.
- [37562] 11114.Zhang, X. M.; Ramalho-Santos, M.; McMahon, A. P.: Smoothened mutantsreveal redundant roles for Shh and

Ihh signaling including regulation of L/R asymmetry by the mouse node. *Cell* 105: 781–792, 2001.

- [37563] 11115. Geurts, J. M. W.; Schoenmakers, E. F. P. M.; Roijer, E.; Astrom, A.-K.; Stenman, G.; van de Ven, W. J. M.: Identification of NFIB as recurrent translocation partner gene of HMGIC in pleomorphic adenomas. *Oncogene* 16:865–872, 1998.
- [37564] 11116. Fiedorek, F. T., Jr.; Kay, E. S.: Mapping of the insulin promoter factor 1 gene (*Ip1*) to distal mouse chromosome 5. *Genomics* 28:581–584, 1995.
- [37565] 11117. Hani, E. H.; Stoffers, D. A.; Chevre, J.-C.; Durand, E.; Stanojevic, V.; Dina, C.; Habener, J. F.; Froguel, P.: Defective mutations in the insulin promoter factor-1 (IPF-1) gene in late-onset type 2 diabetes mellitus. *J. Clin. Invest.* 104: R41–R48, 1999.
- [37566] 11118. Tang, J. Y.; Hwang, B. J.; Ford, J. M.; Hanawalt, P. C.; Chu, G.: Xeroderma pigmentosum p48 gene enhances global genomic repair and suppresses UV-induced mutagenesis. *Molec. Cell* 5: 737–744, 2000.
- [37567] 11119. Bermingham, J. R., Jr.; Arden, K. C.; Naumova, A. K.; Sapienza, C.; Viars, C. S.; Fu, X.-D.; Khotz, J.; Manley, J. L.; Rosenfeld, M. G.: Chromosomal localization of mouse and human genes encoding the splicing factors ASF/SF2

(SFRS1) and SC-35 (SFRS2). *Genomics* 29:70–79, 1995.

- [37568] 11120. Wang, J.; Takagaki, Y.; Manley, J. L.: Targeted disruption of an essential vertebrate gene: ASF/SF2 is required for cell viability. *Genes Dev.* 10: 2588–2599, 1996.
- [37569] 11121. Hopfner, K.-P.; Karcher, A.; Craig, L.; Woo, T. T.; Carney, J.P.; Tainer, J. A.: Structural biochemistry and interaction architecture of the DNA double-strand break repair Mre11 nuclease and Rad50-ATPase. *Cell* 105:473–485, 2001.
- [37570] 11122. Stracker, T. H.; Carson, C. T.; Weitzman, M. D.: Adenovirus oncoproteins inactivate the Mre11-Rad50-NBS1 DNA repair complex. *Nature* 418:348–352, 2002.
- [37571] 11123. Zhong, Q.; Chen, C.-F.; Li, S.; Chen, Y.; Wang, C.-C.; Xiao, J.; Chen, P.-L.; Sharp, Z. D.; Lee, W.-H.: Association of BRCA1 with the hRad50-hMre11-p95 complex and the DNA damage response. *Science* 285:747–750, 1999.
- [37572] 11124. Zhu, X.-D.; Kuster, B.; Mann, M.; Petrini, J. H. J.; de Lange, T.: Cell-cycle-regulated association of RAD50/MRE11/NBS1 with TRF2 and human telomeres. *Nature Genet.* 25: 347–352, 2000.
- [37573] 11125. Virbasius, C. A.; Virbasius, J. V.; Scarpulla, R. C.:

NRF-1, an activator involved in nuclear-mitochondrial interactions, utilizes a new DNA-binding domain conserved in a family of developmental regulators. *Genes Dev.* 7: 2431-2445, 1993.

- [37574] 11126. Choi, S. S.; Park, I.-C.; Yun, J. W.; Sung, Y. C.; Hong, S.-I.; Shin, H.-S.: A novel Bcl-2 related gene, Bfl-1, is overexpressed in stomach cancer and preferentially expressed in bone marrow. *Oncogene* 11:1693-1698, 1995.
- [37575] 11127. Choi, S. S.; Park, S. H.; Kim, U.-J.; Shin, H.-S.: Bfl-1, a Bcl-2-related gene, is the human homolog of the murine A1, and maps to chromosome 15q24.3. *Mammalian Genome* 8: 781-782, 1997.
- [37576] 11128. D'Sa-Eipper, C.; Subramanian, T.; Chinnadurai, G.: Bfl-1, a bcl-2 homologue, suppresses p53-induced apoptosis and exhibits potent cooperative transforming activity. *Cancer Res.* 56: 3879-3882, 1996.
- [37577] 11129. Lin, E. Y.; Kozak, C. A.; Orlofsky, A.; Prystowsky, M. B.: The bcl-2 family member, Bcl2a1, maps to mouse chromosome 9 and human chromosome 15. *Mammalian Genome* 8: 293-294, 1997.
- [37578] 11130. Lin, E. Y.; Orlofsky, A.; Berger, M. S.; Prystowsky, M. B.: Characterization of A1, a novel hemopoietic-specific early-response gene with sequence similarity to bcl-2. *J.*

Immun. 151: 1979–1988, 1993.

- [37579] 11131.Lin, E. Y.; Orlofsky, A.; Wang, H.-G.; Reed, J. C.; Prystowsky,M. B.: A1, a Bcl-2 family member, prolongs cell survival and permitsmyeloid differentiation. Blood 87: 983–992, 1996.
- [37580] 11132.Xiang, Z.; Ahmed, A. A.; Moller, C.; Nakayama, K.; Hatakeyama,S.; Nilsson, G.: Essential role of the prosurvival bcl-2 homologueA1 in mast cell survival after allergic activation. J. Exp. Med. 194:1561–1569, 2001.
- [37581] 11133.Bass, B. L.; Weintraub, H.: An unwinding activity that covalentlymodifies its double-stranded RNA substrate. Cell 55: 1089–1098,1988.
- [37582] 11134.Kim, U.; Wang, Y.; Sanford, T.; Zeng, Y.; Nishikura, K.: Molecularcloning of cDNA for double-stranded RNA adenosine deaminase, a candidateenzyme for nuclear RNA editing. Proc. Nat. Acad. Sci. 91: 11457–11461,1994.
- [37583] 11135.O'Connell, M. A.; Krause, S.; Higuchi, M.; Hsuan, J. J.; Totty,N. F.; Jenny, A.; Keller, W.: Cloning of cDNAs encoding mammaliandouble-stranded RNA-specific adenosine deaminase. Molec. Cell. Biol. 15:1389–1397, 1995.
- [37584] 11136.Patterson, J. B.; Samuel, C. E.: Expression and regulation byinterferon of a double-stranded-RNA-specific adenosine deaminase fromhuman cells: evidence for two

forms of the deaminase. *Molec. Cell.Biol.* 15: 5376–5388, 1995.

- [37585] 11137.Wang, Q.; Khillan, J.; Gadue, P.; Nishikura, K.: Requirement of the RNA editing deaminase ADAR1 gene for embryonic erythropoiesis. *Science* 290:1765–1768, 2000.
- [37586] 11138.Wang, Y.; Zeng, Y.; Murray, J. M.; Nishikura, K.: Genomic organization and chromosomal location of the human dsRNA adenosine deaminase gene: the enzyme for glutamate-activated ion channel RNA editing. *J. Molec.Biol.* 254: 184–195, 1995.
- [37587] 11139.Weier, H.-U. G.; George, C. X.; Greulich, K. M.; Samuel, C. E.: The interferon-inducible, double-stranded RNA-specific adenosine deaminase gene (DSRAD) maps to human chromosome 1q21.1–21.2. *Genomics* 30:372–375, 1995.
- [37588] 11140.Weier, H.-U. G.; George, C. X.; Lersch, R. A.; Breitwieser, S.; Cheng, J.-F.; Samuel, C. E.: Assignment of the RNA-specific adenosine deaminase gene (Adar) to mouse chromosome 3F2 by in situ hybridization. *Cytogenet.Cell Genet.* 89: 214–215, 2000.
- [37589] 11141.Kawagoe, H.; Soma, O.; Goji, J.; Nishimura, N.; Narita, M.; Inazawa, J.; Nakamura, H.; Sano, K.: Molecular cloning and chromosomal assignment of the human brain-

type phosphodiesterase I/nucleotide pyrophosphatase gene (PDNP2). *Genomics* 30: 380–384, 1995.

- [37590] 11142. Murata, J.; Lee, H. Y.; Clair, T.; Krutzsch, H. C.; Arestad, A. A.; Sobel, M. E.; Liotta, L. A.; Stracke, M. L.: cDNA cloning of human tumor motility-stimulating protein, autotaxin, reveals a homology with phosphodiesterases. *J. Biol. Chem.* 269: 30479–30484, 1994.
- [37591] 11143. Narita, M.; Goji, J.; Nakamura, H.; Sano, K. Molecular cloning, expression, and localization of a brain-specific phosphodiesterase I/nucleotide (PD-I- α) from rat brain. *J. Biol. Chem.* 269: 28235–28242, 1994.
- [37592] 11144. Piao, J.-H.; Matsuda, Y.; Nakamura, H.; Sano, K.: Assignment of Pdn2, the gene encoding phosphodiesterase I/nucleotide pyrophosphatase 2, to mouse chromosome 15D2. *Cytogenet. Cell Genet.* 87: 172–174, 1999.
- [37593] 11145. Tokumura, A.; Majima, E.; Kariya, Y.; Tominaga, K.; Kogure, K.; Yasuda, K.; Fukuzawa, K.: Identification of human plasma lysophospholipase D, a lysophosphatidic acid-producing enzyme, as autotaxin, a multifunctional phosphodiesterase. *J. Biol. Chem.* 277: 39436–39442, 2002.
- [37594] 11146. Lin, Q.; Schwarz, J.; Bucana, C.; Olson, E. N.: Control of mouse cardiac morphogenesis and myogenesis by transcription factor MEF2C. *Science* 276: 1404–1407,

1997.

- [37595] 11147. Erickson, H. P.: A tenascin knockout with a phenotype. *Nature Genet.* 17: 5–7, 1997.
- [37596] 11148. Akama, T. O.; Nakagawa, H.; Sugihara, K.; Narisawa, S.; Ohyama, C.; Nishimura, S.-I.; O'Brien, D. A.; Moremen, K. W.; Millan, J. L.; Fukuda, M. N.: Germ cell survival through carbohydrate-mediated interaction with Sertoli cells. *Science* 295: 124–127, 2002.
- [37597] 11149. Bai, R.-Y.; Koester, C.; Ouyang, T.; Hahn, S. A.; Hammerschmidt, M.; Peschel, C.; Duyster, J.: SMIF, a Smad4-interacting protein that functions as a co-activator in TGF-beta signalling. *Nature Cell Biol.* 4: 181–190, 2002.
- [37598] 11150. Akiyama, Y.; Tsubouchi, N.; Yuasa, Y.: Frequent somatic mutations of hMSH3 with reference to microsatellite instability in hereditary nonpolyposis colorectal cancer. *Biochem. Biophys. Res. Commun.* 236: 248–252, 1997.
- [37599] 11151. Chittenden, T.; Harrington, E. A.; O'Connor, R.; Flemington, C.; Lutz, R. J.; Evan, G. I.; Guild, B. C.: Induction of apoptosis by the Bcl-2 homologue Bak. *Nature* 374: 733–736, 1995.
- [37600] 11152. Herberg, J. A.; Phillips, S.; Beck, S.; Jones, T.; Sheer, D.; Wu, J. J.; Prochazka, V.; Barr, P. J.; Kiefer, M. C.; Trowsdale, J.: Genomic structure and domain organisation of the

human Bak gene. *Gene* 211:87–94, 1998.

[37601] 11153.Kiefer, M. C.; Brauer, M. J.; Powers, V. C.; Wu, J. J.; Umansky, S. R.; Tomei, L. D.; Barr, P. J.: Modulation of apoptosis by the widely distributed Bcl–2 homologue Bak. *Nature* 374: 736–739, 1995.

[37602] 11154.Ulrich, E.; Kauffmann–Zeh, A.; Hueber, A.–O.; Williamson, J.; Chittenden, T.; Ma, A.; Evan, G.: Gene structure, cDNA sequence, and expression of murine Bak, a proapoptotic Bcl–2 family member. *Genomics* 44:195–200, 1997.

[37603] 11155.Higuchi, M.; Maas, S.; Single, F. N.; Hartner, J.; Rozov, A.; Burnashev, N.; Feldmeyer, D.; Sprengel, R.; Seeburg, P. H.: Point mutation in an AMPA receptor gene rescues lethality in mice deficient in the RNA–editing enzyme ADAR2. *Nature* 406: 78–81, 2000.

[37604] 11156.Lai, F.; Chen, C.–X.; Carter, K. C.; Nishikura, K.: Editing of glutamate receptor B subunit ion channel RNAs by four alternatively spliced DRADA2 double–stranded RNA adenosine deaminases. *Molec. Cell. Biol.* 17: 2413–2424, 1997.

[37605] 11157.Melcher, T.; Maas, S.; Herb, A.; Sprengel, R.; Seeburg, P. H.; Higuchi, M.: A mammalian RNA editing enzyme. *Nature* 379: 460–463, 1996.

- [37606] 11158.Mittaz, L.; Scott, H. S.; Rossier, C.; Seeburg, P. H.; Higuchi,M.; Antonarakis, S. E.: Cloning of a human RNA editing deaminase(ADARB1) of glutamate receptors that maps to chromosome 21q22.3. *Genomics* 41:210–217, 1997.
- [37607] 11159.O'Connell, M. A.; Gerber, A.; Keller, W.: Purification of humandouble–stranded RNA–specific editase 1 (hRED1) involved in editingof brain glutamate receptor B pre–mRNA. *J. Biol. Chem.* 272: 473–478,1997.
- [37608] 11160.Villard, L.; Tassone, F.; Haymowicz, M.; Welborn, R.; Gardiner,K.: Map location, genomic organization and expression patterns ofthe human RED1 RNA editase. *Somat. Cell Molec. Genet.* 23: 135–145,1997.
- [37609] 11161.Yang, J.–H.; Sklar, P.; Axel, R.; Maniatis, T.: Purification andcharacterization of a human RNA adenosine deaminase for glutamatereceptor B pre–mRNA editing. *Proc. Nat. Acad. Sci.* 94: 4354–4359,1997.
- [37610] 11162.Lu, C.–M.; Han, J.; Rado, T. A.; Brown, G. B.: Differential expressionof two sodium channel subtypes in human brain. *FEBS Letters* 303:53–58, 1992.
- [37611] 11163.Zhao, Z.; Lee, C.–C.; Monckton, D. G.; Yazdani, A.; Coolbaugh,M. I.; Li, X.; Bailey, J.; Shen, Y.; Caskey, C. T.: Characterizationand genomic mapping of genes and

pseudogenes of a new human protein tyrosine phosphatase. *Genomics* 35: 172–181, 1996.

[37612] 11164. Chopra, V. S.; Metzler, M.; Rasper, D. M.; Engqvist-Goldstein, A. E. Y.; Singaraja, R.; Gan, L.; Fichter, K. M.; McCutcheon, K.; Drubin, D.; Nicholson, D. W.; Hayden, M. R.: HIP12 is a non-proapoptotic member of a gene family including HIP1, an interacting protein with huntingtin. *Mammalian Genome* 11: 1006–1015, 2000.

[37613] 11165. Borrás, T.; Rowlette, L. L. S.; Tamm, E. R.; Gottanka, J.; Epstein, D. L.: Effects of elevated intraocular pressure on outflow facility and TIGR/MYOC expression in perfused human anterior segments. *Invest. Ophthalm. Vis. Sci.* 43: 33–40, 2002.

[37614] 11166. Cerretti, D. P.; Nelson, N.: Characterization of the genes from mouse LERK-3/Ephrin-A3 (Epl3), mouse LERK-4/Ephrin-A4 (Epl4), and human LERK-6/Ephrin-A2 (EPLG6): conservation of intron/exon structure. *Genomics* 47: 131–135, 1998.

[37615] 11167. Ma, T.; Yang, B.; Kuo, W.-L.; Verkman, A. S.: cDNA cloning and gene structure of a novel water channel expressed exclusively in human kidney: evidence for a gene cluster of aquaporins at chromosome locus 12q13. *Genomics* 35: 543–550, 1996.

- [37616] 11168.Yasui, M.; Kwon, T.-H.; Knepper, M. A.; Nielsen, S.; Agre, P.:Aquaporin-6: an intracellular vesicle water channel protein in renalepithelia. *Proc. Nat. Acad. Sci.* 96: 5808-5813, 1999.
- [37617] 11169.Shan, X.; Bourdeau, A.; Rhoton, A.; Wells, D. E.; Cohen, E. H.;Landgraf, B. E.; Palfree, R. G. E.: Characterization and mappingto human chromosome 8q24.3 of Ly-6-related gene 9804 encoding an apparenthomologue of mouse TSA-1. *J. Immun.* 160: 197-208, 1998.
- [37618] 11170.Bova, G. S.; MacGrogan, D.; Levy, A.; Pin, S. S.; Bookstein, R.;Isaacs, W. B.: Physical mapping of chromosome 8p22 markers and theirhomozygous deletion in a metastatic prostate cancer. *Genomics* 35:46-54, 1996.
- [37619] 11171.An, S.; Yang, J.; Xia, M.; Goetzl, E. J.: Cloning and expressionof the EP2 subtype of human receptors for prostaglandin E2. *Biochem.Biophys. Res. Comm.* 197: 263-270, 1993.
- [37620] 11172.Bastien, L.; Sawyer, N.; Grygorczyk, R.; Metters, K. M.; Adam,M.: Cloning, functional expression and characterization of the humanprostaglandin E2 receptor EP2 subtype. *J. Biol. Chem.* 269: 11873-11877,1994.
- [37621] 11173.Coleman, R. A.; Grix, S. P.; Head, S. A.; Louttit, J. B.; Mallett,A.; Sheldrick, R. L. G.: A novel inhibitory

prostanoid receptor in piglet saphenous vein.

Prostaglandins 47: 151–168, 1994.

- [37622] 11174. Foord, S. M.; Marks, B.; Stolz, M.; Bufflier, E.; Fraser, N. J.; Lee, M. G.: The structure of the prostaglandin EP4 receptor gene and related pseudogenes. *Genomics* 35: 182–188, 1996.
- [37623] 11175. Mori, K.; Tanaka, I.; Kotani, M.; Miyaoka, F.; Sando, T.; Muro, S.; Sasaki, Y.; Nakagawa, O.; Ogawa, Y.; Usui, T.; Ozaki, S.; Ichikawa, A.; Narumiya, S.; Nakao, K.: Gene expression of the human prostaglandin E receptor EP4 subtype: differential regulation in monocytoid and lymphoid lineage cells by phorbol ester. *J. Molec. Med.* 74: 333–336, 1996.
- [37624] 11176. Regan, J. W.; Bailey, T. J.; Pepperl, D. J.; Pierce, K. L.; Bogardus, A. M.; Donello, J. E.; Fairbairn, C. E.; Kedzie, K. M.; Woodward, D. F.; Gil, D. W.: Cloning of a novel human prostaglandin receptor with characteristics of the pharmacologically defined EP2 subtype. *Molec. Pharm.* 46: 213–220, 1994.
- [37625] 11177. Segi, E.; Sugimoto, Y.; Yamasaki, A.; Aze, Y.; Oida, H.; Nishimura, T.; Murata, T.; Matsuoka, T.; Ushikubi, F.; Hirose, M.; Tanaka, T.; Yoshida, N.; Narumiya, S.; Ichikawa, A.: Patent ductus arteriosus and neonatal death in

prostaglandin receptor EP4-deficient mice.

Biochem.Biophys. Res. Comm. 246: 7-12, 1998.

- [37626] 11178.Yoshida, K.; Oida, H.; Kobayashi, T.; Maruyama, T.; Tanaka, M.;Katayama, T.; Yamaguchi, K.; Segi, E.; Tsuboyama, T.; Matsushita,M.; Ito, K.; Ito, Y.; Sugimoto, Y.; Ushikubi, F.; Ohuchida, S.; Kondo,K.; Nakamura, T.; Narumiya, S.: Stimulation of bone formation and prevention of bone loss by prostaglandin E EP4 receptor activation. Proc.Nat. Acad. Sci. 99: 4580-4585, 2002.

- [37627] 11179.Jadayel, D. M.; Osborne, L. R.; Coignet, L. J. A.; Zani, V. J.;Tsui, L.-C.; Scherer, S. W.; Dyer, M. J. S.: The BCL7 gene family:deletion of BCL7B in Williams syndrome. Gene 224: 35-44, 1998.

- [37628] 11180.Arima, T.; Drewell, R. A.; Arney, K. L.; Inoue, J.; Makita, Y.;Hata, A.; Oshimura, M.; Wake, N.; Surani, M. A.: A conserved imprinting control region at the HYMAI/ZAC domain is implicated in transient neonatal diabetes mellitus. Hum. Molec. Genet. 10: 1475-1483, 2001.

- [37629] 11181.Joung, I.; Strominger, J. L.; Shin, J.: Molecular cloning of aphosphotyrosine-independent ligand of the p56-lck SH2 domain. Proc.Nat. Acad. Sci. 93: 5991-5995, 1996.

- [37630] 11182.Laurin, N.; Brown, J. P.; Morissette, J.; Raymond, V.:

Recurrent mutation of the gene encoding sequestosome 1 (SQSTM1/p62) in Paget disease of bone. *Am. J. Hum. Genet.* 70: 1582–1588, 2002.

[37631] 11183. Park, I.; Chung, J.; Walsh, C. T.; Yun, Y.; Strominger, J. L.; Shin, J.: Phosphotyrosine-independent binding of a 62-kDa protein to the src homology 2 (SH2) domain of p56-lck and its regulation by phosphorylation of ser-59 in the lck unique N-terminal region. *Proc. Nat. Acad. Sci.* 92: 12338–12342, 1995.

[37632] 11184. Vadlamudi, R. K.; Joung, I.; Strominger, J. L.; Shin, J.: p62, a phosphotyrosine-independent ligand of the SH2 domain of p56-lck, belongs to a new class of ubiquitin-binding proteins. *J. Biol. Chem.* 271: 20235–20237, 1996.

[37633] 11185. Akbar, G. K. M.; Dasari, V. R.; Webb, T. E.; Ayyanathan, K.; Pillarisetti, K.; Sandhu, A. K.; Athwal, R. S.; Daniel, J. L.; Ashby, B.; Barnard, E. A.; Kunapuli, S. P.: Molecular cloning of a novel P2 purinoceptor from human erythroleukemia cells. *J. Biol. Chem.* 271: 18363–18367, 1996.

[37634] 11186. Chen, X.-S.; Sheller, J. R.; Johnson, E. N.; Funk, C. D.: Role of leukotrienes revealed by targeted disruption of 5-lipoxygenase gene. *Nature* 372: 179–182, 1994.

[37635] 11187. Devchand, P. R.; Keller, H.; Peters, J. M.; Vazquez,

M.; Gonzalez, F. J.; Wahli, W.: The PPAR alpha-leukotriene B₄ pathway to inflammation control. *Nature* 384: 39–43, 1996.

[37636] 11188. Kato, K.; Yokomizo, T.; Izumi, T.; Shimizu, T.: Cell-specific transcriptional regulation of human leukotriene B₄ receptor gene. *J. Exp. Med.* 192: 413–420, 2000.

[37637] 11189. Owman, C.; Nilsson, C.; Lolait, S. J.: Cloning of cDNA encoding a putative chemoattractant receptor. *Genomics* 37: 187–194, 1996.

[37638] 11190. Samuelsson, B.; Dahlen, S. E.; Lindgren, J. A.; Rouzer, C. A.; Serhan, C. N.: Leukotrienes and lipoxins: structures, biosynthesis, and biological effects. *Science* 237: 1171–1176, 1987.

[37639] 11191. Yokomizo, T.; Izumi, T.; Chang, K.; Takuwa, Y.; Shimizu, T.: A G-protein-coupled receptor for leukotriene B₄ that mediates chemotaxis. *Nature* 387: 620–624, 1997.

[37640] 11192. Kubo, Y.; Reuveny, E.; Slesinger, P. A.; Jan, Y. N.; Jan, L. Y.: Primary structure and functional expression of a rat G-protein-coupled muscarinic potassium channel. *Nature* 364: 802–806, 1993.

[37641] 11193. Schoots, O.; Voskoglou, T.; Van Tol, H. H. M.: Genomic organization and promoter analysis of the human G-protein-coupled K⁺ channel Kir3.1 (KCNJ3/HGIRK1).

Genomics 39: 279–288, 1997.

- [37642] 11194.Schoots, O.; Yue, K.-T.; MacDonald, J. F.; Hampson, D. R.; Nobrega, J. N.; Dixon, L. M.; Van Tol, H. H. M.: Cloning of a G protein-activated inwardly rectifying potassium channel from human cerebellum. *Molec.Brain Res.* 39: 23–30, 1996.
- [37643] 11195.Stoffel, M.; Espinosa, R., III; Powell, K. L.; Philipson, L. H.; Le Beau, M. M.; Bell, G. I.: Human G-protein-coupled inwardly rectifying potassium channel (GIRK1) gene (KCNJ3): localization to chromosome 2 and identification of a simple tandem repeat polymorphism. *Genomics* 21:254–256, 1994.
- [37644] 11196.Phillips, H. A.; Scheffer, I. E.; Berkovic, S. F.; Hollway, G.E.; Sutherland, G. R.; Mulley, J. C.: Localization of a gene for autosomal dominant nocturnal frontal lobe epilepsy to chromosome 20q13.2. *Nature Genet.* 10: 117–118, 1995.
- [37645] 11197.Phillips, H. A.; Scheffer, I. E.; Crossland, K. M.; Bhatta, K.P.; Fish, D. R.; Marsden, C. D.; Howell, S. J. L.; Stephenson, J.B. P.; Tolmie, J.; Plazzi, G.; Eeg-Olofsson, O.; Singh, R.; Lopes-Cendes, I.; Andermann, E.; Andermann, F.; Berkovic, S. F.; Mulley, J. C.: Autosomal dominant nocturnal frontal-lobe epilepsy: genetic heterogeneity and

evidence for a second locus at 15q24. *Am. J. Hum. Genet.* 63:1108–1116, 1998.

- [37646] 11198.D'Arcangelo, G.: Personal Communication. Nutley, N. J. 6/2/1995.
- [37647] 11199.D'Arcangelo, G.; Homayouni, R.; Keshvara, L.; Rice, D. S.; Sheldon,M.; Curran, T.: Reelin is a ligand for lipoprotein receptors. *Neuron* 24:471–479, 1999.
- [37648] 11200.D'Arcangelo, G.; Miao, G. G.; Chen, S.–C.; Soares, H. D.; Morgan,J. I.; Curran, T.: A protein related to extra-cellular matrix proteinsdeleted in the mouse mutant reeler. *Nature* 374: 719–723, 1995.
- [37649] 11201.DeSilva, U.; D'Arcangelo, G.; Braden, V. V.; Chen, J.; Miao, G.G.; Curran, T.; Green, E. D.: The human reelin gene: isolation, sequencing,and mapping on chromosome 7. *Genome Res.* 7: 157–164, 1997.
- [37650] 11202.Green, M. C.: Catalog of mutant genes and polymorphic loci.In:Lyon, M. F.; Searle, A. G.: *Genetic Variants and Strains of the LaboratoryMouse*. Oxford: Oxford Univ. Press (pub.) (2nd ed.): 1989.
- [37651] 11203.Hack, I.; Bancila, M.; Loulier, K.; Carroll, P.; Cremer, H.: Reelinis a detachment signal in tangential chain-migration during postnatalneurogenesis. *Nature Neurosci.* 5: 939–945, 2002.

- [37652] 11204.Hirotsune, S.; Takahara, T.; Sasaki, N.; Hirose, K.; Yoshiki, A.;Ohashi, T.; Kusakabe, M.; Murakami, Y.; Muramatsu, M.; Watanabe, S.;Nakao, K.; Katsuki, M.; Hayashizaki, Y.: The reeler gene encodes a protein with an EGF-like motif expressed by pioneer neurons. *Nature-Genet.* 10: 77–83, 1995.
- [37653] 11205.Hong, S. E.; Shugart, Y. Y.; Huang, D. T.; Al Shahan, S.; Grant,P. E.; Hourihane, J. O.; Martin, N. D. T.; Walsh, C. A.: Autosomalrecessive lissencephaly with cerebellar hypoplasia is associated with human RELN mutations. *Nature Genet.* 26: 93–96, 2000. Note: Erratum:*Nature Genet.* 27: 225 only, 2001.
- [37654] 11206.Hourihane, J. O.; Bennett, C. P.; Chaudhuri, R.; Robb, S. A.;Martin, N. D. T.: A sibship with a neuronal migration defect, cerebellarhypoplasia and congenital lymphedema. *Neuropediatrics* 24: 43–46,1993.
- [37655] 11207.Impagnatiello, F.; Guidotti, A. R.; Pesold, C.; Dwivedi, Y.; Caruncho,H.; Pisu, M. G.; Uzunov, D. P.; Smalheiser, N. R.; Davis, J. M.; Pandey,G. N.; Pappas, G. D.; Tueting, P.; Sharma, R. P.; Costa, E.: A decrease of reelin expression as a putative vulnerability factor in schizophrenia. *Proc.Nat. Acad. Sci.* 95: 15718–15723, 1998.

- [37656] 11208.Magdaleno, S.; Keshvara, L.; Curran, T.: Rescue of ataxia andpreplate splitting by ectopic expression of reelin in reeler mice. *Neuron* 33:573–586, 2002.
- [37657] 11209.Royaux, I.; Lambert de Rouvroit, C.; D'Arcangelo, G.; Demirov,D.; Goffinet, A. M.: Genomic organization of the mouse reelin gene. *Genomics* 46:240–250, 1997.
- [37658] 11210.Yip, J. W.; Yip, Y. P. L.; Nakajima, K.; Capriotti, C.: Reelincontrols position of autonomic neurons in the spinal cord. *Proc.Nat. Acad. Sci.* 97: 8612–8616, 2000.
- [37659] 11211.Cattaneo, M.; Lecchi, A.; Randi, A. M.; McGregor, J. L.; Mannucci,P. M.: Identification of a new congenital defect of platelet functioncharacterized by severe impairment of platelet responses to adenosinediphosphate. *Blood* 80: 2787–2796, 1992.
- [37660] 11212.Pawson, T.; Gish, G. D.: SH2 and SH3 domains: from structure tofunction. *Cell* 71: 359–362, 1992.
- [37661] 11213.Welsh, M.; Mares, J.; Karlsson, T.; Lavergne, C.; Breant, B.; Claesson–Welsh,L.: Shb is a ubiquitously expressed Src homology 2 protein. *Oncogene* 9:19–27, 1994.
- [37662] 11214.Yulug, I. G.; Hillermann, R.; Fisher, E. M. C.: The SHB adaptorprotein maps to human chromosome 9. *Genomics* 24: 615–617, 1994.

- [37663] 11215.Bansal-Pakala, P.; Jember, A. G.-H.; Croft, M.: Signaling through OX40 (CD134) breaks peripheral T-cell tolerance. *Nature Med.* 7:907-912, 2001.
- [37664] 11216.Birkeland, M. L.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N.A.; Barclay, A. N.: Gene structure and chromosomal localization of the mouse homologue of rat OX40 protein. *Europ. J. Immun.* 25: 926-930, 1995.
- [37665] 11217.Jember, A. G.-H.; Zuberi, R.; Liu, F.-T.; Croft, M.: Development of allergic inflammation in a murine model of asthma is dependent on the costimulatory receptor OX40. *J. Exp. Med.* 193: 387-392, 2001.
- [37666] 11218.Latza, U.; Durkop, H.; Schnittger, S.; Ringeling, J.; Eitelbach, F.; Hummel, M.; Fonatsch, C.; Stein, H.: The human OX40 homolog: cDNA structure, expression and chromosomal assignment of the ACT35 antigen. *Europ. J. Immun.* 24: 677-683, 1994.
- [37667] 11219.Anderson, D. W.; Probst, F. J.; Belyantseva, I. A.; Fridell, R.A.; Beyer, L.; Martin, D. M.; Wu, D.; Kachar, B.; Friedman, T. B.; Raphael, Y.; Camper, S. A.: The motor and tail regions of myosin XV are critical for normal structure and function of auditory and vestibular hair cells. *Hum. Molec. Genet.* 9: 1729-1738, 2000.
- [37668] 11220.Probst, F. J.; Fridell, R. A.; Raphael, Y.; Saunders, T.

L.; Wang,A.; Liang, Y.; Morell, R. J.; Touchman, J. W.; Lyons, R. H.; Noben–Trauth,K.; Friedman, T. B.; Camper, S. A.: Correction of deafness in shaker–2mice by an unconventional myosin in a BAC transgene. *Science* 280:1444–1447, 1998.

[37669] 11221.Wang, A.; Liang, Y.; Fridell, R. A.; Probst, F. J.; Wilcox, E.R.; Touchman, J. W.; Morton, C. C.; Morell, R. J.; Noben–Trauth, K.;Camper, S. A.; Friedman, T. B.: Association of unconventional myosinMYO15 mutations with human nonsyndromic deafness DFNB3. *Science* 280:1447–1451, 1998.

[37670] 11222.Wilton, S. D.; Lim, L.; Dorosz, S. D.; Gunn, H. C.; Eyre, H. J.;Callen, D. F.; Laing, N. G.: Assignment of the human alpha–tropomyosin gene TPM4 to band 19p13.1 by fluorescence in situ hybridization. *Cytogenet.Cell Genet.* 72: 294–296, 1996.

[37671] 11223.Lazzaro, M. A.; Picketts, D. J.: Cloning and characterizationof the murine Imitation Switch (ISWI) genes: differential expressionpatterns suggest distinct developmental roles for Snf2h and Snf2l. *J.Neurochem.* 77: 1145–1156, 2001.

[37672] 11224.Okabe, I.; Bailey, L. C.; Attree, O.; Srinivasan, S.; Perkel, J.M.; Laurent, B. C.; Carlson, M.; Nelson, D. L.;

Nussbaum, R. L.:Cloning of human and bovine homologs of SNF2/SWI2: a global activatorof transcription in yeast *S. cerevisiae*. *Nucleic Acids Res.* 20:4649–4655, 1992.

[37673] 11225.Stahl, W. L.; Eakin, T. J.; Owens, J. W. M., Jr.; Breining, J.F.; Filuk, P. E.; Anderson, W. R.: Plasma membrane $\text{Ca}(2+)$ –ATPaseisoforms: distribution of mRNAs in rat brain by in situ hybridization.*Molec. Brain Res.* 16: 223–231, 1992.

[37674] 11226.De Plaen, E.; Arden, K.; Traversari, C.; Gaforio, J. J.; Szikora,J.–P.; De Smet, C.; Brasseur, F.; van der Bruggen, P.; Lethe, B.;Lurquin, C.; Brasseur, R.; Chomez, P.; De Backer, O.; Cavenee, W.;Boon, T.: Structure, chromosomal localization, and expression of12 genes of the MAGE family. *Immunogenetics* 40: 360–369, 1994.

[37675] 11227.Rogner, U. C.; Wilke, K.; Steck, E.; Korn, B.; Poustka, A.: Themelanoma antigen gene (MAGE) family is clustered in the chromosomalband Xq28. *Genomics* 29: 725–731, 1995.

[37676] 11228.Chakarova, C.; Wehnert, M. S.; Uhl, K.; Sakthivel, S.; Vosberg,H.–P.; van der Ven, P. F. M.; Furst, D. O.: Genomic structure andfine mapping of the two human filamin gene paralogues FLNB and FLNCand comparative analysis of the filamin gene family. *Hum. Genet.* 107:597–611, 2000.

- [37677] 11229.Fink, J. M.; Dobyns, W. B.; Guerrini, R.; Hirsch, B. A.: Identification of a duplication of Xq28 associated with bilateral periventricular nodular heterotopia. *Am. J. Hum. Genet.* 61: 379–387, 1997.
- [37678] 11230.Fox, J. W.; Lamperti, E. D.; Eksioglu, Y. Z.; Hong, S. E.; Feng, Y.; Graham, D. A.; Scheffer, I. E.; Dobyns, W. B.; Hirsch, B. A.; Radtke, R. A.; Berkovic, S. F.; Huttenlocher, P. R.; Walsh, C. A.: Mutations in filamin 1 prevent migration of cerebral cortical neurons in human periventricular heterotopia. *Neuron* 21: 1315–1325, 1998.
- [37679] 11231.Gariboldi, M.; Maestrini, E.; Canzian, F.; Manenti, G.; De Gregorio, L.; Rivella, S.; Chatterjee, A.; Herman, G. E.; Archidiacono, N.; Antonacci, R.; Pierotti, M. A.; Dragani, T. A.; Toniolo, D.: Comparative mapping of the actin-binding protein 280 genes in human and mouse. *Genomics* 21:428–430, 1994.
- [37680] 11232.Bashir, R.; Britton, S.; Strachan, T.; Keers, S.; Vafiadaki, E.; Lako, M.; Richard, I.; Marchand, S.; Bourg, N.; Argov, Z.; Sadeh, M.; Mahjneh, I.; Marconi, G.; Passos-Bueno, M. R.; Moreira, E. S.; Zatz, M.; Beckmann, J. S.; Bushby, K.: A gene related to *Caenorhabditis elegans* spermatogenesis factor *fer-1* is mutated in limb-girdle muscular dystrophy type 2B. *Nature Genet.* 20: 37–42, 1998.

- [37681] 11233.Liu, J.; Aoki, M.; Illa, I.; Wu, C.; Fardeau, M.; Angelini, C.;Serrano, C.; Urtizberea, J. A.; Hentati, F.; Ben Hamida, M.; Bohlega,S.; Culper, E. J.; Amato, A. A.; Bossie, K.; Oeltjen, J.; Bejaoui,K.; McKenna–Yasek, D.; Hosler, B. A.; Schurr, E.; Arahata, K.; deJong, P. J.; Brown, R. H., Jr.: Dysferlin, a novel skeletal musclegene, is mutated in Miyoshi myopathy and limb girdle muscular dystrophy. NatureGenet. 20: 31–36, 1998.
- [37682] 11234.Aghai, E.; Yaniv, I.; David, M.: Factor XI deficiency in an ArabMoslem family in Israel. Scand. J. Haemat. 32: 327–331, 1984.
- [37683] 11235.Asakai, R.; Chung, D. W.; Davie, E. W.; Seligsohn, U.: FactorXI deficiency in Ashkenazi Jews in Israel. New Eng. J. Med. 325:153–158, 1991.
- [37684] 11236.Asakai, R.; Chung, D. W.; Ratnoff, O. D.; Davie, E. W.: FactorXI (plasma thromboplastin antecedent) deficiency in Ashkenazi Jewsis a bleeding disorder that can result from three types of point mutations. Proc.Nat. Acad. Sci. 86: 7667–7671, 1989.
- [37685] 11237.Asakai, R.; Davie, E. W.; Chung, D. W.: Organization of the genefor human factor XI. Biochemistry 26: 7221–7228, 1987.
- [37686] 11238.Bauduer, F.; Dupreuilh, F.; Ducout, L.; Marti, B.:

Factor XI deficiency in the French Basque country.

Haemophilia 5: 187–190, 1999.

[37687] 11239. Bertina, R. M.; Veltkamp, J. J.: A genetic variant of factor XI with decreased capacity for Ca ion binding. Brit. J. Haemat. 42:623–635, 1979.

[37688] 11240. Biggs, R.; MacFarlane, R. G.: Human Blood Coagulation and Its Disorders. Oxford: Blackwell (pub.) (3rd ed.): 1962.

[37689] 11241. Bolton-Maggs, P. H. B.: Factor XI deficiency. Bailliere's Clin. Haemat. 9: 355–368, 1996.

[37690] 11242. Braunstein, K. M.; Noyes, C. M.; Griffith, M. J.; Lundblad, R. L.; Roberts, H. R.: Characterization of the defect in activation of factor IX by human factor XIa. J. Clin. Invest. 68:1420–1426, 1981.

[37691] 11243. Buetow, K. H.; Shiang, R.; Yang, P.; Nakamura, Y.; Lathrop, G. M.; White, R.; Wasmuth, J. J.; Wood, S.; Berdahl, L. D.; Leysens, N. J.: A detailed multipoint map of human chromosome 4 provides evidence for linkage heterogeneity and position-specific recombination rates. Am. J. Hum. Genet. 48: 911–925, 1991.

[37692] 11244. Clarkson, K.; Rosenfeld, B.; Fair, J.; Klein, A.; Bell, W.: Factor XI deficiency acquired by liver transplantation. Ann. Intern. Med. 115:877–879, 1991.

- [37693] 11245.Dodds, W. J.; Kull, J. E.: Canine factor XI (plasma thromboplastinantecedent) deficiency. J. Lab. Clin. Med. 78: 746–752, 1971.
- [37694] 11246.Dzik, W. H.; Arkin, C. F.; Jenkins, R. L.: Transfer of congenital factor XI deficiency from a donor to a recipient by liver transplantation.(Letter) New Eng. J. Med. 316: 1217–1218, 1987.
- [37695] 11247.Fujikawa, K.; Chung, D. W.; Hendrickson, L. E.; Davie, E. W.:Amino acid sequence of human factor XI, a blood coagulation factor with four tandem repeats that are highly homologous with plasma prekallikrein. Biochemistry 25:2417–2424, 1986.
- [37696] 11248.Imanaka, Y.; Lal, K.; Nishimura, T.; Bolton–Maggs, P. H. B.; Tuddenham, E. G. D.; McVey, J. H.: Identification of two novel mutations in non–Jewish factor XI deficiency. Brit. J. Haemat. 90: 916–920, 1995.
- [37697] 11249.Kato, A.; Asakai, R.; Davie, E. W.; Aoki, N.: Factor XI gene(F11) is located on the distal end of the long arm of human chromosome 4. Cytogenet. Cell Genet. 52: 77–78, 1989.
- [37698] 11250.Litz, C. E.; Swaim, W. R.; Dalmaso, A. P.: Factor XI deficiency:genetic and clinical studies of a single kindred. Am. J. Hemat. 28:8–12, 1988.

- [37699] 11251. Mannhalter, C.; Hellstern, P.; Deutsch, E.: Identification of a defective factor XI cross-reacting material in a factor XI-deficient patient. *Blood* 70: 31–37, 1987.
- [37700] 11252. Meijers, J. C. M.; Davie, E. W.; Chung, D. W.: Expression of human blood coagulation factor XI: characterization of the defective factor XI type III deficiency. *Blood* 79: 1435–1440, 1992.
- [37701] 11253. Mitchell, M.; Cutler, J.; Thompson, S.; Moore, G.; Rees, E. J.; Smith, M.; Savidge, G.; Alhaq, A.: Heterozygous factor XI deficiency associated with three novel mutations. *Brit. J. Haemat.* 107: 763–765, 1999.
- [37702] 11254. Muir, W. A.; Ratnoff, O. D.: The prevalence of plasma thromboplastin antecedent (PTA factor XI) deficiency. *Blood* 44: 569–570, 1974.
- [37703] 11255. Niskanen, E. O.; Saito, H.; Cline, M. J.: Plasma thromboplastin antecedent (factor XI) deficiency in a black family. *Arch. Intern. Med.* 141: 936–941, 1981.
- [37704] 11256. Peretz, U.; Zivelin, A.; Usher, S.; Eichel, R.; Seligsohn, U.: Identification of a new mutation in the factor XI gene of an Ashkenazi-Jew with severe factor XI deficiency. (Abstract) *Blood* 82 (suppl. 1): 66a only, 1993.
- [37705] 11257. Pugh, R. E.; McVey, J. H.; Tuddenham, E. G. D.; Hancock, J. F.: Six point mutations that cause factor XI de-

ficiency. Blood 85:1509–1516, 1995.

- [37706] 11258.Ragni, M. V.; Sinha, D.; Seaman, F.; Lewis, J. H.; Spero, J. A.;Walsh, P. N.: Comparison of bleeding tendency, factor XI coagulantactivity, and factor XI antigen in 25 factor XI-deficient kindreds. Blood 65:719–724, 1985.
- [37707] 11259.Rapaport, S. I.; Proctor, R. R.; Patch, M. J.; Yettra, M.: Themode of inheritance of PTA deficiency: evidence for the existenceof major PTA deficiency and minor PTA deficiency. Blood 18: 149–165,1961.
- [37708] 11260.Rimon, A.; Schiffman, S.; Feinstein, D. I.; Rapaport, S. I.:Factor–XI activity and factor–XI antigen in homozygous and heterozygousfactor–XI deficiency. Blood 48: 165–174, 1976.
- [37709] 11261.Foresti, V.; Ferrari, C.: Central hypothyroidism: isolated thyrotropin–releasinghormone deficiency or resistance of pituitary thyrotropes?. (Letter) J.Endocr. Invest. 8: 577 only, 1985.
- [37710] 11262.Katakami, H.; Kato, Y.; Inada, M.; Imura, H.: Hypothalamic hypothyroidismdue to isolated thyrotropin–releasing hormone (TRH) deficiency. J.Endocr. Invest. 7: 231–233, 1984.
- [37711] 11263.Niimi, H.; Inomata, H.; Sasaki, N.; Nakajima, H.: Congenital isolatedthyrotrophin releasing hormone defi–

ciency. Arch. Dis. Child. 57:877–878, 1982.

[37712] 11264. Roller, M. L.; Camper, S. A.: Localization of the thyrotropin–releasing hormone gene, Trh, on mouse chromosome 6. Mammalian Genome 6: 443–444, 1995.

[37713] 11265. Yamada, M.; Radovick, S.; Wondisford, F. E.; Nakayama, Y.; Weintraub, B. D.; Wilber, J. F.: Cloning and structure of human genomic DNA and hypothalamic cDNA encoding human preprothyrotropin–releasing hormone. Molec. Endocr. 4: 551–556, 1990.

[37714] 11266. Yamada, M.; Satoh, T.; Monden, T.; Mori, M.: Assignment of the thyrotropin–releasing hormone gene (TRH) to human chromosome 3q13.3–q21 by in situ hybridization. Cytogenet. Cell Genet. 87: 275 only, 1999.

[37715] 11267. Yamada, M.; Wondisford, F. E.; Radovick, S.; Nakayama, Y.; Weintraub, B. D.; Wilber, J. F.: Assignment of human preprothyrotropin–releasing hormone (TRH) gene to chromosome 3. Somat. Cell Molec. Genet. 17:97–100, 1991.

[37716] 11268. McDermott, J. C.; Cardoso, M. C.; Yu, Y.–T.; Andres, V.; Leifer, D.; Krainc, D.; Lipton, S. A.; Nadal-Ginard, B.: hMEF2C gene encodes skeletal muscle– and brain–specific transcription factors. Molec. Cell. Biol. 13: 2564–2577, 1993.

- [37717] 11269. Anderson, G. J.; Murphy, T. L.; Cowley, L.; Evans, B. A.; Halliday, J. W.; McLaren, G. D.: Mapping the gene for sex-linked anemia: an inherited defect of intestinal iron absorption in the mouse. *Genomics* 48:34–39, 1998.
- [37718] 11270. Bannerman, R. M.: Genetic defects of iron transport. *Fed. Proc.* 35:2281–2285, 1976.
- [37719] 11271. Edwards, J. A.; Hoke, J. E.; Mattioli, M.; Reichlin, M.: Ferritin distribution and synthesis in sex-linked anemia. *J. Lab. Clin. Med.* 90:68–76, 1977.
- [37720] 11272. Falconer, D. S.; Isaacson, J. H.: The genetics of sex-linked anaemia in the mouse. *Genet. Res.* 3: 248–250, 1962.
- [37721] 11273. Grewal, M. S.: A sex-linked anaemia in the mouse. *Genet. Res.* 3:238–247, 1962.
- [37722] 11274. Lee, G. R.; Nacht, S.; Lukens, J. N.; Cartwright, G. E.: Iron metabolism in copper-deficient swine. *J. Clin. Invest.* 47: 2058–2069, 1968.
- [37723] 11275. Vulpe, C. D.; Kuo, Y.-M.; Murphy, T. L.; Cowley, L.; Askwith, C.; Libina, N.; Gitschier, J.; Anderson, G. J.: Hephaestin, a ceruloplasmin homologue implicated in intestinal iron transport, is defective in the sla mouse. *Nature Genet.* 21: 195–199, 1999.
- [37724] 11276. Karumanchi, S. A.; Jha, V.; Ramchandran, R.; Kari-

haloo, A.; Tsiokas, L.; Chan, B.; Dhanabal, M.; Hanai, J.; Venkataraman, G.; Shriver, Z.; Keiser, N.; Kalluri, R.; and 9 others: Cell surface glypicans are low-affinity endostatin receptors. *Molec. Cell* 7: 811–822, 2001.

[37725] 11277. Veugelers, M.; Vermeesch, J.; Watanabe, K.; Yamaguchi, Y.; Marynen, P.; David, G.: GPC4, the gene for human K-glypican, flanks GPC3 on Xq26: deletion of the GPC3–GPC4 gene cluster in one family with Simpson–Golabi–Behrmels syndrome. *Genomics* 53: 1–11, 1998.

[37726] 11278. Emes, R. D.; Ponting, C. P.: A new sequence motif linking lissencephaly, Treacher Collins and oral-facial-digital type 1 syndromes, microtubule dynamics and cell migration. *Hum. Molec. Genet.* 10: 2813–2820, 2001.

[37727] 11279. Copley, L. M.; Zhao, W. D.; Kopacz, K.; Herman, G. E.; Kioschis, P.; Poustka, A.; Taudien, S.; Platzer, M.: Exclusion of mutations in the MTMR1 gene as a frequent cause of X-linked myotubular myopathy. (Letter) *Am. J. Med. Genet.* 107: 256–258, 2002.

[37728] 11280. Kioschis, P.; Wiemann, S.; Heiss, N. S.; Francis, F.; Gotz, C.; Poustka, A.; Taudien, S.; Platzer, M.; Wiehe, T.; Beckmann, G.; Weber, J.; Nordsiek, G.; Rosenthal, A.: Genomic organization of a 225-kb region in Xq28 containing the gene for X-linked myotubular myopathy (MTM1) and a

related gene (MTMR1). Genomics 54: 256–266, 1998.

[37729] 11281.Laporte, J.; Blondeau, F.; Buj-Bello, A.; Tentler, D.; Kretz, C.;Dahl, N.; Mandel, J.–L.: Characterization of the myotubularin dualspecificity phosphatase gene family from yeast to human. Hum. Molec.Genet. 7: 1703–1712, 1998.

- [37730] 11282. Gorlin, J. B.; Henske, E.; Warren, S. T.; Kunst, C. B.; D'Urso, M.; Palmieri, G.; Hartwig, J. H.; Bruns, G.; Kwiatkowski, D. J.: Actin-binding protein (ABP-280) filamin gene (FLN) maps telomeric to the color vision locus (R/GCP) and centromeric to G6PD in Xq28. *Genomics* 17:496-498, 1993.
- [37731] 11283. Jager, R. J.; Anvret, M.; Hall, K.; Scherer, G.: A human XY female with a frame shift mutation in the candidate testis-determining gene SRY. *Nature* 348: 452-454, 1990.
- [37732] 11284. Koopman, P.; Gubbay, J.; Vivian, N.; Goodfellow, P.; Lovell-Badge, R.: Male development of chromosomally female mice transgenic for Sry. *Nature* 351: 117-121, 1991.
- [37733] 11285. Koopman, P.; Munsterberg, A.; Capel, B.; Vivian, N.; Lovell-Badge, R.: Expression of a candidate sex-determining gene during mouse testis differentiation. *Nature* 348: 450-452, 1990.
- [37734] 11286. McElreavey, K. D.; Vilain, E.; Boucek, C.; Vidaud, M.; Jaubert, F.; Richaud, F.; Fellous, M.: XY sex reversal associated with a nonsense mutation in SRY. *Genomics* 13: 838-840, 1992.
- [37735] 11287. Page, D. C.; Mosher, R.; Simpson, E. M.; Fisher, E. M. C.; Mardon, G.; Pollack, J.; McGillivray, B.; de la

Chapelle, A.; Brown, L. G.: The sex-determining region of the human Y chromosome encodes a fingerprotein. *Cell* 51: 1091–1104, 1987.

[37736] 11288. Taylor, H.; Barter, R. H.; Jacobson, C. B.: Neoplasms of dysgenetic gonads. *Am. J. Obstet. Gynec.* 96: 816–823, 1966.

[37737] 11289. Vilain, E.; McElreavey, K.; Jaubert, F.; Raymond, J.-P.; Richaud, F.; Fellous, M.: Familial case with sequence variant in the testis-determining region associated with two sex phenotypes. *Am. J. Hum. Genet.* 50:1008–1011, 1992.

[37738] 11290. Gorlin, J. B.; Yamin, R.; Egan, S.; Stewart, M.; Stos-
sel, T. P.; Kwiatkowski, D. J.; Hartwig, J. H.: Human en-
dothelial actin-binding protein (ABP-280, nonmuscle fil-
amin): a molecular leaf spring. *J. Cell Biol.* 111:
1089–1105, 1990.

[37739] 11291. Huttenlocher, P. R.; Taravath, S.; Mojtahedi, S.:
Periventricular heterotopia and epilepsy. *Neurology* 44:
51–55, 1994.

[37740] 11292. Kunst, C. B.; Henske, E.; Hartwig, J. H.;
Kwiatkowski, D. J.; D'Urso, M.; Bruns, G.; Warren, S. T.;
Gorlin, J. B.: The dystrophin-like actin binding protein 280
gene maps between DXS52 and G6PD overlapping the

Emery–Dreifuss muscular dystrophy locus. (Abstract) Am. J. Hum. Genet. 51: A21, 1992.

[37741] 11293. Maestrini, E.; Patrosso, C.; Mancini, M.; Rivella, S.; Rocchi, M.; Repetto, M.; Villa, A.; Frattini, A.; Zoppe, M.; Vezzoni, P.; Toniolo, D.: Mapping of two genes encoding isoforms of the actin-binding protein ABP-280, a dystrophin like protein, to Xq28 and to chromosome 7. Hum. Molec. Genet. 2: 761–766, 1993.

[37742] 11294. Maestrini, E.; Rivella, S.; Tribioli, C.; Purtilo, D.; Rocchi, M.; Archidiacono, N.; Toniolo, D.: Probes for CpG islands on the distal long arm of the human X chromosome are clustered in Xq24 and Xq28. Genomics 8: 664–670, 1990.

[37743] 11295. Patrosso, M. C.; Repetto, M.; Villa, A.; Milanese, L.; Frattini, A.; Faranda, S.; Mancini, M.; Maestrini, E.; Toniolo, D.; Vezzoni, P.: The exon–intron organization of the human X-linked gene (FLN1) encoding actin-binding protein 280. Genomics 21: 71–76, 1994.

[37744] 11296. Sheen, V. L.; Dixon, P. H.; Fox, J. W.; Hong, S. E.; Kinton, L.; Sisodiya, S. M.; Duncan, J. S.; Dubeau, F.; Schaffer, I. E.; Schachter, S. C.; Wilner, A.; Henchy, R.; and 18 others: Mutations in the X-linked filamin 1 gene cause periventricular nodular heterotopia in males as well as in

females. Hum. Molec. Genet. 10: 1775–1783,2001.

[37745] 11297.Small, K.; Wagener, M.; Warren, S. T.: Isolation and characterization of the complete mouse emerin gene.

Mammalian Genome 8: 337–341,1997.

[37746] 11298.Vadlamudi, R. K.; Li, F.; Adam, L.; Nguyen, D.; Ohta, Y.; Stossel,T. P.; Kumar, R.: Filamin is essential in actin cytoskeletal assembly mediated by p21-activated kinase 1. Nature Cell Biol. 4: 681–690,2002.

[37747] 11299.Faranda, S.; Frattini, A.; Vezzoni, P.: The human genes encodingrenin-binding protein and host cell factor are closely linked in Xq28and transcribed in the same direction. Gene 155: 237–239, 1995.

[37748] 11300.Frattini, A.; Chatterjee, A.; Faranda, S.; Sacco, M. G.; Villa,A.; Herman, G. E.; Vezzoni, P.: The chromosome localization and theHCF repeats of the human host cell factor gene (HCFC1) are conserved in the mouse homologue. Genomics 32: 277–280, 1996.

[37749] 11301.Frattini, A.; Faranda, S.; Redolfi, E.; Zucchi, I.; Villa, A.;Patrosso, M. C.; Strina, D.; Susani, L.; Vezzoni, P.: Genomic organization of the human VP16 accessory protein, a housekeeping gene (HCFC1) mapping to Xq28. Genomics 23: 30–35, 1994.

[37750] 11302.Wilson, A. C.; LaMarco, K.; Peterson, M. G.; Herr,

W.: The VP16accessory protein HCF is a family of polypeptides processed from a large precursor protein. *Cell* 74: 115–125, 1993.

[37751] 11303. Wilson, A. C.; Parrish, J. E.; Massa, H. F.; Nelson, D. L.; Trask, B. J.; Herr, W.: The gene encoding the VP16–accessory protein HCF(HCFC1) resides in human Xq28 and is highly expressed in fetal tissues and the adult kidney. *Genomics* 25: 462–468, 1995.

[37752] 11304. Zoppe, M.; Frattini, A.; Faranda, S.; Vezzoni, P.: The complete sequence of the host cell factor 1 (HCFC1) gene and its promoter: a role for YY1 transcription factor in the regulation of its expression. *Genomics* 34: 85–91, 1996.

[37753] 11305. Maestrini, E.; Tamagnone, L.; Longati, P.; Cremona, O.; Gulisano, M.; Bione, S.; Tamanini, F.; Neel, B. G.; Toniolo, D.; Comoglio, P. M.: A family of transmembrane proteins with homology to the MET–hepatocyte growth factor receptor. *Proc. Nat. Acad. Sci.* 93: 674–678, 1996.

[37754] 11306. Tamagnone, L.; Artigiani, S.; Chen, H.; He, Z.; Ming, G.; Song, H.; Chedotal, A.; Winberg, M. L.; Goodman, C. S.; Poo, M.; Tessier-Lavigne, M.; Comoglio, P. M.: Plexins are a large family of receptors for transmembrane, secreted, and GPI–anchored semaphorins in vertebrates. *Cell* 99: 71–80, 1999.

- [37755] 11307.Becker, P. E.: Two new families of benign sex-linked recessivemuscular dystrophy. *Rev. Canad. Biol.* 21: 551–566, 1962.
- [37756] 11308.Becker, P. E.: Eine neue X-chromosomale Muskeldystrophie. *ActaPsychiat. Neurol. Scand.* 193: 427, 1955.
- [37757] 11309.Becker, P. E.: Neue Ergebnisse der Genetik der Muskeldystrophien. *ActaGenet. Statist. Med.* 7: 303–310, 1957.
- [37758] 11310.Bushby, K. M. D.; Cleghorn, N. J.; Curtis, A.; Haggerty, I. D.;Nicholson, L. V. B.; Johnson, M. A.; Harris, J. B.; Bhattacharya,S. S.: Identification of a mutation in the promoter region of thedystrophin gene in a patient with atypical Becker muscular dystrophy. *Hum.Genet.* 88: 195–199, 1991.
- [37759] 11311.Doriguzzi, C.; Palmucci, L.; Mongini, T.; Chiado-Piat, L.; Restagno,G.; Ferrone, M.: Exercise intolerance and recurrent myoglobinuriaas the only expression of Xp21 Becker type muscular dystrophy. *J.Neurol.* 240: 269–271, 1993.
- [37760] 11312.England, S. B.; Nicholson, L. V. B.; Johnson, M. A.; Forrest,S. M.; Love, D. R.; Zubrzycka-Gaarn, E. E.; Bulman, D. E.; Harris,J. B.; Davies, K. E.: Very mild muscular dystro–

phy associated with the deletion of 46% dystrophin. *Nature* 343: 180–182, 1990.

[37761] 11313. Derry, J. M. J.; Barnard, P. J.: Physical linkage of the A-raf-1, properdin, synapsin I, and TIMP genes on the human and mouse X chromosomes. *Genomics* 12:632–638, 1992.

[37762] 11314. Aronsson, F. C.; Magnusson, P.; Andersson, B.; Karsten, S. L.; Shibasaki, Y.; Lendon, C. L.; Goate, A. M.; Brookes, A. J: The NIK protein kinase and C17orf1 genes: chromosomal mapping, gene structures and mutational screening in frontotemporal dementia and parkinsonism-linked to chromosome 17. *Hum. Genet.* 103: 340–345, 1998.

[37763] 11315. Malinin, N. L.; Boldin, M. P.; Kovalenko, A. V.; Wallach, D.: MAP3K-related kinase involved in NF- κ B induction by TNF, CD95 and IL-1. *Nature* 385: 540–544, 1997.

[37764] 11316. Shinkura, R.; Kitada, K.; Matsuda, F.; Tashiro, K.; Ikuta, K.; Suzuki, M.; Kogishi, K.; Serikawa, T.; Honjo, T.: Alymphoplasia is caused by a point mutation in the mouse gene encoding Nf- κ B-inducing kinase. *Nature Genet.* 22: 74–77, 1999.

[37765] 11317. Smith, C.; Andreakos, E.; Crawley, J. B.; Brennan, F.

M.; Feldmann, M.; Foxwell, B. M. J.: NF-kappa-B-inducing kinase is dispensable for activation of NF-kappa-B in inflammatory settings but essential for lymphotoxin beta receptor activation of NF-kappa-B in primary human fibroblasts. *J. Immun.* 167: 5895–5903, 2001.

[37766] 11318. Yin, L.; Wu, L.; Wesche, H.; Arthur, C. D.; White, J. M.; Goeddel, D. V.; Schreiber, R. D.: Defective lymphotoxin-beta receptor-induced NF-kappa-B transcriptional activity in NIK-deficient mice. *Science* 291: 2162–2165, 2001.

[37767] 11319. Aronsson, F. C.; Magnusson, P.; Andersson, B.; Karsten, S. L.; Shibasaki, Y.; Lendon, C. L.; Goate, A. M.; Brookes, A. J.: The NIK protein kinase and C17orf1 genes: chromosomal mapping, gene structures and mutational screening in frontotemporal dementia and parkinsonism-linked to chromosome 17. *Hum. Genet.* 103: 340–345, 1998.

[37768] 11320. Yayoshi-Yamamoto, S.; Taniuchi, I.; Watanabe, T.: FRL, a novel formin-related protein, binds to Rac and regulates cell motility and survival of macrophages. *Molec. Cell. Biol.* 20: 6872–6881, 2000.

[37769] 11321. Gress, T. M.; Muller-Pillasch, F.; Geng, M.; Zimmerhackl, F.; Zehetner, G.; Friess, H.; Buchler, M.; Adler, G.;

Lehrach, H.: A pancreaticcancer-specific expression profile. *Oncogene* 13: 1819–1830, 1996.

[37770] 11322.Muller-Pillasch, F.; Wallrapp, C.; Lacher, U.; Friess, H.; Buchler,M.; Adler, G.; Gress, T. M.: Identification of a new tumour-associatedantigen TM4SF5 and its expression in human cancer. *Gene* 208: 25–30,1998.

[37771] 11323.Borggreffe, T.; Masat, L.; Wabl, M.; Riwar, B.; Cattoretti, G.;Jessberger, R.: Cellular, intracellular, and developmental expressionpatterns of murine SWAP-70. *Europ. J. Immun.* 29: 1812–1822, 1999.

[37772] 11324.Borggreffe, T.; Wabl, M.; Akhmedov, A. T.; Jessberger, R.: A B-cell-specificDNA recombination complex. *J. Biol. Chem.* 273: 17025–17035, 1998.

[37773] 11325.Masat, L.; Caldwell, J.; Armstrong, R.; Khoshnevisan, H.; Jessberger,R.; Herndier, B.; Wabl, M.; Ferrick, D.: Association of SWAP-70 withthe B cell antigen receptor complex. *Proc. Nat. Acad. Sci.* 97: 2180–2184,2000.

[37774] 11326.Masat, L.; Liddell, R. A.; Mock, B. A.; Kuo, W.-L.; Jessberger,R.; Wabl, M.; Morse, H. C., III: Mapping of the SWAP70 gene to mousechromosome 7 and human chromosome 11p15. *Immunogenetics* 51: 16–19,2000.

[37775] 11327.Shinohara, M.; Terada, Y.; Iwamatsu, A.; Shinohara, A.; Mochizuki,N.; Higuchi, M.; Gotoh, Y.; Ihara, S.; Nagata,

S.; Itoh, H.; Fukui, Y.; Jessberger, R.: SWAP-70 is a guanine-nucleotide-exchange factor that mediates signalling of membrane ruffling. *Nature* 416: 759–763, 2002.

[37776] 11328. Miura, K.; Suzuki, K.; Tokino, T.; Isomura, M.; Inazawa, J.; Matsuno, S.; Nakamura, Y.: Detailed deletion mapping in squamous cell carcinomas of the esophagus narrows a region containing a putative tumor suppressor-gene to about 200 kilobases on distal chromosome 9q. *Cancer Res.* 56:1629–1634, 1996.

[37777] 11329. Nishiwaki, T.; Daigo, Y.; Kawasoe, T.; Nakamura, Y.: Isolation and mutational analysis of a novel human cDNA, DEC1 (deleted in esophageal cancer 1), derived from the tumor suppressor locus in 9q32. *Genes Chromosomes Cancer* 27: 169–176, 2000.

[37778] 11330. Nemoto, Y.; Yamamoto, T.; Takada, S.; Matsui, Y.; Obinata, M.: Antisense RNA of the latent period gene (MER5) inhibits the differentiation of murine erythroleukemia cells. *Gene* 91: 261–265, 1990.

[37779] 11331. Shih, S.-F.; Wu, Y.-H.; Hung, C.-H.; Yang, H.-Y.; Lin, J.-Y.: Abrin triggers cell death by inactivating a thiol-specific antioxidant protein. *J. Biol. Chem.* 276: 21870–21877, 2001.

[37780] 11332. Tsuji, K.; Copeland, N. G.; Jenkins, N. A.; Obinata,

M.: Mammalian antioxidant protein complements alkylhydroperoxide reductase (ahpC) mutation in *Escherichia coli*. *Biochem. J.* 307: 377–381, 1995.

- [37781] 11333. Wonsey, D. R.; Zeller, K. I.; Dang, C. V.: The c-Myc target gene PRDX3 is required for mitochondrial homeostasis and neoplastic transformation. *Proc. Nat. Acad. Sci.* 99: 6649–6654, 2002.
- [37782] 11334. Kamimoto, T.; Zama, T.; Aoki, R.; Muro, Y.; Hagiwara, M.: Identification of a novel kinesin-related protein, KRMP1, as a target for mitotic peptidyl-prolyl isomerase Pin1. *J. Biol. Chem.* 276: 37520–37528, 2001.
- [37783] 11335. Akagi, T.; Motegi, M.; Tamura, A.; Suzuki, R.; Hosokawa, Y.; Suzuki, H.; Ota, H.; Nakamura, S.; Morishima, Y.; Taniwaki, M.; Seto, M.: A novel gene, MALT1 at 18q21, is involved in t(11;18) (q21;q21) found in low-grade B-cell lymphoma of mucosa-associated lymphoid tissue. *Oncogene* 18: 5785–5794, 1999.
- [37784] 11336. Dierlamm, J.; Baens, M.; Wlodarska, I.; Stefanova-Ouzounova, M.; Hernandez, J. M.; Hossfeld, D. K.; De Wolf-Peeters, C.; Hagemeijer, A.; Van den Berghe, H.; Marynen, P.: The apoptosis inhibitor gene API2 and a novel 18q gene, MLT, are recurrently rearranged in the t(11;18)(q21;q21) associated with mucosa-associated

lymphoid tissue lymphomas. *Blood* 93: 3601–3609, 1999.

- [37785] 11337. Ott, G.; Katzenberger, T.; Greiner, A.; Kalla, J.; Rosenwald, A.; Heinrich, U.; Ott, M. M.; Muller-Hermelink, H. K.: The t(11;18)(q21;q21) chromosome translocation is a frequent and specific aberration in low-grade but not high-grade malignant non-Hodgkin's lymphomas of the mucosa-associated lymphoid tissue (MALT-) type. *Cancer Res.* 57:3944–3948, 1997.
- [37786] 11338. Sato, Y.; Akiyama, Y.; Tanizawa, T.; Shibata, T.; Saito, K.; Mori, S.; Kamiyama, R.; Yuasa, Y.: Molecular characterization of the genomic breakpoint junction in the t(11;18)(q21;q21) translocation of a gastric MALT lymphoma. *Biochem. Biophys. Res. Commun.* 280: 301–306, 2001.
- [37787] 11339. Valladeau, J.; Clair-Moninot, V.; Dezutter-Dambuyant, C.; Pin, J.-J.; Kissenpfennig, A.; Mattei, M.-G.; Ait-Yahia, S.; Bates, E. E. M.; Malissen, B.; Koch, F.; Fossiez, F.; Romani, N.; Lebecque, S.; Saeland, S.: Identification of mouse langerin/CD207 in Langerhans cells and some dendritic cells of lymphoid tissues. *J. Immun.* 168:782–792, 2002.
- [37788] 11340. Valladeau, J.; Ravel, O.; Dezutter-Dambuyant, C.; Moore, K.; Kleijmeer, M.; Liu, Y.; Duvert-Frances, V.; Vin-

cent, C.; Schmitt, D.; Davoust, J.; Caux, C.; Lebecque, S.; Saeland, S.: Langerin, a novel C-type lectin specific to Langerhans cells, is an endocytic receptor that induces the formation of Birbeck granules. *Immunity* 12: 71–81, 2000.

[37789] 11341. Reichenberger, E.; Tiziani, V.; Watanabe, S.; Park, L.; Ueki, Y.; Santanna, C.; Baur, S. T.; Shiang, R.; Grange, D. K.; Beighton, P.; Gardner, J.; Hamersma, H.; Sellars, S.; Ramesar, R.; Lidral, A. C.; Sommer, A.; Raposo do Amaral, C. M.; Gorlin, R. J.; Mulliken, J. B.; Olsen, B. R.: Autosomal dominant craniometaphyseal dysplasia is caused by mutations in the transmembrane protein ANK. *Am. J. Hum. Genet.* 68:1321–1326, 2001.

[37790] 11342. Ansel, K. M.; Harris, R. B. S.; Cyster, J. G.: CXCL13 is required for B1 cell homing, natural antibody production, and body cavity immunity. *Immunity* 16:67–76, 2002.

[37791] 11343. Gunn, M. D.; Ngo, V. N.; Ansel, K. M.; Ekland, E. H.; Cyster, J. G.; Williams, L. T.: A B-cell-homing chemokine made in lymphoid follicles activates Burkitt's lymphoma receptor-1. *Nature* 391: 799–803, 1998.

[37792] 11344. Legler, D. F.; Loetscher, M.; Roos, R. S.; Clark-Lewis, I.; Baggiolini, M.; Moser, B.: B cell-attracting chemokine 1, a human CXC chemokine expressed in lym-

phoid tissues, selectively attracts B lymphocytes via BLR1/CXCR5. *J. Exp. Med.* 187: 655–660, 1998.

[37793] 11345. Derst, C.; Engel, H.; Grzeschik, K.-H.; Daut, J.: Genomic structure and chromosome mapping of human and mouse RAMP genes. *Cytogenet. Cell Genet.* 90: 115–118, 2000.

[37794] 11346. Amler, L. C.; Bauer, A.; Corvi, R.; Dihlmann, S.; Praml, C.; Cavenee, W. K.; Schwab, M.; Hampton, G. M.: Identification and characterization of novel genes located at the t(1;15)(p36.2;q24) translocation breakpoint in the neuroblastoma cell line NGP. *Genomics* 64: 195–202, 2000.

[37795] 11347. Yanagisawa, H.; Bundo, M.; Miyashita, T.; Okamura-Oho, Y.; Tadokoro, K.; Tokunaga, K.; Yamada, M.: Protein binding of a DRPLA family through arginine-glutamic acid dipeptide repeats is enhanced by extended polyglutamine. *Hum. Molec. Genet.* 9: 1433–1442, 2000.

[37796] 11348. Oh, J.; Takahashi, R.; Kondo, S.; Mizoguchi, A.; Adachi, E.; Sasahara, R. M.; Nishimura, S.; Imamura, Y.; Kitayama, H.; Alexander, D. B.; Ide, C.; Horan, T. P.; Arakawa, T.; Yoshida, H.; Nishikawa, S.; Itoh, Y.; Seiki, M.; Itohara, S.; Takahashi, C.; Noda, M.: The membrane-anchored MMP inhibitor RECK is a key regulator of extracellular matrix

integrity and angiogenesis. *Cell* 107: 789–800, 2001.

- [37797] 11349. Takahashi, C.; Sheng, Z.; Horan, T. P.; Kitayama, H.; Maki, M.; Hitomi, K.; Kitaura, Y.; Takai, S.; Sasahara, R. M.; Horimoto, A.; Ikawa, Y.; Ratzkin, B. J.; Arakawa, T.; Noda, M.: Regulation of matrix metalloproteinase-9 and inhibition of tumor invasion by the membrane-anchored glycoprotein RECK. *Proc. Nat. Acad. Sci.* 95: 13221–13226, 1998.
- [37798] 11350. Nagase, T.; Ishikawa, I.; Nakajima, D.; Ohira, M.; Seki, N.; Miyajima, N.; Tanaka, A.; Kotani, H.; Nomura, N.; O'Hara, O.: Prediction of the coding sequences of unidentified human genes. VII. The complete sequences of 100 new cDNA clones from brain which can code for large proteins in vitro. *DNA Res.* 4: 141–150, 1997.
- [37799] 11351. Koseki, T.; Inohara, N.; Chen, S.; Nunez, G.: ARC, an inhibitor of apoptosis expressed in skeletal muscle and heart that interacts selectively with caspases. *Proc. Nat. Acad. Sci.* 95: 5156–5160, 1998.
- [37800] 11352. Li, P.-F.; Li, J.; Muller, E.-C.; Otto, A.; Dietz, R.; von Harsdorf, R.: Phosphorylation by protein kinase CK2: a signaling switch for the caspase-inhibiting protein ARC. *Molec. Cell* 10: 247–258, 2002.
- [37801] 11353. Muto, A.; Hoshino, H.; Madisen, L.; Yanai, N.; Obi-

nata, M.; Karasuyama, H.; Hayashi, N.; Nakauchi, H.; Yamamoto, M.; Groudine, M.; Igarashi, K.: Identification of Bach2 as a B-cell-specific partner for small Maf proteins that negatively regulate the immunoglobulin heavy chain-gene 3-prime enhancer. EMBO J. 17: 5734–5743, 1998.

[37802] 11354. Sasaki, S.; Ito, E.; Toki, T.; Maekawa, T.; Kanezaki, R.; Umenai, T.; Muto, A.; Nagai, H.; Kinoshita, T.; Yamamoto, M.; Inazawa, J.; Taketo, M. M.; Nakahata, T.; Igarashi, K.; Yokoyama, M.: Cloning and expression of human B cell-specific transcription factor BACH2 mapped to chromosome 6q15. Oncogene 19: 3739–3749, 2000.

[37803] 11355. Gayle, M. A.; Slack, J. L.; Bonnert, T. P.; Renshaw, B. R.; Sonoda, G.; Taguchi, T.; Testa, J. R.; Dower, S. K.; Sims, J. E.: Cloning of a putative ligand for the T1/ST2 receptor. J. Biol. Chem. 271: 5784–5789, 1996.

[37804] 11356. Shimaoka, T.; Kume, N.; Minami, M.; Hayashida, K.; Kataoka, H.; Kita, T.; Yonehara, S.: Molecular cloning of a novel scavenger receptor for oxidized low density lipoprotein, SR-PSOX, on macrophages. J. Biol. Chem. 275: 40663–40666, 2000.

[37805] 11357. Wilbanks, A.; Zondlo, S. C.; Murphy, K.; Mak, S.; Soler, D.; Langdon, P.; Andrew, D. P.; Wu, L.; Briskin, M.: Expression cloning of the STRL33/BONZO/TYMSTR ligand

reveals elements of CC, CXC, CX3C chemokines. *J.Immun.* 166: 5145–5154, 2001.

- [37806] 11358.Brill, S.; Li, S.; Lyman, C. W.; Church, D. M.; Wasmuth, J. J.; Weissbach, L.; Bernards, A.; Snijders, A. J.: The Ras GTPase-activating-protein-related human protein IQ-GAP2 harbors a potential actin binding domain and interacts with calmodulin and Rho family GTPases. *Molec. Cell. Biol.* 16:4869–4878, 1996.
- [37807] 11359.Bitner-Glindzicz, M.; Turnpenny, P.; Hoglund, P.; Kaariainen, H.; Sankila, E.-M.; van der Maarel, S. M.; de Kok, Y. J. M.; Ropers, H.-H.; Cremers, F. P. M.; Pembrey, M.; Malcolm, S.: Further mutations in brain 4 (POU3F4) clarify the phenotype in the X-linked deafness, DFN3. *Hum.Molec. Genet.* 4: 1467–1469, 1995.
- [37808] 11360.de Kok, Y. J. M.; Cremers, C. W. R. J.; Ropers, H.-H.; Cremers, F. P. M.: The molecular basis of X-linked deafness type 3 (DFN3) in two sporadic cases: identification of a somatic mosaicism for a POU3F4 missense mutation. *Hum. Mutat.* 10: 207–211, 1997.
- [37809] 11361.de Kok, Y. J. M.; Merckx, G. F. M.; van der Maarel, S. M.; Huber, I.; Malcolm, S.; Ropers, H.-H.; Cremers, F. P. M.: A duplication/paracentric inversion associated with familial X-linked deafness (DFN3) suggests the presence of a reg-

ulatory element more than 400 kb upstream of the POU3F4 gene. Hum. Molec. Genet. 4: 2145–2150, 1995.

- [37810] 11362. de Kok, Y. J. M.; van der Maarel, S. M.; Bitner-Glindzicz, M.; Huber, I.; Monaco, A. P.; Malcolm, S.; Pembrey, M. E.; Ropers, H.-H.; Cremers, F. P. M.: Association between X-linked mixed deafness and mutations in the POU domain gene POU3F4. Science 267: 685–688, 1995.
- [37811] 11363. Douville, P. J.; Atanasoski, S.; Tobler, A.; Fontana, A.; Schwab, M. E.: The brain-specific POU-box gene Brn4 is a sex-linked transcription factor located on the human and mouse X chromosomes. Mammalian Genome 5: 180–182, 1994.
- [37812] 11364. Friedman, R. A.; Bykhovskaya, Y.; Tu, G.; Talbot, J. M.; Wilson, D. F.; Parnes, L. S.; Fischel-Ghodsian, N.: Molecular analysis of the POU3F4 gene in patients with clinical and radiographic evidence of X-linked mixed deafness with perilymphatic gusher. Ann. Otol. Rhinol. Laryng. 106: 320–325, 1997.
- [37813] 11365. Minowa, O.; Ikeda, K.; Sugitani, Y.; Oshima, T.; Nakai, S.; Katori, Y.; Suzuki, M.; Furukawa, M.; Kawase, T.; Zheng, Y.; Ogura, M.; Asada, Y.; Watanabe, K.; Yamanaka, H.; Gotoh, S.; Nishi-Takeshima, M.; Sugimoto, T.; Kikuchi, T.; Takasaka, T.; Noda, T.: Altered cochlear fibrocytes in a

mouse model of DFN3 nonsyndromic deafness. *Science* 285: 1408–1411, 1999.

[37814] 11366. Phelps, P. D.; Reardon, W.; Pembrey, M.; Bellman, S.; Luxon, L.: X-linked deafness, stapes gushers and a distinctive defect of the inner ear. *Neuroradiology* 33: 326–330, 1991.

[37815] 11367. Phippard, D.; Boyd, Y.; Reed, V.; Fisher, G.; Masson, W. K.; Evans, E. P.; Saunders, J. C.; Crenshaw, E. B., III: The sex-linked fidget mutation abolishes Brn4/Pou3f4 gene expression in the embryonic inner ear. *Hum. Molec. Genet.* 9: 79–85, 2000.

[37816] 11368. Brown, C. J.; Miller, A. P.; Carrel, L.; Rupert, J. L.; Davies, K. E.; Willard, H. F.: The DXS423E gene in Xp11.21 escapes X chromosome inactivation. *Hum. Molec. Genet.* 4: 251–255, 1995.

[37817] 11369. Sumara, I.; Vorlaufer, E.; Gieffers, C.; Peters, B. H.; Peters, J.-M.: Characterization of vertebrate cohesin complexes and their regulation in prophase. *J. Cell Biol.* 151: 749–761, 2000.

[37818] 11370. Lowe, D. G.; Dizhoor, A. M.; Liu, K.; Gu, Q.; Spencer, M.; Laura, R.; Lu, L.; Hurley, J. B.: Cloning and expression of a second photoreceptor-specific membrane retina guanylyl cyclase (RetGC), RetGC-2. *Proc. Nat.*

Acad.Sci. 92: 5535–5539, 1995.

[37819] 11371.Teague, P. W.; Aldred, M. A.; Jay, M.; Dempster, M.; Harrison,C.; Carothers, A. D.; Hardwick, L. J.; Evans, H. J.; Strain, L.; Brock,D. J. H.; Bunday, S.; Jay, B.; Bird, A. C.; Bhattacharya, S. S.; Wright,A. F.: Heterogeneity analysis in 40 X-linked retinitis pigmentosafamilies. Am. J. Hum. Genet. 55: 105–111, 1994.

[37820] 11372.Bodrug, S. E.; Ray, P. N.; Gonzalez, I. L.; Schmickel, R. D.;Sylvester, J. E.; Worton, R. G.: Molecular analysis of a constitutionalX-autosome translocation in a female with muscular dystrophy. Science 237:1620–1624, 1987.

[37821] 11373.Boyce, F. M.; Beggs, A. H.; Feener, C.; Kunkel, L. M.: Dystrophinis transcribed in brain from a distant upstream promoter. Proc. Nat.Acad. Sci. 88: 1276–1280, 1991.

[37822] 11374.Boyd, Y.; Buckle, V. J.: Cytogenetic heterogeneity of translocationsassociated with Duchenne muscular dystrophy. Clin. Genet. 29: 108–115,1986.

[37823] 11375.Bulman, D. E.; Gangopadhyay, S. B.; Bebchuck, K. G.; Worton, R.G.; Ray, P. N.: Point mutation in the human dystrophin gene: identificationthrough Western blot analysis. Genomics 10: 457–460, 1991.

[37824] 11376.Burke, J. F.; Mogg, A. E.: Suppression of a nonsense mutationin mammalian cells in vivo by the aminoglycoside

antibiotics G-418 and paromomycin. *Nucleic Acids Res.* 13: 6265–6272, 1985.

- [37825] 11377. Burnette, W. N.: 'Western blotting': electrophoretic transfer of proteins from sodium dodecyl sulfate–polyacrylamide gels to unmodified nitrocellulose and radiographic detection with antibody and radioiodinated–protein A. *Anal. Biochem.* 112: 195–203, 1981.
- [37826] 11378. Chamberlain, J. S.; Pearlman, J. A.; Muzny, D. M.; Gibbs, R. A.; Ranier, J. E.; Reeves, A. A.; Caskey, C. T.: Expression of the murine Duchenne muscular dystrophy gene in muscle and brain. *Science* 239:1416–1418, 1988.
- [37827] 11379. Chelly, J.; Concorde, J.-P.; Kaplan, J.-C.; Kahn, A.: Illegitimate transcription: transcription of any gene in any cell type. *Proc. Nat. Acad. Sci.* 86: 2617–2621, 1989.
- [37828] 11380. Chelly, J.; Gilgenkrantz, H.; Hugnot, J. P.; Hamard, G.; Lambert, M.; Recan, D.; Akli, S.; Cometto, M.; Kahn, A.; Kaplan, J. C.: Illegitimate transcription: application to the analysis of truncated transcripts of the dystrophin gene in nonmuscle cultured cells from Duchenne and Becker patients. *J. Clin. Invest.* 88: 1161–1166, 1991.
- [37829] 11381. Chelly, J.; Hamard, G.; Koulakoff, A.; Kaplan, J.-C.; Kahn, A.; Berwald-Netter, Y.: Dystrophin gene transcribed from different promoters in neuronal and glial cells. *Nature*

344: 64–65, 1990.

- [37830] 11382.Chelly, J.; Kaplan, J.-C.; Maire, P.; Gautron, S.; Kahn, A.:Transcription of the dystrophin gene in human muscle and non-muscle tissues. *Nature* 333: 858–860, 1988.
- [37831] 11383.Clemens, P. R.; Ward, P. A.; Caskey, C. T.; Bulman, D. E.; Fenwick, R. G.: Premature chain termination mutation causing Duchenne muscular dystrophy. *Neurology* 42: 1775–1782, 1992.
- [37832] 11384.Cooper, B. J.; Valentine, B. A.; Wilson, S.; Patterson, D. F.; Concannon, P. W.: Canine muscular dystrophy: confirmation of X-linked inheritance. *J. Hered.* 79: 405–408, 1988.
- [37833] 11385.Covone, A. E.; Lerone, M.; Romeo, G.: Genotype–phenotype correlation and germline mosaicism in DMD/BMD patients with deletions of the dystrophin gene. *Hum. Genet.* 87: 353–360, 1991.
- [37834] 11386.Cox, G. A.; Cole, N. M.; Matsumura, K.; Phelps, S. F.; Hauschka, S. D.; Campbell, K. P.; Faulkner, J. A.; Chamberlain, J. S.: Overexpression of dystrophin in transgenic mdx mice eliminates dystrophic symptoms without toxicity. *Nature* 364: 725–729, 1993.
- [37835] 11387.Cox, G. A.; Sunada, Y.; Campbell, K. P.; Chamber-

lain, J. S.:Dp71 can restore the dystrophin-associated glycoprotein complex in muscle but fails to prevent dystrophy. *Nature Genet.* 8: 333–339,1994.

[37836] 11388.Comi, G. P.; Ciafaloni, E.; de Silva, H. A. R.; Prella, A.; Bardoni,A.; Rigoletto, C.; Robotti, M.; Bresolin, N.; Moggio, M.; Fortunato,F.; Ciscato, P.; Turconi, A.; Rose, A. D.; Scarlato, G.: A G(+1)-to-Atransversion at the 5-prime splice site of intron 69 of the dystrophingene causing the absence of peripheral nerve Dp116 and severe clinical involvement in a DMD patient. *Hum. Molec. Genet.* 4: 2171–2174, 1995.

[37837] 11389.Crawford, G. E.; Lu, Q. L.; Partridge, T. A.; Chamberlain, J.S.: Suppression of revertant fibers in mdx mice by expression of a functional dystrophin. *Hum. Molec. Genet.* 10: 2745–2750, 2001.

[37838] 11390.Darras, B. T.; Blattner, P.; Harper, J. F.; Spiro, A. J.; Alter,S.; Francke, U.: Intragenic deletions in 21 Duchenne muscular dystrophy(DMD)/Becker muscular dystrophy (BMD) families studied with the dystrophin cDNA: location of breakpoints on HindIII and BglII exon-containing fragment maps, meiotic and mitotic origin of the mutations. *Am. J.Hum. Genet.* 43: 620–629, 1988.

[37839] 11391.Darras, B. T.; Francke, U.: Normal human genomic

restriction-fragment patterns and polymorphisms revealed by hybridization with the entire dystrophin cDNA. *Am. J. Hum. Genet.* 43: 612-619, 1988.

[37840] 11392. Darras, B. T.; Francke, U.: A partial deletion of the muscular dystrophy gene transmitted twice by an unaffected male. *Nature* 329:556-558, 1987.

[37841] 11393. Davies, K. E.; Smith, T. J.; Bunday, S.; Read, A. P.; Flint, T.; Bell, M.; Speer, A.: Mild and severe muscular dystrophy associated with deletions in Xp21 of the human X chromosome. *J. Med. Genet.* 25:9-13, 1988.

[37842] 11394. De Angelis, F. G.; Sthandier, O.; Berarducci, B.; Toso, S.; Galluzzi, G.; Ricci, E.; Cossu, G.; Bozzoni, I.: Chimeric snRNA molecules carrying antisense sequences against the splice junctions of exon 51 of the dystrophin pre-mRNA induce exon skipping and restoration of a dystrophin synthesis in delta-48-50 DMD cells. *Proc Nat. Acad. Sci.* 99: 9456-9461, 2002.

[37843] 11395. den Dunnen, J. T.; Bakker, E.; Klein Breteler, E. G.; Pearson, P. L.; van Ommen, G. J. B.: Direct mutation of more than 50% of the Duchenne muscular dystrophy mutations by field inversion gels. *Nature* 329:640-642, 1987.

[37844] 11396. Dickson, G.; Pizzey, J. A.; Elsom, V. E.; Love, D.;

Davies, K.E.; Walsh, F. S.: Distinct dystrophin mRNA species are expressed in embryonic and adult mouse skeletal muscle. FEBS Lett. 242: 47–52, 1988.

- [37845] 11397. Dominguez–Steglich, M.; Meng, G.; Bettecken, T.; Muller, C. R.; Schmid, M.: The dystrophin gene is autosomally located on a microchromosome in chicken. Genomics 8: 536–540, 1990.
- [37846] 11398. Doolittle, R. F.: Similar amino acid sequences: chance or common ancestry? Science 214: 149–159, 1981.
- [37847] 11399. Bassi, M. T.; Bergen, A. A. B.; Bitoun, P.; Charles, S. J.; Clementi, M.; Gosselin, R.; Hurst, J.; Lewis, R. A.; Lorenz, B.; Meitinger, T.; Messiaen, L.; Ramesar, R. S.; Ballabio, A.; Schiaffino, M. V.: Diverse prevalence of large deletions within the OA1 gene in ocular albinism type 1 patients from Europe and North America. Hum. Genet. 108: 51–54, 2001.
- [37848] 11400. Bassi, M. T.; Schiaffino, M. V.; Renieri, A.; De Nigris, F.; Galli, L.; Bruttini, M.; Gebbia, M.; Bergen, A. A. B.; Lewis, R. A.; Ballabio, A.: Cloning of the gene for ocular albinism type 1 from the distal short arm of the X chromosome. Nature Genet. 10: 13–19, 1995.
- [37849] 11401. Bergen, A. A. B.; Samanns, C.; Schuurman, E. J. M.; van Osch, L.; van Dorp, D. B.; Pinckers, A. J. L. G.; Bakker,

E.; Gal, A.; van Ommen, G. J. B.; Bleeker-Wagemakers, E. M.: Multipoint linkage analysis in X-linked ocular albinism of the Nettleship-Falls type. *Hum. Genet.* 88:162-166, 1991.

[37850] 11402. Bergen, A. A. B.; Schuurman, E. J. M.; van den Born, L. I.; Samanns, C.; van Dorp, D. B.; Pinckers, A. J. L. G.; Bakker, E.; van Ommen, G. J. B.; Gal, A.; Bleeker-Wagemakers, E. M.: Carrier detection in X-linked ocular albinism of the Nettleship-Falls type by DNA analysis. *Clin. Genet.* 41: 135-138, 1992.

[37851] 11403. Bergen, A. A. B.; Zijp, P.; Schuurman, E. J. M.; Bleeker-Wagemakers, E. M.; Apkarian, P.; van Ommen, G.-J. B.: Refinement of the localization of the X-linked ocular albinism gene. *Genomics* 16: 272-273, 1993.

[37852] 11404. Bouloux, P.-M. G.; Kirk, J.; Munroe, P.; Duke, V.; Meindl, A.; Hilson, A.; Grant, D.; Carter, N.; Betts, D.; Meitinger, T.; Besser, G. M.: Deletion analysis maps ocular albinism proximal to the steroid sulphatase locus. *Clin. Genet.* 43: 169-173, 1993.

[37853] 11405. Charles, S. J.; Green, J. S.; Moore, A. T.; Barton, D. E.; Yates, J. R. W.: Genetic mapping of X-linked ocular albinism: linkage analysis in a large Newfoundland kindred. *Genomics* 16: 259-261, 1993.

- [37854] 11406.Charles, S. J.; Moore, A. T.; Yates, J. R. W.: Genetic mapping of X linked ocular albinism: linkage analysis in British families. *J.Med. Genet.* 29: 552–554, 1992.
- [37855] 11407.Cooper, D. N.; Krawczak, M.: Mechanisms of insertional mutagenesis in human genes causing genetic disease. *Hum. Genet.* 87: 409–415, 1991.
- [37856] 11408.Creel, D.; O'Donnell, F. E., Jr.; Witkop, C. J., Jr.: Visual system anomalies in human ocular albinos. *Science* 201: 931–933, 1978.
- [37857] 11409.d'Addio, M.; Pizzigoni, A.; Bassi, M. T.; Baschiroto, C.; Valetti, C.; Incerti, B.; Clementi, M.; De Luca, M.; Bal-labio, A.; Schiaffino, M. V.: Defective intracellular transport and processing of OA1 is a major cause of ocular albinism type 1. *Hum. Molec. Genet.* 9: 3011–3018, 2000.
- [37858] 11410.Engelhard, C. F.: Eine Familie mit hereditaerem Nystagmus. *Z.Ges. Neurol. Psychiat.* 28: 319–338, 1915.
- [37859] 11411.Fialkow, P. J.; Giblett, E. R.; Motulsky, A. G.: Measurable linkage between ocular albinism and Xg. *Am. J. Hum. Genet.* 19: 63–69, 1967.
- [37860] 11412.Francois, J.; Deweer, J. P.: Albinisme oculaire lie au sexe et alterations caracteristiques du fond d'oeil chez les femmes heterozygotes. *Ophthalmologica* 126:209–221, 1953.

- [37861] 11413. Gillespie, F. D.: Ocular albinism with report of a family with female carriers. *Arch. Ophthalmol.* 66: 774–777, 1961.
- [37862] 11414. Incerti, B.; Cortese, K.; Pizzigoni, A.; Surace, E. M.; Varani, S.; Coppola, M.; Jeffery, G.; Seeliger, M.; Jaissle, G.; Bennett, D. C.; Marigo, V.; Schiaffino, M. V.; Tacchetti, C.; Ballabio, A.: Oa1 knock-out: new insights on the pathogenesis of ocular albinism type 1. *Hum. Molec. Genet.* 9: 2781–2788, 2000.
- [37863] 11415. Jaeger, C.; Jay, B.: X-linked ocular albinism: a family containing a manifesting heterozygote, and an affected male married to a female with autosomal recessive ocular albinism. *Hum. Genet.* 56: 299–304, 1981.
- [37864] 11416. Johnson, G. J.; Gillan, J. G.; Pearce, W. G.: Ocular albinism in Newfoundland. *Can. J. Ophthalmol.* 6: 237–248, 1971.
- [37865] 11417. Kidd, J. R.; Castiglione, C. M.; Davies, K. E.; Pakstis, A. J.; Gusella, J.; Sparkes, R. S.; Pearson, P.; Willard, H.; Kidd, K. K.: Mapping the locus for X-linked ocular albinism (OA). (Abstract) *Am. J. Hum. Genet.* 37: A161 only, 1985.
- [37866] 11418. Krawczak, M.; Reiss, J.; Cooper, D. N.: The mutational spectrum of single base-pair substitutions in mRNA

splice junctions of humangenes: causes and consequences. Hum. Genet. 90: 41–54, 1992.

[37867] 11419.Lein, J. N.; Stewart, C. T.; Moll, F. C.: Sex-linked hereditary nystagmus. Pediatrics 18: 214–217, 1956.

[37868] 11420.Lyon, M. F.: Sex chromatin and gene action in the mammalian X-chromosome. Am.J. Hum. Genet. 14: 135–148, 1962.

[37869] 11421.Meindl, A.; Hosenfeld, D.; Bruckl, W.; Schuffenhauer, S.; Jenderny, J.; Bacskulin, A.; Oppermann, H.-C.; Swensson, O.; Bouloux, P.; Meitinger, T.: Analysis of a terminal Xp22.3 deletion in a patient with six monogenic disorders: implications for the mapping of X linked ocular albinism. J. Med. Genet. 30: 838–842, 1993.

[37870] 11422.Negrelli, B. C.: L'albinisme oculaire lie au sexe dans le cadre du depistage des heterozygotes en ophtalmologie. J. Genet. Hum. 8:108 only, 1959.

[37871] 11423.Newton, J. M.; Orlow, S. J.; Barsh, G. S.: Isolation and characterization of a mouse homolog of the X-linked ocular albinism (OA1) gene. Genomics 37:219–225, 1996.

[37872] 11424.O'Donnell, F. E., Jr.; Green, W. R.; Fleishman, J. A.; Hambrick, G. W.: X-linked ocular albinism in Blacks: ocular albinism cum pigmento. Arch.Ophthal. 96: 1189–1192, 1978.

- [37873] 11425.O'Donnell, F. E., Jr.; Hambrick, G. W., Jr.; Green, W. R.; Iliff,W. J.; Stone, D. L.: X-linked ocular albinism: an oculocutaneousmacromelanosomal disorder. Arch. Ophthalmol. 94: 1883-1892, 1976.
- [37874] 11426.Oetting, W. S.: New insights into ocular albinism type 1 (OA1):mutations and polymorphisms of the OA1 gene. Hum. Mutat. 19: 85-92,2002.
- [37875] 11427.Pearce, W. G.; Johnson, G. J.; Gillan, J. G.: Nystagmus in afemale carrier of ocular albinism. J. Med. Genet. 9: 126-128, 1972.
- [37876] 11428.Pearce, W. G.; Johnson, G. J.; Sanger, R.: Ocular albinism andXg. (Letter) Lancet I: 1072 only, 1971.
- [37877] 11429.Pearce, W. G.; Sanger, R.: X mapping in man: evidence againstdirect measurable linkage between ocular albinism and deutan colourblindness. J. Med. Genet. 13: 319 only, 1976.
- [37878] 11430.Gaugitsch, H. W.; Prieschl, E. E.; Kalthoff, F.; Huber, N. E.;Baumruker, T.: A novel transiently expressed, integral membrane proteinlinked to cell activation: molecular cloning via the rapid degradationsignal AUUUA. J. Biol. Chem 267: 11267-11273, 1992.
- [37879] 11431.Kanai, Y.; Segawa, H.; Miyamoto, K.; Uchino, H.; Takeda, H.; Endou,H.: Expression cloning and characteri-

zation of a transporter for large neutral amino acids activated by the heavy chain of 4F2 antigen (CD98). J. Biol. Chem. 273: 23629–23632, 1998.

[37880] 11432. Takahara, K.; Kessler, E.; Biniaminov, L.; Brusel, M.; Eddy, R.L.; Jani-Sait, S.; Shows, T. B.; Greenspan, D. S.: Type I procollagen COOH-terminal proteinase enhancer protein: identification, primary structure, and chromosomal localization of the cognate human gene (PCOLCE). J. Biol. Chem. 269: 26280–26285, 1994.

[37881] 11433. Takahara, K.; Osborne, L.; Elliott, R. W.; Tsui, L.-C.; Scherer, S. W.; Greenspan, D. S.: Fine mapping of the human and mouse genes for the type I procollagen COOH-terminal proteinase enhancer protein. Genomics 31:253–256, 1996.

[37882] 11434. Amagai, M.; Wang, Y.; Minoshima, S.; Kawamura, K.; Green, K. J.; Nishikawa, T.; Shimizu, N.: Assignment of the human genes for desmocollin 3 (DSC3) and desmocollin 4 (DSC4) to chromosome 18q12. Genomics 25:330–332, 1995.

[37883] 11435. Kawamura, K.; Watanabe, K.; Suzuki, T.; Yamakawa, T.; Kamiyama, T.; Nakagawa, H.; Tsurufuji, S.: cDNA cloning and expression of a novel human desmocollin. J. Biol. Chem. 269: 26295–26302, 1994.

- [37884] 11436.King, I. A.; Sullivan, K. H.; Bennett, R., Jr.; Buxton, R. S.:The desmocollins of human foreskin epidermis: identification and chromosomal assignment of a third gene and expression patterns of the three isoforms. *J.Invest. Derm.* 105: 314–321, 1995.
- [37885] 11437.Warburg, O.: On the origin of cancer cells. *Science* 123: 309–314,1956.
- [37886] 11438.Yang, R.–B.; Foster, D. C.; Garbers, D. L.; Fulle, H.–J.: Twomembrane forms of guanylyl cyclase found in the eye. *Proc. Nat. Acad.Sci.* 92: 602–606, 1995.
- [37887] 11439.Dubrovsky, A. L.; Taratuto, A. L.; Sevelever, G.; Schultz, M.;Pegoraro, E.; Hoop, R. C.; Hoffman, E. P.: Duchenne muscular dystrophyand myotonic dystrophy in the same patient. *Am. J. Med. Genet.* 55:342–348, 1995.
- [37888] 11440.Cazzola, M.; Bergamaschi, G.: X-linked Wiskott–Aldrich syndromein a girl. (Letter) *New Eng. J. Med.* 338: 1850 only, 1998.
- [37889] 11441.Emery, A. E. H.: *Duchenne Muscular Dystrophy*. Oxford, UK: OxfordUniversity Press (pub.) (2nd ed.): 1993.
- [37890] 11442.Fabb, S. A.; Wells, D. J.; Serpente, P.; Dickson, G.: Adeno–associatedvirus vector gene transfer and sar–colemmal expression of a 144 kDamicro–dystrophin effectively restores the dystrophin–associated proteincom–

plex and inhibits myofibre degeneration in nude/mdx mice. *Hum.Molec. Genet.* 11: 733–741, 2002.

[37891] 11443.Feener, C. A.; Boyce, F. M.; Kunkel, L. M.: Rapid detection of CA polymorphisms in cloned DNA: application to the 5–prime region of the dystrophin gene. *Am. J. Hum. Genet.* 48: 621–627, 1991.

[37892] 11444.Ferlini, A.; Galie, N.; Merlini, L.; Sewry, C.; Branzi, A.; Muntoni, F.: A novel Alu–like element rearranged in the dystrophin gene causes a splicing mutation in a family with X–linked dilated cardiomyopathy. *Am.J. Hum. Genet.* 63: 436–446, 1998.

[37893] 11445.Finnegan, D. J.: Eukaryotic transposable elements and genome evolution. *Trends Genet.* 5: 103–107, 1989.

[37894] 11446.Forrest, S. M.; Cross, G. S.; Speer, A.; Gardner–Medwin, D.; Burn, J.; Davies, K. E.: Preferential deletion of exons in Duchenne and Becker muscular dystrophies. *Nature* 329: 638–640, 1987.

[37895] 11447.Francke, U.; Ochs, H. D.; de Martinville, B.; Giacalone, J.; Lindgren, V.; Distèche, C.; Pagon, R. A.; Hofker, M. H.; van Ommen, G.–J. B.; Pearson, P. L.; Wedgwood, R. J.: Minor Xp21 chromosome deletion in a male associated with expression of Duchenne muscular dystrophy, chronic granulomatous disease, retinitis pigmentosa, and McLeod

syndrome. Am.J. Hum. Genet. 37: 250–267, 1985.

- [37896] 11448.Furst, D.; Nave, R.; Osborn, M.; Weber, K.; Bardosi, A.; Archidiacono,N.; Ferro, M.; Romano, V.; Romeo, G.: Nebulin and titin expressionin Duchenne muscular dystrophy appears normal. FEBS Lett. 224: 49–53,1987.
- [37897] 11449.Giacalone, J. P.; Francke, U.: Common sequence motifs at therearrangement sites of a constitutional X/ autosome translocation andassociated deletion. Am. J. Hum. Genet. 50: 725–741, 1992.
- [37898] 11450.Gillard, E. F.; Chamberlain, J. S.; Murphy, E. G.; Duff, C. L.;Smith, B.; Burghes, A. H. M.; Thompson, M. W.; Sutherland, J.; Oss,I.; Bodrug, S. E.; Klamut, H. J.; Ray, P. N.; Worton, R. G.: Molecularand phenotypic analysis of patients with deletions within the deletion–richregion of the Duchenne muscular dystrophy (DMD) gene. Am. J. Hum.Genet. 45: 507–520, 1989.
- [37899] 11451.Ginjaar, I. B.; Kneppers, A. L. J.; Meulen, J.–D. M.; Anderson,L. V. B.; Bremmer–Bout, M.; van Deutekom, J. C. T.; Weegenaar, J.;den Dunnen, J. T.; Bakker, E.: Dystrophin nonsense mutation inducesdifferent levels of exon 29 skipping and leads to variable phenotypeswithin one BMD family. Europ. J. Hum. Genet. 8: 793–796, 2000.
- [37900] 11452.Greenberg, D. S.; Sunada, Y.; Campbell, K. P.;

Yaffe, D.; Nudel, U.: Exogenous Dp71 restores the levels of dystrophin associated proteins but does not alleviate muscle damage in mdx mice. *Nature Genet.* 8:340–344, 1994.

[37901] 11453. Gussoni, E.; Soneoka, Y.; Strickland, C. D.; Buzney, E. A.; Khan, M. K.; Flint, A. F.; Kunkel, L. M.; Mulligan, R. C.: Dystrophin expression in the mdx mouse restored by stem cell transplantation. *Nature* 401:390–394, 1999.

[37902] 11454. Hagiwara, Y.; Mizuno, Y.; Takemitsu, M.; Matsuzaki, T.; Nonaka, I.; Ozawa, E.: Dystrophin-positive muscle fibers following C2 myoblast transplantation into mdx nude mice. *Acta Neuropath.* 90: 592–600, 1995.

[37903] 11455. Hagiwara, Y.; Nishio, H.; Kitoh, Y.; Takeshima, Y.; Narita, N.; Wada, H.; Yokoyama, M.; Nakamura, H.; Matsuo, M.: A novel point mutation (G(–1) to T) in a 5-prime splice donor site of intron 13 of the dystrophin gene results in exon skipping and is responsible for Becker muscular dystrophy. *Am. J. Hum. Genet.* 54: 53–61, 1994.

[37904] 11456. Hammonds, R. G., Jr.: Protein sequence of DMD gene is related to actin-binding domain of alpha-actinin. (Letter) *Cell* 51: 1, 1987.

[37905] 11457. Harper, S. Q.; Hauser, M. A.; Dello Russo, C.; Duan, D.; Crawford, R. W.; Phelps, S. F.; Harper, H. A.; Robinson, A. S.; Engelhardt, J. F.; Brooks, S. V.; Chamberlain, J. S.:

Modular flexibility of dystrophin: implications for gene therapy of Duchenne muscular dystrophy. *Nature Med.* 8: 253–261, 2002.

- [37906] 11458. Hart, K. A.; Hodgson, S.; Walker, A.; Cole, C. G.; Johnson, L.; Dubowitz, V.; Bobrow, M.: DNA deletions in mild and severe Becker muscular dystrophy. *Hum. Genet.* 75: 281–285, 1987.
- [37907] 11459. Hodgson, S. V.; Abbs, S.; Clark, S.; Manzur, A.; Heckmatt, J. Z. H.; Dubowitz, V.; Bobrow, M.: Correlation of clinical and deletion data in Duchenne and Becker muscular dystrophy, with special reference to mental ability. *Neuromusc. Disord.* 2: 269–276, 1992.
- [37908] 11460. Hoffman, E. P.; Brown, R. H., Jr.; Kunkel, L. M.: The protein product of the Duchenne muscular dystrophy locus. *Cell* 51: 919–928, 1987.
- [37909] 11461. Hoffman, E. P.; Knudson, C. M.; Campbell, K. P.; Kunkel, L. M.: Subcellular fractionation of dystrophin to the triads of skeletal muscle. *Nature* 330: 754–758, 1987.
- [37910] 11462. Hoffman, E. P.; Monaco, A. P.; Feener, C. C.; Kunkel, L. M.: Conservation of the Duchenne muscular dystrophy gene in mice and humans. *Science* 238: 347–350, 1987.
- [37911] 11463. Hoop, R. C.; Russo, L. S.; Riconda, D. L.; Schwartz,

L. S.; Hoffman, E. P.: Restoration of half the normal dystrophin sequence in a double-deletion Duchenne muscular dystrophy family. *Am. J. Med. Genet.* 49: 323–327, 1994.

[37912] 11464. Hoffman, E. P.; Fischbeck, K. H.; Brown, R. H.; Johnson, M.; Medori, R.; Loike, J. D.; Harris, J. B.; Waterston, R.; Brooke, M.; Specht, L.; Kopsky, W.; Chamberlain, J.; Caskey, C. T.; Shapiro, F.; Kunkel, L. M.: Characterization of dystrophin in muscle-biopsy specimens from patients with Duchenne's or Becker's muscular dystrophy. *New Eng. J. Med.* 318: 1363–1368, 1988.

[37913] 11465. Howard, P. L.; Dally, G. Y.; Wong, M. H.; Ho, A.; Weleber, R. G.; Pillers, D.-A. M.; Ray, P. N.: Localization of dystrophin isoform Dp71 to the inner limiting membrane of the retina suggests a unique functional contribution of Dp71 in the retina. *Hum. Molec. Genet.* 7:1385–1391, 1998.

[37914] 11466. Hu, X.; Burghes, A. H. M.; Bulman, D. E.; Ray, P. N.; Worton, R. G.: Evidence for mutation by unequal sister chromatid exchange in the Duchenne muscular dystrophy gene. *Am. J. Hum. Genet.* 44:855–863, 1989.

[37915] 11467. Birnbaumer, M.: Vasopressin receptors. *TEM* 11: 406–410, 2000.

[37916] 11468. Kools, P.; Vanhalst, K.; Wan den Eynde, E.; van Roy,

F.: The humancadherin-10 gene: complete coding sequence, predominant expression in the brain and mapping on chromosome 5p13-13. FEBS Lett. 452:328-334, 1999.

[37917] 11469.Leder, S.; Weber, Y.; Altafaj, X.; Estivill, X.; Joost, H.-G.;Becker, W.: Cloning and characterization of DYRK1B, a novel member of the DYRK family of protein kinases.

Biochem. Biophys. Res. Commun. 254:474-479, 1999.

[37918] 11470.Lee, K.; Deng, X.; Friedman, E.: Mirk protein kinase is a mitogen-activated protein kinase substrate that mediates survival of colon cancer cells. Cancer Res. 60: 3631-3637, 2000.

[37919] 11471.Coyle, A. J.; Lehar, S.; Lloyd, C.; Tian, J.; Delaney, T.; Manning, S.; Nguyen, T.; Burwell, T.; Schneider, H.; Gonzalo, J. A.; Gosselin, M.; Owen, L. R.; Rudd, C. E.; Gutierrez-Ramos, J. C.: The CD28-related molecule ICOS is required for effective T cell-dependent immune responses. Immunity 13:95-105, 2000.

[37920] 11472.Dong, C.; Juedes, A. E.; Temann, U. A.; Shresta, S.; Allison, J.P.; Ruddle, N. H.; Flavell, R. A.: ICOS co-stimulatory receptor is essential for T-cell activation and function. Nature 409: 97-101, 2001.

[37921] 11473.Haimila, K. E.; Partanen, J. A.; Holopainen, P. M.: Genetic polymorphism of the human ICOS gene. Immuno-

genetics 53: 1028–1032, 2002.

- [37922] 11474.Hutloff, A.; Dittrich, A. M.; Beier, K. C.; Eljaschewitsch, B.;Kraft, R.; Anagnostopoulos, I.; Kroczeck, R. A.: ICOS is an inducibleT-cell co-stimulator structurally and functionally related to CD28. Nature 397:263–266, 1999.
- [37923] 11475.McAdam, A. J.; Greenwald, R. J.; Levin, M. A.; Chernova, T.; Malenkovich,N.; Ling, V.; Freeman, G. J.; Sharpe, A. H.: ICOS is critical forCD40-mediated antibody class switching. Nature 409: 102–105, 2001.
- [37924] 11476.Pack, S. D.; Pak, E.; Tanigami, A.; Ledbetter, D. H.; Fukuda, M.N.: Assignment of the bystin gene BYSL to human chromosome band 6p21.1by in situ hybridization. Cytogenet. Cell Genet. 83: 76–77, 1998.
- [37925] 11477.Medina, M.; Marinescu, R. C.; Overhauser, J.; Kosik, K. S.: Hemizygosityof delta-catenin (CTNND2) is associated with severe mental retardationin cri-du-chat syndrome. Genomics 63: 157–164, 2000.
- [37926] 11478.Paffenholz, R.; Franke, W. W.: Identification and localizationof a neurally expressed member of the plakoglobin/armadillo multigenefamily. Differentiation 61: 293–304, 1997.
- [37927] 11479.Zhou, J.; Liyanage, U.; Medina, M.; Ho, C.; Simmons, A. D.; Lovett.M.; Kosik, K. S.: Presenilin 1 interac-

tion in the brain with a novel member of the armadillo family. *Neuroreport* 8: 2085–2090, 1997.

- [37928] 11480. Hatzfeld, M.; Nachtsheim, C.: Cloning and characterization of a new armadillo family member, p0071, associated with the junctional plaque: evidence for a subfamily of closely related proteins. *J. Cell Sci.* 109: 2767–2778, 1996.
- [37929] 11481. Burger, J.; Fonknechten, N.; Hoeltzenbein, M.; Neumann, L.; Bratanoff, E.; Hazan, J.; Reis, A.: Hereditary spastic paraplegia caused by mutations in the SPG4 gene. *Eur. J. Hum. Genet.* 8: 771–776, 2000.
- [37930] 11482. Burger, J.; Metzke, H.; Paternotte, C.; Schilling, F.; Hazan, J.; Reis, A.: Autosomal dominant spastic paraplegia with anticipation maps to a 4-cM interval on chromosome 2p21–p24 in a large German family. *Hum. Genet.* 98: 371–375, 1996.
- [37931] 11483. Errico, A.; Ballabio, A.; Rugarli, E. I.: Spastin, the protein mutated in autosomal dominant hereditary spastic paraplegia, is involved in microtubule dynamics. *Hum. Molec. Genet.* 11: 153–163, 2002.
- [37932] 11484. Lindsey, J. C.; Lusher, M. E.; McDermott, C. J.; White, K. D.; Reid, E.; Rubinsztein, D. C.; Bashir, R.; Hazan, J.; Shaw, P. J.; Bushby, K. M. D.: Mutation analysis of the

spastin gene (SPG4) inpatients with hereditary spastic paraplegia. *J. Med. Genet.* 37:759–765, 2000.

- [37933] 11485. Takahashi, N.; Tuiki, H.; Saya, H.; Kaibuchi, K.: Localization of the gene coding for ROCK II/Rho kinase on human chromosome 2p24. *Genomics* 55:235–237, 1999.
- [37934] 11486. Gruber, A. D.; Pauli, B. U.: Clustering of the human CLCA gene family on the short arm of chromosome 1 (1p22–31). *Genome* 42: 1030–1032, 1999.
- [37935] 11487. Kedra, D.; Pan, H.-Q.; Seroussi, E.; Fransson, I.; Guilbaud, C.; Collins, J. E.; Dunham, I.; Blennow, E.; Roe, B. A.; Piehl, F.; Dumanski, J. P.: Characterization of the human synaptogyrin gene family. *Hum. Genet.* 103: 131–141, 1998.
- [37936] 11488. Hazan, J.; Davoine, C. S.; Mavel, D.; Fonknechten, N.; Paternotte, C.; Fizames, C.; Cruaud, C.; Samson, D.; Muselet, D.; Vega-Czarny, N.; Brice, A.; Gyapay, G.; Heilig, R.; Fontaine, B.; Weissenbach, J.: A fine integrated map of the SPG4 locus excludes an expanded CAG repeat in chromosome 2p-linked autosomal dominant spastic paraplegia. *Genomics* 60:309–319, 1999.
- [37937] 11489. Hazan, J.; Fonknechten, N.; Mavel, D.; Paternotte, C.; Samson, D.; Artiguenave, F.; Davoine, C.-S.; Cruaud, C.; Durr, A.; Wincker, P.; Brottier, P.; Cattolico, L.; Barbe, V.;

Burgunder, J.-M.; Prud'homme, J.-F.; Brice, A.; Fontaine, B.; Heilig, R.; Weissenbach, J.: Spastin, a new AAA protein, is altered in the most frequent form of autosomal dominant spastic paraplegia. *Nature Genet.* 23: 296–303, 1999.

[37938] 11490. Bedford, F. K.; Ashworth, A.; Enver, T.; Wiedemann, L. M.: HEX: a novel homeobox gene expressed during haematopoiesis and conserved between mouse and human. *Nucleic Acids Res.* 21: 1245–1249, 1993.

[37939] 11491. Crompton, M. R.; Bartlett, T. J.; MacGregor, A. D.; Manfioletti, G.; Buratti, E.; Giancotti, V.; Goodwin, G. H.: Identification of a novel vertebrate homeobox gene expressed in haematopoietic cells. *Nucleic Acids Res.* 20: 5661–5667, 1992.

[37940] 11492. D'Elia, A. V.; Tell, G.; Russo, D.; Arturi, F.; Puglisi, F.; Manfioletti, G.; Gattei, V.; Mack, D. L.; Cataldi, P.; Filetti, S.; Di Loreto, C.; Damante, G.: Expression and localization of the homeodomain-containing protein HEX in human thyroid tumors. *J. Clin. Endocr. Metab.* 87: 1376–1383, 2002.

[37941] 11493. Hromas, R.; Radich, J.; Collins, S.: PCR cloning of an orphan homeobox gene (PRH) preferentially expressed in myeloid and liver cells. *Biochem. Biophys. Res. Commun.* 195: 976–983, 1993.

- [37942] 11494.Morgutti, M.; Demori, E.; Pecile, V.; Amoroso, A.; Rustighi, A.;Manfioletti, G.: Genomic organization and chromosome mapping of thehuman homeobox gene HHEX. *Cytogenet. Cell Genet.* 94: 30–32, 2001.
- [37943] 11495.Tanaka, T.; Inazu, T.; Yamada, K.; Myint, Z.; Keng, V. W; Inoue,Y.; Taniguchi, N.; Noguchi, T.: cDNA cloning and expression of rathomeobox gene, Hex, and functional characterization of the protein. *Biochem.J.* 339: 111–117, 1999.
- [37944] 11496.Miles, M. F.; Barhite, S.; Sganga, M.; Elliott, M.: Phosducin–likeprotein: an ethanol–responsive potential modulator of guanine nucleotide–bindingprotein function. *Proc. Nat. Acad. Sci.* 90: 10831–10835, 1993.
- [37945] 11497.Thibault, C.; Wang, J. F.; Charnas, R.; Mirel, D.; Barhite, S.;Miles, M. F.: Cloning and characterization of the rat and human phosducin–likeprotein genes: structure, expression and chromosomal localization. *Biochim.Biophys. Acta* 1444: 346–354, 1999.
- [37946] 11498.Pope, R. K.; Pestonjamas, K. N.; Smith, K. P.; Wulfkuhle, J. D.;Strassel, C. P.; Lawrence, J. B.; Luna, E. J.: Cloning, characterization,and chromosomal localization of human supervillin (SVIL). *Genomics* 52:342–351, 1998.
- [37947] 11499.Ting, H.–J.; Yeh, S.; Nishimura, K.; Chang, C.: Su–

pervillin associates with androgen receptor and modulates its transcriptional activity. Proc.Nat. Acad. Sci. 99: 661–666, 2002.

- [37948] 11500. Bianchi, V.; Robles, R.; Alberio, L.; Furlan, M.; Lammle, B.: Von Willebrand factor–cleaving protease (ADAMTS13) in thrombocytopenic disorders: a severely deficient activity is specific for thrombotic thrombocytopenic purpura. Blood 100: 710–713, 2002.
- [37949] 11501. Fujikawa, K.; Suzuki, H.; McMullen, B.; Chung, D.: Purification of human von Willebrand factor–cleaving protease and its identification as a new member of the metalloproteinase family. Blood 98: 1662–1666, 2001.
- [37950] 11502. Furlan, M.; Robles, R.; Solenthaler, M.; Wassmer, M.; Sandoz, P.; Lammle, B.: Deficient activity of von Willebrand factor–cleaving protease in chronic relapsing thrombotic thrombocytopenic purpura. Blood 89: 3097–3103, 1997.
- [37951] 11503. Gerritsen, H. E.; Robles, R.; Lammle, B.; Furlan, M.: Partial amino acid sequence of purified von Willebrand factor–cleaving protease. Blood 98: 1654–1661, 2001.
- [37952] 11504. Kokame, K.; Matsumoto, M.; Soejima, K.; Yagi, H.; Ishizashi, H.; Funato, M.; Tamai, H.; Konno, M.; Kamide, K.; Kawano, Y.; Miyata, T.; Fujimura, Y.: Mutations and com–

mon polymorphisms in ADAMTS13 gene responsible for von Willebrand factor-cleaving protease activity. *Proc. Nat. Acad. Sci.* 99: 11902–11907, 2002.

- [37953] 11505. Alexopoulou, L.; Holt, A. C.; Medzhitov, R.; and Flavell, R. A.: Recognition of double stranded RNA and activation of NF κ B by Toll-like receptor 3. *Nature* 413: 732–738, 2001.
- [37954] 11506. Advani, R. J.; Bae, H.-R.; Bock, J. B.; Chao, D. S.; Doung, Y.-C.; Prekeris, R.; Yoo, J.-S.; Scheller, R. H.: Seven novel mammalian SNARE proteins localize to distinct membrane compartments. *J. Biol. Chem.* 273: 10317–10324, 1998.
- [37955] 11507. Bock, J. B.; Scheller, R. H.: A fusion of new ideas. *Nature* 387: 133–135, 1997.
- [37956] 11508. Bui, T. D.; Wong, S. H.; Lu, L.; Hong, W.: Endobrevin maps to chromosome 2 in human and chromosome 6 in mouse. *Genomics* 54: 579–580, 1998.
- [37957] 11509. Wong, S. H.; Zhang, T.; Xu, Y.; Subramaniam, V. N.; Griffiths, G.; Hong, W.: Endobrevin, a novel synaptobrevin/VAMP-like protein preferentially associated with the early endosome. *Molec. Biol. Cell* 9: 1549–1563, 1998.
- [37958] 11510. Hewett-Emmett, D.; Tashian, R. E.: Functional diversity, conservation, and convergence in the evolution of

the alpha-, beta- and gamma- carbonicanhydrase gene families. *Molec. Phylogenet. Evol.* 5: 50–77, 1996.

[37959] 11511.Nakagawa, Y.; Uemura, H.; Hirao, Y.; Yoshida, K.; Saga, S.; Yoshikawa,K.: Radiation hybrid mapping of the human MN/CA9 locus to chromosomeband 9p12–p13. *Genomics* 53: 118–119, 1998.

[37960] 11512.Opavsky, R.; Pastorekova, S.; Zelnik, V.; Gibadulinova, A.; Stanbridge,E. J.; Zavada, J.; Kettmann, R.; Pastorek, J.: Human MN/CA9 gene,a novel member of the carbonic anhydrase family: structure and exonto protein domain relationships. *Genomics* 33: 480–487, 1996.

[37961] 11513.Soilleux, E. J.; Barten, R.; Trowsdale, J.: Cutting edge: DC–SIGN;a related gene, DC–SIGNR; and CD23 form a cluster on 19p13. *J. Immun.* 165:2937–2942, 2000.

[37962] 11514.Matoskova, B.; Wong, W. T.; Nomura, N.; Robbins, K. C.; Di Fiore,P. P.: RN–tre specifically binds to the SH3 domain of eps8 with highaffinity and confers growth advantage to NIH3T3 upon carboxy–terminaltruncation. *Oncogene* 12: 2679–2688, 1996.

[37963] 11515.Matoskova, B.; Wong, W. T.; Seki, N.; Nagase, T.; Nomura, N.; Robbins,K. C.; Di Fiore, P. P.: RN–tre identifies a family of tre–relatedproteins displaying a novel potential protein binding domain. *Oncogene* 12:2563–2571, 1996.

- [37964] 11516.Lee, M. P.; Brandenburg, S.; Landes, G. M.; Adams, M.; Miller,G.; Feinberg, A. P.: Two novel genes in the center of the 11p15 imprinted domain escape genomic imprinting. *Hum. Molec. Genet.* 8: 683–690,1999.
- [37965] 11517.Nakamura, M.; Masuda, H.; Horii, J.; Kuma, K.; Yokoyama, N.; Ohba,T.; Nishitani, H.; Miyata, T.; Tanaka, M.; Nishimoto, T.: When overexpressed,a novel centrosomal protein, RanBPM, causes ectopic microtubule nucleations similar to gamma-tubulin. *J. Cell Biol.* 143: 1041–1052, 1998.
- [37966] 11518.Nishitani, H.; Hirose, E.; Uchimura, Y.; Nakamura, M.; Umeda,M.; Nishii, K.; Mori, N.; Nishimoto, T.: Full-sized RanBPM cDNA encodes a protein possessing a long stretch of proline and glutamine within the N-terminal region, comprising a large protein complex. *Gene* 272:25–33, 2001.
- [37967] 11519.Miyamoto, T.; Kanazawa, N.; Kato, S.; Kawakami, M.; Inoue, Y.;Kuhara, T.; Inoue, T.; Takeshita, K.; Tsujino, S.: Diagnosis of Japanese patients with HHH syndrome by molecular genetic analysis: a common mutation, R179X. *J. Hum. Genet.* 46: 260–262, 2001.
- [37968] 11520.Nakajima, M.; Ishii, S.; Mito, T.; Takeshita, K.; Takashima, S.;Takakura, H.; Inoue, I.; Saheki, T.; Akiyoshi,

H.; Ichihara, K.:Clinical, biochemical and ultrastructural study on the pathogenesis of hyperornithinemia–hyperammonemia–homocitrullinuria syndrome. *BrainDev.* 10: 181–185, 1988.

[37969] 11521.Tsujino, S.; Kanazawa, N.; Ohashi, T.; Eto, Y.; Saito, T.; Kira,J.; Yamada, T.: Three novel mutations (G27E, in-sAAC, R179X) in theORNT1 gene of Japanese patients with hyperornithinemia, hyperammonemia,and homocitrullinuria syndrome. *Ann. Neurol.* 47: 625–631, 2000.

[37970] 11522.Grand, R. J. A.; Milner, A. E.; Mustoe, T.; Johnson, G. D.; Owen,D.; Grant, M. L.; Gregory, C. D.: A novel protein expressed in mammalian cells undergoing apoptosis. *Exp. Cell Res.* 218: 439–451, 1995.

[37971] 11523.Twist, C. J.; Beier, D. R.; Disteché, C. M.; Edelhoff, S.; Tedder,T. F.: The mouse Cd83 gene: structure, domain organization, and chromosomal localization. *Immunogenetics* 48: 383–393, 1998.

[37972] 11524.Lee, S. B.; Rao, A. K.; Lee, K.–H.; Yang, X.; Bae, Y. S.; Rhee,S. G.: Decreased expression of phospholipase C–b2 isozyme in human platelets with impaired function. *Blood* 88: 1684–1691, 1996.

[37973] 11525.Mao, G. F.; Vaidyula, V. R.; Kunapuli, S. P.; Rao, A. K.: Lineage–specific defect in gene expression in human

platelet phospholipase C-beta-2 deficiency. *Blood* 99: 905-911, 2002.

[37974] 11526. Park, D.; Jhon, D.-Y.; Kriz, R.; Knopf, J.; Rhee, S. G.: Cloning, sequencing, expression, and Gq-independent activation of phospholipase C-beta-2. *J. Biol. Chem.* 267: 16048-16055, 1992.

[37975] 11527. Park, S. H.; Ryu, S. H.; Suh, P. G.; Kim, H.: Assignment of human PLCB2 encoding PLC-beta-2 to human chromosome 15q15 by fluorescence in situ hybridization. *Cytogenet. Cell Genet.* 83: 48-49, 1998.

[37976] 11528. Rao, A. K.; Kowalska, M. A.; Disa, J.: Impaired cytoplasmic ionized calcium mobilization in inherited platelet secretion defects. *Blood* 74:664-672, 1989.

[37977] 11529. Yang, X.; Sun, L.; Ghosh, S.; Rao, A. K.: Human platelet signaling defect characterized by impaired production of 1,4,5 inositol triphosphate and phosphatidic acid, and diminished pleckstrin phosphorylation. Evidence for defective phospholipase C activation. *Blood* 88: 1676-1683, 1996.

[37978] 11530. Gillen, C. M.; Brill, S.; Payne, J. A.; Forbush, B., III: Molecular cloning and functional expression of the K-Cl cotransporter from rabbit, rat, and human: a new member of the cation-chloride cotransporter family. *J. Biol. Chem.*

271: 16237–16244, 1996.

- [37979] 11531.Larsen, F.; Solheim, J.; Kristensen, T.; Kolsto, A. B.; Prydz,H.: A tight cluster of five unrelated human genes on chromosome 16q22.1. *Hum.Molec. Genet.* 2: 1589–1595, 1993.
- [37980] 11532.Pellegrino, C. M.; Rybicki, A. C.; Musto, S.; Nagel, R. L.; Schwartz,R. S.: Molecular identification and expression of erythroid K:Clcotransporter in human and mouse erythroleukemic cells. *Blood CellsMolec. Dis.* 24: 31–40, 1998.
- [37981] 11533.Hammond, E. M.; Brunet, C. L.; Johnson, G. D.; Parkhill, J.; Milner,A. E.; Brady, G.; Gregory, C. D.; Grand, R. J. A.: Homology betweena human apoptosis specific protein and the product of APG5, a geneinvolved in autophagy in yeast. *FEBS Lett.* 425: 391–395, 1998.
- [37982] 11534.Schmeiser, K.; Armstrong, S.; Hammond, E. M.; Grand, R. J. A.:Assignment of the yeast APG5 human homologue APG5L to chromosome band6q21 by fluorescence in situ hybridisation. *Cytogenet. Cell Genet.* 87:213–214, 1999.
- [37983] 11535.Nakayama, M.; Nakajima, D.; Nagase, T.; Nomura, N.; Seki, N.; Ohara,O.: Identification of high-molecular-weight proteins with multipleEGF-like motifs by motif-

trap screening. *Genomics* 51: 27–34, 1998.

- [37984] 11536.Wu, Q.; Maniatis, T.: Large exons encoding multiple ectodomains are a characteristic feature of protocadherin genes. *Proc. Nat. Acad.Sci.* 97: 3124–3129, 2000.
- [37985] 11537.Falcon–Perez, J. M.; Starcevic, M.; Gautam, R.; Dell'Angelica, E. C.: BLOC–1, a novel complex containing the pallidin and muted proteins involved in the biogenesis of melanosomes and platelet–dense granules. *J. Biol. Chem.* 277: 28191–28199, 2002.
- [37986] 11538.Huang, L.: Personal Communication. San Francisco, Calif. 2/3/2000.
- [37987] 11539.Huang, L.; Kuo, Y.–M.; Gitschier, J.: The pallid gene encodes a novel, syntaxin 13–interacting protein involved in platelet storage pool deficiency. *Nature Genet.* 23: 329–332, 1999.
- [37988] 11540.Paine–Saunders, S.; Viviano, B. L.; Saunders, S.: GPC6, a novel member of the glypican gene family, encodes a product structurally related to GPC4 and is colocalized with GPC5 on human chromosome 13. *Genomics* 57:455–458, 1999.
- [37989] 11541.Veugelers, M.; De Cat, B.; Ceulemans, H.; Bruystens, A. M.; Coomans, C.; Durr, J.; Vermeesch, J.; Marynen, P.; David, G.: Glypican–6, a new member of the glypican

family of cell surface heparan sulfate proteoglycans. J. Biol. Chem. 274: 26968–26977, 1999.

- [37990] 11542. Takei, Y.; Sasaki, S.; Fujiwara, T.; Takahashi, E.; Muto, T.; Nakamura, Y.: Molecular cloning of a novel gene similar to myeloid antigen CD33 and its specific expression in placenta. Cytogenet. Cell Genet. 78:295–300, 1997.
- [37991] 11543. Jacobs, S.; Schurmann, A.; Becker, W.; Bockers, T. M.; Copeland, N. G.; Jenkins, N. A.; Joost, H.–G.: The mouse ADP–ribosylation factor–like 4 gene: two separate promoters direct specific transcription in tissues and testicular germ cell. Biochem. J. 335: 259–265, 1998.
- [37992] 11544. Bootcov, M. R.; Bauskin, A. R.; Valenzuela, S. M.; Moore, A. G.; Bansal, M.; He, X. Y.; Zhang, H. P.; Donnellan, M.; Mahler, S.; Pryor, K.; Walsh, B. J.; Nicholson, R. C.; Fairlie, W. D.; Por, S. B.; Robbins, J. M.; Breit, S. N.: MIC–1, a novel macrophage inhibitory cytokine, is a divergent member of the TGF–beta superfamily. Proc. Nat. Acad. Sci. 94: 11514–11519, 1997.
- [37993] 11545. Bottner, M.; Laaff, M.; Schechinger, B.; Rappold, G.; Unsicker, K.; Suter–Crazzolara, C.: Characterization of the rat, mouse, and human genes of growth/differentiation factor–15/macrophage inhibiting cytokine–1

(GDF-15/MIC-1). *Gene* 237: 105-111, 1999.

[37994] 11546. Brown, D. A.; Breit, S. N.; Buring, J.; Fairlie, W. D.; Bauskin, A. R.; Liu, T.; Ridker, P. M.: Concentration in plasma of macrophageinhibitory cytokine-1 and risk of cardiovascular events in women: a nested case-control study. *Lancet* 359: 2159-2163, 2002.

[37995] 11547. Fairlie, W. D.; Russell, P. K.; Wu, W. M.; Moore, A. G.; Zhang, H.-P.; Brown, P. K.; Bauskin, A. R.; Breit, S. N.: Epitope mapping of the transforming growth factor-beta superfamily protein, macrophageinhibitory cytokine-1 (MIC-1): identification of at least five distinct epitope specificities. *Biochemistry* 40: 65-73, 2001.

[37996] 11548. Hromas, R.; Hufford, M.; Sutton, J.; Xu, D.; Li, Y.; Lu, L.: PLAB, a novel placental bone morphogenetic protein. *Biochim. Biophys. Acta* 1354: 40-44, 1997.

[37997] 11549. Moore, A. G.; Brown, D. A.; Fairlie, W. D.; Bauskin, A. R.; Brown, P. K.; Munier, M. L. C.; Russell, P. K.; Salamonsen, L. A.; Wallace, E. M.; Breit, S. N.: The transforming growth factor-beta superfamily cytokine macrophage inhibitory cytokine-1 is present in high concentrations in the serum of pregnant women. *J. Clin. Endocr. Metab.* 85: 4781-4788, 2000.

[37998] 11550. Paralkar, V. M.; Vail, A. L.; Grasser, W. A.; Brown, T.

A.; Xu, H.; Vukicevic, S.; Ke, H. Z.; Qi, H.; Owen, T. A.; Thompson, D. D.: Cloning and characterization of a novel member of the transforming growth factor- β /bone morphogenetic protein family. *J. Biol. Chem.* 273:13760–13767, 1998.

- [37999] 11551. Yokoyama–Kobayashi, M.; Saeki, M.; Sekine, S.; Kato, S.: Human cDNA encoding a novel TGF- β super-family protein highly expressed in placenta. *J. Biochem.* 122: 622–626, 1997.
- [38000] 11552. Conklin, D. C.; Rixon, M. W.; Kuestner, R. E.; Maurer, M. F.; Whitmore, T. E.; Millar, R. P.: Cloning and gene expression of a novel human ribonucleoprotein. *Biochim. Biophys. Acta* 1492: 465–469, 2000.
- [38001] 11553. Kataoka, N.; Yong, J.; Kim, V. N.; Velazquez, F.; Perkinson, R. A.; Wang, F.; Dreyfuss, G.: Pre-mRNA splicing imprints mRNA in the nucleus with a novel RNA-binding protein that persists in the cytoplasm. *Molec. Cell* 6: 673–682, 2000.
- [38002] 11554. Salicioni, A. M.; Xi, M.; Vanderveer, L. A.; Balsara, B.; Testa, J. R.; Dunbrack, R. L., Jr.; Godwin, A. K.: Identification and structural analysis of human RBM8A and RBM8B: two highly conserved RNA-binding motif proteins that interact with OVCA1, a candidate tumor suppressor.

Genomics 69:54–62, 2000.

[38003] 11555.Zhao, X.-F.; Nowak, N. J.; Shows, T. B.; Aplan, P. D.: MAGOH interacts with a novel RNA-binding protein. Genomics 63: 145–148, 2000.

[38004] 11556.Fischle, W.; Emiliani, S.; Hendzel, M. J.; Nagase, T.; Nomura, N.; Voelter, W.; Verdin, E.: A new family of human histone deacetylases related to *Saccharomyces cerevisiae* HDA1p. J. Biol. Chem. 274: 11713–11720, 1999.

[38005] 11557.Pazin, M. J.; Kadonaga, J. T.: What's up and down with histone deacetylation and transcription? Cell 89: 325–328, 1997.

[38006] 11558.Zhou, L.-J.; Tedder, T. F.: Human blood dendritic cells selectively express CD83, a member of the immunoglobulin superfamily. J. Immun. 154:3821–3835, 1995.

[38007] 11559.Zhou, L. J.; Schwarting, R.; Smith, H. M.; Tedder, T. F.: A novel cell-surface molecule expressed by human interdigitating reticulum cells, Langerhans cells, and activated lymphocytes is a new member of the Ig superfamily. J. Immun. 149: 735–742, 1992.

[38008] 11560.Bost-Usinger, L.; Chen, R. J.; Hillman, D.; Park, P.; Burnside, B.: Multiple kinesin family members expressed in teleost retina and RPE include a novel C-terminal kinesin.

Exp. Eye Res. 64: 781–794,1997.

- [38009] 11561.Hoang, E. H.; Whitehead, J. L.; Dose, A. C.; Burnside, B.: Cloning of a novel C-terminal kinesin (KIFC3) that maps to human chromosome 16q13–q21 and thus is a candidate gene for Bardet–Biedl syndrome. Genomics 52:219–222, 1998.
- [38010] 11562.Norman, D. A. M.; Barton, P. J. R.: Isolation, sequence, and chromosomal localisation of the human I(kappa)BR gene. Ann. Hum. Genet. 64:15–23, 2000.
- [38011] 11563.Ray, P.; Zhang, D.–H.; Elias, J. A.; Ray, A.: Cloning of a differentially expressed I–kappa–B–related protein. J. Biol. Chem. 270: 10680–10685,1995.
- [38012] 11564.Uchiumi, T.; Hinoshita, E.; Haga, S.; Nakamura, T.; Tanaka, T.;Toh, S.; Furukawa, M.; Kawabe, T.; Wada, M.; Kagotani, K.; Okumura,K.; Kohno, K.; Akiyama, S.; Kuwano, M.: Isolation of a novel human canalicular multi-specific organic anion transporter, cMOAT2/MRP3, and its expression in cisplatin-resistant cancer cells with decreased ATP-dependent drug transport. Biochem. Biophys. Res. Commun. 252:103–110, 1998.
- [38013] 11565.Bruhn, S. L.; Pil, P. M.; Essigmann, J. M.; Housman, D. E.; Lippard,S. J.: Isolation and characterization of human cDNA clones encoding a high mobility group box pro-

tein that recognizes structural distortions to DNA caused by binding of the anticancer agent cisplatin. *Proc. Nat. Acad. Sci.* 89: 2307–2311, 1992.

- [38014] 11566. Orphanides, G.; LeRoy, G.; Chang, C.-H.; Luse, D. S.; Reinberg, D.: FACT, a factor that facilitates transcript elongation through nucleosomes. *Cell* 92: 105–116, 1998.
- [38015] 11567. Acker, J.; Wintzerith, M.; Vigneron, M.; Keding, C.: A 14.4KDa acidic subunit of human RNA polymerase II with a putative leucine-zipper. *DNASEq.* 4: 329–331, 1994.
- [38016] 11568. Pusch, C.; Wang, Z.; Roe, B.; Blin, N.: Genomic structure of the RNA polymerase II small subunit (hRPB14.4) locus (POLRF) (sic) and mapping to 22q13.1 by sequence identity. *Genomics* 34: 440–442, 1996.
- [38017] 11569. Caira, F.; Antonson, P.; Pelto-Huikko, M.; Treuter, E.; Gustafsson, J.-A.: Cloning and characterization of RAP250, a novel nuclear receptor coactivator. *J. Biol. Chem.* 275: 5308–5317, 2000.
- [38018] 11570. Guan, X. Y.; Xu, J.; Anzick, S. L.; Zhang, H.; Trent, J. M.; Meltzer, P. S.: Hybrid selection of transcribed sequences from microdissected DNA: isolation of genes within amplified region at 20q11–q13.2 in breast cancer. *Cancer Res.* 56: 3446–3450, 1996.

- [38019] 11571.Ko, L.; Cardona, G. R.; Chin, W. W.: Thyroid hormone receptor-binding protein, an LXXLL motif-containing protein, functions as a general coactivator. *Proc. Nat. Acad. Sci.* 97: 6212–6217, 2000.
- [38020] 11572.Lee, S.-K.; Anzick, S. L.; Choi, J.-E.; Bubendorf, L.; Guan, X.-Y.; Jung, Y.-K.; Kallioniemi, O. P.; Kononen, J.; Trent, J. M.; Azorsa, D.; Jhun, B.-H.; Cheong, J. H.; Lee, Y. C.; Meltzer, P. S.; Lee, J.W.: A nuclear factor, ASC-2, as a cancer-amplified transcriptional coactivator essential for ligand-dependent transactivation by nuclear receptors in vivo. *J. Biol. Chem.* 274: 34283–34293, 1999.
- [38021] 11573.Mahajan, M. A.; Samuels, H. H: A new family of nuclear receptor coregulators that integrate nuclear receptor signaling through CREB-binding protein. *Molec. Cell. Biol.* 20: 5048–5063, 2000.
- [38022] 11574.Zhu, Y.; Kan, L.; Qi, C.; Kanwar, Y. S.; Yeldandi, A. V.; Rao, M. S.; Reddy, J. K.: Isolation and characterization of peroxisome proliferator-activated receptor (PPAR) interacting protein (PRIP) as a coactivator for PPAR. *J. Biol. Chem.* 275: 13510–13516, 2000.
- [38023] 11575.Glockner, G.; Scherer, S.; Schattevoy, R.; Borigt, A.; Weber, J.; Tsui, L.-C.; Rosenthal, A.: Large-scale sequencing of two regions in human chromosome 7q22:

analysis of 650 kb of genomic sequence around the EPO and CUTL1 loci reveals 17 genes. *Genome Res.* 8: 1060–1073, 1998.

[38024] 11576. Yokouchi, M.; Suzuki, R.; Masuhara, M.; Komiya, S.; Inoue, A.; Yoshimura, A.: Cloning and characterization of APS, an adaptor molecule containing PH and SH2 domains that is tyrosine phosphorylated upon B-cell receptor stimulation. *Oncogene* 15: 7–15, 1997.

[38025] 11577. Still, I. H.; Hamilton, M.; Vince, P.; Wolfman, A.; Cowell, J. K.: Cloning of TACC1, an embryonically expressed, potentially transforming coiled coil containing gene, from the 8p11 breast cancer amplicon. *Oncogene* 18: 4032–4038, 1999.

[38026] 11578. Still, I. H.; Vince, P.; Cowell, J. K.: The third member of the transforming acidic coiled coil-containing gene family, TACC3, maps to 4p16, close to translocation breakpoints in multiple myeloma, and is upregulated in various cancer cell lines. *Genomics* 58: 165–170, 1999.

[38027] 11579. Burmester, T.; Weich, B.; Reinhardt, S.; Hankeln, T.: A vertebrate globin expressed in the brain. *Nature* 407: 520–523, 2000.

[38028] 11580. Moens, L.; Dewilde, S.: Globins in the brain. *Nature* 407: 461–462, 2000.

- [38029] 11581.Scott, A. F.: Personal Communication. Baltimore, Md. 9/28/2000.
- [38030] 11582.Vilches, C.; Gardiner, C. M.; Parham, P.: Gene structure and promoter variation of expressed and nonexpressed variants of the KIR2DL5 gene. *J.Immun.* 165: 6416–6421, 2000.
- [38031] 11583.Vilches, C.; Rajalingam, R.; Uhrberg, M.; Gardiner, C. M.; Young, N. T.; Parham, P.: KIR2DL5, a novel killer-cell receptor with a D0–D2 configuration of Ig-like domains. *J. Immun.* 164: 5797–5804, 2000.
- [38032] 11584.Bates, E. E. M.; Fournier, N.; Garcia, E.; Valladeau, J.; Durand, I.; Pin, J.-J.; Zurawski, S. M.; Patel, S.; Abrams, J. S.; Lebecque, S.; Garrone, P.; Saeland, S.: APCs express DCIR, a novel C-type lectin surface receptor containing an immunoreceptor tyrosine-based inhibitory motif. *J. Immun.* 163: 1973–1983, 1999.
- [38033] 11585.Huang, X.; Yuan, Z.; Chen, G.; Zhang, M.; Zhang, W.; Yu, Y.; Cao, X.: Cloning and characterization of a novel ITIM containing lectin-like immunoreceptor LLIR and its two transmembrane region deletion variants. *Biochem.Biophys. Res. Commun.* 281: 131–140, 2001.
- [38034] 11586.Seeger, M.; Kraft, R.; Ferrell, K.; Bech-Otschir, D.; Dumdey, R.; Schade, R.; Gordon, C.; Naumann, M.; Dubiel,

W.: A novel protein complex involved in signal transduction possessing similarities to 26S proteasome subunits. *FASEB J.* 12: 469–478, 1998.

- [38035] 11587. Ann, K.; Kowalchuk, J. A.; Loyet, K. M.; Martin, T. F. J.: Novel Ca^{2+} -binding protein (CAPS) related to UNC-31 required for Ca^{2+} -activated exocytosis. *J. Biol. Chem.* 272: 19637–19640, 1997.
- [38036] 11588. Berwin, B.; Floor, E.; Martin, T. F. J.: CAPS (mammalian UNC-31) protein localizes to membranes involved in dense-core vesicle exocytosis. *Neuron* 21: 137–145, 1998.
- [38037] 11589. Hirose, M.; Nagase, T.; Ishikawa, K.; Kikuno, R.; Nomura, N.; Ohara, O.: Characterization of cDNA clones selected by the GeneMark analysis from size-fractionated cDNA libraries from human brain. *DNAREs.* 6: 329–336, 1999.
- [38038] 11590. Walent, J. H.; Porter, B. W.; Martin, T. F. J.: A novel 145 kDa brain cytosolic protein reconstitutes Ca^{2+} -regulated secretion in permeable neuroendocrine cells. *Cell* 70: 765–775, 1992.
- [38039] 11591. Veldhuis, B.; Spruit, L.; Dauwerse, H. G.; Breuning, M. H.; Peters, D. J.: Genes homologous to the autosomal dominant polycystic kidney disease genes (PKD1 and

PKD2). *Europ. J. Hum. Genet.* 7: 860–872, 1999.

- [38040] 11592. Hughes, J.; Ward, C. J.; Aspinwall, R.; Butler, R.; Harris, P.C.: Identification of a human homologue of the sea urchin receptor for egg jelly: a polycystic kidney disease-like protein. *Hum. Molec. Genet.* 8: 543–549, 1999.
- [38041] 11593. Curtis, B. M.; Scharnowske, S.; Watson, A. J.: Sequence and expression of a membrane-associated C-type lectin that exhibits CD4-independent binding of human immunodeficiency virus envelope glycoprotein gp120. *Proc. Nat. Acad. Sci.* 89: 8356–8360, 1992.
- [38042] 11594. Feinberg, H.; Mitchell, D. A.; Drickamer, K.; Weis, W. I.: Structural basis for selective recognition of oligosaccharides by DC-SIGN and DC-SIGNR. *Science* 294: 2163–2166, 2001.
- [38043] 11595. Geijtenbeek, T. B. H.; Kwon, D. S.; Torensma, R.; van Vliet, S. J.; van Duijnhoven, G. C. F.; Middel, J.; Cornelissen, I. L. M. H. A.; Nottet, H. S. L. M.; KewalRamani, V. N.; Littman, D. R.; Figdor, C. G.; van Kooyk, Y.: DC-SIGN, a dendritic cell-specific HIV-1-binding protein that enhances trans-infection of T cells. *Cell* 100: 587–597, 2000.
- [38044] 11596. Geijtenbeek, T. B. H.; Torensma, R.; van Vliet, S. J.; van Duijnhoven, G. C. F.; Adema, G. J.; van Kooyk, Y.; Figdor, C. G.: Identification of DC-SIGN, a novel dendritic cell-

specific ICAM-3 receptor that supports primary immune responses. *Cell* 100: 575-585, 2000.

[38045] 11597. Battini, J.-L.; Rasko, J. E. J.; Miller, A. D.: A human cell-surface receptor for xenotropic and polytropic murine leukemia viruses: possible role in G protein-coupled signal transduction. *Proc. Nat. Acad. Sci.* 96:1385-1390, 1999.

[38046] 11598. Levy, J. A.: Xenotropism: the elusive viral receptor finally uncovered. *Proc. Nat. Acad. Sci.* 96: 802-804, 1999.

[38047] 11599. Taylor, C. S.; Nouri, A.; Lee, C. G.; Kozak, C.; Kabat, D.: Cloning and characterization of a cell surface receptor for xenotropic and polytropic murine leukemia viruses. *Proc. Nat. Acad. Sci.* 96: 927-932, 1999.

[38048] 11600. Yang, Y.-L.; Guo, L.; Xu, S.; Holland, C. A.; Kitamura, T.; Hunter, K.; Cunningham, J. M.: Receptors for polytropic and xenotropic mouse leukemia viruses encoded by a single gene at Rmc1. *Nature Genet.* 21:216-219, 1999.

[38049] 11601. Aksoy, S.; Raftogianis, R.; Weinshilboum, R.: Human histamine N-methyltransferase gene: structural characterization and chromosomal localization. *Biochem. Biophys. Res. Commun.* 219: 548-554, 1996.

[38050] 11602. Preuss, C. V.; Wood, T. C.; Szumlanski, C. L.; Raftogianis, R. B.; Otterness, D. M.; Girard, B.; Scott, M. C.;

Weinshilboum, R. M.: Human histamine N-methyltransferase pharmacogenetics: common genetic polymorphisms that alter activity. *Molec. Pharm.* 53: 708–717, 1998.

[38051] 11603. Price, R. A.; Scott, M. C.; Weinshilboum, R. M.: Genetic segregation analysis of red blood cell (RBC) histamine N-methyltransferase (HNMT) activity. *Genet. Epidemiol.* 10: 123–131, 1993.

[38052] 11604. Scott, M. C.; Van Loon, J. A.; Weinshilboum, R. M.: Pharmacogenetics of N-methylation: heritability of human erythrocyte histamine N-methyltransferase activity. *Clin. Pharm. Ther.* 43: 256–262, 1988.

[38053] 11605. Yamauchi, K.; Sekizawa, K.; Suzuki, H.; Nakazawa, H.; Ohkawara, Y.; Katayose, D.; Ohtsu, H.; Tamura, G.; Shibahara, S.; Takemura, M.; Maeyama, K.; Watanabe, T.; Sasaki, H.; Shirato, K.; Takishima, T.: Structure and function of human histamine N-methyltransferase: critical enzyme in histamine metabolism in airway. *Am. J. Physiol.* 267: L342–L349, 1994.

[38054] 11606. Yan, L.; Galinsky, R. E.; Bernstein, J. A.; Liggett, S. B.; Weinshilboum, R. M.: Histamine N-methyltransferase pharmacogenetics: association of a common functional polymorphism with asthma. *Pharmacogenetics*

10:261–266, 2000.

- [38055] 11607. Bartoloni, L.; Blouin, J.-L. C.; Sainsbury, A. J.; Gos, A.; Morris, M. A.; Affara, N. A.; DeLozier-Blanchet, C. D.; Antonarakis, S. E.: Assignment of the human dynein heavy chain gene DNAH17L to human chromosome 17p12 by in situ hybridization and radiation hybrid mapping. *Cytogenet. Cell Genet.* 84: 188–189, 1999.
- [38056] 11608. Bartoloni, L.; Blouin, J.-L.; Maiti, A. K.; Sainsbury, A.; Rossier, C.; Gehrig, C.; She, J.-X.; Marron, M. P.; Lander, E. S.; Meeks, M.; Chung, E.; Armengot, M.; Jorissen, M.; Scott, H. S.; DeLozier-Blanchet, C. D.; Gardiner, R. M.; Antonarakis, S. E.: Axonemal beta heavy chain dynein DNAH9: cDNA sequence, genomic structure, and investigation of its role in primary ciliary dyskinesia. *Genomics* 72: 21–33, 2001.
- [38057] 11609. Maiti, A. K.; Mattei, M.-G.; Jorissen, M.; Volz, A.; Zeigler, A.; Bouvagnet, P.: Identification, tissue specific expression, and chromosomal localisation of several human dynein heavy chain genes. *Europ. J. Hum. Genet.* 8: 923–932, 2000.
- [38058] 11610. Milisav, I.; Jones, M. H.; Affara, N. A.: Characterization of a novel human dynein-related gene that is specifically expressed in testis. *Mammalian Genome* 7: 667–672,

1996.

- [38059] 11611.Hart, M. J.; Callow, M. G.; Souza, B.; Polakis, P.: IQ-GAP1, acalmodulin-binding protein with a rasGAP-related domain, is a potentialeffector for cdc42Hs. EMBO J. 15: 2997-3005, 1996.
- [38060] 11612.Sugimoto, N.; Imoto, I.; Fukuda, Y.; Kurihara, N.; Kuroda, S.;Tanigami, A.; Kaibuchi, K.; Kamiyama, R.; Inazawa, J.: IQGAP1, anegative regulator of cell-cell adhesion, is upregulated by gene amplificationat 15q26 in gastric cancer cell lines HSC39 and 40A. J. Hum. Genet. 46:21-25, 2001.
- [38061] 11613.Weissbach, L.; Settleman, J.; Kalady, M. F.; Snijders, A. J.; Murthy,A. E.; Yan, Y.-X.; Bernards, A.: Identification of a human RasGAP-relatedprotein containing calmodulin-binding motifs. J. Biol. Chem. 269:20517-20521, 1994.
- [38062] 11614.Harrington, J. J.; Lieber, M. R.: Functional domains within FEN-1and RAD2 define a family of structure-specific endonucleases: implicationsfor nucleotide excision repair. Genes Dev. 8: 1344-1355, 1994.
- [38063] 11615.Kato, H.; Torigoe, T.: Radioimmunoassay for tumor antigen of humancervical squamous cell carcinoma. Cancer 40: 1621-1628, 1977.

- [38064] 11616.Kuwano, A.; Kondo, I.; Kishi, F.; Suminami, Y.; Kato, H.: Assignment of the squamous cell carcinoma antigen locus (SCC) to 18q21 by insitu hybridization. *Genomics* 30: 626, 1995.
- [38065] 11617.Suminami, Y.; Kishi, F.; Sekiguchi, K.; Kato, H.: Squamous cell carcinoma antigen is a new member of the serine protease inhibitors. *Biochem.Biophys. Res. Commun.* 181: 51–58, 1991.
- [38066] 11618.Barnes, R. C.; Worrall, D. M.: Identification of a novel human serpin gene; cloning sequencing and expression of leupin. *FEBS Lett.* 373:61–65, 1995.
- [38067] 11619.Schick, C.; Kamachi, Y.; Bartuski, A. J.; Cataltepe, S.; Schechter, N. M.; Pemberton, P. A.; Silverman, G. A.: Squamous cell carcinoma antigen 2 is a novel serpin that inhibits the chymotrypsin-like proteinases cathepsin G and mast cell chymase. *J. Biol. Chem.* 272: 1849–1855, 1997.
- [38068] 11620.Dahl, M. R.; Thiel, S.; Matsushita, M.; Fujita, T.; Willis, A.C.; Christensen, T.; Vorup-Jensen, T.; Jensenius, J. C.: MASP-3 and its association with distinct complexes of the mannan-binding lectin complement activation pathway. *Immunity* 15: 127–135, 2001.
- [38069] 11621.Endo, Y.; Sato, T.; Matsushita, M.; Fujita, T.: Exon structure of the gene encoding the human mannose-bind-

ing protein-associated serine protease light chain: comparison with complement C1r and C1s genes. *Int. Immun.* 8: 1355–1358, 1996.

[38070] 11622. Matsushita, M.; Thiel, S.; Jensenius, J. C.; Terai, I.; Fujita, T.: Proteolytic activities of two types of mannose-binding lectin-associated serine protease. *J. Immun.* 165: 2637–2642, 2000.

[38071] 11623. Sato, T.; Endo, Y.; Matsushita, M.; Fujita, T.: Molecular characterization of a novel serine protease involved in activation of the complement system by mannose-binding protein. *Int. Immun.* 6: 665–669, 1994.

[38072] 11624. Takada, F.; Seki, N.; Matsuda, Y.; Takayama, Y.; Kawakami, M.: Localization of the genes for the 100-kDa complement-activating component of C3a-reactive factor (CRARF and Crarf) to human 3q27–q28 and mouse 16B2–B3. *Genomics* 25: 757–759, 1995.

[38073] 11625. Takada, F.; Takayama, Y.; Hatsuse, H.; Kawakami, M.: A new member of the C1s family of complement proteins found in a bactericidal factor, C3a-reactive factor, in human serum. *Biochem. Biophys. Res. Commun.* 196: 1003–1009, 1993.

[38074] 11626. Takayama, Y.; Takada, F.; Nowatari, M.; Kawakami, M.; Matsuura, N.: Gene structure of the P100 serine-

protease component of the humanRa-reactive factor.

Molec. Immun. 36: 505–514, 1999.

[38075] 11627.Dessen, A.; Tang, J.; Schmidt, H.; Stahl, M.; Clark, J. D.; Seehra,J.; Somers, W. S.: Crystal structure of human cytosolic phospholipaseA(2) reveals a novel topology and catalytic mechanism. Cell 97:349–360, 1999.

[38076] 11628.Hansen, L.; Urioste, S.; Petersen, H. V.; Jensen, J. N.; Eiberg,H.; Barbetti, F.; Serup, P.; Hansen, T.; Pedersen, O.: Missense mutationsin the human insulin promoter factor–1 gene and their relation to maturity-onset diabetes of the young and late-onset type 2 diabetes mellitus in Caucasians. J. Clin. Endocr. Metab. 85: 1323–1326, 2000.

[38077] 11629.Johansson, K. A.; Grapin–Botton, A.: Development and diseases of the pancreas. Clin. Genet. 62: 14–23, 2002.

[38078] 11630.Jonsson, J.; Carlsson, L.; Edlund, T.; Edlund, H.: Insulin–promoter–factor1 is required for pancreas development in mice. Nature 371: 606–609,1994.

[38079] 11631.Leonard, J.; Peers, B.; Johnson, T.; Ferreri, I.; Lee, K.; Montminy,M. R.: Characterization of somatostatin transactivating factor–1,a novel homeobox factor that stimulates somatostatin expression in pancreatic islet cells. Molec. Endocr. 7: 1275–1283, 1993.

- [38080] 11632. Macfarlane, W. M.; Frayling, T. M.; Ellard, S.; Evans, J. C.; Allen, L. I. S.; Bulman, M. P.; Ayres, S.; Shepherd, M.; Clark, P.; Millward, A.; Demaine, A.; Wilkin, T.; Docherty, K.; Hattersley, A. T.: Missense mutations in the insulin promoter factor-1 gene predispose to type 2 diabetes. *J. Clin. Invest.* 104: R33–R39, 1999.
- [38081] 11633. Miller, C. P.; McGehee, R. E., Jr.; Habener, J. F.: IDX-1: a new homeodomain transcription factor expressed in rat pancreatic islets and duodenum that transactivates the somatostatin gene. *EMBO J.* 13: 1145–1156, 1994.
- [38082] 11634. Ohlsson, H.; Karlsson, K.; Edlund, T.: IPF1, a homeodomain-containing transactivator of the insulin gene. *EMBO J.* 12: 4251–4259, 1993.
- [38083] 11635. Sharma, S.; Jhala, U. S.; Johnson, T.; Ferreri, K.; Leonard, J.; Montminy, M.: Hormonal regulation of an islet-specific enhancer in the pancreatic homeobox gene STF-1. *Molec. Cell. Biol.* 17: 2598–2604, 1997.
- [38084] 11636. Stoffel, M.; Stein, R.; Wright, C. V. E.; Espinosa, R., III; LeBeau, M. M.; Bell, G. I.: Localization of human homeodomain transcription factor insulin promoter factor 1 (IPF1) to chromosome band 13q12.1. *Genomics* 28: 125–126, 1995.
- [38085] 11637. Stoffers, D. A.; Ferrer, J.; Clarke, W. L.; Habener, J.

F.: Early-onset type-II diabetes mellitus (MODY4) linked to IPF1. (Letter) *Nature Genet.* 17: 138–141, 1997.

[38086] 11638. Stoffers, D. A.; Stanojevic, V.; Habener, J. F.: Insulin promoter factor-1 gene mutation linked to early-onset type 2 diabetes mellitus directs expression of a dominant negative isoform. *J. Clin. Invest.* 102:232–241, 1998.

[38087] 11639. Watada, H.; Kajimoto, Y.; Kaneto, H.; Matsuoka, T.; Fujitani, Y.; Miyazaki, J.; Yamasaki, Y.: Involvement of the homeodomain-containing transcription factor PDX-1 in islet amyloid polypeptide gene transcription. *Biochem. Biophys. Res. Commun.* 229: 746–751, 1996.

[38088] 11640. Wright, N. M.; Metzger, D. L.; Borowitz, S. M.; Clarke, W. L.: Permanent neonatal diabetes mellitus and pancreatic exocrine insufficiency resulting from congenital pancreatic agenesis. *Am. J. Dis. Child.* 147:607–609, 1993.

[38089] 11641. Ashford, M. L. J.; Bond, C. T.; Blair, T. A.; Adelman, J. P.: Cloning and functional expression of a rat heart KATP channel. *Nature* 370:456–459, 1994.

[38090] 11642. Bond, C. T.; Pessia, M.; Xia, X.-M.; Lagrutta, A.; Kavanaugh, M. P.; Adelman, J. P.: Cloning and expression of a family of inward rectifier potassium channels. *Receptors Channels* 2: 183–191, 1994.

- [38091] 11643. Tucker, S. J.; James, M. R.; Adelman, J. P.: Assignment of K(ATP)-1, the cardiac ATP-sensitive potassium channel gene (KCNJ5), to human chromosome 11q24. *Genomics* 28: 127-128, 1995.
- [38092] 11644. Wickman, K.; Seldin, M. F.; Gendler, S. J.; Clapham, D. E.: Partial structure, chromosome localization, and expression of the mouse *Girk4* gene. *Genomics* 40: 395-401, 1997.
- [38093] 11645. Jenkins, N. A.: Personal Communication. Frederick, Md. 5/12/1997.
- [38094] 11646. Khurana, T. S.; Engle, E. C.; Bennett, R. R.; Silverman, G. A.; Selig, S.; Bruns, G. A. P.; Kunkel, L. M.: (CA) repeat polymorphism in the chromosome 18 encoded dystrophin-like protein. *Hum. Molec. Genet.* 3: 841 only, 1994.
- [38095] 11647. Eisenberg, I.; Avidan, N.; Potikha, T.; Hochner, H.; Chen, M.; Olender, T.; Barash, M.; Shemesh, M.; Sadeh, M.; Grabov-Nardini, G.; Shmylevich, I.; Friedmann, A.; Karpati, G.; Bradley, W. G.; Baumbach, L.; Lancet, D.; Ben Asher, E.; Beckmann, J. S.; Argov, Z.; Mitrani-Rosenbaum, S.: The UDP-N-acetylglucosamine 2-epimerase/N-acetylmannosamine kinase gene is mutated in recessive hereditary inclusion body myopathy.

NatureGenet. 29: 83–87, 2001.

- [38096] 11648.Metzinger, L.; Blake, D. J.; Squier, M. V.; Anderson, L. V. B.;Deconinck, A. E.; Nawrotzki, R.; Hilton–Jones, D.; Davies, K. E.:Dystrobrevin deficiency at the sarcolemma of patients with muscular dystrophy. Hum. Molec. Genet. 6: 1185–1191, 1997.
- [38097] 11649.Sadoulet–Puccio, H. M.; Feener, C. A.; Schaid, D. J.; Thibodeau,S. N.; Michels, V. V.; Kunkel, L. M.: The genomic organization of human dystrobrevin. Neurogenetics 1: 37–42, 1997.
- [38098] 11650.Sadoulet–Puccio, H. M.; Khurana, T. S.; Cohen, J. B.; Kunkel, L.M.: Cloning and characterization of the human homologue of a dystrophinrelated phosphoprotein found at the Torpedo electric organ post–synaptic membrane. Hum. Molec. Genet. 5: 489–496, 1996.
- [38099] 11651.Yoshida, M.; Hama, H.; Ishikawa–Sakurai, M.; Ima–mura, M.; Mizuno,Y.; Araishi, K.; Wakabayashi–Takai, E.; Noguchi, S.; Sasaoka, T.;Ozawa, E.: Biochemical evidence for association of dystrobrevin with the sarcoglycan–sar–cospan complex as a basis for understanding sarcogly–canopathy. Hum.Molec. Genet. 9: 1033–1040, 2000.
- [38100] 11652.Bauer, W. R.; Hayes, J. J.; White, J. H.; Wolffe, A. P.: Nucleosome structural changes due to acetylation. J.

Molec. Biol. 236: 685–690,1994.

- [38101] 11653.Furukawa, Y.; Kawakami, T.; Sudo, K.; Inazawa, J.; Matsumine, A.;Akiyama, T.; Nakamura, Y.: Isolation and mapping of a human gene(RPD3L1) that is homologous to RPD3, a transcription factor in *Saccharomyces cerevisiae*. Cytogenet. Cell Genet. 73: 130–133, 1996.
- [38102] 11654.Lee, D. Y.; Hayes, J. J.; Pruss, D.; Wolffe, A. P.: A positive role for histone acetylation in transcription factor access to nucleosomal DNA. Cell 72: 73–84, 1993.
- [38103] 11655.Luo, J.; Su, F.; Chen, D.; Shiloh, A.; Gu, W.: Deacetylation of p53 modulates its effect on cell growth and apoptosis. Nature 408:377–381, 2000.
- [38104] 11656.Magnaghi-Jaulin, L.; Groisman, R.; Naguibneva, I.; Robin, P.; Lorain, S.; Le Villain, J. P.; Troalen, F.; Trouche, D.; Harel-Bellan, A.: Retinoblastoma protein represses transcription by recruiting a histone deacetylase. Nature 391: 601–605, 1998.
- [38105] 11657.Taunton, J.; Hassig, C. A.; Schreiber, S. L.: A mammalian histone deacetylase related to the yeast transcriptional regulator Rpd3p. Science 272:408–411, 1996.
- [38106] 11658.Vidal, M.; Gaber, R. F.: RPD3 encodes a second factor required to achieve maximum positive and negative transcriptional states in *Saccharomyces cerevisiae*. Molec.

Cell. Biol. 11: 6317–6327, 1991.

- [38107] 11659. Vidal, M.; Strich, R.; Esposito, R. E.; Gaber, R. F.: RPD1 (SIN3/UME4) is required for maximal activation and repression of diverse yeast genes. *Molec. Cell. Biol.* 11: 6306–6316, 1991.
- [38108] 11660. Wade, P. A.: Transcriptional control at regulatory checkpoints by histone deacetylases: molecular connections between cancer and chromatin. *Hum. Molec. Genet.* 10: 693–698, 2001.
- [38109] 11661. Wolffe, A. P.: Histone deacetylase: a regulator of transcription. *Science* 272:371–372, 1996.
- [38110] 11662. Zhang, Y.; Sun, Z.-W.; Iratni, R.; Erdjument-Bromage, H.; Tempst, P.; Hampsey, M.; Reinberg, D.: SAP30, a novel protein conserved between human and yeast, is a component of a histone deacetylase complex. *Molec. Cell* 1: 1021–1031, 1998.
- [38111] 11663. Chappell, T. G.; Welch, W. J.; Schlossman, D. M.; Palter, K. B.; Schlesinger, M. J.; Rothman, J. E.: Uncoating ATPase is a member of the 70 kilodalton family of stress proteins. *Cell* 45: 3–13, 1986.
- [38112] 11664. Dworniczak, B.; Mirault, M.-E.: Structure and expression of a human gene coding for a 71 kd heat shock 'cognate' protein. *Nucleic Acids Res.* 15: 5181–5197,

1987.

- [38113] 11665.Tavaria, M.; Gabriele, T.; Anderson, R. L.; Mirault, M.-E.; Baker,E.; Sutherland, G.; Kola, I.: Localization of the gene encoding thehuman heat shock cognate protein, HSP73, to chromosome 11. *Genomics* 29:266–268, 1995.
- [38114] 11666.Ungewickell, E.: The 70-kd mammalian heat shock proteins are structurallyand functionally related to the un-coating protein that releases clathrintriskelia from coated vesicles. *EMBO J.* 4: 3385–3391, 1985.
- [38115] 11667.Camoretti–Mercado, B.; Forsythe, S. M.; LeBeau, M. M.; Espinosa,R., III; Vieira, J. E.; Halayko, A. J.; Willadsen, S.; Kurtz, B.;Ober, C.; Evans, G. A.; Thweatt, R.; Shapiro, S.; Niu, Q.; Qin, Y.;Padrid, P. A.; Solway, J.: Expression and cytogenetic localizationof the human SM22 gene (TAGLN). *Genomics* 49: 452–457, 1998.
- [38116] 11668.Lawson, D.; Harrison, M.; Shapland, C.: Fibroblast transgelinand smooth muscle SM22–alpha are the same protein, the expressionof which is down–regulated in many cell lines. *Cell Motil. Cytoskeleton* 38:250–257, 1997.
- [38117] 11669.Lees–Miller, J. P.; Heeley, D. H.; Smillie, L. B.; Kay, C. M.:Isolation and characterization of an abundant and novel 22–kDa protein(SM22) from chicken gizzard smooth

muscle. J. Biol. Chem. 262: 2988–2993, 1987.

[38118] 11670. Nishida, W.; Kitami, Y.; Abe, M.; Hiwada, K.: Gene cloning and nucleotide sequence of SM22- α from the chicken gizzard smooth muscle. Biochem. Int. 23: 663–668, 1991.

[38119] 11671. Shapland, C.; Hsuan, J. J.; Totty, N. F.; Lawson, D.: Purification and properties of transgelin: a transformation and shape change sensitive actin-gelling protein. J. Cell Biol. 121: 1065–1073, 1993.

[38120] 11672. Shapland, C.; Lowings, P.; Lawson, D.: Identification of new actin-associated polypeptides that are modified by viral transformation and changes in cell shape. J. Cell Biol. 107: 153–161, 1988.

[38121] 11673. Solway, J.; Seltzer, J.; Samaha, F. F.; Kim, S.; Alger, L. E.; Niu, Q.; Morrissey, E. E.; Ip, H. S.; Parmacek, M. S.: Structure and expression of a smooth muscle cell-specific gene, SM22- α . J. Biol. Chem. 270: 13460–13469, 1995.

[38122] 11674. Stanier, P.; Abu-Hayyeh, S.; Murdoch, J. N.; Eddleston, J.; Copp, A. J.: Paralogous Sm22- α (Tagln) genes map to mouse chromosomes 1 and 9: further evidence for a paralogous relationship. Genomics 51: 144–147, 1998.

[38123] 11675. Thweatt, R.; Lumpkin, C. K., Jr.; Goldstein, S.: A

novel gene encoding a smooth muscle protein is overexpressed in senescent human fibroblasts. *Biochem. Biophys. Res. Commun.* 187: 1–7, 1992.

[38124] 11676. Tamanini, F.; Bontekoe, C.; Bakker, C. E.; van Unen, L.; Anar, B.; Willemsen, R.; Yoshida, M.; Galjaard, H.; Oostra, B. A.; Hoogeveen, A. T.: Different targets for the fragile X-related proteins revealed by their distinct nuclear localizations. *Hum. Molec. Genet.* 8: 863–869, 1999.

[38125] 11677. Tamanini, F.; Kirkpatrick, L. L.; Schonkeren, J.; van Unen, L.; Bontekoe, C.; Bakker, C.; Nelson, D. L.; Galjaard, H.; Oostra, B. A.; Hoogeveen, A. T.: The fragile X-related proteins FXR1P and FXR2P contain a functional nucleolar-targeting signal equivalent to the HIV-1 regulatory proteins. *Hum. Molec. Genet.* 9: 1487–1493, 2000.

[38126] 11678. Radice, P.; Pensotti, V.; Jones, C.; Perry, H.; Pierotti, M. A.; Tunnacliffe, A.: The human archain gene, ARCN1, has highly conserved homologs in rice and *Drosophila*. *Genomics* 26: 101–106, 1995.

[38127] 11679. Tunnacliffe, A.; van de Vrugt, H.; Pensotti, V.; Radice, P.: The coatamer protein delta-COP, encoded by the archain gene, is conserved across diverse eukaryotes. *Mammalian Genome* 7: 784–786, 1996.

[38128] 11680. Hawtin, S. R.; Wesley, V. J.; Parslow, R. A.; Simms,

J.; Miles, A.; McEwan, K.; Wheatley, M.: A single residue (arg46) located within the N-terminus of the V(1a) vasopressin receptor is critical for binding vasopressin but not peptide or nonpeptide antagonists. *Molec. Endocr.* 16:600–609, 2002.

[38129] 11681. Morel, A.; O'Carroll, A.-M.; Brownstein, M. J.; Lolait, S. J.: Molecular cloning and expression of a rat V1a arginine vasopressin receptor. *Nature* 356: 523–526, 1992.

[38130] 11682. Thibonnier, M.; Auzan, C.; Madhun, Z.; Wilkins, P.; Berti-Mattera, L.; Clauser, E.: Molecular cloning, sequencing, and functional expression of a cDNA encoding the human V1a vasopressin receptor. *J. Biol. Chem.* 269:3304–3310, 1994.

[38131] 11683. Thibonnier, M.; Graves, M. K.; Wagner, M. S.; Auzan, C.; Clauser, E.; Willard, H. F.: Structure, sequence, expression, and chromosomal localization of the human V(1a) vasopressin receptor gene. *Genomics* 31:327–334, 1996.

[38132] 11684. Young, L. J.; Nilsen, R.; Waymire, K. G.; MacGregor, G. R.; Insel, T. R.: Increased affiliative response to vasopressin in mice expressing the V(1A) receptor from a monogamous vole. *Nature* 400: 766–768, 1999.

[38133] 11685. Langer, L. O., Jr.; Cervenka, J.; Camargo, M.: A se-

vere autosomalrecessive acromesomelic dysplasia, the Hunter–Thompson type, and comparison with the Grebe type. *Hum. Genet.* 81: 323–328, 1989.

[38134] 11686. Robin, N. H.: Personal Communication. Cleveland, Ohio 11/3/1997.

[38135] 11687. Runner, M. N.: Linkage of brachypodism: a new member of linkage group V of the house mouse. *J. Hered.* 50: 81–84, 1959.

[38136] 11688. Thomas, J. T.; Lin, K.; Nandedkar, M.; Camargo, M.; Cervenka, J.; Luyten, F. P.: A human chondrodysplasia due to a mutation in a TGF–beta superfamily member. *Nature Genet.* 12: 315–317, 1996.

[38137] 11689. Tsumaki, N.; Tanaka, K.; Arikawa–Hirasawa, E.; Nakase, T.; Kimura, T.; Thomas, J. T.; Ochi, T.; Luyten, F. P.; Yamada, Y.: Role of CDMP–1 in skeletal morphogenesis: promotion of mesenchymal cell recruitment and chondrocyte differentiation. *J. Cell Biol.* 144: 161–173, 1999.

[38138] 11690. Huh, T.–L.; Kim, Y.–O.; Oh, I.–U.; Song, B. J.; Inazawa, J.: Assignment of the human mitochondrial NAD(+)-specific isocitrate dehydrogenase alpha subunit (IDH3A) gene to 15q25.1–q25.2 by in situ hybridization. *Genomics* 31:295–296, 1996.

[38139] 11691. Kim, Y.–O.; Oh, I.–U.; Park, H.–S.; Jeng, J.; Song, B.

J.; Huh, T.-L.: Characterization of a cDNA clone for human NAD(+)-specific isocitrate dehydrogenase alpha-subunit and structural comparison with its isoenzymes from different species. *Biochem. J.* 308: 63–68, 1995.

[38140] 11692. Amann, J.; Kidd, V. J.; Lahti, J. M.: Characterization of putative human homologues of the yeast chromosome transmission fidelity gene, CHL1. *J. Biol. Chem.* 272: 3823–3832, 1997.

[38141] 11693. Amann, J.; Valentine, M.; Kidd, V. J.; Lahti, J. M.: Localization of Chl1-related helicase genes to human chromosome regions 12p11 and 12p13: similarity between parts of these genes and conserved human telomeric-associated DNA. *Genomics* 32: 260–265, 1996.

[38142] 11694. Frank, S.; Werner, S.: The human homologue of the yeast CHL1 gene is a novel keratinocyte growth factor-regulated gene. *J. Biol. Chem.* 271: 24337–24340, 1996.

[38143] 11695. Barnes, L. D.; Garrison, P. N.; Siprashvili, Z.; Guranowski, A.; Robinson, A. K.; Ingram, S. W.; Croce, C. M.; Ohta, M.; Huebner, K.: Fhit, a putative tumor suppressor in humans, is a dinucleoside 5'-prime, 5'-tripleprime-P(1), P(3)-triphosphate hydrolase. *Biochemistry* 35: 11529–11535, 1996.

[38144] 11696. Bernar, J.; Funderburk, S. J.; Sparkes, R. S.: The in-

ducible fragilesite on chromosome 3. (Letter) Hum. Genet. 66: 373 only, 1984.

- [38145] 11697.Jin, C.-J.; Miners, J. O.; Lillywhite, K. J.; Mackenzie, P. I.: cDNA cloning and expression of two new members of the human liverUDP-glucuronosyltransferase 2B subfamily. Biochem. Biophys. Res.Comm. 194: 496–503, 1993.
- [38146] 11698.Krasnewich, D.; Kozak, C. A.; Nebert, D. W.; Mackenzie, P. I.:Localization of UDP glucuronosyltransferase gene(s) on mouse chromosome
- [38147] 11699.Somat. Cell Molec. Genet. 13: 179–182, 1987.5. Monaghan, G.; Clarke, D. J.; Povey, S.; See, C. G.; Boxer, M.;Burchell, B.: Isolation of a human YAC contig encompassing a clusterof UGT2 genes and its regional localization to chromosome 4q13. Genomics 23:496–499, 1994.
- [38148] 11700.Monaghan, G.; Povey, S.; Burchell, B.; Boxer, M.: Localizationof a bile acid UDP-glucuronosyltransferase gene (UGT2B) to chromosome4 using the polymerase chain reaction. Genomics 13: 908–909, 1992.
- [38149] 11701.Riedy, M.; Wang, J. Y.; Miller, A. P.; Buckler, A.; Hall, J.; Guida,M.: Genomic organization of the UGT2b gene cluster on human chromosome4q13. Pharmacogenetics 10: 251–260, 2000.
- [38150] 11702.Buc-Caron, M. H.; Condamine, H.; Kerjaschki, D.:

Rat Heymann nephritis antigen is closely related to brushin, a glycoprotein present in early mouse embryo epithelia. *Ann. Inst. Pasteur/Immun.* 138: 707–722, 1987.

[38151] 11703. Chatelet, F.; Brianti, E.; Ronco, P.; Roland, J.; Verroust, P.: Ultrastructural localization by monoclonal antibodies of brush border antigens expressed by glomeruli: II. Extrarenal distribution. *Am. J. Path.* 122: 512–519, 1986.

[38152] 11704. Chowdhary, B. P.; Lundgren, S.; Johansson, M.; Hjalmar, G.; Akerstrom, G.; Gustavsson, I.; Rask, L.: In situ hybridization mapping of a 500-kDa calcium-sensing protein gene (LRP2) to human chromosome region 2q31–q32.1 and porcine chromosome region 15q22–q24. *Cytogenet. Cell Genet.* 71: 120–123, 1995.

[38153] 11705. Farquhar, M. G.: The unfolding story of megalin (gp330): now recognized as a drug receptor (Editorial) *J. Clin. Invest.* 96: 1184 only, 1995.

[38154] 11706. Farquhar, M. G.; Saito, A.; Kerjaschki, D.; Orlando, R. A.: The Heymann nephritis antigenic complex: megalin (gp330) and RAP. *J. Am. Soc. Nephrol.* 6: 35–47, 1995.

[38155] 11707. Hjalmar, G.; Murray, E.; Crumley, G.; Harazim, W.; Lundgren, S.; Onyango, I.; Bo, E. K.; Larsson, M.; Juhlin, C.; Hellman, P.; Davis, H.; Akerstrom, G.; Rask, L.; Morse, B.:

Cloning and sequencing of human gp330, a Ca(2+)-binding receptor with potential intracellular signaling properties. *Europ. J. Biochem.* 239: 132–137, 1996.

- [38156] 11708. Arinami, T.; Kondo, I.; Hamaguchi, H.; Nakajima, S.: Multifocal meningiomas in a patient with a constitutional ring chromosome 22. *J. Med. Genet.* 23: 178–180, 1986.
- [38157] 11709. Petrella, R.; Levine, S.; Wilmot, P. L.; Ashar, K. D.; Casamassima, A. C.; Shapiro, L. R.: Multiple meningiomas in a patient with constitutional ring chromosome 22. *Am. J. Med. Genet.* 47: 184–186, 1993.
- [38158] 11710. Peyrard, M.; Fransson, I.; Xie, Y.-G.; Han, F.-Y.; Rutledge, M. H.; Swahn, S.; Collins, J. E.; Dunham, I.; Collins, V. P.; Dumanski, J. P.: Characterization of a new member of the human beta-adaptin gene family from chromosome 22q12, a candidate meningioma gene. *Hum. Molec. Genet.* 3: 1393–1399, 1994.
- [38159] 11711. Peyrard, M.; Pan, H.-Q.; Kedra, D.; Fransson, I.; Swahn, S.; Hartman, K.; Clifton, S. W.; Roe, B. A.; Dumanski, J. P.: Structure of the promoter and genomic organization of the human beta-prime-adaptin gene (BAM22) from chromosome 22q12. *Genomics* 36: 112–117, 1996.
- [38160] 11712. Zankl, H.; Zang, K. D.: Correlations between clinical and cytogenetical data in 180 human meningiomas. *Can-*

cer Genet. Cytogenet. 1: 351–356,1980.

- [38161] 11713.Bai, C.; Sen, P.; Hofmann, K.; Ma, L.; Goebel, M.; Harper, J. W.; Elledge, S. J.: SKP1 connects cell cycle regulators to the ubiquitinproteolysis machinery through a novel motif, the F-box. Cell 86:263–274, 1996.
- [38162] 11714.Kraus, B.; Pohlschmidt, M.; Leung, A. L. S.; Germino, G. G.; Snarey,A.; Schneider, M. C.; Reeders, S. T.; Frischauf, A.–M.: A novel cyclin gene (CCNF) in the region of the polycystic kidney disease gene (PKD1). Genomics 24:27–33, 1994.
- [38163] 11715.Obermayr, F.; Sutherland, H. F.; Kraus, B.; Frischauf, A.–M.: Mouse cyclin F maps to a conserved linkage group on mouse chromosome17. Mammalian Genome 6: 149–150, 1995.
- [38164] 11716.Donaldson, S. H.; Hirsh, A.; Li, D. C.; Holloway, G.; Chao, J.;Boucher, R. C.; Gabriel, S. E.: Regulation of the epithelial sodiumchannel by serine proteases in human airways. J. Biol. Chem. 277:8338–8345, 2002.
- [38165] 11717.Harvey, K. F.; Dinudom, A.; Cook, D. I.; Kumar, S.: The Nedd4–likeprotein KIAA0439 is a potential regulator of the epithelial sodiumchannel. J. Biol. Chem. 276: 8597–8601, 2001.
- [38166] 11718.Arriza, J. L.; Kavanaugh, M. P.; Fairman, W. A.; Wu,

Y.-N.; Murdoch, G. H.; North, R. A.; Amara, S. G.: Cloning and expression of a human neutral amino acid transporter with structural similarity to the glutamate transporter gene family. *J. Biol. Chem.* 268: 15329–15332, 1993.

[38167] 11719. Hofmann, K.; Duker, M.; Fink, T.; Lichter, P.; Stoffel, W.: Human neutral amino acid transporter ASCT1: structure of the gene (SLC1A4) and localization to chromosome 2p13–p15. *Genomics* 24: 20–26, 1994.

[38168] 11720. Shafqat, S.; Tamarappoo, B. K.; Kilberg, M. S.; Purnam, R. S.; McNamara, J. O.; Guadano-Ferraz, A.; Freneau, R. T., Jr.: Cloning and expression of a novel Na(+)-dependent neutral amino acid transporter structurally related to mammalian Na(+)/glutamate cotransporters. *J. Biol. Chem.* 268: 15351–15355, 1993.

[38169] 11721. Zerangue, N.; Kavanaugh, M. P.: ASCT-1 is a neutral amino acid exchanger with chloride channel activity. *J. Biol. Chem.* 271: 27991–27994, 1996.

[38170] 11722. Albertin, G.; Rossi, G. P.; Majone, F.; Tiso, N.; Mattara, A.; Danieli, G. A.; Pessina, A. C.; Palu, G.: Fine mapping of the human endothelin-converting enzyme gene by fluorescent in situ hybridization and radiation hybrids. *Biochem. Biophys. Res. Commun.* 221: 682–687, 1996.

[38171] 11723. Sharp, J. D.; Pickard, R. T.; Chiou, X. G.; Manetta, J.

V.; Kovacevic,S.; Miller, J. R.; Varshavsky, A. D.; Roberts, E. F.; Strifler, B.A.; Brems, D. N.; Kramer, R. M.: Serine 228 is essential for catalyticactivities of 85–kDa cytosolic phospholipase A2. *J. Biol. Chem.* 269:23250–23254, 1994.

[38172] 11724.Tamimi, R.; Steingrimsson, E.; Copeland, N. G.; Dyer–Montgomery,K.; Lee, J. E.; Hernandez, R.; Jenkins, N. A.; Tapscott, S. J.: TheNEUROD gene maps to human chromosome 2q32 and mouse chromosome 2. *Genomics* 34:418–421, 1996.

[38173] 11725.Newton, J. S.; Deed, R. W.; Mitchell, E. L. D.; Murphy, J. J.;Norton, J. D.: A B cell specific immediate early human gene is locatedon chromosome band 1q31 and encodes an alpha helical basic phosphoprotein. *Biochim.Biophys. Acta* 1216: 314–316, 1993.

[38174] 11726.Sierra, D. A.; Gilbert, D. J.; Householder, D.; Grishin, N. V.;Yu, K.; Ukidwe, P.; Barker, S. A.; He, W.; Wensel, T. G.; Otero, G.;Brown, G.; Copeland, N. G.; Jenkins, N. A.; Wilkie, T. M.: Evolutionof the regulators of G–protein signaling multigene family in mouseand human. *Genomics* 79: 177–185, 2002.

[38175] 11727.Hofstra, R. M. W.; Valdenaire, O.; Arch, E.; Osinga, J.; Kroes,H.; Loffler, B.–M.; Hamosh, A.; Meijers, C.; Buys, C. H. C. M.: Aloss–of–function mutation in the endothelin–

converting enzyme 1 (ECE-1) associated with Hirschsprung disease, cardiac defects, and autonomic dysfunction.

(Letter) Am. J. Hum. Genet. 64: 304–308, 1999.

- [38176] 11728. Matsuoka, R.; Sawamura, T.; Yamada, K.; Yoshida, M.; Furutani, Y.; Ikura, T.; Shiraki, T.; Hoshikawa, H.; Shimada, K.; Tanzawa, K.; Masaki, T.: Human endothelin converting enzyme gene (ECE1) mapped to chromosomal region 1p36.1. Cytogenet. Cell Genet. 72: 322–324, 1996.
- [38177] 11729. Schmidt, M.; Kroger, B.; Jacob, E.; Seulberger, H.; Subkowski, T.; Otter, R.; Meyer, T.; Schmalzing, G.; Hillen, H.: Molecular characterization of human and bovine endothelin converting enzyme (ECE-1). FEBS Lett. 356:238–243, 1994.
- [38178] 11730. Shimada, K.; Matsushita, Y.; Wakabayashi, K.; Takahashi, M.; Matsubara, A.; Iijima, Y.; Tanzawa, K.: Cloning and functional expression of human endothelin-converting enzyme cDNA. Biochem. Biophys. Res. Commun. 207:807–812, 1995.
- [38179] 11731. Valdenaire, O.; Rohrbacher, E.; Mattei, M.-G.: Organization of the gene encoding the human endothelin-converting enzyme (ECE-1). J. Biol. Chem. 270: 29794–29798, 1995.
- [38180] 11732. Yorimitsu, K.; Moroi, K.; Inagaki, N.; Saito, T.; Ma-

suda, Y.;Masaki, T.; Seino, S.; Kimura, S.: Cloning and sequencing of a humanendothelin converting enzyme in renal adenocarcinoma (ACHN) cellsproducing endothelin-2. Biochem. Biophys. Res. Commun. 208: 721–727,1995.

[38181] 11733.Ivey–Hoyle, M.; Conroy, R.; Huber, H. E.; Goodhart, P. J.; Oliff,A.; Heimbrook, D. C.: Cloning and characterization of E2F–2, a novelprotein with the biochemical properties of transcription factor E2F. Molec.Cell. Biol. 13: 7802–7812, 1993.

[38182] 11734.Lees, J. A.; Saito, M.; Vidal, M.; Valentine, M.; Look, T.; Harlow,E.; Dyson, N.; Helin, K.: The retinoblastoma protein binds to a familyof E2F transcription factors. Molec. Cell. Biol. 13: 7813–7825,1993.

[38183] 11735.Cloud, J. E.; Rogers, C.; Reza, T. L.; Ziebold, U.; Stone, J. R.;Picard, M. H.; Caron, A. M.; Bronson, R. T.; Lees, J. A.: Mutantmouse models reveal the relative roles of E2F1 and E2F3 in vivo. Molec.Cell. Biol. 22: 2663–2672, 2002.

[38184] 11736.He, Y.; Armanious, M. K.; Thomas, M. J.; Cress, W. D.: Identificationof E2F–3B, an alternative form of E2F–3 lacking a conserved N–terminalregion. Oncogene 19: 3422–3433, 2000.

[38185] 11737.Sharp, J. D.; White, D. L.; Chiou, X. G.; Goodson, T.;

Gamboa, G. C.; McClure, D.; Burgett, S.; Hoskins, J.; Skatrud, P. L.; Sportsman, J. R.; Becker, G. W.; Kang, L. H.; Roberts, E. F.; Kramer, R. M.: Molecular cloning and expression of human Ca(2+)-sensitive cytosolic phospholipase A2. *J. Biol. Chem.* 266: 14850–14853, 1991.

[38186] 11738. Sheridan, A. M.; Force, T.; Yoon, H.-J.; O'Leary, E.; Choukroun, G.; Taheri, M. R.; Bonventre, J. V.: PLIP, a novel splice variant of Tip60, interacts with group IV cytosolic phospholipase A2, induces apoptosis, and potentiates prostaglandin production. *Molec. Cell. Biol.* 21: 4470–4481, 2001.

[38187] 11739. Skorecki, K. L.: Personal Communication. Toronto, Canada 4/19/1995.

[38188] 11740. Tay, A.; Simon, J. S.; Squire, J.; Hamel, K.; Jacob, H. J.; Skorecki, K.: Cytosolic phospholipase A2 gene in human and rat: chromosomal localization and polymorphic markers. *Genomics* 26: 138–141, 1995.

[38189] 11741. Halford, M. M.; Armes, J.; Buchert, M.; Meskenaite, V.; Grail, D.; Hibbs, M. L.; Wilks, A. F.; Farlie, P. G.; Newgreen, D. F.; Hovens, C. M.; Stacker, S. A.: Ryk-deficient mice exhibit craniofacial defects associated with perturbed Eph receptor crosstalk. *Nature Genet.* 25: 414–418, 2000.

[38190] 11742. Nakamura, S.; Stock, D. W.; Wydner, K. L.;

Bollekens, J. A.; Takeshita, K.; Nagai, B. M.; Chiba, S.; Kitamura, T.; Freeland, T. M.; Zhao, Z.; Minowada, J.; Lawrence, J. B.; Weiss, K. M.; Ruddle, F. H.: Genomic analysis of a new mammalian distal-less gene: *Dlx7*. *Genomics* 38:314–324, 1996.

[38191] 11743. Singh, J.; Itahana, Y.; Parrinello, S.; Murata, K.; Desprez, P.-Y.: Molecular cloning and characterization of a zinc finger protein involved in Id-1-stimulated mammary epithelial cell growth. *J. Biol. Chem.* 276: 11852–11858, 2001.

[38192] 11744. Kumar, S.; Kinoshita, M.; Noda, M.; Copeland, N. G.; Jenkins, N. A.: Induction of apoptosis by the mouse *Nedd2* gene, which encodes a protein similar to the product of the *Caenorhabditis elegans* cell death gene *ced-3* and the mammalian IL-1-beta-converting enzyme. *Genes Dev.* 8: 1613–1626, 1994.

[38193] 11745. Bollag, R. J.; Siegfried, Z.; Cebra-Thomas, J. A.; Garvey, N.; Davison, E. M.; Silver, L. M.: An ancient family of embryonically expressed mouse genes sharing a conserved protein motif with the *T* locus. *Nature Genet.* 7: 383–389, 1994.

[38194] 11746. Campbell, C.; Goodrich, K.; Casey, G.; Beatty, B.: Cloning and mapping of a human gene (*TBX2*) sharing a

highly conserved protein motif with the *Drosophila omb* gene. *Genomics* 28: 255–260, 1995.

- [38195] 11747. Campbell, C. E.; Casey, G.; Goodrich, K.: Genomic structure of *TBX2* indicates conservation with distantly related T-box genes. *Mammalian Genome* 9: 70–73, 1998.
- [38196] 11748. Jacobs, J. J. L.; Keblusek, P.; Robanus-Maandag, E.; Kristel, P.; Lingbeek, M.; Nederlof, P. M.; van Welsem, T.; van de Vijver, M. J.; Koh, E. Y.; Daley, G. Q.; van Lohuizen, M.: Senescence bypass screen identifies *TBX2*, which represses *Cdkn2a*(p19ARF) and is amplified in a subset of human breast cancers. *Nature Genet.* 26: 291–299, 2000.
- [38197] 11749. Law, D. J.; Gebhur, T.; Garvey, N.; Agulnik, S. I.; Silver, L.M.: Identification, characterization, and localization to chromosome 17q21–22 of the human *TBX2* homolog, member of a conserved developmental gene family. *Mammalian Genome* 6: 793–797, 1995.
- [38198] 11750. Pflugfelder, G. O.; Roth, H.; Poeck, B.: A homology domain shared between *Drosophila optomotor-blind* and mouse *Brachyury* is involved in DNA binding. *Biochem. Biophys. Res. Commun.* 186: 918–925, 1992.
- [38199] 11751. Sowden, J. C.; Holt, J. K. L.; Meins, M.; Smith, H. K.; Bhattacharya, S. S.: Expression of *Drosophila omb*-related T-box genes in the developing human and mouse neural

retina. Invest. Ophthalm. Vis. Sci. 42: 3095–3102,2001.

- [38200] 11752.Walter, L.; Dirks, B.; Rothermel, E.; Heyens, M.; Szpirer, C.;Levan, G.; Gunther, E.: A novel, conserved gene of the rat that isdevelopmentally regulated in the testis. Mammalian Genome 5: 216–221,1994.
- [38201] 11753.Walter, L.; Marynen, P.; Szpirer, J.; Levan, G.; Gunther, E.:Identification of a novel conserved human gene, TEGT. Genomics 28:301–304, 1995.
- [38202] 11754.Xu, Q.; Reed, J. C.: Bax inhibitor–1, a mammalian apoptosis suppressoridentified by functional screening in yeast. Molec. Cell 1: 337–346,1998.
- [38203] 11755.Hsu, Y.–C.; Perin, M. S.: Human neuronal pentraxin II (NPTX2):conservation, genomic structure, and chromosomal localization. Genomics 28:220–227, 1995.
- [38204] 11756.Schlimgen, A. K.; Helms, J. A.; Vogel, H.; Perin, M. S.: Neuronalpentraxin, a secreted protein with homology to acute phase proteinsof the immune system. Neuron 14: 519–526, 1995.
- [38205] 11757.Kumar, S.; White, D. L.; Takai, S.; Turczynowicz, S.; Juttner,C. A.; Hughes, T. P.: Apoptosis regulatory gene NEDD2 maps to humanchromosome segment 7q34–35, a region frequently affected in haematologicalneoplasms. Hum. Genet. 95: 641–644, 1995.

- [38206] 11758.Lassus, P.; Opitz-Araya, X.; Lazebnik, Y.: Requirement for caspase-2 in stress-induced apoptosis before mitochondrial permeabilization. *Science* 297:1352–1354, 2002.
- [38207] 11759.Aksoy, I. A.; Callen, D. F.; Apostolou, S.; Her, C.; Weinshilboum, R. M.: Thermolabile phenol sulfotransferase gene (STM) localization to human chromosome 16p11.2. *Genomics* 23: 275–277, 1994.
- [38208] 11760.Aksoy, I. A.; Weinshilboum, R. M.: Human thermolabile phenol sulfotransferase gene (STM): molecular cloning and structural characterization. *Biochem. Biophys. Res. Commun.* 208: 786–795, 1995.
- [38209] 11761.Wood, T. C.; Aksoy, I. A.; Aksoy, S.; Weinshilboum, R. M.: Human liver thermolabile phenol sulfotransferase: cDNA cloning, expression and characterization. *Biochem. Biophys. Res. Commun.* 198: 1119–1127, 1994.
- [38210] 11762.Zhu, X.; Veronese, M. E.; Bernard, C. C. A.; Sansom, L. N.; McManus, M. E.: Identification of two human brain aryl sulfotransferase cDNAs. *Biochem. Biophys. Res. Commun.* 195: 120–127, 1993.
- [38211] 11763.Fuhlbrigge, R. C.; Kieffer, J. D.; Armerding, D.; Kupper, T. S.: Cutaneous lymphocyte antigen is a specialized form of PSGL-1 expressed on skin-homing T cells.

Nature 389: 978–981, 1997.

- [38212] 11764.Herron, M. J.; Nelson, C. M.; Larson, J.; Snapp, K. R.; Kansas, G. S.; Goodman, J. L.: Intracellular parasitism by the human granulocyte ehrlichiosis bacterium through the P-selectin ligand, PSGL-1. Science 288:1653–1656, 2000.
- [38213] 11765.Veldman, G. M.; Bean, K. M.; Cumming, D. A.; Eddy, R. L.; Sait, S. N. J.; Shows, T. B.: Genomic organization and chromosomal localization of the gene encoding human P-selectin glycoprotein ligand. J. Biol.Chem. 270: 16470–16475, 1995.
- [38214] 11766.Yang, J.; Galipeau, J.; Kozak, C. A.; Furie, B. C.; Furie, B.: Mouse P-selectin glycoprotein ligand-1: molecular cloning, chromosomal localization, and expression of a functional P-selectin receptor. Blood 87:4176–4186, 1996.
- [38215] 11767.Brown, C. B.; Boyer, A. S.; Runyan, R. B.; Barnett, J. V.: Requirement of type III TGF-beta receptor for endocardial cell transformation in the heart. Science 283: 2080–2082, 1999.
- [38216] 11768.Lewis, K. A.; Gray, P. C.; Blount, A. L.; MacConell, L. A.; Wiater, E.; Bilezikjian, L. M.; Vale, W.: Betaglycan binds inhibin and can mediate functional antagonism of activin signalling. Nature 404:411–414, 2000.

- [38217] 11769.Hu, Y.-F.; Luscher, B.; Admon, A.; Mermod, N.; Tjian, R.: Transcriptionfactor AP-4 contains multiple dimerization domains that regulate dimerspecificity. *Genes Dev.* 4: 1741-1752, 1990.
- [38218] 11770.Mermod, N.; Williams, T. J.; Tjian, R.: Enhancer binding factorsAP-4 and AP-1 act in concert to activate SV40 late transcription invitro. *Nature* 332: 557-561, 1988.
- [38219] 11771.Carr, C. S.; Sharp, P. A.: A helix-loop-helix protein relatedto the immunoglobulin E box-binding proteins. *Molec. Cell. Biol.* 10:4384-4388, 1990.
- [38220] 11772.Bonner, C. A.; Loftus, S. K.; Wasmuth, J. J.: Isolation, characterization,and precise physical localization of human CDX1, a caudal-type homeoboxgene. *Genomics* 28: 206-211, 1995.
- [38221] 11773.Subramanian, V.; Meyer, B. I.; Gruss, P.: Disruption of the murinehomeobox gene Cdx1 affects axial skeletal identities by altering themesodermal expression domains of Hox genes. *Cell* 83: 641-653, 1995.
- [38222] 11774.Henske, E. P.; Short, M. P.; Jozwiak, S.; Bovey, C. M.; Ramlakhan,S.; Haines, J. L.; Kwiatkowski, D. J.: Identification of VAV2 on9q34 and its exclusion as the tuberous sclerosis gene TSC1. *Ann.Hum. Genet.* 59: 25-37, 1995.

- [38223] 11775.Schwientek, T.; Nomoto, M.; Levery, S. B.; et al:
Control of O-glycanbranch formation. J. Biol. Chem. 274:
4504-4512, 1999.
- [38224] 11776.Walczak, H.; Degli-Esposti, M. A.; Johnson, R. S.;
Smolak, P. J.; Waugh, J. Y.; Boiani, N.; Timour, M. S.; Ger-
hart, M. J.; Schooley, K. A.; Smith, C. A.; Goodwin, R. G.;
Rauch, C. T.: TRAIL-R2: a novel apoptosis-mediating re-
ceptor for TRAIL. EMBO J. 16: 5386-5397, 1997.
- [38225] 11777.Wu, G. S.; Burns, T. F.; McDonald, E. R., III; Jiang,
W.; Meng, R.; Krantz, I. D.; Kao, G.; Gan, D.-D.; Zhou, J.-Y.;
Muschel, R.; Hamilton, S. R.; Spinner, N. B.; Markowitz, S.;
Wu, G.; El-Deiry, W. S.: KILLER/DR5 is a DNA damage-in-
ducible p53-regulated death receptor gene. Nature Genet.
17: 141-143, 1997.
- [38226] 11778.Degli-Esposti, M. A.; Smolak, P. J.; Walczak, H.;
Waugh, J.; Huang, C.-P.; DuBose, R. F.; Goodwin, R. G.;
Smith, C. A.: Cloning and characterization of TRAIL-R3, a
novel member of the emerging TRAIL receptor family.
J. Exp. Med. 186: 1165-1170, 1997.
- [38227] 11779.Kanaar, R.; Troelstra, C.; Swagemakers, S. M. A.;
Essers, J.; Smit, B.; Franssen, J.-H.; Pastink, A.; Bezzubova,
O. Y.; Buerstedde, J.-M.; Clever, B.; Heyer, W.-D.; Hoeij-
makers, J. H. J.: Human and mouse homologs of the Sac-

charomyces cerevisiae RAD54 DNA repair gene: evidence for functional conservation. Curr. Biol. 6: 828–838, 1996.

[38228] 11780. Matsuda, M.; Miyagawa, K.; Takahashi, M.; Fukuda, T.; Kataoka, T.; Asahara, T.; Inui, H.; Watatani, M.; Yasutomi, M.; Kamada, N.; Dohi, K.; Kamiya, K.: Mutations in the RAD54 recombination gene in primary cancers. Oncogene 18: 3427–3430, 1999.

[38229] 11781. Tucker, J. E.; Winkfein, R. J.; Cooper, C. B.; Schnetkamp, P. P.: cDNA cloning of the human retinal rod Na–Ca + K exchanger: comparison with a revised bovine sequence. Invest. Ophthalmol. Vis. Sci. 39: 435–440, 1998.

[38230] 11782. Tucker, J. E.; Winkfein, R. J.; Murthy, S. K.; Friedman, J. S.; Walter, M. A.; Demetrick, D. J.; Schnetkamp, P. P. M.: Chromosomal localization and genomic organization of the human retinal rod Na–Ca+K exchanger. Hum. Genet. 103: 411–414, 1998.

[38231] 11783. Dennis, J. A.; Healy, P. J.; Beaudet, A. L.; O'Brien, W. E.: Molecular definition of bovine argininosuccinate synthetase deficiency. Proc. Nat. Acad. Sci. 86: 7947–7951, 1989.

[38232] 11784. McCarrey, J. R.; Riggs, A. D.: Determinator–inhibitor pairs as a mechanism for threshold setting in development: a possible function for pseudogenes. Proc. Nat.

Acad. Sci. 83: 679–683, 1986.

- [38233] 11785.Su, T.-S.; Nussbaum, R. L.; Airhart, S.; Ledbetter, D. H.; Mohandas,T.; O'Brien, W. E.; Beaudet, A. L.: Human chromosomal assignmentsfor 14 argininosuccinate synthetase pseudogenes: cloned DNAs as reagentsfor cytogenetic analysis. *Am. J. Hum. Genet.* 36: 954–964, 1984.
- [38234] 11786.Hara, Y.; Wakamori, M.; Ishii, M.; Maeno, E.; Nishida, M.; Yoshida,T.; Yamada, H.; Shimizu, S.; Mori, E.; Kudoh, J.; Shimizu, S.; Kurose,H.; Okada, Y.; Imoto, K.; Mori, Y.: LTRPC2 Ca(2+)-permeable channelactivated by changes in redox status confers susceptibility to celldeath. *Molec. Cell* 9: 163–173, 2002.
- [38235] 11787.Harteneck, C.; Plant, T. D.; Schultz, G.: From worm to man: threesubfamilies of TRP channels. *Trends Neurosci.* 23: 159–166, 2000.
- [38236] 11788.Kudoh, J.; Nagamine, K.; Asakawa, S.; Abe, I.; Kawasaki, K.; Maeda,H.; Tsujimoto, S.; Minoshima, S.; Ito, F.; Shimizu, N.: Localizationof 16 exons to a 450-kb region involved in the autoimmune polyglandulardisease type I (APECED) on human chromosome 21q22.3. *DNA Res.* 4:45–52, 1997.
- [38237] 11789.Nagamine, K.; Kudoh, J.; Minoshima, S.; Kawasaki, K.; Asakawa,S.; Ito, F.; Shimizu, N.: Molecular cloning of a

novel putative Ca(2+)channel protein (TRPC7) highly expressed in brain. Genomics 54:124–131, 1998.

- [38238] 11790.Perraud, A.–L.; Fleig, A.; Dunn, C. A.; Bagley, L. A.; Launay,P.; Schmitz, C.; Stokes, A. J.; Zhu, Q.; Bessman, M. J.; Penner, R.;Kinet, J.–P.; Scharenberg, A. M.: ADP–ribose gating of the calcium–permeableLTRPC2 channel revealed by Nudix motif homology. Nature 411: 595–599,2001.
- [38239] 11791.Sano, Y.; Inamura, K.; Miyake, A.; Mochizuki, S.; Yokoi, H.; Matsushime,H.; Furuichi, K.: Immunocyte Ca(2+) influx system mediated by LTRPC2. Science 293:1327–1330, 2001.
- [38240] 11792.Koyama, N.; Ishibashi, K.; Kuwahara, M.; Inase, N.; Ichioka, M.;Sasaki, S.; Marumo, F.: Cloning and functional expression of humanaquaporin8 cDNA and analysis of its gene. Genomics 54: 169–172,1998.
- [38241] 11793.Viggiano, L.; Rocchi, M.; Svelto, M.; Calamita, G.: Assignmentof the aquaporin–8 water channel gene (AQP8) to human chromosome 16p12. Cytogenet.Cell Genet. 84: 208–210, 1999.
- [38242] 11794.Graler, M. H.; Bernhardt, G.; Lipp, M.: EDG6, a novel G–protein–coupledreceptor related to receptors for bioactive lysophospholipids, isspecifically expressed in lymphoid tissue. Genomics 53: 164–169,1998.

- [38243] 11795.Jedlicka, A. E.; Taylor, E. W.; Meyers, D. A.; Liu, Z.; Levitt,R. C.: Localization of the highly polymorphic locus D19S120 to 19p13.3by linkage. *Cytogenet. Cell. Genet.* 65: 140 only, 1994.
- [38244] 11796.Koegl, M.; Hoppe, T.; Schlenker, S.; Ulrich, H. D.; Mayer, T. U.;Jentsch, S.: A novel ubiquitination factor, E4, is involved in multiubiquitinchain assembly. *Cell* 96: 635–644, 1999.
- [38245] 11797.Nonaka, S.; Tanaka, Y.; Okada, Y.; Takeda, S.; Harada, A.; Kanai,Y.; Kido, M.; Hirokawa, N.: Randomization of left–right asymmetrydue to loss of nodal cilia generating leftward flow of extraembryonicfluid in mice lacking KIF3B motor protein. *Cell* 95: 829–837, 1998.
- [38246] 11798.Yamazaki, H.; Nakata, T.; Okada, Y.; Hirokawa, N.: KIF3A/B: aheterodimeric kinesin superfamily protein that works as a microtubuleplus end–directed motor for membrane organelle transport. *J. CellBiol.* 130: 1387–1399, 1995.
- [38247] 11799.Allikmets, R.; Schriml, L. M.; Hutchinson, A.; Romano–Spica, V.;Dean, M.: A human placenta–specific ATP–binding cassette gene (ABCP)on chromosome 4q22 that is involved in multidrug resistance. *CancerRes.* 58: 5337–5339, 1998.

- [38248] 11800.Bailey–Dell, K. J.; Hassel, B.; Doyle, L. A.; Ross, D. D.: Promoter characterization and genomic organization of the human breast cancer resistance protein (ATP-binding cassette transporter G2) gene. *Biochim.Biophys. Acta* 1520: 234–241, 2001.
- [38249] 11801.Doyle, L. A.; Yang, W.; Abruzzo, L. V.; Krogmann, T.; Gao, Y.;Rishi, A. K.; Ross, D. D.: A multidrug resistance transporter from human MCF–7 breast cancer cells. *Proc. Nat. Acad. Sci.* 95: 15665–15670,1998.
- [38250] 11802.Suzuki, N.; Zara, J.; Sato, T.; Ong, E.; Bakhiet, N.; Oshima, R.G.; Watson, K. L.; Fukuda, M. N.: A cytoplasmic protein, bystin, interacts with trophinin, tastin, and cytokeratin and may be involved in trophinin–mediated cell adhesion between trophoblast and endometrial epithelial cells. *Proc. Nat. Acad. Sci.* 95: 5027–5032, 1998.
- [38251] 11803.Cerutti, A.; Schaffer, A.; Goodwin, R. G.; Shah, S.; Zan, H.; Ely,S.; Casali, P.: Engagement of CD153 (CD30 ligand) by CD30–positive T cells inhibits class switch DNA recombination and antibody production in human IgD–positive IgM–positive B cells. *J. Immun.* 165: 786–794,2000.
- [38252] 11804.Croager, E. J.; Abraham, L. J.: Characterisation of the human CD30 ligand gene structure. *Biochim. Biophys.*

Acta 1353: 231–235,1997.

[38253] 11805.Smith, C. A.; Gruss, H.-J.; Davis, T.; Anderson, D.; Farrah, T.;Baker, E.; Sutherland, G. R.; Brannan, C. I.; Copeland, N. G.; Jenkins,N. A.; Grabstein, K. H.; Gliniak, B.; and 9 others: CD30 antigen,a marker for Hodgkin's lymphoma, is a receptor whose ligand definesan emerging family of cytokines with homology to TNF. Cell 73: 1349–1360,1993.

[38254] 11806.Hurskainen, T. L.; Hirohata, S.; Seldin, M. F.; Apte, S. S.: ADAM–TS5,ADAM–TS6, and ADAM–TS7, novel members of a new family of zinc metalloproteases:general features and genomic distribution of the ADAM–TS family. J.Biol. Chem. 274: 25555–25563, 1999.

[38255] 11807.Tang, B. L.; Hong, W.: ADAMTS: a novel family of proteases withan ADAM protease domain and thrombospondin 1 repeats. FEBS Lett. 445:223–225, 1999.

[38256] 11808.Tortorella, M. D.; Burn, T. C.; Pratta, M. A.; Abbaszade, I.; Hollis,J. M.; Liu, R.; Rosenfeld, S. A.; Copeland, R. A.; Decicco, C. P.;Wynn, R.; Rockwell, A.; Yang, F.; and 16 others: Purification andcloning of aggrecanase–1: a member of the ADAMTS family of proteins. Science 284:1664–1666, 1999.

[38257] 11809.Higashide, T.; Inana, G.: Characterization of the

gene for HRG4(UNC119), a novel photoreceptor synaptic protein homologous to unc-119. *Genomics* 57:446–450, 1999.

[38258] 11810.Higashide, T.; McLaren, M. J.; Inana, G.: Localization of HRG4,a photoreceptor protein homologous to unc-119, in ribbon synapse. *Invest.Ophthal. Vis. Sci.* 39: 690–698, 1998.

[38259] 11811.Higashide, T.; Murakami, A.; McLaren, M. J.; Inana, G.: Cloning of the cDNA for a novel photoreceptor protein. *J. Biol. Chem.* 271:1797–1804, 1996.

[38260] 11812.Swanson, D. A.; Chang, J. T.; Campochiaro, P. A.; Zack, D. J.;Valle, D.: Mammalian orthologs of *C. elegans* unc-119 highly expressed in photoreceptors. *Invest. Ophthal. Vis. Sci.* 39: 2085–2094, 1998.

[38261] 11813.Gundelfinger, E.: Personal Communication. Madgeburg, FRG. 1/8/1999.

[38262] 11814.Hashida, H.; Goto, J.; Zhao, N.; Takahashi, N.; Hirai, M.; Kanazawa,I.; Sakaki, Y.: Cloning and mapping of ZNF231, a novel brain-specific gene encoding neuronal double zinc finger protein whose expression is enhanced in a neurodegenerative disorder, multiple system atrophy(MSA). *Genomics* 54: 50–58, 1998.

[38263] 11815.Santorelli, F. M.; Patrono, C.; Fortini, D.; Tessa, A.;

Comanducci,G.; Bertini, E.; Pierallini, A.; Amabile, G. A.; Casali, C.: Intrafamilialvariability in hereditary spastic paraplegia associated with an SPG4gene mutation. Neu-
rology 55: 702–705, 2000.

[38264] 11816.Sauter, S.; Mitterski, B.; Klimpe, S.; Bonsch, D.; Schols, L.;Visbeck, A.; Papke, T.; Hopf, H. C.; Engel, W.; Deufel, T.; Epplen,J. T.; Neesen, J.: Mutation analysis of the spastin gene (SPG4) inpatients in Germany with autosomal dominant hereditary spastic paraplegia. Hum.Mutat. 20: 127–132, 2002.

[38265] 11817.Svenson, I. K.; Ashley–Koch, A. E.; Pericak–Vance, M. A.; Marchuk,D. A.: A second leaky splice–site mutation in the spastin gene. (Letter) Am.J. Hum. Genet. 69: 1407–1409, 2001.

[38266] 11818.Flannery, C. R.; Hughes, C. E.; Schumacher, B. L.; Tudor, D.; Aydelotte,M. B.; Kuettner, K. E.; Caterson, B.: Articular cartilage superficialzone protein (SZP) is homolo-
gous to megakaryocyte stimulating factorprecursor and is a multifunctional proteoglycan with potential growth-
promoting,cytoprotective, and lubricating properties in cartilage metabolism. Biochem.Biophys. Res. Commun. 254: 535–541, 1999.

[38267] 11819.Ikegawa, S.; Sano, M.; Koshizuka, Y.; Nakamura, Y.:

Isolation, characterization and mapping of the mouse and human PRG4 (proteoglycan4) genes. *Cytogenet. Cell Genet.* 90: 291–297, 2000.

[38268] 11820. Merberg, D. M.; Fitz, L. J.; Temple, P.; Giannotti, J.; Murtha, P.; Fitzgerald, M.; Scaltreto, H.; Kelleher, K.; Preissner, K.; Kriz, R.; Jacobs, K.; Turner, K.: In: Preissner, K. T.; Rosenblatt, S.; Kost, C.; Wegerhoff, J.; Mosher, D. F. (eds.): *Biology of Vitronectins and Their Receptors*. Elsevier Science, B.V. 1993. Pp. 45–53.

[38269] 11821. Schumacher, B. L.; Block, J. A.; Schmid, T. M.; Aydelotte, M. B.; Kuettner, K. E.: A novel proteoglycan synthesized and secreted by chondrocytes of the superficial zone of articular cartilage. *Arch. Biochem. Biophys.* 15: 144–152, 1994.

[38270] 11822. Pennica, D.; Swanson, T. A.; Welsh, J. W.; Roy, M. A.; Lawrence, D. A.; Lee, J.; Brush, J.; Taneyhill, L. A.; Deuel, B.; Lew, M.; Watanabe, C.; Cohen, R. L.; Melhem, M. F.; Finley, G. G.; Quirke, P.; Goddard, A. D.; Hillan, K. J.; Gurney, A. L.; Botstein, D.; Levine, A. J.: WISP genes are members of the connective tissue growth factor family that are up-regulated in Wnt-1-transformed cells and aberrantly expressed in human colon tumors. *Proc. Nat. Acad. Sci.* 95: 14717–14722, 1998.

- [38271] 11823.Tanaka, S.; Sugimachi, K.; Saeki, H.; Kinoshita, J.; Ohga, T.;Shimada, M.; Maehara, Y.; Sugimachi, K.: A novel variant of WISP1lacking a von Willebrand type C module overexpressed in scirrhousgastric carcinoma. *Oncogene* 20: 5525–5532, 2001.
- [38272] 11824.Gee, S.; Krauss, S. W.; Miller, E.; Aoyagi, K.; Arenas, J.; Conboy,J. G.: Cloning of mDEAH9, a putative RNA heli-case and mammalian homologueof *Saccharomyces cere-visiae* splicing factor Prp43. *Proc. Nat. Acad.Sci.* 94: 11803–11807, 1997.
- [38273] 11825.Imamura, O.; Sugawara, M.; Furuichi, Y.: Cloning and characterizationof a putative human RNA helicase gene of the DEAH–box protein family. *Biochem.Biophys. Res. Commun.* 240: 335–340, 1997.
- [38274] 11826.Sano, K.; Tanihara, H.; Heimark, R. L.; Obata, S.; Davidson, M.;St. John, T.; Taketani, S.; Suzuki, S.: Proto-cadherins: a large familyof cadherin–related molecules in central nervous system. *EMBO J.* 12:2249–2256, 1993.
- [38275] 11827.Eisenblatter, T.; Galla, H.–J.: A new multidrug re-sistance proteinat the blood–brain barrier. *Biochem. Bio-phys. Res. Commun.* 293:1273–1278, 2002.
- [38276] 11828.Miyake, K.; Mickley, L.; Litman, T.; Zhan, Z.; Robey, R.; Cristensen,B.; Brangi, M.; Greenberger, L.; Dean, M.;

Fojo, T.; Bates, S. E.: Molecular cloning of cDNAs which are highly overexpressed in mitoxantrone-resistant cells: demonstration of homology to ABC transport genes. *Cancer Res.* 59:8–13, 1999.

[38277] 11829.Ozvegy, C.; Litman, T.; Szakacs, G.; Nagy, Z.; Bates, S.; Varadi,A.; Sarkadi, B.: Functional characterization of the human multidrugtransporter, ABCG2, expressed in insect cells. *Biochem. Biophys.Res. Commun.* 285: 111–117, 2001.

[38278] 11830.Dominguez, O.; Ashhab, Y.; Sabater, L.; Belloso, E.; Caro, P.;Pujol–Borrell, R.: Cloning of ARE-containing genes by AU–motif–directeddisplay. *Genomics* 54: 278–286, 1998.

[38279] 11831.Kostrub, C. F.; Knudsen, K.; Subramani, S.; Enoch, T.: Hus1p,a conserved fission yeast checkpoint protein, interacts with Rad1pand is phosphorylated in response to DNA damage. *EMBO J.* 17: 2055–2066,1998.

[38280] 11832.Lieberman, H. B.; Hopkins, K. M.; Nass, M.; Demetrick, D.; Davey,S.: A human homolog of the *Schizosaccharomyces pombe rad9+* checkpointcontrol gene. *Proc. Nat. Acad. Sci.* 93: 13890–13895, 1996.

[38281] 11833.Katashima, R.; Iwahana, H.; Fujimura, M.; Yamaoka, T.; Ishizuka,T.; Tatibana, M.; Itakura, M.: Molecular

cloning of a human cDNA for the 41-kDa phosphoribosylpyrophosphate synthetase-associated protein.

Biochim. Biophys. Acta 1396: 245–250, 1998.

- [38282] 11834. Katashima, R.; Iwahana, H.; Fujimura, M.; Yamaoka, T.; Itakura, M.: Assignment of the human phosphoribosylpyrophosphate synthetase-associated protein 41 gene (PRPSAP2) to 17p11.2–p12. Genomics 54: 180–181, 1998.
- [38283] 11835. Ando, A.; Kikuti, Y. Y.; Kawata, H.; Okamoto, N.; Imai, T.; Eki, T.; Yokoyama, K.; Soeda, E.; Ikemura, T.; Abe, K.; Inoko, H.: Cloning of a new kinesin-related gene located at the centromeric end of the human MHC region. Immunogenetics 39: 194–200, 1994.
- [38284] 11836. Endow, S. A.; Higuchi, H.: A mutant of the motor protein kinesin that moves in both directions on microtubules. Nature 406: 913–916, 2000.
- [38285] 11837. Hoyt, M. A.; He, L.; Totis, L.; Saunders, W. S.: Loss of function of *Saccharomyces cerevisiae* kinesin-related CIN8 and KIP1 is suppressed by KAR3 motor domain mutations. Genetics 135: 35–44, 1993.
- [38286] 11838. Janatipour, M.; Naumov, Y.; Ando, A.; Sugimura, K.; Okamoto, N.; Tsuji, K.; Abe, K.; Inoko, H.: Search for MHC-associated genes in human: five new genes centromeric to HLA-DP with yet unknown functions. Immunogenetics

35:272–278, 1992.

- [38287] 11839.Koike, N.; Hida, A.; Numano, R.; Hirose, M.; Sakaki, Y.; Tei, H.: Identification of the mammalian homologues of the *Drosophila* timeless gene, timeless1. *FEBS Lett.* 441: 427–431, 1998.
- [38288] 11840.Sangoram, A. M.; Saez, L.; Antoch, M. P.; Gekakis, N.; Staknis, D.; Whiteley, A.; Fruechte, E. M.; Vitaterna, M. H.; Shimomura, K.; King, D. P.; Young, M. W.; Weitz, C. J.; Takahashi, J. S.: Mammalian circadian autoregulatory loop: a timeless ortholog and mPer1 interact and negatively regulate CLOCK–BMAL1–induced transcription. *Neuron* 21:1101–1113, 1998.
- [38289] 11841.Zylka, M. J.; Shearman, L. P.; Levine, J. D.; Jin, X.; Weaver, D. R.; Reppert, S. M.: Molecular analysis of mammalian timeless. *Neuron* 21:1115–1122, 1998.
- [38290] 11842.Ardley, H. C.; Rose, S. A.; Tan, N.; Leek, J. P.; Markham, A. F.; Robinson, P. A.: Genomic organization of the human ubiquitin–conjugating enzyme gene, UBE2L6 on chromosome 11q12. *Cytogenet. Cell Genet.* 89:137–140, 2000.
- [38291] 11843.Kumar, S.; Kao, W. H.; Howley, P. M.: Physical interaction between specific E2 and Hect E3 enzymes determines functional cooperativity. *J. Biol. Chem.* 272:

13548–13554, 1997.

- [38292] 11844.tom Dieck, S.; Sanmarti-Vila, L.; Langnaese, K.; Richter, K.; Kindler, S.; Soyke, A.; Wex, H.; Smalla, K.-H.; Kampf, U.; Franzer, J.-T.; Stumm, M.; Garner, C. C.; Gundelfinger, E. D.: Bassoon, a novel zinc-fingerCAG/glutamine-repeat protein selectively localized at the active zone of presynaptic nerve terminals. *J. Cell Biol.* 142: 499–509, 1998.
- [38293] 11845.Winter, c.; tom Dieck, S.; Boeckers, T. M.; Bockmann, J.; Kampf, U.; Sanmarti-Vila, L.; Langnaese, K.; Altmann, W.; Stumm, M.; Soyke, A.; Wieacker, P.; Garner, C. C.; Gundelfinger, E. D.: The presynaptic cytomatrix protein Bassoon: sequence and chromosomal localization of the human BSN gene. *Genomics* 57: 389–397, 1999.
- [38294] 11846.Bui, T. D.; Levy, E. R.; Subramaniam, V. N.; Lowe, S. L.; Hong, W.: cDNA characterization and chromosomal mapping of human Golgi SNARE GS27 and GS28 to chromosome 17. *Genomics* 57: 285–288, 1999.
- [38295] 11847.Lowe, S. L.; Peter, F.; Subramaniam, V. N.; Wong, S. H.; Hong, W.: A SNARE involved in protein transport through the Golgi apparatus. *Nature* 389:881–884, 1997.
- [38296] 11848.Nagahama, M.; Orci, L.; Ravazzola, M.; Amherdt, M.; Lacomis, L.; Tempst, P.; Rothman, J. E.; Sollner, T. H.: A

v-SNARE implicated in intra-Golgi transport. *J. Cell Biol.* 133: 507–516, 1996.

[38297] 11849. Subramaniam, V. N.; Peter, F.; Philp, R.; Wong, S. H.; Hong, W.: GS28, a 28-kilodalton Golgi SNARE that participates in ER-Golgi transport. *Science* 272: 1161–1163, 1996.

[38298] 11850. Hay, J. C.; Chao, D. S.; Kuo, C. S.; Scheller, R. H.: Protein interactions regulating vesicle transport between the endoplasmic reticulum and Golgi apparatus in mammalian cells. *Cell* 89: 149–158, 1997.

[38299] 11851. Li, J.; Ding, S.-F.; Habib, N. A.; Fermor, B. F.; Wood, C. B.; Gilmour, R. S.: Partial characterization of a cDNA for human stearyl-CoA desaturase and changes in its mRNA expression in some normal and malignant tissues. *Int. J. Cancer* 57: 348–352, 1994.

[38300] 11852. Ntambi, J. M.; Miyazaki, M.; Stoehr, J. P.; Lan, H.; Kendziora, C. M.; Yandell, B. S.; Song, Y.; Cohen, P.; Friedman, J. M.; Attie, A. D.: Loss of stearyl-CoA desaturase-1 function protects mice against adiposity. *Proc. Nat. Acad. Sci.* 99: 11482–11486, 2002.

[38301] 11853. Thiede, M. A.; Ozols, J.; Strittmatter, P.: Construction and sequence of cDNA for rat liver stearyl coenzyme A desaturase. *J. Biol. Chem.* 261: 13230–13235, 1986.

- [38302] 11854.Zhang, L.; Ge, L.; Parimoo, S.; Stenn, K.; Prouty, S. M.: Humanstearoyl-CoA desaturase: alternative transcripts generated from a single gene by usage of tandem polyadenylation sites. *Biochem. J.* 340:255–264, 1999.
- [38303] 11855.Zheng, Y.; Eilertsen, K. J.; Ge, L.; Zhang, L.; Sundberg, J. P.; Prouty, S. M.; Stenn, K. S.; Parimoo, S.: Scd1 is expressed in sebaceous glands and is disrupted in the asebia mouse. (Letter) *Nature Genet.* 23:268–270, 1999.
- [38304] 11856.Duh, F.-M.; Latif, F.; Weng, Y.; Geil, L.; Modi, W.; Stackhouse, T.; Matsumura, F.; Duan, D. R.; Linehan, W. M.; Lerman, M. I.; Gnarr, J. R.: cDNA cloning and expression of the human homolog of the sea urchin fascin and *Drosophila* singed genes which encodes an actin-bundling protein. *DNA Cell Biol.* 13: 821–827, 1994.
- [38305] 11857.Mosialos, G.; Birkenbach, M.; Ayehunie, S.; Matsumura, F.; Pinkus, G. S.; Kieff, E.; Langhoffer, E.: Circulating human dendritic cells differentially express high levels of a 55-kd actin-bundling protein. *Am.J. Path.* 148: 593–600, 1996.
- [38306] 11858.Ono, S.; Yamakita, Y.; Yamashiro, S.; Matsudaira, P. T.; Gnarr, J. R.; Obinata, T.; Matsumura, F.: Identification of an actin binding region and a protein kinase C phosphorylation site on human fascin. *J.Biol. Chem.* 272:

2527–2533, 1997.

- [38307] 11859. Pinkus, G. S.; Pinkus, J. L.; Langhoff, E.; Matsumura, F.; Yamashiro, S.; Mosialos, G.; Said, J. W.: Fascin, a sensitive new marker for Reed–Sternberg cells of Hodgkin's disease: evidence for a dendritic or B cell derivation? *Am. J. Path.* 150: 543–562, 1997.
- [38308] 11860. Sonderbye, L.; Magerstadt, R.; Blatman, R. N.; Prefer, F. I.; Langhoff, E.: Selective expression of human fascin (p55) by dendritic leukocytes. *Adv. Exp. Med. Biol.* 471: 41–46, 1997.
- [38309] 11861. Yamakita, Y.; Ono, S.; Matsumura, F.; Yamashiro, S.: Phosphorylation of human fascin inhibits its actin binding and bundling activities. *J. Biol. Chem.* 271: 12632–12638, 1996.
- [38310] 11862. Yamashiro–Matsumura, S.; Matsumura, F.: Intracellular localization of the 55–kD actin–bundling protein in cultured cells: spatial relationships with actin, alpha-actinin, tropomyosin, and fimbrin. *J. Cell Biol.* 103:631–640, 1986.
- [38311] 11863. Yamashiro–Matsumura, S.; Matsumura, F.: Purification and characterization of an F–actin–bundling 55–kilodalton protein from HeLa cells. *J. Biol. Chem.* 260: 5087–5097, 1985.

- [38312] 11864.Carapeti, M.; Aguiar, R. C. T.; Chase, A.; Goldman, J. M.; Cross,N. C. P.: Assignment of the steroid receptor coactivator-1 (SRC-1)gene to human chromosome band 2p23. *Genomics* 52: 242-244, 1998.
- [38313] 11865.Hayashi, Y.; Ohmori, S.; Ito, T.; Seo, H.: A splicing variantof steroid receptor coactivator-1 (SRC-1E): the major isoform of SRC-1to mediate thyroid hormone action. *Biochem. Biophys. Res. Commun.* 236:83-87, 1997.
- [38314] 11866.Jenster, G.; Spencer, T. E.; Burcin, M. M.; Tsai, S. Y.; Tsai,M.-J.; O'Malley, B. W.: Steroid receptor induction of gene transcription:a two-step model. *Proc. Nat. Acad. Sci.* 94: 7879-7884, 1997.
- [38315] 11867.Liu, Z.; Wong, J.; Tsai, S. Y.; Tsai, M. J.; O'Malley, B. W.:Steroid receptor coactivator-1 (SRC-1) enhances ligand-dependent andreceptor-dependent cell-free transcription of chromatin. *Proc. Nat.Acad. Sci.* 96: 9485-9490, 1999.
- [38316] 11868.Onate, S. A.; Tsai, S. Y.; Tsai, M.-J.; O'Malley, B. W.: Sequenceand characterization of a coactivator for the steroid hormone receptorsuperfamily. *Science* 270: 1354-1357, 1995.
- [38317] 11869.Takeshita, A.; Yen, P. M.; Misiti, S.; Cardona, G. R.; Liu, Y.;Chin, W. W.: Molecular cloning and properties of a

full-length putative thyroid hormone receptor coactivator. *Endocrinology* 137: 3594–3597, 1996.

[38318] 11870. Masson, J.-Y.; Stasiak, A. Z.; Stasiak, A.; Benson, F. E.; West, S. C.: Complex formation by the human RAD51C and XRCC3 recombination repair proteins. *Proc. Nat. Acad. Sci.* 98: 8440–8446, 2001.

[38319] 11871. Selfors, L. M.; Schutzman, J. L.; Borland, C. Z.; Stern, M. J.: Soc-2 encodes a leucine-rich repeat protein implicated in fibroblast growth factor receptor signaling. *Proc. Nat. Acad. Sci.* 95: 6903–6908, 1998.

[38320] 11872. Sieburth, D. S.; Sun, Q.; Han, M.: SUR-8, a conserved Ras-binding protein with leucine-rich repeats, positively regulates Ras-mediated signaling in *C. elegans*. *Cell* 94: 119–130, 1998.

[38321] 11873. Gibbs, P. E. M.; McGregor, W. G.; Maher, V. M.; Nis-son, P.; Lawrence, C. W.: A human homolog of the *Saccharomyces cerevisiae* REV3 gene, which encodes the catalytic subunit of DNA polymerase zeta. *Proc. Nat. Acad. Sci.* 95: 6876–6880, 1998.

[38322] 11874. Morelli, C.; Mungall, A. J.; Negrini, M.; Barbanti-Brodano, G.; Croce, C. M.: Alternative splicing, genomic structure, and fine chromosomal localization of REV3L. *Cytogenet. Cell Genet.* 83: 18–20, 1998.

- [38323] 11875.Xiao, W.; Lechler, T.; Chow, B. L.; Fontanie, T.; Augustus, M.;Carter, K. C.; Wei, Y.-F.: Identification, chromosomal mapping and tissue-specific expression of hREV3 encoding a putative human DNA polymerase zeta. *Carcinogenesis* 19: 945–949, 1998.
- [38324] 11876.Kahn, M. L.; Zheng, Y.-W.; Huang, W.; Bigornia, V.; Zeng, D.; Moff,S.; Farese, R. V., Jr.; Tam, C.; Coughlin, S. R.: A dual thrombin receptor system for platelet activation. *Nature* 394: 690–694, 1998.
- [38325] 11877.Xu, W.-F.; Andersen, H.; Whitmore, T. E.; Presnell, S. R.; Yee,D. P.; Ching, A.; Gilbert, T.; Davie, E. W.; Foster, D. C.: Cloning and characterization of human protease-activated receptor 4. *Proc.Nat. Acad. Sci.* 95: 6642–6646, 1998.
- [38326] 11878.Santoro, B.; Grant, S. G. N.; Bartsch, D.; Kandel, E. R.: Interactive cloning with the SH3 domain of N-src identifies a new brain specific ion channel protein, with homology to Eag and cyclic nucleotide-gated channels. *Proc. Nat. Acad. Sci.* 94: 14815–14820, 1997.
- [38327] 11879.Santoro, B.; Liu, D. T.; Yao, H.; Bartsch, D.; Kandel, E. R.; Siegelbaum,S. A.; Tibbs, G. R.: Identification of a gene encoding a hyperpolarization-activated pacemaker channel of brain. *Cell* 93: 717–729, 1998.

- [38328] 11880.Stevens, D. R.; Seifert, R.; Bufe, B.; Muller, F.; Kremmer, E.;Gauss, R.; Meyerhof, W.; Kaupp, U. B.; Lindemann, B.: Hyperpolarization-activatedchannels HCN1 and HCN4 mediate responses to sour stimuli. *Nature* 413:631–635, 2001.
- [38329] 11881.Wainger, B. J.; DeGennaro, M.; Santoro, B.; Siegelbaum, S. A.;Tibbs, G. R.: Molecular mechanism of cAMP modulation of HCN pacemakerchannels. *Nature* 411: 805–810, 2001.
- [38330] 11882.Ludwig, A.; Zong, X.; Stieber, J.; Hullin, R.; Hofmann, F.; Biel,M.: Two pacemaker channels from human heart with profoundly differentactivation kinetics. *EMBO J.* 18: 2323–2329, 1999.
- [38331] 11883.Casari, G.; De Fusco, M.; Ciarmatori, S.; Zeviani, M.; Mora, M.;Fernandez, P.; De Michele, G.; Filla, A.; Coccozza, S.; Marconi, R.;Durr, A.; Fontaine, B.; Ballabio, A.: Spastic paraplegia and OXPHOSimpairment caused by mutations in paraplegin, a nuclear-encoded mitochondrialmetalloprotease. *Cell* 93: 973–983, 1998.
- [38332] 11884.De Michele, G.; De Fusco, M.; Cavalcanti, F.; Filla, A.; Marconi,R.; Volpe, G.; Monticelli, A.; Ballabio, A.; Casari, G.; Coccozza,S.: A new locus for autosomal recessive hereditary spastic paraplegiamaps to chromosome

16q24.3. Am. J. Hum. Genet. 63: 135–139, 1998.

[38333] 11885. Settasatian, C.; Whitmore, S. A.; Crawford, J.; Bilton, R. L.; Cleton-Jansen, A.-M.; Sutherland, G. R.; Callen, D. F.: Genomic structure and expression analysis of the spastic paraplegia gene, SPG7. Hum. Genet. 105: 139–144, 1999.

[38334] 11886. de Lecea, L.; Criado, J. R.; Prospero-Garcia, O.; Gautvik, K. M.; Schweitzer, P.; Danielson, P. E.; Dunlop, C. L. M.; Siggins, G. R.; Henriksen, S. J.; Sutcliffe, J. G.: A cortical neuropeptide with neuronal depressant and sleep-modulating properties. Nature 381: 242–245, 1996.

[38335] 11887. Kerjaschki, D.; Farquhar, M. G.: Immunocytochemical localization of the Heymann nephritis antigen (GP330) in glomerular epithelial cells of normal Lewis rats. J. Exp. Med. 157: 667–686, 1983.

[38336] 11888. Korenberg, J. R.; Argraves, K. M.; Chen, X.-N.; Tran, H.; Strickland, D.; Argraves, W. S.: Chromosomal localization of human genes for the LDL receptor family member glycoprotein 330 (LRP2) and its associated protein RAP (LRPAP1). Genomics 22: 88–93, 1994.

[38337] 11889. Kounnas, M. Z.; Loukinova, E. B.; Stefansson, S.; Harmony, J. A. K.; Brewer, B. H.; Strickland, D. K.; Argraves, W. S.: Identification of glycoprotein 330 as an endocytic

receptor for apolipoprotein J/clusterin. *J. Biol. Chem.* 270: 13070–13075, 1995.

- [38338] 11890. Leheste, J.-R.; Rolinski, B.; Vorum, H.; Hilpert, J.; Nykjaer, A.; Jacobsen, C.; Aucouturier, P.; Moskaug, J. O.; Otto, A.; Christensen, E. I.; Willnow, T. E.: Megalin knock-out mice as an animal model of low molecular weight proteinuria. *Am. J. Path.* 155: 1361–1370, 1999.
- [38339] 11891. Marino, M.; Chiovato, L.; Friedlander, J. A.; Latrofa, F.; Pinchera, A.; McCluskey, R. T.: Serum antibodies against megalin (GP330) in patients with autoimmune thyroiditis. *J. Clin. Endocr. Metab.* 84: 2468–2474, 1999.
- [38340] 11892. Moestrup, S. K.; Cui, S.; Vorum, H.; Bregengard, C.; Bjorn, S. E.; Norris, K.; Gliemann, J.; Christensen, E. I.: Evidence that epithelial glycoprotein 330/megalin mediates uptake of polybasic drugs. *J. Clin. Invest.* 96: 1404–1413, 1995.
- [38341] 11893. Nykjaer, A.; Dragun, D.; Walther, D.; Vorum, H.; Jacobsen, C.; Herz, J.; Melsen, F.; Christensen, E. I.; Willnow, T. E.: An endocytic pathway essential for renal uptake and activation of the steroid 25-(OH) vitamin D₃. *Cell* 96: 507–515, 1999.
- [38342] 11894. Raychowdhury, R.; Niles, J. L.; McCluskey, R. T.; Smith, J. A.: Autoimmune target in Heymann nephritis is a

glycoprotein with homology to the LDL receptor. *Science* 244: 1163–1165, 1989.

- [38343] 11895. Saito, A.; Pietromonaco, S.; Loo, A. K.-C.; Farquhar, M. G.: Complete cloning and sequencing of rat gp330/`megalin,' a distinctive member of the low density lipoprotein receptor gene family. *Proc. Nat. Acad. Sci.* 91: 9725–9729, 1994.
- [38344] 11896. Schmitz, C.; Hilpert, J.; Jacobsen, C.; Boensch, C.; Christensen, E. I.; Luft, F. C.; Willnow, T. E.: Megalin deficiency offers protection from renal aminoglycoside accumulation. *J. Biol. Chem.* 277: 618–622, 2002.
- [38345] 11897. Willnow, T. E.; Hilpert, J.; Armstrong, S. A.; Rohlmann, A.; Hammer, R. E.; Burns, D. K.; Herz, J.: Defective forebrain development in mice lacking gp330/megalin. *Proc. Nat. Acad. Sci.* 93: 8460–8464, 1996.
- [38346] 11898. Gostout, B.; Liu, Q.; Sommer, S. S.: 'Cryptic' repeating triplets of purines and pyrimidines (cRRY(i)) are frequent and polymorphic: analysis of coding cRRY(i) in the proopiomelanocortin (POMC) and TATA-binding protein (TBP) genes. *Am. J. Hum. Genet.* 52: 1182–1190, 1993.
- [38347] 11899. Hobbs, N. K.; Bondareva, A. A.; Barnett, S.; Capecchi, M. R.; Schmidt, E. E.: Removing the vertebrate-specific

TBP N terminus disrupts placental beta-2M-dependent interactions with the maternal immune system. *Cell* 110:43–54, 2002.

- [38348] 11900. Imbert, G.; Trottier, Y.; Beckmann, J.; Mandel, J. L.: The gene for the TATA binding protein (TBP) that contains a highly polymorphic protein coding CAG repeat maps to 6q27. *Genomics* 21: 667–668, 1994.
- [38349] 11901. Jones, A. L.; Middle, F.; Guy, C.; Spurlock, G.; Cairns, N. J.; McGuffin, P.; Craddock, N.; Owen, M.; O'Donovan, M. C.: No evidence for expanded polyglutamine sequences in bipolar disorder and schizophrenia. *Molec. Psychiat.* 2: 478–482, 1997.
- [38350] 11902. Kao, C. C.; Lieberman, P. M.; Schmidt, M. C.; Zhou, Q.; Pei, R.; Berk, A. J.: Cloning of a transcriptionally active human TATA binding factor. *Science* 248: 1646–1650, 1990.
- [38351] 11903. Koide, R.; Kobayashi, S.; Shimohata, T.; Ikeuchi, T.; Maruyama, M.; Saito, M.; Yamada, M.; Takahashi, H.; Tsuji, S.: A neurological disease caused by an expanded CAG trinucleotide repeat in the TATA-binding protein gene: a new polyglutamine disease? *Hum. Molec. Genet.* 8:2047–2053, 1999.
- [38352] 11904. Nakamura, K.; Jeong, S.-Y.; Uchihara, T.; Anno, M.;

Nagashima,K.; Nagashima, T.; Ikeda, S.; Tsuji, S.; Kanazawa, I.: SCA17, a novel autosomal dominant cerebellar ataxia caused by an expanded polyglutamine in TATA-binding protein. *Hum. Molec. Genet.* 10: 1441–1448, 2001.

[38353] 11905. Nikolov, D. B.; Hu, S.-H.; Lin, J.; Gasch, A.; Hoffmann, A.; Horikoshi, M.; Chua, N.-H.; Roeder, R. G.; Burley, S. K.: Crystal structure of TFIID TATA-box binding protein. *Nature* 360: 40–46, 1992.

[38354] 11906. Peterson, M. G.; Tanese, N.; Pugh, B. F.; Tjian, R.: Functional domains and upstream activation properties of cloned human TATA binding protein. *Science* 248: 1625–1630, 1990.

[38355] 11907. Polymeropoulos, M. H.; Rath, D. S.; Xiao, H.; Merrill, C. R.: Trinucleotide repeat polymorphism at the human transcription factor IID gene. *Nucleic Acids Res.* 19: 4307 only, 1991.

[38356] 11908. Rosen, D. R.; Trofatter, J. A.; Brown, R. H., Jr.: Mapping of the human TATA-binding protein gene (TBP) to chromosome 6qter. *Cytogenet. Cell Genet.* 69: 279–280, 1995.

[38357] 11909. Rubinsztein, D. C.; Leggo, J.; Crow, T. J.; DeLisi, L. E.; Walsh, C.; Jain, S.; Paykel, E. S.: Analysis of polyglu-

tamine-coding repeats in the TATA-binding protein in different human populations and in patients with schizophrenia and bipolar affective disorder. *Am. J. Med. Genet.* 67: 495–498, 1996.

- [38358] 11910. Saito, F.; Yamamoto, T.; Horikoshi, M.; Ikeuchi, T.: Direct mapping of the human TATA box-binding protein (TBP) gene to 6q27 by fluorescence in situ hybridization. *Jpn. J. Hum. Genet.* 39: 421–425, 1994.
- [38359] 11911. Serrano, M.; Hannon, G. J.; Beach, D.: A new regulatory motif in cell-cycle control causing specific inhibition of cyclin D/CDK4. *Nature* 366: 704–707, 1993.
- [38360] 11912. Testa, J. R.; Zhou, J.; Bell, D. W.; Yen, T. J.: Chromosomal localization of the genes encoding the kinetochore proteins CENPE and CENPF to human chromosomes 4q24–q25 and 1q32–q41, respectively, by fluorescence in situ hybridization. *Genomics* 23: 691–693, 1994.
- [38361] 11913. Pennica, D.; Swanson, T. A.; Shaw, K. J.; Kuang, W.-J.; Gray, C. L.; Beatty, B. G.; Wood, W. I.: Human cardiotrophin-1: protein and gene structure, biological and binding activities, and chromosomal localization. *Cytokine* 8: 183–189, 1996.
- [38362] 11914. Fukudome, K.; Esmon, C. T.: Identification, cloning, and regulation of a novel endothelial cell protein C/

activated protein C receptor. J.Biol. Chem. 269:
26486–26491, 1994.

[38363] 11915.Hayashi, T.; Nakamura, H.; Okada, A.; Takebayashi, S.; Wakita,T.; Yuasa, H.; Okumura, K.; Suzuki, K.: Organization and chromosomallocalization of the human endothelial protein C receptor gene. Gene 238:367–373, 1999.

[38364] 11916.Mincheva, A.; Rothbarth, K.; Werner, D.; Lichter, P.: Assignmentof the gene encoding centrosome–associated protein CCD41 to mousechromosome 2H. Mammalian Genome 6: 444 only, 1995.

[38365] 11917.Rothbarth, K.; Dabaghian, A. R. H.; Stammer, H.; Werner, D.: Onesingle mRNA encodes the centrosomal protein CCD41 and the endothelialcell protein C receptor (EPCR). FEBS Lett. 458: 77–80, 1999.

[38366] 11918.Rothbarth, K.; Petzelt, C.; Lu, X.; Todorov, I. T.; Joswig, G.;Pepperkok, R.; Ansorge, W.; Werner, D.: cDNA–derived molecular characteristicsand antibodies to a new centrosome–associated and G2/M phase–prevalentprotein. J. Cell Sci. 104: 19–30, 1993.

[38367] 11919.Simmonds, R. E.; Lane, D. A.: Structural and functional implicationsof the intron/exon organization of the human endothelial cell proteinC/activated protein C re–

ceptor (EPCR) gene: comparison with the structure of CD1/major histocompatibility complex alpha-1 and alpha-2 domains. *Blood* 94:632–641, 1999.

[38368] 11920. Ye, X.; Fukudome, K.; Tsuneyoshi, N.; Satoh, T.; Tokunaga, O.; Sugawara, K.; Mizokami, H.; Kimoto, M.: The endothelial cell protein C receptor (EPCR) functions as a primary receptor for protein C activation on endothelial cells in arteries, veins, and capillaries. *Biochem. Biophys. Res. Commun.* 259: 671–677, 1999.

[38369] 11921. Purrello, M.; Pietro, C. D.; Mirabile, E.; Rapisarda, A.; Rimini, R.; Tine, A.; Pavone, L.; Motta, S.; Grzeschik, K.-H.; Sichel, G.: Physical mapping at 6q27 of the locus for the TATA box-binding protein, the DNA-binding subunit of TFIID and a component of SL1 and TFIIB, strongly suggests that it is single copy in the human genome. *Genomics* 22:94–100, 1994.

[38370] 11922. Candia, A. F.; Hu, J.; Crosby, J.; Lalley, P. A.; Noden, D.; Nadeau, J. H.; Wright, C. V. E.: Mox-1 and Mox-2 define a novel homeobox gene subfamily and are differentially expressed during early mesodermal patterning in mouse embryos. *Development* 116: 1123–1136, 1992.

[38371] 11923. Gorski, D. H.; LePage, D. F.; Patel, C. V.; Copeland, N. G.; Jenkins, N. A.; Walsh, K.: Molecular cloning of a di-

verged homeobox gene that is rapidly down-regulated during the G0/G1 transition in vascular smooth muscle cells. *Molec. Cell Biol.* 13: 3722–3733, 1993.

- [38372] 11924. Grigoriou, M.; Kastrinaki, M.-C.; Modi, W. S.; Theodorakis, K.; Mankoo, B.; Pachnis, V.; Karagogeos, D.: Isolation of the human MOX2 homeobox gene and localization to chromosome 7p22.1–p21.3. *Genomics* 26:550–555, 1995.
- [38373] 11925. LePage, D. F.; Altomare, D. A.; Testa, J. R.; Walsh, K.: Molecular cloning and localization of the human GAX gene to 7p21. *Genomics* 24:535–540, 1994.
- [38374] 11926. Mankoo, B. S.; Collins, N. S.; Ashby, P.; Grigorieva, E.; Pevny, L. H.; Candia, A.; Wright, C. V. E.; Rigby, P. W. J.; Pachnis, V.: Mox2 is a component of the genetic hierarchy controlling limb muscle development. *Nature* 400: 69–73, 1999.
- [38375] 11927. Lindor, N. M.; Furuichi, Y.; Kitao, S.; Shimamoto, A.; Arndt, C.; Jalal, S.: Rothmund–Thomson syndrome due to RECQ4 helicase mutations: report and clinical and molecular comparisons with Bloom syndrome and Werner syndrome. *Am. J. Med. Genet.* 90: 223–228, 2000.
- [38376] 11928. Puranam, K. L.; Blackshear, P. J.: Cloning and characterization of RECQL, a potential human homologue of

the Escherichia coli DNA helicase RecQ. J. Biol. Chem. 269: 29838–29845, 1994.

[38377] 11929. Puranam, K. L.; Kennington, E.; J.-Sait, S. N.; Shows, T. B.; Rochelle, J. M.; Seldin, M. F.; Blackshear, P. J.: Chromosomal localization of the gene encoding the human DNA helicase RECQL and its mouse homologue. Genomics 26:595–598, 1995.

[38378] 11930. Gollner, H.; Bouwman, P.; Mangold, M.; Karis, A.; Braun, H.; Rohner, I.; Del Rey, A.; Besedovsky, H.-O.; Meinhardt, A.; van den Broek, M.; Cutforth, T.; Grosveld, F.; Philipsen, S.; Suske, G.: Complex phenotype of mice homozygous for a null mutation in the Sp4 transcription factor gene. Genes Cells 6: 689–697, 2001.

[38379] 11931. Hagen, G.; Dennig, J.; Preiss, M.; Beato, M.; Suske, G.: Functional analyses of the transcription factor Sp4 reveal properties distinct from Sp1 and Sp3. J. Biol. Chem. 270: 24989–24994, 1995.

[38380] 11932. Hagen, G.; Muller, S.; Beato, M.; Suske, G.: Cloning by recognition site screening of two novel GT box binding proteins: a family of Sp1 related genes. Nucleic Acids Res. 20: 5519–5525, 1992.

[38381] 11933. Kalff-Suske, M.; Kunz, J.; Grzeschik, K.-H.; Suske, G.: Human Sp4 transcription factor gene (SP4) maps to

chromosome 7p15. Genomics 26:631–633, 1995.

- [38382] 11934. Nguyen–Tran, V. T. B.; Kubalak, S. W.; Minamisawa, S.; Fiset, C.; Wollert, K. C.; Brown, A. B.; Ruiz–Lozano, P.; Barrere–Lemaire, S.; Kondo, R.; Norman, L. W.; Gourdie, R. G.; Rahme, M. M.; Feld, G. K.; Clark, R. B.; Giles, W. R.; Chien, K. R.: A novel genetic pathway for sudden cardiac death via defects in the transition between ventricular and conduction system cell lineages. Cell 102: 671–682, 2000.
- [38383] 11935. Lamour, V.; Lecluse, Y.; Desmaze, C.; Spector, M.; Bodescot, M.; Aurias, A.; Osley, M. A.; Lipinski, M.: A human homolog of the *S.cerevisiae* HIR1 and HIR2 transcriptional repressors cloned from the DiGeorge syndrome critical region. Hum. Molec. Genet. 4: 791–799, 1995.
- [38384] 11936. Llevadot, R.; Scambler, P.; Estivill, X.; Pritchard, M.: Genomic organization of TUPLE1/HIRA: a gene implicated in DiGeorge syndrome. Mammalian Genome 7: 911–914, 1996.
- [38385] 11937. Lorain, S.; Quivy, J.–P.; Monier–Gavelle, F.; Scamps, C.; Lecluse, Y.; Almouzni, G.; Lipinski, M.: Core histones and HIRIP3, a novel histone–binding protein, directly interact with WD repeat protein HIRA. Molec. Cell. Biol. 18: 5546–5556, 1998.

- [38386] 11938.Magnaghi, P.; Roberts, C.; Lorain, S.; Lipinski, M.; Scambler,P. J.: HIRA, a mammalian homologue of *Saccharomyces cerevisiae* transcriptionalco-repressors, interacts with Pax3. *Nature Genet.* 20: 74–77, 1998.
- [38387] 11939.Mattei, M.–G.; Halford, S.; Scambler, P. J.: Mapping of the *Tuple1*gene to mouse chromosome 16A–B1. *Genomics* 23: 717–718, 1994.
- [38388] 11940.Roberts, C.; Daw, S. C. M.; Halford, S.; Scambler, P. J.: Cloningand developmental expression analysis of chick Hira (Chira), a candidategene for DiGeorge syndrome. *Hum. Molec. Genet.* 6: 237–245, 1997.
- [38389] 11941.Wilming, L. G.; Snoeren, C. A. S.; van Rijswijk, A.; Grosveld,F.; Meijers, C.: The murine homologue of HIRA, a DiGeorge syndromecandidate gene, is expressed in embryonic structures affected in humanCATCH22 patients. *Hum. Molec. Genet.* 6: 247–258, 1997.
- [38390] 11942.Kim, I.–G.; Gorman, J. J.; Park, S.–C.; Chung, S.–I.; Steinert,P. M.: The deduced sequence of the novel pro-transglutaminase E (TGase3)of human and mouse. *J. Biol. Chem.* 268: 12682–12690, 1993.
- [38391] 11943.Kim, I.–G.; Lee, S.–C.; Lee, J.–H.; Yang, J.–M.; Chung, S.–I.;Steinert, P. M.: Structure and organization of the human transglutaminase3 gene: evolutionary relation–

ship to the transglutaminase family. *J. Invest. Derm.* 103: 137–142, 1994.

[38392] 11944. Marchese, A.; Docherty, J. M.; Nguyen, T.; Heiber, M.; Cheng, R.; Heng, H. H. Q.; Tsui, L.-C.; Shi, X.; George, S. R.; O'Dowd, B. F.: Cloning of human genes encoding novel G protein-coupled receptors. *Genomics* 23:609–618, 1994.

[38393] 11945. Homey, B.; Alenius, H.; Muller, A.; Soto, H.; Bowman, E. P.; Yuan, W.; McEvoy, L.; Lauerma, A. I.; Assmann, T.; Bunemann, E.; Lehto, M.; Wolff, H.; Yen, D.; Marxhausen, H.; To, W.; Sedgwick, J.; Ruzicka, T.; Lehmann, P.; Zlotnik, A.: CCL27–CCR10 interactions regulate T cell-mediated skin inflammation. *Nature Med.* 8: 157–165, 2002.

[38394] 11946. Homey, B.; Wang, W.; Soto, H.; Buchanan, M. E.; Wiesenborn, A.; Catron, D.; Muller, A.; McClanahan, T. K.; Dieu-Nosjean, M.-C.; Orozco, R.; Ruzicka, T.; Lehmann, P.; Oldham, E.; Zlotnik, A.: Cutting edge: the orphan chemokine receptor G protein-coupled receptor-2 (GPR-2, CCR10) binds the skin-associated chemokine CCL27 (CTACK/ALP/ILC). *J. Immun.* 164: 3465–3470, 2000.

[38395] 11947. Jarmin, D. I.; Rits, M.; Bota, D.; Gerard, N. P.; Gra-

ham, G. J.; Clark-Lewis, I.; Gerard, C.: Cutting edge: identification of the orphan receptor G-protein-coupled receptor 2 as CCR10, a specific receptor for the chemokine Eotaxin. J. Immun. 164: 3460–3464, 2000.

[38396] 11948. Wang, W.; Soto, H.; Oldham, E. R.; Buchanan, M. E.; Homey, B.; Catron, D.; Jenkins, N.; Copeland, N. G.; Gilbert, D. J.; Nguyen, N.; Abrams, J.; Kershenovich, D.; Smith, K.; McClanahan, T.; Vicari, A. P.; Zlotnik, A.: Identification of a novel chemokine (CCL28), which binds CCR10 (GPR2). J. Biol. Chem. 275: 22313–22323, 2000.

[38397] 11949. Akao, Y.; Matsuda, Y.: Identification and chromosome mapping of the mouse homologue of the human gene (DDX6) that encodes a putative RNA helicase of the DEAD box protein family. Cytogenet. Cell Genet. 75:38–44, 1996.

[38398] 11950. Akao, Y.; Seto, M.; Takahashi, T.; Kubonishi, I.; Miyoshi, I.; Nakazawa, S.; Tsujimoto, Y.; Croce, C. M.; Ueda, R.: Molecular cloning of the chromosomal breakpoint of a B-cell lymphoma with the t(11;14)(q23;q32) chromosome translocation. Cancer Res. 51: 1574–1576, 1991.

[38399] 11951. Akao, Y.; Seto, M.; Yamamoto, K.; Iida, S.; Nakazawa, S.; Inazawa, J.; Abe, T.; Takahashi, T.; Ueda, R.: The RCK gene associated with t(11;14) translocation is dis-

tinct from the MLL/ALL-1 gene with t(4;11) and t(11;19) translocations. Cancer Res. 52: 6083–6087, 1992.

- [38400] 11952. Akao, Y.; Tsujimoto, Y.; Finan, J.; Nowell, P. C.; Croce, C. M.: Molecular characterization of a t(11;14)(q23;q32) chromosome translocation in a B-cell lymphoma. Cancer Res. 50: 4856–4859, 1990.
- [38401] 11953. Lu, D.; Yunis, J. J.: Cloning, expression and localization of an RNA helicase gene from a human lymphoid cell line with chromosomal breakpoint 11q23.3. Nucleic Acids Res. 20: 1967–1972, 1992.
- [38402] 11954. Seto, M.; Yamamoto, K.; Takahashi, T.; Ueda, R.: Cloning and expression of a murine cDNA homologous to the human RCK/P54, a lymphoma-linked chromosomal translocation junction gene on 11q23. Gene 166: 293–296, 1995.
- [38403] 11955. Tunnacliffe, A.; Perry, H.; Radice, P.; Budarf, M. L.; Emanuel, B. S.: A panel of sequence tagged sites for chromosome band 11q23. Genomics 17: 744–747, 1993.
- [38404] 11956. Hallier, E.; Jager, R.; Deutschmann, S.; Bolt, H. M.; Peter, H.: Glutathione conjugation and cytochrome P-450 metabolism of methylchloride in vitro. Toxicol. in Vitro 4: 513–517, 1990.
- [38405] 11957. Pemble, S.; Schroeder, K. R.; Spencer, S. R.; Meyer,

D. J.; Hallier, E.; Bolt, H. M.; Ketterer, B.; Taylor, J. B.: Human glutathione S-transferase theta (GSTT1): cDNA cloning and the characterization of a genetic polymorphism. *Biochem. J.* 300: 271–276, 1994.

[38406] 11958. Peter, H.; Deutschmann, S.; Reichel, C.; Hallier, E.: Metabolism of methyl chloride by human erythrocytes. *Arch. Toxicol.* 63: 351–355, 1989.

[38407] 11959. Schroder, K. R.; Wiebel, F. A.; Reich, S.; Dannappel, D.; Bolt, H. M.; Hallier, E.: Glutathione S-transferase (GST) theta polymorphism influences background SCE rate. *Arch. Toxicol.* 69: 505–507, 1995.

[38408] 11960. Webb, G.; Vaska, V.; Coggan, M.; Board, P.: Chromosomal localization of the gene for the human theta class glutathione transferase (GSTT1). *Genomics* 33: 121–123, 1996.

[38409] 11961. Wiebel, F. A.; Dommermuth, A.; Thier, R.: The hereditary transmission of the glutathione transferase hGSTT1-1 conjugator phenotype in a large family. *Pharmacogenetics* 9: 251–256, 1999.

[38410] 11962. Dumon, K. R.; Ishii, H.; Fong, L. Y. Y.; Zanesi, N.; Fidanza, V.; Mancini, R.; Vecchione, A.; Baffa, R.; Trapasso, F.; During, M. J.; Huebner, K.; Croce, C. M.: FHIT gene therapy prevents tumor development in Fhit-deficient mice.

Proc. Nat. Acad. Sci. 98: 3346–3351, 2001.

[38411] 11963.Ikeda, A.; Zheng, Q. Y.; Zuberi, A. R.; Johnson, K. R.; Naggert, J. K.; Nishina, P. M.: Microtubule-associated protein 1A is a modifier of tubby hearing (moth1). Nature Genet. 30: 401–405, 2002.

[38412] 11964.Roy, S. K.; Hu, J.; Meng, Q.; Xia, Y.; Shapiro, P. S.; Reddy, S.P. M.; Platanius, L. C.; Lindner, D. J.; Johnson, P. F.; Pritchard, C.; Pages, G.; Pouyssegur, J.; Kalvakolanu, D. V.: MEKK1 plays a critical role in activating the transcription factor C/EBP-beta-dependent gene expression in response to IFN-gamma. Proc. Nat. Acad. Sci. 99:7945–7950, 2002.

[38413] 11965.Chou, D.; Miyashita, T.; Mohrenweiser, H. W.; Ueki, K.; Kastury, K.; Druck, T.; von Deimling, A.; Huebner, K.; Reed, J. C.; Louis, D. N.: The BAX gene maps to the glioma candidate region at 19q13.3, but is not altered in human gliomas. Cancer Genet. Cytogenet. 88:136–140, 1996.

[38414] 11966.Deckwerth, T. L.; Elliott, J. L.; Knudson, C. M.; Johnson, E. M., Jr.; Snider, W. D.; Korsmeyer, S. J.: BAX is required for neuronal death after trophic factor deprivation and during development. Neuron 17:401–411, 1996.

[38415] 11967.Knudson, C. M.; Tung, K. S. K.; Tourtellotte, W. G.; Brown, G.A. J.; Korsmeyer, S. J.: Bax-deficient mice with

lymphoid hyperplasia and male germ cell death. *Science* 270: 96–99, 1995.

- [38416] 11968. LeBlanc, H.; Lawrence, D.; Varfolomeev, E.; Totpal, K.; Morlan, J.; Schow, P.; Fong, S.; Schwall, R.; Sinicropi, D.; Ashkenazi, A.: Tumor–cell resistance to death receptor–induced apoptosis through mutational inactivation of the proapoptotic Bcl–2 homolog Bax. *Nature Med.* 8: 274–281, 2002.
- [38417] 11969. Lindsten, T.; Ross, A. J.; King, A.; Zong, W.–X.; Rathmell, J. C.; Shiels, H. A.; Ulrich, E.; Waymire, K. G.; Mahar, P.; Frauwirth, K.; Chen, Y.; Wei, M.; and 9 others: The combined functions of proapoptotic Bcl–2 family members Bak and Bax are essential for normal development of multiple tissues. *Molec. Cell* 6: 1389–1399, 2000.
- [38418] 11970. Marzo, I.; Brenner, C.; Zamzami, N.; Jurgensmeier, J. M.; Susin, S. A.; Vieira, H. L. A.; Prevost, M.–C.; Xie, Z.; Matsuyama, S.; Reed, J. C.; Kroemer, G.: Bax and adenine nucleotide translocator cooperate in the mitochondrial control of apoptosis. *Science* 281: 2027–2031, 1998.
- [38419] 11971. Matikainen, T.; Perez, G. I.; Jurisicova, A.; Pru, J. K.; Schlezinger, J. J.; Ryu, H.–Y.; Laine, J.; Sakai, T.; Korsmeyer, S. J.; Casper, R. F.; Sherr, D. H.; Tilly, J. L.: Aromatic hydrocarbon receptor–driven Bax gene expression is required

for premature ovarian failure caused by biohazardous environmental chemicals. *Nature Genet.* 28: 355–360, 2001.

[38420] 1972. Matsuda, Y.; Kusano, H.; Tsujimoto, Y.: Chromosomal assignment of the Bcl2-related genes, Bcl2l and Bax, in the mouse and rat. *Cytogenet. Cell Genet.* 74: 107–110, 1996.

[38421] 1973. Meijerink, J. P. P.; Mensink, E. J. B. M.; Wang, K.; Sedlak, T. W.; Sloetjes, A. W.; de Witte, T.; Waksman, G.; Korsmeyer, S. J.: Hematopoietic malignancies demonstrate loss-of-function mutations of BAX. *Blood* 91: 2991–2997, 1998.

[38422] 1974. Miyashita, T.; Reed, J. C.: Tumor suppressor p53 is a direct transcriptional activator of the human bax gene. *Cell* 80: 293–299, 1995.

[38423] 1975. Oltvai, Z. N.; Millman, C. L.; Korsmeyer, S. J.: Bcl-2 heterodimers in vivo with a conserved homolog, Bax, that accelerates programmed cell death. *Cell* 74: 609–619, 1993.

[38424] 1976. Perez, G. I.; Robles, R.; Knudson, C. M.; Flaws, J. A.; Korsmeyer, S. J.; Tilly, J. L.: Prolongation of ovarian lifespan into advanced chronological age by Bax-deficiency. *Nature Genet.* 21: 200–203, 1999.

[38425] 1977. Rampino, N.; Yamamoto, H.; Ionov, Y.; Li, Y.; Sawai,

H.; Reed, J. C.; Perucho, M.: Somatic frameshift mutations in the BAX gene in colon cancers of the microsatellite mutator phenotype. *Science* 275:967–969, 1997.

[38426] 1978. Suzuki, M.; Youle, R. J.; Tjandra, N.: Structure of Bax: coregulation of dimer formation and intracellular localization. *Cell* 103: 645–654, 2000.

[38427] 1979. Wei, M. C.; Zong, W.-X.; Cheng, E. H.-Y.; Lindsten, T.; Panoutsakopoulou, V.; Ross, A. J.; Roth, K. A.; MacGregor, G. R.; Thompson, C. B.; Korsmeyer, S. J.: Proapoptotic BAX or BAK: a requisite gateway to mitochondrial dysfunction and death. *Science* 292: 727–730, 2001.

[38428] 1980. Zhang, L.; Yu, J.; Park, B. H.; Kinzler, K. W.; Vogelstein, B.: Role of BAX in the apoptotic response to anti-cancer agents. *Science* 290:989–992, 2000.

[38429] 1981. Dasari, V. R.; Sandhu, A. K.; Mills, D. C. B.; Athwal, R. S.; Kunapuli, S. P.: Mapping of the P2U purinergic receptor gene to human chromosome 11q13.5–14.1. *Somat. Cell Molec. Genet.* 22: 75–79, 1996.

[38430] 1982. Katzur, A. C.; Koshimizu, T.-A.; Tomic, M.; Schultze-Mosgau, A.; Ortmann, O.; Stojilkovic, S. S.: Expression and responsiveness of P2Y₂ receptors in human endometrial cancer cell lines. *J. Clin. Endocr. Metab.* 84: 4085–4091, 1999.

- [38431] 11983.Parr, C. E.; Sullivan, D. M.; Paradiso, A. M.; Lazarowski, E. R.;Burch, L. H.; Olsen, J. C.; Erb, L.; Weisman, G. A.; Boucher, R. C.;Turner, J. T.: Cloning and expression of a human P(2U) nucleotidereceptor, a target for cystic fibrosis pharmacotherapy. *Proc. Nat.Acad. Sci.* 91: 3275–3279, 1994.
- [38432] 11984.Somers, G. R.; Hammet, F.; Woollatt, E.; Richards, R. I.; Southey,M. C.; Venter, D. J.: Chromosomal localization of the human P2Y(6)purinoceptor gene and phylogenetic analysis of the P2y purinoceptorfamily. *Genomics* 44: 127–130, 1997.
- [38433] 11985.Tai, C.–J.; Kang, S. K.; Cheng, K. W.; Choi, K.–C.; Nathwani, P.S.; Leung, P. C. K.: Expression and regulation of P2U–purinergicreceptor in human granulosa–luteal cells. *J. Clin. Endocr. Metab.* 85:1591–1597, 2000.
- [38434] 11986.Santamarina–Fojo, S.; Peterson, K.; Knapper, C.; Qiu, Y.; Freeman,L.; Cheng, J.–F.; Osorio, J.; Remaley, A.; Yang, X.–P.; Haudenschild,C.; Prades, C.; Chimini, G.; Blackmon, E.; Francois, T.; Duverger,N.; Rubin, E. M.; Rosier, M.; Denefle, P.; Fredrickson, D. S.; Brewer,H. B., Jr.: Complete genomic sequence of the human ABCA1 gene: analysisof the human and mouse ATP–binding cassette A promoter. *Proc. Nat.Acad. Sci.* 97: 7987–7992, 2000.

Note: Erratum: Proc. Nat. Acad. Sci.99: 1098 only, 2002.

- [38435] 11987.Szakacs, G.; Langmann, T.; Ozvegy, C.; Orso, E.; Schmitz, G.;Varadi, A.; Sarkadi, B.: Characterization of the ATPase cycle ofhuman ABCA1: implications for its function as a regulator rather thanan active transporter. Biochem. Biophys. Res. Commun. 288: 1258–1264,2001.
- [38436] 11988.Utech, M.; Hobbel, G.; Rust, S.; Reinecke, H.; Assmann, G.; Walter,M.: Accumulation of RhoA, RhoB, RhoG, and Rac1 in fibroblasts fromTangier disease subjects suggests a regulatory role of Rho familyproteins in cholesterol efflux. Biochem. Biophys. Res. Commun. 280:229–236, 2001.
- [38437] 11989.Zhao, L.-X.; Zhou, C.-J.; Tanaka, A.; Nakata, M.; Hirabayashi,T.; Amachi, T.; Shioda, S.; Ueda, K.; Inagaki, N.: Cloning, characterizationand tissue distribution of the rat ATP-binding cassette (ABC) transporterABC2/ABCA2. Biochem J. 350: 865–872, 2000.
- [38438] 11990.Zwarts, K. Y.; Clee, S. M.; Zwinderman, A. H.; Engert, J. C.;Singaraja, R.; Loubser, O.; James, E.; Roomp, K.; Hudson, T. J.; Jukema,J. W.; Kastelein, J. J. P.; Hayden, M. R.: ABCA1 regulatory variantsinfluence coronary artery disease independent of effects on plasmalipid levels. Clin. Genet. 61: 115–125, 2002.

- [38439] 11991.Fears, S.; Mathieu, C.; Zeleznik-Le, N.; Huang, S.; Rowley, J.D.; Nucifora, G.: Intergenic splicing of MDS1 and EVI1 occurs in normal tissues as well as in myeloid leukemia and produces a new member of the PR domain family. *Proc. Nat. Acad. Sci.* 93: 1642–1647, 1996.
- [38440] 11992.Mochizuki, N.; Shimizu, S.; Nagasawa, T.; Tanaka, H.; Taniwaki, M.; Yokota, J.; Morishita, K.: A novel gene, MEL1, mapped to 1p36.3 is highly homologous to the MDS1/EVI1 gene and is transcriptionally activated in t(1;3)(p36;q21)–positive leukemia cells. *Blood* 96:3209–3214, 2000.
- [38441] 11993.Ing, Y. L.; Leung, I. W. L.; Heng, H. H. Q.; Tsui, L.-C.; Lassam, N. J.: MLK-3: identification of a widely-expressed protein kinase bearing an SH3 domain and a leucine zipper–basic region domain. *Oncogene* 9:1745–1750, 1994.
- [38442] 11994.Bernard, O. A.; Mauchauffe, M.; Mecucci, C.; Van Den Berghe, H.; Berger, R.: A novel gene, AF-1p, fused to HRX in t(1;11)(p32;q23), is not related to AF-4, AF-9 nor ENL. *Oncogene* 9: 1039–1045, 1994.
- [38443] 11995.Fazioli, F.; Minichiello, L.; Matoska, V.; Castagnino, P.; Miki, T.; Wong, W. T.; Di Fiore, P. P.: Eps8, a substrate for the epidermal growth factor receptor kinase, enhances

EGF-dependent mitogenic signals. *EMBO J.* 12:
3799–3808, 1993.

[38444] 11996. Fazioli, F.; Minichiello, L.; Matoskova, B.; Wong, W. T.; Di Fiore, P. P.: eps 15, a novel tyrosine kinase substrate, exhibits transforming activity. *Molec. Cell. Biol.* 13:
5814–5828, 1993.

[38445] 11997. Fazioli, F.; Wong, W. T.; Ullrich, S. J.; Sakaguchi, K.; Appella, E.; Di Fiore, P. P.: The ezrin-like family of tyrosine kinase substrates: receptor-specific pattern of tyrosine phosphorylation and relationship to malignant transformation. *Oncogene* 8: 1335–1345, 1993.

[38446] 11998. Wong, W. T.; Kraus, M. H.; Carlomagno, F.; Zelano, A.; Druck, T.; Croce, C. M.; Huebner, K.; Di Fiore, P. P.: The human eps15 gene, encoding a tyrosine kinase substrate, is conserved in evolution and maps to 1p31–p32. *Oncogene* 9: 1591–1597, 1994.

[38447] 11999. Kozu, T.; Henrich, B.; Schafer, K. P.: Structure and expression of the gene (HNRPA2B1) encoding the human hnRNP protein A2/B1. *Genomics* 25:365–371, 1995.

[38448] 12000. Kordeli, E.; Lambert, S.; Bennett, V.: Ankyrin-G: a new ankyrin gene with neural-specific isoforms localized at the axonal initial segment and node of Ranvier. *J. Biol. Chem.* 270: 2352–2359, 1995.

- [38449] 12001.Hsu, L. C.; Chang, W.-C.; Yoshida, A.: Human aldehyde dehydrogenase genes, ALDH7 and ALDH8: genomic organization and gene structure comparison. *Gene* 189:89–94, 1997.
- [38450] 12002.Hsu, L. C.; Chang, W.-C.; Yoshida, A.: Cloning of a cDNA encoding human ALDH7, a new member of the aldehyde dehydrogenase family. *Gene* 151:285–289, 1994.
- [38451] 12003.Aruga, J.; Yokota, N.; Hashimoto, M.; Furuichi, T.; Fukuda, M.; Mikoshiba, K.: A novel zinc finger protein, Zic, is involved in neurogenesis, especially in the cell lineage of cerebellar granule cells. *J. Neurochem.* 63:1880–1890, 1994.
- [38452] 12004.Salero, E.; Perez-Sen, R.; Aruga, J.; Gimenez, C.; Zafra, F.: Transcription factors Zic1 and Zic2 bind and transactivate the apolipoprotein E gene promoter. *J. Biol. Chem.* 276: 1881–1888, 2001.
- [38453] 12005.Yokota, N.; Aruga, J.; Takai, S.; Yamada, K.; Hamazaki, M.; Iwase, T.; Sugimura, H.; Mikoshiba, K.: Pre-dominant expression of human Zic in cerebellar granule cell lineage and medulloblastoma. *Cancer Res.* 56: 377–383, 1996.
- [38454] 12006.Chen, N. N.; Chang, C.-F.; Gallia, G. L.; Kerr, D. A.; Johnson, E. M.; Krachmarov, C. P.; Barr, S. M.; Frisque, R. J.;

Bollag, B.;Khalili, K.: Cooperative action of cellular proteins YB-1 and Pur-alpha with the tumor antigen of the human JC polyomavirus determines their interaction with the viral lytic control element. Proc. Nat. Acad.Sci. 92: 1087-1091, 1995.

[38455] 12007.Gallia, G. L.; Johnson, E. M.; Khalili, K.: Pur-alpha: a multifunctional single-stranded DNA- and RNA-binding protein. Nucleic Acids Res. 28:3197-3205, 2000.

[38456] 12008.Kennedy, G. C.; Rutter, W. J.: Pur-1, a zinc-finger protein that binds to purine-rich sequences, transactivates an insulin promoter in heterologous cells. Proc. Nat. Acad. Sci. 89: 11498-11502, 1992.

[38457] 12009.Ma, Z.-W.; Pejovic, T.; Najfeld, V.; Ward, D. C.; Johnson, E. M.: Localization of PURA, the gene encoding the sequence-specific single-stranded-DNA-binding protein Pur-alpha, to chromosome band 5q31. Cytogenet. Cell Genet. 71:64-67, 1995.

[38458] 12010.Agerberth, B.; Gunne, H.; Odeberg, J.; Kogner, P.; Boman, H. G.; Gudmundsson, G. H.: FALL-39, a putative human peptide antibiotic, is cysteine-free and expressed in bone marrow and testis. Proc. Nat. Acad. Sci. 92: 195-199, 1995.

[38459] 12011.Frohm, M.; Agerberth, B.; Ahangari, G.; Stahle-

Backdahl, M.; Liden, S.; Wigzell, H.; Gudmundsson, G. H.: The expression of the gene coding for the antibacterial peptide LL-37 is induced in human keratinocytes during inflammatory disorders. *J. Biol. Chem.* 272: 15258–15263, 1997.

[38460] 12012. Gudmundsson, G. H.; Agerberth, B.; Odeberg, J.; Bergman, T.; Olsson, B.; Salcedo, R.: The human gene FALL39 and processing of the cathelin precursor to the antibacterial peptide LL-37 in granulocytes. *Europ. J. Biochem.* 238: 325–332, 1996.

[38461] 12013. Kirikae, T.; Hirata, M.; Yamasu, H.; Kirikae, F.; Tamura, H.; Kayama, F.; Nakatsuka, K.; Yokochi, T.; Nakano, M.: Protective effects of a human 18-kilodalton cationic antimicrobial protein (CAP18)-derived peptide against murine endotoxemia. *Infect. Immun.* 66: 1861–1868, 1998.

[38462] 12014. Matsuzono, Y.; Kinoshita, N.; Tamura, S.; Shimozawa, N.; Hamasaki, M.; Ghaedi, K.; Wanders, R. J. A.; Suzuki, Y.; Kondo, N.; Fujiki, Y.: Human PEX19: cDNA cloning by functional complementation, mutation analysis in a patient with Zellweger syndrome, and potential role in peroxisomal membrane assembly. *Proc. Nat. Acad. Sci.* 96: 2116–2121, 1999.

- [38463] 12015.Kinoshita, N.; Ghaedi, K.; Shimozawa, N.; Wanders, R. J. A.; Matsuzono, Y.; Imanaka, T.; Okumoto, K.; Suzuki, Y.; Kondo, N.; Fujiki, Y.: Newly identified Chinese hamster ovary cell mutants are defective in biogenesis of peroxisomal membrane vesicles (peroxisomal ghosts), representing a novel complementation group in mammals J. Biol. Chem. 273:24122–24130, 1998.
- [38464] 12016.Chen, K.-S.; Gunaratne, P. H.; Hoheisel, J. D.; Young, I. G.; Gabor Miklos, G. L.; Greenberg, F.; Shaffer, L. G.; Campbell, H. D.; Lupski, J. R.: The human homologue of the *Drosophila melanogaster* flightless-I gene (*flil*) maps within the Smith–Magenis microdeletion critical region on 17p11.2. Am. J. Hum. Genet. 56: 175–182, 1995.
- [38465] 12017.Heisterkamp, N.; Kaartinen, V.; van Soest, S.; Bokoch, G. M.; Groffen, J.: Human ABR encodes a protein with GAP–rac activity and homology to the DBL nucleotide exchange factor domain. J. Biol. Chem. 268:16903–16906, 1993.
- [38466] 12018.Heisterkamp, N.; Morris, C.; Groffen, J.: ABR, an active BCR–related gene. Nucleic Acids Res. 17: 8821–8831, 1989.
- [38467] 12019.McDonald, J. D.; Daneshvar, L.; Willert, J. R.; Matsuura, K.; Waldman, F.; Cogen, P. H.: Physical mapping of

chromosome 17p13.3 in the region of a putative tumor suppressor gene important in medulloblastoma. *Genomics* 23:229–232, 1994.

- [38468] 12020. Shimomura, H.; Sanke, T.; Hanabusa, T.; Tsunoda, K.; Furuta, H.; Nanjo, K.: Nonsense mutation of islet-1 gene (Q310X) found in a type 2 diabetic patient with a strong family history. *Diabetes* 49: 1597–1600, 2000.
- [38469] 12021. Tanizawa, Y.; Riggs, A. C.; Dagogo-Jack, S.; Vaxillaire, M.; Froguel, P.; Liu, L.; Donis-Keller, H.; Permutt, M. A.: Isolation of the human LIM/homeodomain gene islet-1 and identification of a simple sequence repeat 1. *Diabetes* 43: 935–941, 1994.
- [38470] 12022. Kleiman, F. E.; Manley, J. L.: Functional interaction of BRCA1-associated BARD1 with polyadenylation factor CstF-50. *Science* 285: 1576–1579, 1999.
- [38471] 12023. Ishibashi, T.; Bottaro, D. P.; Chan, A.; Miki, T.; Aaronson, S. A.: Expression cloning of a human dual-specificity phosphatase. *Proc. Nat. Acad. Sci.* 89: 12170–12174, 1992.
- [38472] 12024. Kamb, A.; Futreal, P. A.; Rosenthal, J.; Cochran, C.; Harshman, K. D.; Liu, Q.; Phelps, R. S.; Tavtigian, S. V.; Tran, T.; Hussey, C.; Bell, R.; Miki, Y.; Swensen, J.; Hobbs, M. R.; Marks, J.; Bennett, L. M.; Barrett, J. C.; Wiseman, R.

W.; Shattuck-Eidens, D.: Localization of the VHR phosphatase gene and its analysis as a candidate for BRCA1. *Genomics* 23:163–167, 1994.

[38473] 12025. Corti, O.; Finocchiaro, G.; Rossi, E.; Zuffardi, O.; DiDonato, S.: Molecular cloning of cDNAs encoding human carnitine acetyltransferase and mapping of the corresponding gene to chromosome 9q34.1. *Genomics* 23:94–99, 1994.

[38474] 12026. Kalaria, R. N.; Harik, S. I.: Carnitine acetyltransferase activity in the human brain and its microvessels is decreased in Alzheimer's disease. *Ann. Neurol.* 32: 583–586, 1992.

[38475] 12027. van der Leij, F. R.; Huijkman, N. C. A.; Boomsma, C.; Kuipers, J. R. G.; Bartelds, B.: Genomics of the human carnitine acyltransferase genes. *Molec. Genet. Metab.* 71: 139–153, 2000.

[38476] 12028. Couch, F. J.; Rommens, J. M.; Neuhausen, S. L.; Belanger, C.; Dumont, M.; Abel, K.; Bell, R.; Berry, S.; Bogden, R.; Cannon-Albright, L.; Farid, L.; Frye, C.; and 30 others: Generation of an integrated transcription map of the BRCA2 region on chromosome 13q12–q13. *Genomics* 36:86–99, 1996.

[38477] 12029. Nelms, K.; Snow, A. J.; Noben-Trauth, K.: Dok1 en-

coding p62(dok) maps to mouse chromosome 6 and human chromosome 2 in a region of translocation in chronic lymphocytic leukemia. *Genomics* 53: 243–245, 1998.

[38478] 12030. Wisniewski, D.; Strife, A.; Wojchietowicz, D.; Lambek, C.; Clarkson, B.: A 62-kilodalton tyrosine phosphoprotein constitutively present in primary chronic phase chronic myelogenous leukemia enriched lineage-negative blast populations. *Leukemia* 8: 688–693, 1994.

[38479] 12031. Gengyo-Ando, K.; Kitayama, H.; Mukaida, M.; Ikawa, Y.: A murine neural-specific homolog corrects cholinergic defects in *Caenorhabditis elegans* unc-18 mutants. *J. Neurosci.* 16: 6695–6702, 1996.

[38480] 12032. Pevsner, J.; Hsu, S.-C.; Scheller, R. H.: n-Sec1: a neural-specific syntaxin-binding protein. *Proc. Nat. Acad. Sci.* 91: 1445–1449, 1994.

[38481] 12033. Swanson, D. A.; Steel, J. M.; Valle, D.: Identification and characterization of the human ortholog of rat STXBP1, a protein implicated in vesicle trafficking and neurotransmitter release. *Genomics* 48: 373–376, 1998.

[38482] 12034. Verhage, M.; Mala, A. S.; Plomp, J. J.; Brussaard, A. B.; Heeroma, J. H.; Vermeer, H.; Toonen, R. F.; Hammer, R. E.; van den Berg, T. K.; Missler, M.; Geuze, H. J.; Sudhof, T. C.: Synaptic assembly of the brain in the absence of neuro-

transmitter secretion. *Science* 287:864–869, 2000.

[38483] 12035.Cox, P. R.; Zoghbi, H. Y.: Sequencing, expression analysis, and mapping of three unique human tropomodulin genes and their mouse orthologs. *Genomics* 63:97–107, 2000.

[38484] 12036.Watakabe, A.; Kobayashi, R.; Helfman, D. M.: N-tropomodulin: a novel isoform of tropomodulin identified as the major binding protein to brain tropomyosin. *J. Cell Sci.* 109: 2299–2310, 1996.

[38485] 12037.Galvin, K. M.; Donovan, M. J.; Lynch, C. A.; Meyer, R. I.; Paul, R. J.; Lorenz, J. N.; Fairchild–Huntress, V.; Dixon, K. L.; Dunmore, J. H.; Gimbrone, M. A., Jr.; Falb, D.; Huszar, D.: A role for Smad6 in development and homeostasis of the cardiovascular system. *Nature Genet.* 24: 171–174, 2000.

[38486] 12038.Topper, J. N.; Cai, J.; Qiu, Y.; Anderson, K. R.; Xu, Y.–Y.; Deeds, J. D.; Feeley, R.; Gimeno, C. J.; Woolf, E. A.; Tayber, O.; Mays, G. G.; Sampson, B. A.; Schoen, F. J.; Gimbrone, M. A., Jr.; Falb, D.: Vascular MADs: two novel MAD-related genes selectively inducible by flow in human vascular endothelium. *Proc. Nat. Acad. Sci.* 94:9314–9319, 1997.

[38487] 12039.Festing, M. F. W. (ed.): *Inbred strains in biomedical*

research. New York: Oxford University Press , 1979. p. 255.

- [38488] 12040. Illa, I.; Serrano-Munuera, C.; Gallardo, E.; Lasa, A.; Rojas-Garcia, R.; Palmer, J.; Gallano, P.; Baiget, M.; Matsuda, C.; Brown, R. H.: Distal anterior compartment myopathy: a dysferlin mutation causing a new muscular dystrophy phenotype. *Ann. Neurol.* 49: 130–134, 2001.
- [38489] 12041. Illarioshkin, S. N.; Ivanova-Smolenskaya, I. A.; Greenberg, C. R.; Nylen, E.; Sukhorukov, V. S.; Poleshchuk, V. V.; Markova, E. D.; Wrogemann, K.: Identical dysferlin mutation in limb-girdle muscular dystrophy type 2B and distal myopathy. *Neurology* 55: 1931–1933, 2000.
- [38490] 12042. Liu, J.; Wu, C.; Bossie, K.; Bejaoui, K.; Hosler, B. A.; Gingrich, J. C.; Ben Hamida, M.; Hentati, F.; Schurr, E.; de Jong, P. J.; Brown, R. H., Jr.: Generation of 3-Mb PAC contig spanning the Miyoshi myopathy/limb-girdle muscular dystrophy (MM/LGMD2B) locus on chromosome 2p13. *Genomics* 49:23–29, 1998.
- [38491] 12043. Mahjneh, I.; Vannelli, G.; Bushby, K.; Marconi, G. P.: A large inbred Palestinian family with two forms of muscular dystrophy. *Neuromusc. Disord.* 2: 277–283, 1992.
- [38492] 12044. Weiler, T.; Bashir, R.; Anderson, L. V. B.; Davison, K.; Moss, J. A.; Britton, S.; Nylen, E.; Keers, S.; Vafiadaki, E.;

Greenberg,C. R.; Bushby, K. M. D.; Wrogemann, K.: Identical mutation in patients with limb girdle muscular dystrophy type 2B or Miyoshi myopathy suggests a role for modifier gene(s). *Hum. Molec. Genet.* 8: 871–877, 1999.

[38493] 12045. Pennica, D.; Arce, V.; Swanson, T. A.; Vejsada, R.; Pollock, R.A.; Armanini, M.; Dudley, K.; Phillips, H. S.; Rosenthal, A.; Kato, A. C.; Henderson, C. E.: Cardiotrophin-1, a cytokine present in embryonic muscle, supports long-term survival of spinal motoneurons. *Neuron* 17:63–74, 1996.

[38494] 12046. Pennica, D.; King, K. L.; Shaw, K. J.; Luis, E.; Rullamas, J.; Luoh, S.-M.; Darbonne, W. C.; Knutzon, D. S.; Yen, R.; Chien, K. R.; Baker, J. B.; Wood, W. I.: Expression cloning of cardiotrophin 1, a cytokine that induces cardiac myocyte hypertrophy. *Proc. Nat. Acad. Sci.* 92: 1142–1146, 1995.

[38495] 12047. Ghosh, A.: Learning more about NMDA receptor regulation. *Science* 295:449–451, 2002.

[38496] 12048. Grunwald, I. C.; Korte, M.; Wolfer, D.; Wilkinson, G. A.; Unsicker, K.; Lipp, H.-P.; Bonhoeffer, T.; Klein, R.: Kinase-independent requirement of EphB2 receptors in hippocampal synaptic plasticity. *Neuron* 32:1027–1040, 2001.

- [38497] 12049.Henderson, J. T.; Georgiou, J.; Jia, Z.; Robertson, J.; Elowe,S.; Roder, J. C.; Pawson, T.: The receptor tyrosine kinase EphB2regulates NMDA–dependent synaptic function. *Neuron* 32: 1041–1056,2001.
- [38498] 12050.Himanen, J.–P.; Rajashankar, K. R.; Lackmann, M.; Cowan, C. A.;Henkemeyer, M.; Nikolov, D. B.: Crystal structure of an Eph receptor–ephrincomplex. *Nature* 414: 933–938, 2001.
- [38499] 12051.Takasu, M. A.; Dalva, M. B.; Zigmond, R. E.; Greenberg, M. E.: Modulation of NMDA receptor–dependent calcium influx and gene expressionthrough EphB receptors. *Science* 295: 491–495, 2002.
- [38500] 12052.Doudney, K.; Murdoch, J. N.; Paternotte, C.; Bentley, L.; Gregory,S.; Copp, A. J.; Stanier, P.: Comparative physical and transcriptmaps of approximately 1 Mb around loop–tail, a gene for severe neuraltube defects on distal mouse chromosome 1 and human chromosome 1q22–q23. *Genomics* 72:180–192, 2001.
- [38501] 12053.Kibar, Z.; Vogan, K. J.; Groulx, N.; Justice, M. J.; Underhill,D. A.; Gros, P.: Ltap, a mammalian homolog of *Drosophila Strabismus/VanGogh*, is altered in the mouse neural tube mutant loop–tail. *NatureGenet.* 28: 251–255, 2001.

- [38502] 12054.Mullick, A.; Trasler, D.; Gros, P.: High-resolution linkage map in the vicinity of the Lp locus. *Genomics* 26: 479–488, 1995.
- [38503] 12055.Murdoch, J. N.; Doudney, K.; Paternotte, C.; Copp, A. J.; Stanier, P.: Severe neural tube defects in the loop-tail mouse result from mutation of Lpp1, a novel gene involved in floor plate specification. *Hum.Molec. Genet.* 10: 2593–2601, 2001.
- [38504] 12056.Stanier, P.; Henson, J. N.; Eddleston, J.; Moore, G. E.; Copp, A. J.: Genetic basis of neural tube defects: the mouse gene loop-tail maps to a region of chromosome 1 syntenic with human 1q21–q23. *Genomics* 26:473–478, 1995.
- [38505] 12057.Yu, J. X.; Chao, L.; Chao, J.: Prostaticin is a novel human serineproteinase from seminal fluid: purification, tissue distribution, and localization in prostate gland. *J. Biol. Chem.* 269: 18843–18848, 1994.
- [38506] 12058.Yu, J. X.; Chao, L.; Chao, J.: Molecular cloning, tissue-specific expression, and cellular localization of human prostaticin mRNA. *J.Biol. Chem.* 270: 13483–13489, 1995.
- [38507] 12059.Yu, J. X.; Chao, L.; Ward, D. C.; Chao, J.: Structure and chromosomal localization of the human prostaticin (PRSS8) gene. *Genomics* 32: 334–340, 1996.

- [38508] 12060.Arber, S.; Halder, G.; Caroni, P.: Muscle LIM protein, a novelessential regulator of myogenesis, promotes myogenic differentiation. *Cell* 79:221–231, 1994.
- [38509] 12061.Fung, Y. W.; Wang, R. X.; Heng, H. H. Q.; Liew, C. C.: Mapping of a human LIM protein (CLP) to human chromosome 11p15.1 by fluorescence in situ hybridization. *Genomics* 28: 602–603, 1995.
- [38510] 12062.Becker–Andre, M.; Andre, E.; DeLamarter, J. F.: Identification of nuclear receptor mRNAs by RT–PCR amplification of conserved zinc–finger motif sequences. *Biochem. Biophys. Res. Commun.* 194: 1371–1379, 1993.
- [38511] 12063.Carlberg, C.; Hooft van Huijsduijnen, R.; Staple, J. K.; DeLamarter, J. F.; Becker–Andre, M.: RZR_s, a new family of retinoid–related orphan receptors that function as both monomers and homodimers. *Molec. Endocr.* 8:757–770, 1994.
- [38512] 12064.Giguere, V.; Beatty, B.; Squire, J.; Copeland, N. G.; Jenkins, N. A.: The orphan nuclear receptor ROR–alpha (RORA) maps to a conserved region of homology on human chromosome 15q21–q22 and mouse chromosome 9. *Genomics* 28: 596–598, 1995.
- [38513] 12065.Giguere, V.; Tini, M.; Flock, G.; Ong, E.; Evans, R. M.; Otulakowski, G.: Isoform–specific amino–terminal do–

mains dictate DNA-binding properties of ROR- α , a novel family of orphan hormone nuclear receptors.

Genes Dev. 8: 538–553, 1994.

- [38514] 12066. Hamilton, B. A.; Frankel, W. N.; Kerrebrock, A. W.; Hawkins, T. L.; FitzHugh, W.; Kusumi, K.; Russell, L. B.; Mueller, K. L.; vanBerkel, V.; Birren, B. W.; Kruglyak, L.; Lander, E. S.: Disruption of the nuclear hormone receptor ROR- α in staggerer mice. *Nature* 379:736–739, 1996.
- [38515] 12067. Matysiak-Scholze, U.; Nehls, M.: The structural integrity of ROR- α isoforms is mutated in staggerer mice: cerebellar coexpression of ROR- α -1 and ROR- α -4. *Genomics* 43: 78–84, 1997.
- [38516] 12068. Meyer, T.; Kneissel, M.; Mariani, J.; Fournier, B.: In vitro and in vivo evidence for orphan nuclear receptor ROR- α function in bone metabolism. *Proc. Nat. Acad. Sci.* 97: 9197–9202, 2000.
- [38517] 12069. Ueda, H. R.; Chen, W.; Adachi, A.; Wakamatsu, H.; Hayashi, S.; Takasugi, T.; Nagano, M.; Nakahama, K.; Suzuki, Y.; Sugano, S.; Iino, M.; Shigeyoshi, Y.; Hashimoto, S.: A transcription factor response element for gene expression during circadian night. *Nature* 418:534–539, 2002.
- [38518] 12070. Zanjani, H. S.; Herrup, K.; Guastavino, J.-M.; Del-

hayé-Bouchaud,N.; Mariani, J.: Developmental studies of the inferior olivary nucleus in staggerer mutant mice. *Develop. Brain Res.* 82: 18–28, 1994.

[38519] 12071.Masternak, K.; Barras, E.; Zufferey, M.; Conrad, B.; Corthals,G.; Aebersold, R.; Sanchez, J.–C.; Hochstrasser, D. F.; Mach, B.;Reith, W.: A gene encoding a novel RFX–associated transactivator is mutated in the majority of MHC class II deficiency patients. *NatureGenet.* 20: 273–277, 1998.

[38520] 12072.Prange, C. K.; Pennacchio, L. A.; Lieuallen, K.; Fan, W.; Lennon,G. G.: Characterization of the human neurocan gene, CSPG3. *Gene* 221:199–205, 1998.

[38521] 12073.Rauch, U.; Grimpe, B.; Kulbe, G.; Arnold–Ammer, I.; Beier, D. R.;Fassler, R.: Structure and chromosomal localization of the mouse neurocan gene. *Genomics* 28: 405–410, 1995.

[38522] 12074.Rauch, U.; Karthikeyan, L.; Maurel, P.; Margolis, R. U.; Margolis,R. K.: Cloning and primary structure of neurocan, a developmentally regulated, aggregating chondroitin sulfate proteoglycan of brain. *J.Biol. Chem.* 267: 19536–19547, 1992.

[38523] 12075.Clement, S.; Krause, U.; Desmedt, F.; Tanti, J.–F.; Behrends, J.;Pesesse, X.; Sasaki, T.; Penninger, J.; Doherty,

M.; Malaisse, W.; Dumont, J. E.; Le Marchand-Brustel, Y.; Erneux, C.; Hue, L.; Schurmans, S.: The lipid phosphatase SHIP2 controls insulin sensitivity. *Nature* 409:92–97, 2001.

[38524] 12076. Habib, T.; Hejna, J. A.; Moses, R. E.; Decker, S. J.: Growth factors and insulin stimulate tyrosine phosphorylation of the 51C/SHIP2 protein. *J. Biol. Chem.* 273: 18605–18609, 1998.

[38525] 12077. Hejna, J. A.; Saito, H.; Merkens, L. S.; Tittle, T. V.; Jakobs, P. M.; Whitney, M. A.; Grompe, M.; Friedberg, A. S.; Moses, R. E.: Cloning and characterization of a human cDNA (INPPL1) sharing homology with inositol polyphosphate phosphatases. *Genomics* 29: 285–287, 1995.

[38526] 12078. Pesesse, X.; Deleu, S.; De Smedt, F.; Drayer, L.; Erneux, C.: Identification of a second SH2-domain-containing protein closely related to the phosphatidylinositol polyphosphate 5-phosphatase SHIP. *Biochem. Biophys. Res. Commun.* 239: 697–700, 1997.

[38527] 12079. Schurmans, S.; Carrio, R.; Behrends, J.; Pouillon, V.; Merino, J.; Clement, S.: The mouse SHIP2 (Inppl1) gene: complementary DNA, genomic structure, promoter analysis, and gene expression in the embryo and adult mouse. *Genomics* 62: 260–271, 1999.

- [38528] 12080.Drummond, J. T.; Genschel, J.; Wolf, E.; Modrich, P.: DHFR/MSH3 amplification in methotrexate-resistant cells alters the hMutS-alpha/hMutS-beta ratio and reduces the efficiency of base-base mismatch repair. *Proc.Nat. Acad. Sci.* 94: 10144-10149, 1997.
- [38529] 12081.Fujii, H.; Shimada, T.: Isolation and characterization of cDNA clones derived from the divergently transcribed gene in the region upstream from the human dihydrofolate reductase gene. *J. Biol. Chem.* 264:10057-10064, 1989.
- [38530] 12082.Inokuchi, K.; Ikejima, M.; Watanabe, A.; Nakajima, E.; Orimo, H.; Nomura, T.; Shimada, T.: Loss of expression of the human MSH3 gene in hematological malignancies. *Biochem. Biophys. Res. Commun.* 214:171-179, 1995.
- [38531] 12083.Linton, J. P.; Yen, J. Y.-J.; Selby, E.; Chen, Z.; Chinsky, J.M.; Liu, K.; Kellems, R. E.; Crouse, G. F.: Dual bidirectional promoters at the mouse dhfr locus: cloning and characterization of two mRNA classes of the divergently transcribed Rep-1 gene. *Molec. Cell. Biol.* 9:3058-3072, 1989.
- [38532] 12084.Marra, G.; Iaccarino, I.; Lettieri, T.; Roscilli, G.; Delmastro, P.; Jiricny, J.: Mismatch repair deficiency associated with overexpression of the MSH3 gene. *Proc. Nat. Acad.*

Sci. 95: 8568–8573, 1998.

- [38533] 12085. Nakajima, E.; Orimo, H.; Ikejima, M.; Shimada, T.: Nine-bp repeat polymorphism in exon 1 of the hMSH3 gene. *Jpn. J. Hum. Genet.* 40:343–345, 1995.
- [38534] 12086. Risinger, J. I.; Umar, A.; Boyd, J.; Berchuck, A.; Kunkel, T. A.; Barrett, J. C.: Mutation of MSH3 in endometrial cancer and evidence for its functional role in heteroduplex repair. *Nature Genet.* 14:102–109, 1996.
- [38535] 12087. Smith, M. L.; Mitchell, P. J.; Crouse, G. F.: Analysis of the mouse Dhfr/Rep-3 major promoter region by using linker-scanning and internal deletion mutations and DNase I footprinting. *Molec. Cell Biol.* 10: 6003–6012, 1990.
- [38536] 12088. Watanabe, A.; Ikejima, M.; Suzuki, N.; Shimada, T.: Genomic organization and expression of the human MSH3 gene. *Genomics* 31: 311–318, 1996.
- [38537] 12089. Yin, J.; Kong, D.; Wang, S.; Zou, T.–T. Souza, R. F.; Smolinski, K. N.; Lynch, P. M.; Hamilton, S. R.; Sugimura, H.; Powell, S. M.; Young, J.; Abraham, J. M.; Meltzer, S. J.: Mutation of hMSH3 and hMSH6 mismatch repair genes in genetically unstable human colorectal and gastric carcinomas. *Hum. Mutat.* 10: 474–478, 1997.
- [38538] 12090. Chan, A. M–L.; McGovern, E. S.; Catalano, G.; Fleming, T. P.; Miki, T.: Expression cDNA cloning of a novel

oncogene with sequence similarity to regulators of small GTP-binding proteins. *Oncogene* 9: 1057–1063, 1994.

[38539] 12091. Miki, T.; Fleming, T. P.; Crescenzi, M.; Molloy, C. J.; Blam, S. B.; Reynolds, S. H.; Aaronson, S. A.: Development of a highly efficient expression cDNA cloning system: application to oncogene isolation. *Proc. Nat. Acad. Sci.* 88: 5167–5171, 1991.

[38540] 12092. Takai, S.; Chan, A. M.-L.; Yamada, K.; Miki, T.: Assignment of the human TIM proto-oncogene to 7q33–q35. *Cancer Genet. Cytogenet.* 83:87–89, 1995.

[38541] 12093. Burger, B.; Uhlhaas, S.; Mangold, E.; Propping, P.; Friedl, W.; Jenne, D.; Dockter, G.; Back, W.: Novel de novo mutation of MADH4/SMAD4 in a patient with juvenile polyposis. (Letter) *Am. J. Med. Genet.* 110:289–291, 2002.

[38542] 12094. Kurima, K.; Peters, L. M.; Yang, Y.; Riazuddin, S.; Ahmed, Z. M.; Naz, S.; Arnaud, D.; Drury, S.; Mo, J.; Makishima, T.; Ghosh, M.; Menon, P. S. N.; and 13 others: Dominant and recessive deafness caused by mutations of a novel gene, TMC1, required for cochlear hair-cell function. *Nature Genet.* 30: 277–284, 2002.

[38543] 12095. Bryant, P. J.; Huettner, B.; Held, L. I., Jr.; Ryerse, J.; Szidonya, J.: Mutations at the fat locus interfere with cell proliferation control and epithelial morphogenesis in

Drosophila. Dev. Biol. 129:541–554, 1988.

- [38544] 12096. Dunne, J.; Hanby, A. M.; Poulson, R.; Jones, T. A.; Sheer, D.; Chin, W. G.; Da, S. M.; Zhao, Q.; Beverley, P. C. L.; Owen, M. J.: Molecular cloning and tissue expression of FAT, the human homologue of the *Drosophila* fat gene that is located on chromosome 4q34–q35 and encodes a putative adhesion molecule. Genomics 30: 207–223, 1995.
- [38545] 12097. Hortsch, M.; Goodman, C. S.: Cell and substrate adhesion molecules in *Drosophila*. Ann. Rev. Cell. Biol. 7: 505–557, 1991.
- [38546] 12098. Mahoney, P. A.; Weber, U.; Onofrechuk, P.; Biessmann, H.; Bryant, P. J.; Goodman, C. S.: The fat tumor suppressor gene in *Drosophila* encodes a novel member of the cadherin gene superfamily. Cell 67: 853–868, 1991.
- [38547] 12099. Crowe, P. D.; VanArsdale, T. L.; Walter, B. N.; Ware, C. F.; Hession, C.; Ehrenfels, B.; Browning, J. L.; Din, W. S.; Goodwin, R. G.; Smith, C. A.: A lymphotoxin-beta-specific receptor. Science 264: 707–710, 1994.
- [38548] 12100. Nakamura, T.; Tashiro, K.; Nazarea, M.; Nakano, T.; Sasayama, S.; Honjo, T.: The murine lymphotoxin-beta receptor cDNA: isolation by the signal sequence trap and chromosomal mapping. Genomics 30: 312–319, 1995.
- [38549] 12101. Tashiro, K.; Tada, H.; Heilker, R.; Shirozu, M.;

Nakano, T.; Honjo, T.: Signal sequence trap: a cloning strategy for secreted proteins and type I membrane proteins. *Science* 261: 600–603, 1993.

[38550] 12102. George, A.; Sabsay, B.; Simonian, P. A.; Veis, A.: Characterization of a novel dentin matrix acidic phosphoprotein: implications for induction of biomineralization. *J. Biol. Chem.* 268: 12624–12630, 1993.

[38551] 12103. Hirst, K. L.; Simmons, D.; Feng, J.; Aplin, H.; Dixon, M. J.; MacDougall, M.: Elucidation of the sequence and the genomic organization of the human dentin matrix acidic phosphoprotein 1 (DMP1) gene: exclusion of the locus from a causative role in the pathogenesis of dentinogenesis imperfecta type II. *Genomics* 42: 38–45, 1997.

[38552] 12104. MacDougall, M.; DuPont, B. R.; Simmons, D.; Leach, R. J.: Assignment of DMP1 to human chromosome 4 band q21 by in situ hybridization. *Cytogenet. Cell. Genet.* 74: 189 only, 1996.

[38553] 12105. Lu, Z.; Xu, S.; Joazeiro, C.; Cobb, M. H.; Hunter, T.: The PHD domain of MEKK1 acts as an E3 ubiquitin ligase and mediates ubiquitination and degradation of ERK1/2. *Molec. Cell* 9: 945–956, 2002.

[38554] 12106. Bohm, H.; Benndorf, R.; Gaestel, M.; Gross, B.; Nurnberg, P.; Kraft, R.; Otto, A.; Bielka, H.: The growth-re-

lated protein P23 of the Ehrlich ascites tumor: translational control, cloning and primary structure. *Biochem.Int.* 19: 277–286, 1989.

[38555] 12107. Barnard, R. C.; Pascall, J. C.; Brown, K. D.; McKay, I. A.; Williams, N. S.; Bustin, S. A.: Coding sequence of ERF-1, the human homologue of Tis11b/cMG1, members of the Tis11 family of early response genes. *Nucleic Acids Res.* 21: 3580 only, 1993.

[38556] 12108. Bustin, S. A.; Xiao-Feng, N.; Barnard, R. C.; Kumar, V.; Pascall, J. C.; Brown, K. D.; Leigh, I. M.; Williams, N. S.; McKay, I. A.: Cloning and characterisation of ERF1, a human member of the Tis11 family of early-response genes. *DNA Cell Biol.* 13: 449–459, 1994.

[38557] 12109. Maclean, K. N.; See, C. G.; McKay, I. A.; Bustin, S. A.: The human immediate early gene BRF1 maps to chromosome 14q22–q24. *Genomics* 30:89–90, 1995.

[38558] 12110. Ning, Z.-Q.; Norton, J. D.; Li, J.; Murphy, J. J.: Distinct mechanisms for rescue from apoptosis in Ramos human B cells by signaling through CD40 and interleukin-4 receptor: a role for inhibition of an early response gene, Berg36. *Europ. J. Immun.* 26: 2356–2363, 1996.

[38559] 12111. Chihade, J. W.; Brown, J. R.; Schimmel, P. R.; Ribas de Pouplana, L.: Origin of mitochondria in relation to evo-

lutionary history of eukaryotic alanyl-tRNA synthetase.

Proc. Nat. Acad. Sci. 97: 12153–12157, 2000.

- [38560] 12112. Maas, S.; Kim, Y.-G.; Rich, A.: Genomic clustering of tRNA-specific adenosine deaminase ADAT1 and two tRNA synthetases. *Mammalian Genome* 12:387–393, 2001.
- [38561] 12113. Derynck, R.; Gelbart, W. M.; Harland, R. M.; Heldin, C.-H.; Kern, S. E.; Massague, J.; Melton, D. A.; Mlodzik, M.; Padgett, R. W.; Roberts, A. B.; Smith, J.; Thomsen, G. H.; Vogelstein, B.; Wang, X.-F.: Nomenclature: vertebrate mediators of TGF-beta family signals. (Letter) *Cell* 87:173 only, 1996.
- [38562] 12114. Friedl, W.; Kruse, R.; Uhlhaas, S.; Stolte, M.; Scharmann, B.; Keller, K. M.; Jungck, M.; Stern, M.; Loff, S.; Back, W.; Propping, P.; Jenne, D. E.: Frequent 4-bp deletion in exon 9 of the SMAD4/MADH4 gene in familial juvenile polyposis patients. *Genes Chromosomes Cancer* 25:403–406, 1999.
- [38563] 12115. Hahn, S. A.; Schutte, M.; Hoque, T. M. S.; Moskaluk, C. A.; daCosta, L. T.; Rozenblum, E.; Weinstein, C. L.; Fischer, A.; Yeo, C. J.; Hruban, R. H.; Kern, S. E.: DPC4, a candidate tumor suppressor gene at human chromosome 18q21.1. *Science* 271: 350–354, 1996.

- [38564] 12116.Howe, J. R.; Shellnut, J.; Wagner, B.; Ringold, J. C.; Sayed,M. G.; Ahmed, A. F.; Lynch, P. M.; Amos, C. I.; Sistonen, P.; Aaltonen,L. A.: Common deletion of SMAD4 in juvenile polyposis is a mutational hotspot. *Am. J. Hum. Genet.* 70: 1357–1362, 2002.
- [38565] 12117.Inman, G. J.; Nicolas, F. J.; Hill, C. S.: Nucleocytoplasmic shuttling of Smads 2, 3, and 4 permits sensing of TGF-beta receptor activity. *Molec. Cell* 10: 283–294, 2002.
- [38566] 12118.Kim, S. K.; Fan, Y.; Papadimitrakopoulou, V.; Clayman, G.; Hittelman,W. N.; Hong, W. K.; Lotan, R.; Mao, L.: DPC4, a candidate tumor suppressor gene, is altered infrequently in head and neck squamous cell carcinoma. *Cancer Res.* 56: 2519–2521, 1996.
- [38567] 12119.Kinzler, K. W.; Vogelstein, B.: Landscaping the cancer terrain. *Science* 280:1036–1037, 1998.
- [38568] 12120.MacGrogan, D.; et al.; et al: Comparative mutational analysis of DPC4 (Smad4) in prostatic and colorectal carcinomas. *Oncogene* 15:1111–1114, 1997.
- [38569] 12121.Roth, S.; Johansson, M.; Loukola, A.; Peltomaki, P.; Jarvinen,H.; Mecklin, J.-P.; Aaltonen, L. A.: Mutation analysis of SMAD2,SMAD3, and SMAD4 genes in hereditary non-polyposis colorectal cancer. *J.Med. Genet.* 37: 298–300, 2000.

- [38570] 12122.Schutte, M.; Hruban, R. H.; Hedrick, L.; Cho, K. R.; Nadasdy, G. M.; Weinstein, C. L.; Bova, G. S.; Isaacs, W. B.; Cairns, P.; Nawroz, H.; Sidransky, D.; Casero, R. A., Jr.; Meltzer, P. S.; Hahn, S. A.; Kern, S. E.: DPC4 gene in various tumor types. *Cancer Res.* 56:2527–2530, 1996.
- [38571] 12123.Shioda, T.; Lechleider, R. J.; Dunwoodie, S. L.; Li, H.; Yahata, T.; de Caestecker, M. P.; Fenner, M. H.; Roberts, A. B.; Isselbacher, K. J.: Transcriptional activating activity of Smad4: roles of SMAD hetero-oligomerization and enhancement by an associating transactivator. *Proc. Nat. Acad. Sci.* 95: 9785–9790, 1998.
- [38572] 12124.Sirard, C.; de la Pompa, J. L.; Elia, A.; Itie, A.; Mirtos, C.; Cheung, A.; Hahn, S.; Wakeham, A.; Schwartz, L.; Kern, S. E.; Rossant, J.; Mak, T. W.: The tumor suppressor gene Smad4/Dpc4 is required for gastrulation and later for anterior development of the mouse embryo. *Genes Dev.* 12: 107–119, 1998.
- [38573] 12125.Tagaki, Y.; et al.; et al: Somatic alterations of the DPC4 gene in human colorectal cancers in vivo. *Gastroenterology* 111: 1369–1372, 1996.
- [38574] 12126.Takaku, K.; Oshima, M.; Miyoshi, H.; Matsui, M.; Seldin, M. F.; Taketo, M. M.: Intestinal tumorigenesis in compound mutant mice of both Dpc4 (Smad4) and Apc

genes. *Cell* 92: 645–656, 1998.

- [38575] 12127. Thiagalingam, S.; Lengauer, C.; Leach, F. S.; Schutte, M.; Hahn, S. A.; Overhauser, J.; Willson, J. K. V.; Markowitz, S.; Hamilton, S. R.; Kern, S. E.; Kinzler, K. W.; Vogelstein, B.: Evaluation of candidate tumour suppressor genes on chromosome 18 in colorectal cancers. *Nature-Genet.* 13: 343–346, 1996.
- [38576] 12128. Zawel, L.; Dai, J. L.; Buckhaults, P.; Zhou, S.; Kinzler, K. W.; Vogelstein, B.; Kern, S. E.: Human Smad3 and Smad4 are sequence-specific transcription activators. *Molec. Cell* 1: 611–617, 1998.
- [38577] 12129. Zhou, S.; Buckhaults, P.; Zawel, L.; Bunz, F.; Riggs, G.; LeDai, J.; Kern, S. E.; Kinzler, K. W.; Vogelstein, B.: Targeted deletion of Smad4 shows it is required for transforming growth factor beta and activin signaling in colorectal cancer cells. *Proc. Nat. Acad. Sci.* 95: 2412–2416, 1998.
- [38578] 12130. Abbs, S.; Roberts, R. G.; Mathew, C. G.; Bentley, D. R.; Bobrow, M.: Accurate assessment of intragenic recombination frequency within the Duchenne muscular dystrophy gene. *Genomics* 7: 602–606, 1990.
- [38579] 12131. Ahn, A. H.; Kunkel, L. M.: The structural and functional diversity of dystrophin. *Nature Genet.* 3: 283–291,

1993.

- [38580] 12132. Alwine, J. C.; Kemp, D. J.; Stark, G. R.: Method for detection of specific RNAs in agarose gels by transfer to diazobenzyloxymethyl-paper and by hybridization with DNA probes. *Proc. Nat. Acad. Sci.* 74:5350–5354, 1977.
- [38581] 12133. Angelini, C.; Beggs, A. H.; Hoffman, E. P.; Fanin, M.; Kunkel, L. M.: Enormous dystrophin in a patient with Becker muscular dystrophy. *Neurology* 40:808–812, 1990.
- [38582] 12134. Badorff, C.; Berkely, N.; Mehrotra, S.; Talhouk, J. W.; Rhoads, R. E.; Knowlton, K. U.: Enteroviral protease 2A directly cleaves dystrophin and is inhibited by a dystrophin-based substrate analogue. *J. Biol. Chem.* 275: 11191–11197, 2000.
- [38583] 12135. Badorff, C.; Lee, G.-H.; Lamphear, B. J.; Martone, M. E.; Campbell, K. P.; Rhoads, R. E.; Knowlton, K. U.: Enteroviral protease 2A cleaves dystrophin: evidence of cytoskeletal disruption in an acquired cardiomyopathy. *Nature Med.* 5: 320–326, 1999.
- [38584] 12136. Bakker, E.; Pearson, P. L.: Mutation of the Duchenne muscular dystrophy gene associated with meiotic recombination. (Letter) *Clin. Genet.* 30: 347–349, 1986.
- [38585] 12137. Bakker, E.; Hofker, M. H.; Goor, N.; Mandel, J. L.;

Wrogemann,K.; Davies, K. E.; Kunkel, L. M.; Willard, H. F.; Fenton, W. A.; Sandkuyl,L.; Majoor–Krakauer, D.; van Es–sen, A. J.; Jahoda, M. G. J.; Sachs,E. S.; van Ommen, G. J. B.; Pearson, P. L.: Prenatal diagnosis andcarrier detection of Duchenne muscular dystrophy with closely linkedRFLPs. Lancet I: 655–658, 1985.

[38586] 12138.Bakker, E.; Van Broeckhoven, C.; Bonten, E. J.; van de Vooren,M. J.; Veenema, H.; Van Hul, W.; Van Ommen, G. J. B.; Vandenberghe,A.; Pearson, P. L.: Germline mo–saicism and Duchenne muscular dystrophymutations. Na–ture 329: 554–556, 1987.

[38587] 12139.Bar, S.; Barnea, E.; Levy, Z.; Neuman, S.; Yaffe, D.; Nudel, U.: A novel product of the Duchenne muscular dys–trophy gene which greatlydiffers from the known isoforms in its structure and tissue distribution. Biochem.J. 272: 557–560, 1990.

[38588] 12140.Barbieri, A. M.; Soriani, N.; Tubiello, G. M.; Ferrari, M.; Carrera,P.: A nonsense mutation (gln–673–term) in exon 17 of the human dystrophingene detected by het–eroduplex analysis. Hum. Genet. 96: 343–344,1995.

[38589] 12141.Bartlett, R. J.; Pericak–Vance, M. A.; Koh, J.; Ya–maoka, L. H.;Chen, J. C.; Hung, W.–Y.; Speer, M. C.; Wape–naar, M. C.; Van Ommen,G. J. B.; Bakker, E.; Pearson, P. L.;

Kandt, R. S.; Siddique, T.; Gilbert, J. R.; Lee, J. E.; Sirotkin-Roses, M. J.; Roses, A. D.: Duchenne muscular dystrophy: high frequency of deletions. *Neurology* 38:1-4, 1988.

[38590] 12142. Barton-Davis, E. R.; Cordier, L.; Shoturma, D. I.; Leland, S.E.; Sweeney, H. L.: Aminoglycoside antibiotics restore dystrophin function to skeletal muscles of mdx mice. *J. Clin. Invest.* 104:375-381, 1999.

[38591] 12143. Bastianutto, C.; Bestard, J. A.; Lahnakoski, K.; Broere, D.; DeVisser, M.; Zaccolo, M.; Pozzan, T.; Ferlini, A.; Muntoni, F.; Patarnello, T.; Klamut, H. J.: Dystrophin muscle enhancer 1 is implicated in the activation of non-muscle isoforms in the skeletal muscle of patients with X-linked dilated cardiomyopathy. *Hum. Molec. Genet.* 10: 2627-2635, 2001.

[38592] 12144. Baumbach, L. L.; Chamberlain, J. S.; Ward, P. A.; Farwell, N.J.; Caskey, C. T.: Molecular and clinical correlation of deletion leading to Duchenne and Becker muscular dystrophies. *Neurology* 39:465-474, 1989.

[38593] 12145. Baumbach, L. L.; Ward, P. A.; Fenwick, R.; Caskey, C. T.: Analysis of mutations at the Duchenne muscular dystrophy locus provides no evidence for illegitimate recombination in deletion formation. (Abstract) *Am.J. Hum. Genet.* 45 (suppl.): A173, 1989.

- [38594] 12146.Beggs, A. H.; Koenig, M.; Boyce, F. M.; Kunkel, L. M.: Detection of 98% of DMD/BMD gene deletions by polymerase chain reaction. *Hum.Genet.* 86: 45–48, 1990.
- [38595] 12147.Berko, B. A.; Swift, M.: X-linked dilated cardiomyopathy. *NewEng. J. Med.* 316: 1186–1191, 1987.
- [38596] 12148.Bettecken, T.; Muller, C. R.: Identification of a 220-kb insertion into the Duchenne gene in a family with an atypical course of muscular dystrophy. *Genomics* 4: 592–596, 1989.
- [38597] 12149.Bies, R. D.: X-linked dilated cardiomyopathy. (Letter) *New Eng.J. Med.* 330: 368–369, 1994.
- [38598] 12150.Bies, R. D.; Caskey, C. T.; Fenwick, R.: An intact cysteine-rich domain is required for dystrophin function. *J. Clin. Invest.* 90:666–672, 1992.
- [38599] 12151.Bittner, R. E.; Streubel, B.; Shorny, S.; Schaden, G.; Voit, T.; Hoyer, H.: Coisogenic all-plus-one immunization: a model for identifying missing proteins in null-mutant conditions. Antibodies to dystrophin in mdx mouse after transplantation of muscle from normal coisogenic donor. *Neuropediatrics* 25: 176–182, 1994.
- [38600] 12152.Ryu, S.; Zhou, S.; Ladurner, A. G.; Tjian, R.: The transcriptional cofactor complex CRSP is required for activity of the enhancer-binding protein Sp1. *Nature* 397:

446–450, 1999.

- [38601] 12153. Mohapatra, B.; Verma, S.; Shankar, S.; Suri, A.: Molecular cloning of human testis mRNA specifically expressed in haploid germ cells, having structural homology with the A-kinase anchoring proteins. *Biochem. Biophys. Res. Commun.* 244: 540–545, 1998.
- [38602] 12154. Turner, R. M. O.; Johnson, L. R.; Haig-Ladewig, L.; Gerton, G.L.; Moss, S. B.: An X-linked gene encodes a major human sperm fibrous sheath protein, hAKAP82: genomic organization, protein kinase A-R11 binding, and distribution of the precursor in the sperm tail. *J. Biol. Chem.* 273: 32135–32141, 1998.
- [38603] 12155. Dever, T. E.; Wei, C.-L.; Benkowski, L. A.; Browning, K.; Merrick, W. C.; Hershey, J. W. B.: Determination of the amino acid sequence of rabbit, human, and wheat germ protein synthesis factor eIF-4C by cloning and chemical sequencing. *J. Biol. Chem.* 269: 3212–3218, 1994.
- [38604] 12156. Lee, S.-H.; Kim, W.-H.; Kim, H.-K.; Woo, K.-M.; Nam, H.-S.; Kim, H.-S.; Kim, J.-G.; Cho, M.-H.: Altered expression of the fragile histidine triad gene in primary gastric adenocarcinomas. *Biochem. Biophys. Res. Commun.* 284: 850–855, 2001.
- [38605] 12157. Markkanen, A.; Heinonen, K.; Knuutila, S.; de la

Chapelle, A.: Methotrexate-induced increase in gap formation in human chromosome band 3p14. *Hereditas* 96: 317–319, 1982.

[38606] 12158. Markkanen, A.; Knuutila, S.; de la Chapelle, A.: Inducible fragile site on chromosome 3. (Letter) *Hum. Genet.* 65: 217 only, 1983.

[38607] 12159. Anderson, D. M.; Kumaki, S.; Ahdieh, M.; Bertles, J.; Tometsko, M.; Loomis, A.; Giri, J.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Valentine, V.; Shapiro, D. N.; Morris, S. W.; Park, L. S.; Cosman, D.: Functional characterization of the human interleukin-15 receptor alpha chain and close linkage of IL15RA and IL2RA genes. *J. Biol. Chem.* 270: 29862–29869, 1995.

[38608] 12160. Giri, J. G.; Kumaki, S.; Ahdieh, M.; Friend, D. J.; Loomis, A.; Shanebeck, K.; DuBose, R.; Cosman, D.; Park, L. S.; Anderson, D. M.: Identification and cloning of a novel IL-15 binding protein that is structurally related to the alpha of the IL-2 receptor. *EMBO J.* 14:3654–3663, 1995.

[38609] 12161. Yasunaga, S.; Grati, M.; Cohen-Salmon, M.; El-Amraoui, A.; Mustapha, M.; Salem, N.; El-Zir, E.; Loiselet, J.; Petit, C.: A mutation in OTOF, encoding otoferlin, a FER-1-like protein, causes DFNB9, a nonsyndromic form of deafness. *Nature Genet.* 21: 363–369, 1999.

- [38610] 12162. Berry, A.; Scott, H. S.; Kudoh, J.; Talior, I.; Korostishevsky, M.; Wattenhofer, M.; Guipponi, M.; Barras, C.; Rossier, C.; Shibuya, K.; Wang, J.; Kawasaki, K.; Asakawa, S.; Minoshima, S.; Shimizu, N.; Antonarakis, S.; Bonne-Tamir, B.: Refined localization of autosomal recessive non-syndromic deafness DFNB10 locus using 34 novel microsatellite markers, genomic structure, and exclusion of six known genes in the region. *Genomics* 68: 22–29, 2000.
- [38611] 12163. Good, P. J.; Chen, Q.; Warner, S. J.; Herring, D. C.: A family of human RNA-binding proteins related to the *Drosophila* Bruno transcriptional regulator. *J. Biol. Chem.* 275: 28583–28592, 2000.
- [38612] 12164. Roberts, R.; Timchenko, N. A.; Miller, J. W.; Reddy, S.; Caskey, C. T.; Swanson, M. S.; Timchenko, L. T.: Altered phosphorylation and intracellular distribution of a (CUG)_n triplet repeat RNA-binding protein in patients with myotonic dystrophy and in myotonin protein kinase knockout mice. *Proc. Nat. Acad. Sci.* 94: 13221–13226, 1997.
- [38613] 12165. Li, Z.; Jiang, H.; Xie, W.; Zhang, Z.; Smrcka, A. V.; Wu, D.: Roles of PLC- β -2 and - β -3 and PI3K- γ in chemoattractant-mediated signal transduction. *Science* 287: 1046–1049, 2000.
- [38614] 12166. Fowler, K. J.; Saffery, R.; Irvine, D. V.; Trowell, H. E.;

Choo,K. H. A.: Mouse centromere protein F (Cenpf) gene maps to the distalregion of chromosome 1 by interspecific backcross analysis. *Cytogenet.Cell Genet.* 82: 180–181, 1998.

- [38615] 12167.Liao, H.; Winkfein, R. J.; Mack, G.; Rattner, J. B.; Yen, T. J.: CENP–F is a protein of the nuclear matrix that assembles onto kinetochoresat late G2 and is rapidly degraded after mitosis. *J. Cell Biol.* 130:507–518, 1995.
- [38616] 12168.Rattner, J. B.; Rao, A.; Fritzler, M. J.; Valencia, D. W.; Yen,T. J.: CENP–F is a ca 400 kDa kinetochore protein that exhibits acell–cycle dependent localization. *Cell Motil. Cytoskeleton* 26:214–226, 1993.
- [38617] 12169.Panagopoulos, I.; Mencinger, M.; Dietrich, C. U.; Bjerkehagan,B.; Saeter, G.; Mertens, F.; Mandahl, N.; Heim, S.: Fusion of theRBP56 and CHN genes in extraskkeletal myxoid chondrosarcomas with translocation(9;17)(q22;q11). *Oncogene* 18: 7594–7598, 1999.
- [38618] 12170.Gassmann, M.; Casagrande, F.; Orioli, D.; Simon, H.; Lai, C.; Klein,R.; Lemke, G.: Aberrant neural and cardiac development in mice lackingthe ErbB4 neuregulin receptor. *Nature* 378: 390–394, 1995.
- [38619] 12171.Golding, J. P.; Trainor, P.; Krumlauf, R.; Gassmann, M.: Defectsin pathfinding by cranial neural crest cells in

mice lacking the neuregulinreceptor ErbB4. *Nature Cell Biol.* 2: 103–109, 2000.

- [38620] 12172.Zimonjic, D. B.; Alimandi, M.; Miki, T.; Popescu, N. C.; Kraus,M. H.: Localization of the human HER4/erbB–4 gene to chromosome 2. *Oncogene* 10:1235–1237, 1995.
- [38621] 12173.Adibi, S. A.: The oligopeptide transporter (Pept–1) in human intestine:biology and function. *Gastroenterology* 113: 332–340, 1997.
- [38622] 12174.Fei, Y. –J.; Kanai, Y.; Nussberger, S.; Ganapathy, V.; Leibach,F. H.; Romero, M. F.; Singh, S. K.; Boron, W. F.; Hediger, M. A.:Expression cloning of a mammalian proton–coupled oligopeptide transporter. *Nature* 368:563–566, 1994.
- [38623] 12175.Liang, R.; Fei, Y.–J.; Prasad, P. D.; Ramamoorthy, S.; Han, H.;Yang–Feng, T. L.; Hediger, M. A.; Ganapathy, V.; Leibach, F. H.:Human intestinal H(+)/peptide cotransporter: cloning, functional expression,and chromosomal localization. *J. Biol. Chem.* 270: 6456–6463, 1995.
- [38624] 12176.Heiber, M.; Docherty, J. M.; Shah, G.; Nguyen, T.; Cheng, R.; Heng,H. H. Q.; Marchese, A.; Tsui, L.–C.; Shi, X.; George, S. R.; O'Dowd,B. F.: Isolation of three novel human genes encoding G protein–coupledreceptors. *DNA Cell Biol.* 14: 25–35, 1995.

- [38625] 12177. Mahadevan, M. S.; Baird, S.; Bailly, J. E.; Shutler, G. G.; Sabourin, L. A.; Tsilfidis, C.; Neville, C. E.; Narang, M.; Korneluk, R. G.: Isolation of a novel G protein-coupled receptor (GPR4) localized to chromosome 19q13.3. *Genomics* 30: 84–88, 1995.
- [38626] 12178. Ye, R. D.; Prossnitz, E. R.; Zou, A.; Cochrane, C. G.: Characterization of a human cDNA that encodes a functional receptor for platelet activating factor. *Biochem. Biophys. Res. Commun.* 180: 105–111, 1991.
- [38627] 12179. Hossain, A.; Saunders, G. F.: The human sex-determining gene SRY is a direct target of WT1. *J. Biol. Chem.* 276: 16817–16823, 2001.
- [38628] 12180. Iida, T.; Nakahori, Y.; Komaki, R.; Mori, E.; Hayashi, N.; Tsutsumi, O.; Taketani, Y.; Nakagome, Y.: A novel missense mutation in the HMG box of the SRY gene in a patient with XY sex reversal. *Hum. Molec. Genet.* 3: 1437–1438, 1994.
- [38629] 12181. Jager, R. J.; Harley, V. R.; Pfeiffer, R. A.; Goodfellow, P. N.; Scherer, G.: A familial mutation in the testis-determining gene SRY shared by both sexes. *Hum. Genet.* 90: 350–355, 1992.
- [38630] 12182. Kawakura, K.; Miyake, Y.-I.; Murakami, R.-K.; Kondoh, S.; Hirata, T.-I.; Kaneda, Y.: Deletion of the SRY region

on the Y chromosomedetected in bovine gonadal hy-
poplasia (XY female) by PCR. Cytogenet.Cell Genet. 72:
183–184, 1996.

[38631] 12183.Lahr, G.; Maxson, S. C.; Mayer, A.; Just, W.; Pilgrim, C.; Reisert,I.: Transcription of the Y chromosomal gene, Sry, in adult mousebrain. Molec. Brain Res. 33: 179–182, 1995.

[38632] 12184.Li, B.; Zhang, W.; Chan, G.; Jancso–Radek, A.; Liu, S.; Weiss,M. A.: Human sex reversal due to impaired nu-
clear localization ofSRY: a clinical correlation. J. Biol.
Chem. 276: 46480–46484, 2001.

[38633] 12185.Lovell–Badge, R.; Robertson, E.: XY female mice re-
sulting froma heritable mutation in the primary testis–
determining gene, Tdy. Development 109:635–646, 1990.

[38634] 12186.Margarit, E.; Coll, M. D.; Oliva, R.; Gomez, D.; Soler, A.; Ballesta,F.: SRY gene transferred to the long arm of the X chromosome in aY–positive XX true hermaphrodite. Am. J. Med. Genet. 90: 25–28,2000.

[38635] 12187.Mittwoch, U.: Sex determination and sex reversal: genotype, phenotype,dogma and semantics. Hum. Genet. 89: 467–479, 1992.

[38636] 12188.Muller, J.; Schwartz, M.; Skakkebaek, N. E.: Analysis of thesex–determining region of the Y–chromosome (SRY)

in sex reversed patients: point-mutation in SRY causing sex-reversion in a 46,XY female. *J. Clin. Endocr. Metab.* 75: 331–333, 1992.

- [38637] 12189. Peters, R.; King, C.-Y.; Ukiyama, E.; Falsafi, S.; Donahoe, P.K.; Weiss, M. A.: An SRY mutation causing human sex reversal resolves a general mechanism of structure-specific DNA recognition: application to the four-way DNA junction. *Biochemistry* 34: 4569–4576, 1995.
- [38638] 12190. Schaffler, A.; Barth, N.; Winkler, K.; Zietz, B.; Rümmele, P.; Knuchel, R.; Scholmerich, J.; Palitzsch, K.-D.: Identification of a new missense mutation (gly95glu) in a highly conserved codon within the high-mobility group box of the sex-determining region Y gene: report on a 46,XY female with gonadal dysgenesis and yolk-sac tumor. *J. Clin. Endocr. Metab.* 85: 2287–2292, 2000.
- [38639] 12191. Schmitt-Ney, M.; Thiele, H.; Kaltwasser, P.; Bardoni, B.; Cisternino, M.; Scherer, G.: Two novel SRY missense mutations reducing DNA binding identified in XY females and their mosaic fathers. *Am. J. Hum. Genet.* 56: 862–869, 1995.
- [38640] 12192. Sinclair, A. H.; Berta, P.; Palmer, M. S.; Hawkins, R.; Griffiths, B. L.; Smith, M.; Foster, J. W.; Frischau, A.-M.; Lovell-Badge, R.; Goodfellow, P. N.: A gene from the hu-

man sex-determining region encodes a protein with homology to a conserved DNA-binding motif.. Nature 346:240–245, 1990.

- [38641] 12193.Su, H.; Lau, Y.-F. C.: Identification of the transcriptional unit, structural organization, and promoter sequence of the human sex-determining region Y (SRY) gene, using a reverse genetic approach. Am.J. Hum. Genet. 52: 24–38, 1993.
- [38642] 12194.Tucker, P. K.; Lundrigan, B. L.: Rapid evolution of the sex determining locus in Old World mice and rats. Nature 364: 715–717, 1993.
- [38643] 12195.Uehara, S.; Funato, T.; Yaegashi, N.; Suzuki, H.; Sato, J.; Sasaki, T.; Yajima, A.: SRY mutation and tumor formation on the gonads of XY pure gonadal dysgenesis patients. Cancer Genet. Cytogenet. 113:78–84, 1999.
- [38644] 12196.Uehara, S.; Hashiyada, M.; Sato, K.; Nata, M.; Funato, T.; Okamura, K.: Complete XY gonadal dysgenesis and aspects of the SRY genotype and gonadal tumor formation. J. Hum. Genet. 47: 279–284, 2002.
- [38645] 12197.Veitia, R.; Ion, A.; Barboux, S.; Jobling, M. A.; Souleyreau, N.; Ennis, K.; Ostrer, H.; Tosi, M.; Meo, T.; Chibani, J.; Fellous, M.; McElreavey, K.: Mutations and sequence variants in the testis-determining region of the Y

chromosome in individuals with a 46,XY female phenotype. Hum.Genet. 99: 648–652, 1997.

- [38646] 12198.Werner, M. H.; Huth, J. R.; Gronenborn, A. M.; Clore, G. M.:Molecular basis of human 46X,Y (sic) sex reversal revealed from thethree-dimensional solution structure of the human SRY–DNA complex. Cell 81:705–714, 1995.
- [38647] 12199.Whitfield, L. S.; Lovell–Badge, R.; Goodfellow, P. N.: Rapidsequence evolution of the mammalian sex–determining gene SRY. Nature 364:713–715, 1993.
- [38648] 12200.Zeng, Y.; Ren, Z.; Zhang, M.; Huang, Y.; Zeng, F.; Huang, S.:A new de novo mutation (A113T) in HMG box of the SRY gene leads toXY gonadal dysgenesis. J. Med. Genet. 30: 655–657, 1993.
- [38649] 12201.Zenteno, J. C.; Lopez, M.; Vera, C.; Mendez, J. P.; Kofman–Alfaro,S.: Two SRY–negative XX male brothers without genital ambiguity. Hum.Genet. 100: 606–610, 1997.
- [38650] 12202.Henkemeyer, M.; Orioli, D.; Henderson, J. T.; Saxton, T. M.; Roder,J.; Pawson, T.; Klein, R.: Nuk controls pathfinding of commissuralaxons in the mammalian central nervous system. Cell 86: 35–46, 1996.
- [38651] 12203.Ikegaki, N.; Tang, X. X.; Liu, X.–G.; Biegel, J. A.;

Allen, C.; Yoshioka, A.; Sulman, E. P.; Brodeur, G. M.; Pleasure, D. E.: Molecular characterization and chromosomal localization of DRT (EPHT3): a developmentally regulated human protein-tyrosine kinase gene of the EPH family. *Hum.Molec. Genet.* 4: 2033–2045, 1995.

[38652] 12204.Saito, T.; Seki, N.; Matsuda, Y.; Kitahara, M.; Murata, M.; Kanda, N.; Nomura, N.; Yamamoto, T.; Hori, T.: Identification of the human ERK gene as a putative receptor tyrosine kinase and its chromosomal localization to 1p36.1: a comparative mapping of human, mouse, and rat chromosomes. *Genomics* 26: 382–384, 1995.

[38653] 12205.Wybenga-Groot, L. E.; Baskin, B.; Ong, S. H.; Tong, J.; Pawson, T.; Sicheri, F.: Structural basis for autoinhibition of the EphB2 receptor tyrosine kinase by the unphosphorylated juxtamembrane region. *Cell* 106:745–757, 2001.

[38654] 12206.Johnson, D. F.; Hamon, M.; Fischel-Ghodsian, N.: Characterization of the human mitochondrial ribosomal S12 gene. *Genomics* 52: 363–368, 1998.

[38655] 12207.Shah, Z. H.; Migliosi, V.; Miller, S. C. M.; Wang, A.; Friedman, T. B.; Jacobs, H. T.: Chromosomal locations of three human nuclear genes (RPSM12, TUFM, and AFG3L1) specifying putative components of the mitochondrial gene expression apparatus. *Genomics* 48: 384–388, 1998.

- [38656] 12208.Honore, B.; Leffers, H.; Madsen, P.; Celis, J. E.: In-
terferon-gamma up-regulates a unique set of proteins in
human keratinocytes: molecular cloning and expression of
the cDNA encoding the RGD-sequence-containing protein
IGUP I5111. *Europ. J. Biochem.* 218: 421-430, 1993.
- [38657] 12209.Preckel, T.; Fung-Leung, W.-P.; Cai, Z.; Vitiello, A.;
Salter-Cid, L.; Winqvist, O.; Wolfe, T. G.; Von Herrath, M.;
Angulo, A.; Ghazal, P.; Lee, J.-D.; Fourie, A. M.; Wu, Y.;
Pang, J.; Ngo, K.; Peterson, P. A.; Fruh, K.; Yang, Y.: Im-
paired immunoproteasome assembly and immune re-
sponses in PA28 $-/-$ mice. *Science* 286: 2162-2165,
1999.
- [38658] 12210.Realini, C.; Dubiel, W.; Pratt, G.; Ferrell, K.; Rech-
steiner, M.: Molecular cloning and expression of a
gamma-interferon-inducible activator of the multicatalytic
protease. *J. Biol. Chem.* 269: 20727-20732, 1994.
- [38659] 12211.Trachtulec, Z.; Forejt, J.: Synteny of orthologous
genes conserved in mammals, snake, fly, nematode, and
fission yeast. *Mammalian Genome* 12:227-231, 2001.
- [38660] 12212.Reif, K.; Ekland, E. H.; Ohl, L.; Nakano, H.; Lipp, M.;
Forster, R.; Cyster, J. G.: Balanced responsiveness to
chemoattractants from adjacent zones determines B-cell
position. *Nature* 416: 94-99, 2002.

- [38661] 12213.Zhu, K.; Baudhuin, L. M.; Hong, G.; Williams, F. S.; Cristina,K. L.; Kabarowski, J. H. S.; Witte, O. N.; Xu, Y.: Sphingosylphosphorylcholineand lysophosphatidylcholine are ligands for the G protein–coupledreceptor GPR4. J. Biol. Chem. 276: 41325–41335, 2001.
- [38662] 12214.Vinik, B. S.; Kay, E. S.; Fiedorek, F. T., Jr.: Mapping of theMEK kinase gene (Mekk) to mouse chromosome 13 and human chromosome5. Mammalian Genome 6: 782–783, 1995.
- [38663] 12215.Yujiri, T.; Sather, S.; Fanger, G. R.; Johnson, G. L.: Role ofMEKK1 in cell survival and activation of JNK and ERK pathways definedby targeted gene disruption. Science 282: 1911–1914, 1998.
- [38664] 12216.Arriza, J. L.; Weinberger, C.; Cerelli, G.; Glaser, T. M.; Handelin,B. L.; Housman, D. E.; Evans, R. M.: Cloning of human mineralocorticoidreceptor complementary DNA: structural and functional kinship withthe glucocorticoid receptor. Science 237: 268–275, 1987.
- [38665] 12217.Berger, S.; Bleich, M.; Schmid, W.; Cole, T. J.; Peters, J.; Watanabe,H.; Kriz, W.; Warth, R.; Greger, R.; Schutz, G.: Mineralocorticoidreceptor knockout mice: pathophysiology of Na⁺ metabolism. Proc.Nat. Acad. Sci. 95: 9424–9429, 1998.

- [38666] 12218.Fan, Y.-S.; Eddy, R. L.; Byers, M. G.; Haley, L. L.; Henry, W.M.; Nowak, N. J.; Shows, T. B.: The human mineralocorticoid receptor gene (MLR) is located on chromosome 4 at q31.2. *Cytogenet. Cell Genet.* 52:83–84, 1989.
- [38667] 12219.Geller, D. S.; Farhi, A.; Pinkerton, N.; Fradley, M.; Moritz, M.; Spitzer, A.; Meinke, G.; Tsai, F. T. F.; Sigler, P. B.; Lifton, R.P.: Activating mineralocorticoid receptor mutation in hypertension exacerbated by pregnancy. *Science* 289: 119–123, 2000.
- [38668] 12220.Geller, D. S.; Rodriguez–Soriano, J.; Vallo Boado, A.; Schifter, S.; Bayer, M.; Chang, S. S.; Lifton, R. P.: Mutations in the mineralocorticoid receptor gene cause autosomal dominant pseudohypoaldosteronism type 1. *Nature Genet.* 19: 279–281, 1998.
- [38669] 12221.Hellal–Levy, C.; Fagart, J.; Souque, A.; Wurtz, J.-M.; Moras, D.; Rafestin–Oblin, M.-E.: Crucial role of the H11–H12 loop in stabilizing the active conformation of the human mineralocorticoid receptor. *Molec.Endocr.* 14: 1210–1221, 2000.
- [38670] 12222.Le Menuet, D.; Isnard, R.; Bichara, M.; Viengchareun, S.; Muffat–Joly, M.; Walker, F.; Zennaro, M.-C.; Lombes, M.: Alteration of cardiac and renal functions in transgenic mice overexpressing human mineralo-

corticoidreceptor. J. Biol. Chem. 276: 38911–38920, 2001.

[38671] 12223.Morrison, N.; Harrap, S. B.; Arriza, J. L.; Boyd, E.; Connor, J.M.: Regional chromosomal assignment of the human mineralocorticoidreceptor gene to 4q31.1. Hum. Genet. 85: 130–132, 1990.

[38672] 12224.Morrison, N.; Harrap, S. B.; Arriza, J. L.; Boyd, E.; Connor,J. M.: Regional chromosomal assignment of the human mineralocorticoidreceptor gene to 4q31.1. (Abstract) Cytogenet. Cell Genet. 51: 1048,1989.

[38673] 12225.Veenstra, G. J. C.; Weeks, D. L.; Wolffe, A. P.: Distinct rolesfor TBP and TBP–like factor in early embryonic gene transcriptionin Xenopus. Science 290: 2312–2314, 2000.

[38674] 12226.Zuhlke, C.; Hellenbroich, Y.; Dalski, A.; Kononowa, N.; Hagenah,J.; Vieregge, P.; Riess, O.; Klein, C.; Schwinger, E.: Differenttypes of repeat expansion in the TATA–binding protein gene are associatedwith a new form of inherited ataxia. Europ. J. Hum. Genet. 9: 160–164,2001.

[38675] 12227.Charest, A.; Wagner, J.; Muise, E. S.; Heng, H. H. Q.; Tremblay,M. L.: Structure of the murine MPTP–PEST gene: genomic organizationand chromosomal mapping. Ge–

nomics 28: 501–507, 1995.

- [38676] 12228. Takekawa, M.; Itoh, F.; Hinoda, Y.; Adachi, M.; Ariyama, T.; Inazawa, J.; Imai, K.; Yachi, A.: Chromosomal localization of the protein tyrosine phosphatase G1 gene and characterization of the aberrant transcripts in human colon cancer cells. *FEBS Lett.* 339: 222–228, 1994.
- [38677] 12229. Takekawa, M.; Itoh, F.; Hinoda, Y.; Arimura, Y.; Toyota, M.; Sekiya, M.; Adachi, M.; Imai, K.; Yachi, A.: Cloning and characterization of a human cDNA encoding a novel putative cytoplasmic protein-tyrosine-phosphatase. *Biochem. Biophys. Res. Commun.* 189: 1223–1230, 1992.
- [38678] 12230. Yang, Q.; Co, D.; Sommercorn, J.; Tonks, N. K.: Cloning and expression of PTP-PEST: a novel, human, non-transmembrane protein tyrosine phosphatase. *J. Biol. Chem.* 268: 6622–6628, 1993.
- [38679] 12231. Coopman, P. J. P.; Do, M. T. H.; Barth, M.; Bowden, E. T.; Hayes, A. J.; Basyuk, E.; Blancato, J. K.; Vezza, P. R.; McLeskey, S. W.; Mangeat, P. H.; Mueller, S. C.: The Syk tyrosine kinase suppresses malignant growth of human breast cancer cells. *Nature* 406: 742–747, 2000.
- [38680] 12232. Inatome, R.; Yanagi, S.; Takano, T.; Yamamura, H.: A critical role for Syk in endothelial cell proliferation and migration. *Biochem. Biophys. Res. Commun.* 286:

195–199, 2001.

- [38681] 12233.Mocsai, A.; Zhou, M.; Meng, F.; Tybulewicz, V. L.; Lowell, C. A.: Syk is required for integrin signaling in neutrophils. *Immunity* 16:547–558, 2002.
- [38682] 12234.Muller, B.; Cooper, L.; Terhorst, C.: Molecular cloning of the human homologue to the pig protein-tyrosine kinase syk. *Immunogenetics* 39:359–362, 1994.
- [38683] 12235.Taniguchi, T.; Kobayashi, T.; Kondo, J.; Takahashi, K.; Nakamura, H.; Suzuki, J.; Nagai, K.; Yamada, T.; Nakamura, S.; Yamamura, H.: Molecular cloning of a porcine gene syk that encodes a 72-kDa protein-tyrosine kinase showing high susceptibility to proteolysis. *J. Biol. Chem.* 266:15790–15796, 1991.
- [38684] 12236.Zioncheck, T. F.; Harrison, M. L.; Isaacson, C. C.; Geahlen, R.L.: Generation of an active protein-tyrosine kinase from lymphocytes by proteolysis. *J. Biol. Chem.* 263: 19195–19202, 1988.
- [38685] 12237.Satre, M. A.; Zgombic-Knight, M.; Duester, G.: The complete structure of human class IV alcohol dehydrogenase (retinol dehydrogenase) determined from the ADH7 gene. *J. Biol. Chem.* 269: 15606–15612, 1994.
- [38686] 12238.Yokoyama, H.; Baraona, E.; Lieber, C. S.: Molecular cloning and chromosomal localization of the ADH7 gene

encoding human class IV(sigma) ADH. Genomics 31: 243–245, 1996.

[38687] 12239.Zgombic–Knight, M.; Foglio, M. H.; Duester, G.: Genomic structure and expression of the ADH7 gene encoding human class IV alcohol dehydrogenase, the form most efficient for retinol metabolism in vitro. J. Biol.Chem. 270: 4305–4311, 1995.

[38688] 12240.Bashir, M. M.; Abrams, W. R.; Tucker, T.; Sellinger, B.; Budarf, M.; Emanuel, B.; Rosenbloom, J.: Molecular cloning and characterization of the bovine and human tuftelin genes. Connect. Tissue Res. 39:13–24, 1998.

[38689] 12241.Deutsch, D.: Structure and function of enamel gene products. Anat.Rec. 224: 189–210, 1989.

[38690] 12242.Beguin, P.; Nagashima, K.; Gono, T.; Shibasaki, T.; Takahashi, K.; Kashima, Y.; Ozaki, N.; Geering, K.; Iwanaga, T.; Seino, S.: Regulation of Ca(2+) channel expression at the cell surface by the small G-protein kir/Gem. Nature 411: 701–706, 2001.

[38691] 12243.Maguire, J.; Santoro, T.; Jensen, P.; Siebenlist, U.; Yewdell, J.; Kelly, K.: GEM: an induced, immediate early protein belonging to the Ras family. Science 265: 241–244, 1994.

[38692] 12244.Santoro, T.; Maguire, J.; McBride, O. W.; Avraham,

K. B.; Copeland, N. G.; Jenkins, N. A.; Kelly, K.: Chromosomal organization and transcriptional regulation of human GEM and localization of the human and mouse Gemloci encoding an inducible Ras-like protein. *Genomics* 30: 558–564, 1995.

[38693] 12245. Inoue, I.; Taniuchi, I.; Kitamura, D.; Jenkins, N. A.; Gilbert, D. J.; Copeland, N. G.; Watanabe, T.: Characteristics of the mouse genomic histamine H1 receptor gene. *Genomics* 36: 178–181, 1996.

[38694] 12246. Le Coniat, M.; Traiffort, E.; Ruat, M.; Arrang, J.-M.; Berger, R.: Chromosomal localization of the human histamine H1-receptor gene. *Hum. Genet.* 94: 186–188, 1994.

[38695] 12247. Ma, R. Z.; Gao, J.; Meeker, N. D.; Fillmore, P. D.; Tung, K. S. K.; Watanabe, T.; Zachary, J. F.; Offner, H.; Blankenhorn, E. P.; Teuscher, C.: Identification of Bphs, an autoimmune disease locus, as histamine receptor H-1. *Science* 297: 620–623, 2002.

[38696] 12248. Yamashita, M.; Fukui, H.; Sugama, K.; Horio, Y.; Ito, S.; Mizuguchi, H.; Wada, H.: Expression cloning of a cDNA encoding the bovine histamine H1 receptor. *Proc. Nat. Acad. Sci.* 88: 11515–11519, 1991.

[38697] 12249. Bauer, S.; Groh, V.; Wu, J.; Steinle, A.; Phillips, J. H.;

Lanier, L. L.; Spies, T.: Activation of NK cells and T cells by NKG2D, a receptor for stress-inducible MICA. *Science* 285: 727–729, 1999.

[38698] 12250. Diefenbach, A.; Jensen, E. R.; Jamieson, A. M.; Raulet, D. H.: Rae1 and H60 ligands of the NKG2D receptor stimulate tumour immunity. *Nature* 413: 165–171, 2001.

[38699] 12251. Girardi, M.; Oppenheim, D. E.; Steele, C. R.; Lewis, J. M.; Glusac, E.; Filler, R.; Hobby, P.; Sutton, B.; Tigelaar, R. E.; Hayday, A. C.: Regulation of cutaneous malignancy by gamma-delta T cells. *Science* 294: 605–609, 2001.

[38700] 12252. Groh, V.; Rhinehart, R.; Randolph-Habecker, J.; Topp, M. S.; Riddell, S. R.; Spies, T.: Costimulation of CD8-alpha-beta T cells by NKG2D via engagement by MIC induced on virus-infected cells. *Nature Immun.* 2: 255–260, 2001.

[38701] 12253. Groh, V.; Steinle, A.; Bauer, S.; Spies, T.: Recognition of stress-induced MHC molecules by intestinal epithelial gamma-delta T cells. *Science* 279: 1737–1740, 1998.

[38702] 12254. Groh, V.; Wu, J.; Yee, C.; Spies, T.: Tumour-derived soluble MIC ligands impair expression of NKG2D and T-cell activation. *Nature* 419: 734–738, 2002.

[38703] 12255. Li, P.; Morris, D. L.; Willcox, B. E.; Steinle, A.; Spies, T.; Strong, R. K.: Complex structure of the activating im-

munoreceptorNKG2D and its MHC class I-like ligand MICA. *Nature Immun.* 2: 443–451,2001.

- [38704] 12256.Jepsen, K. J.; Wu, F.; Peragallo, J. H.; Paul, J.; Roberts, L.;Ezura, Y.; Oldberg, A.; Birk, D. E.; Chakravarti, S.: A syndromeof joint laxity and impaired tendon integrity in lumican- and fibromodulin-deficientmice. *J. Biol. Chem.* 277: 35532–35540, 2002.
- [38705] 12257.Sztrolovics, R.; Chen, X.-N.; Grover, J.; Roughley, P. J.; Korenberg,J. R.: Localization of the human fibromodulin gene (FMOD) to chromosome1q32 and completion of the cDNA sequence. *Genomics* 23: 715–717,1994.
- [38706] 12258.Dalton, S.; Treisman, R.: Characterization of SAP-1, a proteinrecruited by serum response factor to the c-fos serum response element. *Cell* 68:597–612, 1992.
- [38707] 12259.Mo, Y.; Vaessen, B.; Johnston, K.; Marmorstein, R.: Structuresof SAP-1 bound to DNA targets from the E74 and c-fos promoters: insightsinto DNA sequence discrimination by Ets proteins. *Molec. Cell* 2:201–212, 1998.
- [38708] 12260.Shipley, J.; Sheer, D.; Dalton, S.; Treisman, R.; Patel, K.: Mappingof the human SAP1 (SRF accessory protein 1) gene and SAP2, a geneencoding a related protein, to chromosomal bands 1q32 and 12q23, respectively. *Genomics* 23:710–711, 1994.

- [38709] 12261.Giovane, A.; Pintzas, A.; Maira, S.-M.; Sobieszczuk, P.; Wasylyk,B.: Net, a new ets transcription factor that is activated by Ras. *GenesDev.* 8: 1502–1513, 1994.
- [38710] 12262.Lopez, M.; Oettgen, P.; Akbarali, Y.; Dendorfer, U.; Libermann,T. A.: ERP, a new member of the ets transcription factor/oncoproteinfamily: cloning, characterization, and differential expression duringB–lymphocyte development. *Molec. Cell. Biol.* 14: 3292–3309, 1994.
- [38711] 12263.Kelner, G. S.; Kennedy, J.; Bacon, K. B.; Kleyensteuber, S.; Largaespada,D. A.; Jenkins, N. A.; Copeland, N. G.; Bazan, J. F.; Moore, K. W.;Schall, T. J.; Zlotnik, A.: Lymphotactin: a cytokine that representsa new class of chemokine. *Science* 266: 1395–1399, 1994.
- [38712] 12264.Kennedy, J.; Kelner, G. S.; Kleyensteuber, S.; Schall, T. J.; Weiss,M. C.; Yssel, H.; Schneider, P. V.; Cocks, B. G.; Bacon, K. B.; Zlotnik,A.: Molecular cloning and functional characterization of human lymphotactin. *J.Immun.* 155: 203–209, 1995.
- [38713] 12265.Muller, S.; Dorner, B.; Korthauer, U.; Mages, H. W.; D'Apuzzo,M.; Senger, G.; Kroczeck, R. A.: Cloning of ATAC, an activation–induced,chemokine–related molecule exclusively expressed in CD8+ T lymphocytes. *Europ.J. Immun.* 25: 1744–1748, 1995.

- [38714] 12266.Yoshida, T.; Imai, T.; Kakizaki, M.; Nishimura, M.; Takagi, S.;Yoshie, O.: Identification of single C motif-1/lymphotactin receptorXCR1. J. Biol. Chem. 273: 16551-16554, 1998.
- [38715] 12267.Yoshida, T.; Imai, T.; Kakizaki, M.; Nishimura, M.; Yoshie, O.: Molecular cloning of a novel C or gamma type chemokine, SCM-1. FEBSLett. 360: 155-159, 1995.
- [38716] 12268.Yoshida, T.; Imai, T.; Takagi, S.; Nishimura, M.; Ishikawa, I.;Yaoi, T.; Yoshie, O.: Structure and expression of two highly relatedgenes encoding SCM-1/human lymphotactin. FEBS Lett. 395: 82-88,1996.
- [38717] 12269.Ikeuchi, T.; Asaka, T.; Saito, M.; Tanaka, H.; Higuchi, S.; Tanaka,K.; Saida, K.; Uyama, E.; Mizusawa, H.; Fukuhara, N.; Nonaka, I.;Takamori, M.; Tsuji, S.: Gene locus for autosomal recessive distalmyopathy with rimmed vacuoles maps to chromosome 9. Ann. Neurol. 41:432-437, 1997.
- [38718] 12270.Erickson, J. D.; Varoqui, H.; Schafer, M. K.-H.; Modi, W.; Diebler,M.-F.; Weihe, E.; Rand, J.; Eiden, L. E.; Bonner, T. I.; Usdin, T.B.: Functional identification of a vesicular acetylcholine transporterand its expression from a 'cholinergic' gene locus. J. Biol. Chem. 269:21929-21932, 1994.
- [38719] 12271.Tanahashi, H.; Tabira, T.: Alzheimer's disease-

associated presenilin2 interacts with DRAL, an LIM-domain protein. Hum. Molec. Genet. 9:2281–2289, 2000.

- [38720] 12272. Erkinheimo, T.-L.; Saukkonen, K.; Narko, K.; Jalkanen, J.; Ylikorkala, O.; Ristimäki, A.: Expression of cyclooxygenase-2 and prostanoid receptors by human myometrium. J. Clin. Endocr. Metab. 85: 3468–3475, 2000.
- [38721] 12273. Hla, T.; Neilson, K.: Human cyclooxygenase-2 cDNA. Proc. Nat. Acad. Sci. 89: 7384–7388, 1992.
- [38722] 12274. Jones, D. A.; Carlton, D. P.; McIntyre, T. M.; Zimmerman, G. A.; Prescott, S. M.: Molecular cloning of human prostaglandin endoperoxidase synthase type II and demonstration of expression in response to cytokines. J. Biol. Chem. 268: 9049–9054, 1993.
- [38723] 12275. Kraemer, S. A.; Meade, E. A.; DeWitt, D. L.: Prostaglandin endoperoxidase synthase gene structure: identification of the transcriptional start site and 5-prime-flanking regulatory sequences. Arch. Biochem. Biophys. 293: 391–400, 1992.
- [38724] 12276. Lassus, P.; Wolff, H.; Andersson, S.: Cyclooxygenase-2 in human perinatal lung. Pediatr. Res. 47: 602–605, 2000.
- [38725] 12277. Lim, H.; Paria, B. C.; Das, S. K.; Dinchuk, J. E.; Langenbach, R.; Trzaskos, J. M.; Dey, S. K.: Multiple female re-

productive failures in cyclooxygenase 2-deficient mice.
Cell 91: 197–208, 1997.

- [38726] 12278. Jacquemin, P.; Durviaux, S. M.; Jensen, J.; Godfraind, C.; Gradwohl, G.; Guillemot, F.; Madsen, O. D.; Carmeliet, P.; Dewerchin, M.; Collen, D.; Rousseau, G. G.; Lemaigre, F. P.: Transcription factor hepatocyte nuclear factor 6 regulates pancreatic endocrine cell differentiation and controls expression of the proendocrine gene *ngn3*. *Molec. Cell Biol.* 20: 4445–4454, 2000.
- [38727] 12279. Jacquemin, P.; Lannoy, V. J.; Rousseau, G. G.; Lemaigre, F. P.: OC-2, a novel mammalian member of the ONECUT class of homeodomain transcription factors whose function in liver partially overlaps with that of hepatocyte nuclear factor-6. *J. Biol. Chem.* 274: 2665–2671, 1999.
- [38728] 12280. Lemaigre, F. P.; Durviaux, S. M.; Truong, O.; Lannoy, V. J.; Hsuan, J. J.; Rousseau, G. G.: Hepatocyte nuclear factor 6, a transcription factor that contains a novel type of homeodomain and a single cut domain. *Proc. Nat. Acad. Sci.* 93: 9460–9464, 1996.
- [38729] 12281. Pierreux, C. E.; Stafford, J.; Demonte, D.; Scott, D. K.; Vandenhoute, J.; O'Brien, R. M.; Granner, D. K.; Rousseau, G. G.; Lemaigre, F. P.: Antigluocorticoid activity

of hepatocyte nuclear factor-6. *Proc.Nat. Acad. Sci.* 96: 8961–8966, 1999.

- [38730] 12282.Samadani, U.; Costa, R. H.: The transcriptional activator hepatocytenuclear factor 6 regulates liver gene expression. *Molec. Cell. Biol.* 16:6273–6284, 1996.
- [38731] 12283.Vaisse, C.; Kim, J.; Espinosa, R., III; Le Beau, M. M.; Stoffel,M.: Pancreatic islet expression studies and polymorphic DNA markersin the genes encoding hepatocyte nuclear factor-3-alpha, -3-beta,-4-gamma, and -6. *Diabetes* 46: 1364–1367, 1997.
- [38732] 12284.Iacobazzi, V.; Palmieri, F.; Runswick, M. J.; Walker, J. E.: Sequencesof the human and bovine genes for the mitochondrial 2-oxoglutaratecarrier. *DNA Seq.* 3: 79–88, 1992.
- [38733] 12285.Piccininni, S.; Iacobazzi, V.; Lauria, G.; Rocchi, M.; Palmieri,F.: Assignment of the oxoglutarate carrier gene (SLC20A4) to humanchromosome 17p13.3. *Cytogenet. Cell Genet.* 83: 256–257, 1998.
- [38734] 12286.Abbott, G. W.; Sesti, F.; Splawski, I.; Buck, M. E.; Lehmann, M.H.; Timothy, K. W.; Keating, M. T.; Goldstein, S. A. N.: MiRP1 formsI(Kr) potassium channels with HERG and is associated with cardiacarrhythmia. *Cell* 97: 175–187, 1999.

- [38735] 12287.Schroeder, B. C.; Waldegger, S.; Fehr, S.; Bleich, M.; Warth, R.; Greger, R.; Jentsch, T. J.: A constitutively open potassium channel formed by KCNQ1 and KCNE3. *Nature* 403: 196–199, 2000.
- [38736] 12288.Harvey, T. J.; Hooper, J. D.; Myers, S. A.; Stephenson, S.-A.; Ashworth, L. K.; Clements, J. A.: Tissue-specific expression patterns and fine mapping of the human kallikrein (KLK) locus on proximal 19q13.4. *J.Biol. Chem.* 275: 37397–37406, 2000.
- [38737] 12289.Chaudhry, F. A.; Reimer, R. J.; Krizaj, D.; Barber, D.; Storm-Mathisen, J.; Copenhagen, D. R.; Edwards, R. H.: Molecular analysis of system N suggests novel physiological roles in nitrogen metabolism and synaptic transmission. *Cell* 99: 769–780, 1999.
- [38738] 12290.Yamada, K.; Nishida, K.; Hibi, M.; Hirano, T.; Matsuda, Y.: Comparative FISH mapping of Gab1 and Gab2 genes in human, mouse and rat. *Cytogenet.Cell Genet.* 94: 39–42, 2001.
- [38739] 12291.Afonina, E.; Stauber, R.; Pavlakis, G. N.: The human poly(A)-binding protein 1 shuttles between the nucleus and the cytoplasm. *J. Biol.Chem.* 273: 13015–13021, 1998.
- [38740] 12292.Deo, R. C.; Bonanno, J. B.; Sonenberg, N.; Burley, S.

K.: Recognition of polyadenylate RNA by the poly(A)-binding protein. *Cell* 98: 835–845, 1999.

[38741] 12293. Gorlach, M.; Burd, C. G.; Dreyfuss, G.: The mRNA poly(A)-binding protein: localization, abundance, and RNA-binding specificity. *Exp. Cell Res.* 211: 400–407, 1994.

[38742] 12294. Grange, T.; Martins de Sa, C.; Oddos, J.; Pictet, R.: Human mRNA polyadenylate binding protein: evolutionary conservation of a nucleic acid binding motif. *Nucleic Acids Res.* 15: 4771–4787, 1987.

[38743] 12295. Danpure, C. J.; Purdue, P. E.; Fryer, P.; Griffiths, S.; Allsop, J.; Lumb, M. J.; Guttridge, K. M.; Jennings, P. R.; Scheinman, J. I.; Mauer, S. M.; Davidson, N. O.: Enzymological and mutational analysis of a complex primary hyperoxaluria type I phenotype involving alanine: glyoxylate aminotransferase peroxisome-to-mitochondrion mistargeting and intra-peroxisomal aggregation. *Am. J. Hum. Genet.* 53: 417–432, 1993.

[38744] 12296. Mori, M.; Oda, T.; Nishiyama, K.; Serikawa, T.; Yamada, J.; Ichiyama, A.: A single serine: pyruvate aminotransferase gene on rat chromosome 9q34–q36. *Genomics* 13: 686–689, 1992.

[38745] 12297. Purdue, P. E.; Allsop, J.; Isaya, G.; Rosenberg, L. E.;

Danpure, C. J.: Mistargeting of peroxisomal L-alanine:glyoxylate aminotransferase to mitochondria in primary hyperoxaluria patients depends upon activation of a cryptic mitochondrial targeting sequence by a point mutation. *Proc. Nat. Acad. Sci.* 88: 10900–10904, 1991.

[38746] 12298. Purdue, P. E.; Lumb, M. J.; Allsop, J.; Minatogawa, Y.; Danpure, C. J.: A glycine-to-glutamate substitution abolishes alanine:glyoxylate aminotransferase catalytic activity in a subset of patients with primary hyperoxaluria type 1. *Genomics* 13: 215–218, 1992.

[38747] 12299. Purdue, P. E.; Takada, Y.; Danpure, C. J.: Identification of mutations associated with peroxisome-to-mitochondrion mistargeting of alanine/glyoxylate aminotransferase in primary hyperoxaluria type 1. *J. Cell Biol.* 111: 2341–2351, 1990.

[38748] 12300. Takada, Y.; Kaneko, N.; Esumi, H.; Purdue, P. E.; Danpure, C. J.: Human peroxisomal L-alanine:glyoxylate aminotransferase: evolutionary loss of a mitochondrial targeting signal by point mutation of the initiation codon. *Biochem. J.* 268: 517–520, 1990.

- [38749] 12301.von Schnakenburg, C.; Rumsby, G.: Primary hyperoxaluria type1: a cluster of new mutations in exon 7 of the AGXT gene. *J. Med.Genet.* 34: 489–492, 1997.
- [38750] 12302.Kobayashi, H.; Hino, M.; Shimodahira, M.; Iwakura, T.; Ishihara,T.; Ikekubo, K.; Ogawa, Y.; Nakao, K.; Kurahachi, H.: Missense mutationof TRPS1 in a family of tricho–rhino–phalangeal syndrome type III. *Am.J. Med. Genet.* 107: 26–29, 2002.
- [38751] 12303.Lipkin, S. M.; Moens, P. B.; Wang, V.; Lenzi, M.; Shanmugarajah,D.; Gilgeous, A.; Thomas, J.; Cheng, J.; Touchman, J. W.; Green, E.D.; Schwartzberg, P.; Collins, F. S.; Cohen, P. E.: Meiotic arrestand aneuploidy in MLH3–deficient mice. *Nature Genet.* 31: 385–390,2002.
- [38752] 12304.Lipkin, S. M.; Wang, V.; Jacoby, R.; Banerjee–Basu, S.; Baxevanis,A. D.; Lynch, H. T.; Elliott, R. M.; Collins, F. S.: MLH3: a DNAmismatch repair gene associated with mammalian microsatellite instability. *NatureGenet.* 24: 27–35, 2000.
- [38753] 12305.Lipkin, S. M.; Wang, V.; Stoler, D. L.; Anderson, G. R.; Kirsch,I.; Hadley, D.; Lynch, H. T.; Collins, F. S.: Germline and somaticmutation analyses in the DNA mismatch repair gene MLH3: evidence forsomatic mutation in colorectal cancers. *Hum. Mutat.* 17: 389–396,2001.

- [38754] 12306.Wu, Y.; Berends, M. J. W.; Sijmons, R. H.; Mensink, R. G. J.; Verlind,E.; Kooi, K. A.; van der Sluis, T.; Kempinga, C.; van der Zee, A.G. J.; Hollema, H.; Buys, C. H. C. M.; Kleibeuker, J. H.; Hofstra,R. M. W.: A role for MLH3 in hereditary nonpolyposis colorectal cancer. *NatureGenet.* 29: 137–138, 2001.
- [38755] 12307.Sahara, S.; Aoto, M.; Eguchi, Y.; Imamoto, N.; Yoneda, Y.; Tsujimoto,Y.: Acinus is a caspase–3–activated protein required for apoptoticchromatin condensation. *Nature* 401: 168–173, 1999.
- [38756] 12308.Hata, M.; Ohtsuka, K.: Characterization of HSE sequences in humanHsp40 gene: structural and promoter analysis. *Biochim. Biophys. Acta* 1397:43–55, 1998.
- [38757] 12309.Hata, M.; Okumura, K.; Seto, M.; Ohtsuka, K.: Genomic cloningof a human heat shock protein 40 (Hsp40) gene (HSPF1) and its chromosomallocalization to 19p13.2. *Genomics* 38: 446–449, 1996.
- [38758] 12310.Ohtsuka, K.: Cloning of a cDNA for heat–shock protein hsp40, ahuman homologue of bacterial DnaJ. *Biochem. Biophys. Res. Commun.* 197:235–240, 1993.
- [38759] 12311.Janitz, K.; Wild, A.; Beck, S.; Savasta, S.; Beluffi, G.; Ziegler,A.; Volz, A.: Genomic organization of the HSET locus and the possibleassociation of HLA–linked genes with

immotile cilia syndrome (ICS). Immunogenetics
49:644–652, 1999.

- [38760] 12312. Muravenko, O. V.; Gizatullin, R. Z.; Protopopov, A. I.; Kashuba, V. I.; Zabarovsky, E. R.; Zelenin, A. V.: Assignment of CDK5R2 coding for the cyclin-dependent kinase 5, regulatory subunit 2 (NCK5A1 protein) to human chromosome band 2q35 by fluorescent in situ hybridization. Cytogenet. Cell Genet. 89: 160–161, 2000.
- [38761] 12313. Nilden, F.; Backstrom, A.; Bark, C.: Molecular cloning and characterisation of a mouse gene encoding an isoform of the neuronal cyclin-dependent kinase 5 (CDK5) activator. Biochim. Biophys. Acta 1398: 371–376, 1998.
- [38762] 12314. Tang, D.; Yeung, J.; Lee, K-Y.; Matsushita, M.; Matsui, H.; Tomizawa, K.; Hatase, O.; Wang, J. H.: An isoform of the neuronal cyclin-dependent kinase 5 (Cdk5) activator. J. Biol. Chem. 270: 26897–26903, 1995.
- [38763] 12315. DuPont, B. R.; Hu, C.-C.; Reveles, X.; Simmer, J. P.: Assignment of serine protease 17 (PRSS17) to human chromosome bands 19q13.3–q13.4 by in situ hybridization. Cytogenet. Cell Genet. 86: 212–213, 1999.
- [38764] 12316. Nelson, P. S.; Gan, L.; Ferguson, C.; Moss, P.; Gelinas, R.; Hood, L.; Wang, K.: Molecular cloning and characterization of prostase, an androgen-regulated serine pro-

tease with prostate-restricted expression. Proc.Nat. Acad. Sci. 96: 3114–3119, 1999.

[38765] 12317.Hallas, C.; Pekarsky, Y.; Itoyama, T.; Varnum, J.; Bichi, R.; Rothstein, J. L.; Croce, C. M.: Genomic analysis of human and mouse TCL1 locireveals a complex of tightly clustered genes. Proc. Nat. Acad. Sci. 96:14418–14423, 1999.

[38766] 12318.Pekarsky, Y.; Hallas, C.; Isobe, M.; Russo, G.; Croce, C. M.:Abnormalities at 14q32.1 in T cell malignancies involve two oncogenes. Proc.Nat. Acad. Sci. 96: 2949–2951, 1999.

[38767] 12319.Koga, Y.; Fabrizi, G. M.; Mita, S.; Arnaudo, E.; Lomax, M. I.;Aqua, M. S.; Grossman, L. I.; Schon, E. A.: Sequence of a cDNA specifying subunit VIIc of human cytochrome c oxidase. Nucleic Acids Res. 18:684 only, 1990.

[38768] 12320.Chaudhary, P. M.; Ferguson, C.; Nguyen, V.; Nguyen, O.; Massa, H. F.; Eby, M.; Jasmin, A.; Trask, B. J.; Hood, L.; Nelson, P. S.: Cloning and characterization of two Toll/interleukin–1 receptor–like genes TIL3 and TIL4: evidence for a multi–gene receptor family in humans. Blood 91: 4020–4027, 1998.

[38769] 12321.Smith, M.; Wasmuth, J.; McPherson, J. D.; Wagner,

C.; Grandy, D.; Civelli, O.; Potkin, S.; Litt, M.: Cosegregation of an 11q22.3–9p22 translocation with affective disorder: proximity of the dopamine D2 receptor gene relative to the translocation breakpoint. (Abstract) Am.J. Hum. Genet. 45 (suppl.): A220 only, 1989.

- [38770] 12322. Abuladze, N.; Lee, I.; Newman, D.; Hwang, J.; Boorer, K.; Pushkin, A.; Kurtz, I.: Molecular cloning, chromosomal localization, tissue distribution, and functional expression of the human pancreatic sodium bicarbonate cotransporter. J. Biol. Chem. 273: 17689–17695, 1998.
- [38771] 12323. Burnham, C. E.; Amlal, H.; Wang, Z.; Shull, G. E.; Soleimani, M.: Cloning and functional expression of a human kidney $\text{Na}^+:\text{HCO}_3^-$ cotransporter. J. Biol. Chem. 272: 19111–19114, 1997.
- [38772] 12324. Choi, I.; Romero, M. F.; Khandoudi, N.; Bril, A.; Boron, W. F.: Cloning and characterization of a human electrogenic $\text{Na}(+)\text{--HCO}_3(-)$ cotransporter isoform (hhNBC). Am. J. Physiol. 276: C576–C584, 1999.
- [38773] 12325. Igarashi, T.; Inatomi, J.; Sekine, T.; Cha, S. H.; Kanai, Y.; Kunimi, M.; Tsukamoto, K.; Satoh, H.; Shimadzu, M.; Tozawa, F.; Mori, T.; Shiobara, M.; Seki, G.; Endou, H.: Mutations in SLC4A4 cause permanent isolated proximal renal tubular acidosis with ocular abnormalities. (Letter)

Nature Genet. 23: 264–265, 1999.

- [38774] 12326. Romero, M. F.; Boron, W. F.: Electrogenic Na(+)/HCO(3-) cotransporters: cloning and physiology. Annu. Rev. Physiol. 61: 699–723, 1999.
- [38775] 12327. Soleimani, M.; Burnham, C. E.: Physiologic and molecular aspects of the Na(+):HCO(3-) cotransporter in health and disease processes. Kidney Int. 57: 371–384, 2000.
- [38776] 12328. Usui, T.; et al.; et al.: Pflugers Arch. 438: 458–462, 1999.
- [38777] 12329. Zhou, Y.-D.; Barnard, M.; Tian, H.; Li, X.; Ring, H. Z.; Francke, U.; Shelton, J.; Richardson, J.; Russell, D. W.; McKnight, S. L.: Molecular characterization of two mammalian bHLH-PAS domain proteins selectively expressed in the central nervous system. Proc. Nat. Acad. Sci. 94: 713–718, 1997.
- [38778] 12330. Garcia, J. A.; Zhang, D.; Estill, S. J.; Michnoff, C.; Rutter, J.; Reick, M.; Scott, K.; Diaz-Arrastia, R.; McKnight, S. L.: Impaired cued and contextual memory in NPAS2-deficient mice. Science 288: 2226–2230, 2000.
- [38779] 12331. Spencer, E.; Jiang, J.; Chen, Z. J.: Signal-induced ubiquitination of I-kappa-B-alpha by the F-box protein Slimb/beta-TrCP. Genes Dev. 13: 284–294, 1999.

- [38780] 12332.Yaron, A.; Hatzubai, A.; Davis, M.; Lavon, I.; Amit, S.; Manning,A. M.; Andersen, J. S.; Mann, M.; Mercurio, F.; Ben-Neriah, Y.: Identificationof the receptor component of the I-kappa-B-alpha-ubiquitin ligase. *Nature* 396:590–594, 1998.
- [38781] 12333.Lucas, C. A.; Rughani, A.; Hoh, J. F. Y.: Expression of extraocularmyosin heavy chain in rabbit laryngeal muscle. *J. Muscle Res. CellMotil.* 16: 368–378, 1995.
- [38782] 12334.Wieczorek, D. F.; Periasamy, M.; Butler-Browne, G. S.; Whalen,R. G.; Nadal-Ginard, B.: Co-expression of multiple myosin heavy chaingenesis, in addition to a tissue-specific one, in extraocular musculature. *J.Cell Biol.* 101: 618–629, 1985.
- [38783] 12335.Winters, L. M.; Briggs, M. M.; Schachet, F.: The human extraocularmuscle myosin heavy chain gene (MYH13) maps to the cluster of fastand developmental myosin genes on chromosome 17. *Genomics* 54: 188–189,1998.
- [38784] 12336.Brisken, C.; Heineman, A.; Chavarria, T.; Elenbaas, B.; Tan, J.;Dey, S. K.; McMahon, J. A.; McMahon, A. P.; Weinberg, R. A.: Essentialfunction of Wnt-4 in mammary gland development downstream of progesteronesignaling. *Genes Dev.* 14: 650–654, 2000.
- [38785] 12337.Jordan, B. K.; Mohammed, M.; Ching, S. T.; Delot,

E.; Chen, X.-N.; Dewing, P.; Swain, A.; Rao, P. N.; Elejalde, B. R.; Vilain, E.: Up-regulation of WNT-4 signaling and dosage-sensitive sex reversal in humans. *Am.J. Hum. Genet.* 68: 1102-1109, 2001.

[38786] 12338. Stark, K.; Vainio, S.; Vassileva, G.; McMahon, A. P.: Epithelial transformation of metanephric mesenchyme in the developing kidney regulated by Wnt-4. *Nature* 372: 679-683, 1994.

[38787] 12339. Vainio, S.; Heikkila, M.; Kispert, A.; Chin, N.; McMahon, A. P.: Female development in mammals is regulated by Wnt-4 signalling. *Nature* 397:405-409, 1999.

[38788] 12340. Vilain, A.; Apiou, F.; Dutrillaux, B.; Malfoy, B.: Assignment of candidate DNA methyltransferase gene (DNMT2) to human chromosome band 10p15.1 by in situ hybridization. *Cytogenet. Cell Genet.* 82:120 only, 1998.

[38789] 12341. Yoder, J. A.; Bestor, T. H.: A candidate mammalian DNA methyltransferase related to pmt1p of fission yeast. *Hum. Molec. Genet.* 7: 279-284, 1998.

[38790] 12342. Avraham, K. B.; Cho, B. C.; Gilbert, D.; Fujii, H.; Okamoto, K.; Shimazaki, T.; Ito, T.; Shoji, H.; Wakamatsu, Y.; Kondoh, H.; Takahashi, N.; Muramatsu, M.; Hamada, H.; Copeland, N. G.; Jenkins, N. A.: Murine chromosomal location of four class III POU transcription factors. *Genomics*

18:131–133, 1993.

- [38791] 12343.Egan, S.; Herbrick, J.-A.; Tsui, L.-C.; Cohen, B.; Flock, G.; Beatty, B.; Scherer, S. W.: Mapping of the human lunatic fringe (LFNG) gene to 7p22 and manic fringe (MFNG) to 22q12. *Genomics* 54: 576–577, 1998.
- [38792] 12344.Johnston, S. H.; Rauskolb, C.; Wilson, R.; Prabhakaran, B.; Irvine, K. D.; Vogt, T. F.: A family of mammalian Fringe genes implicated in boundary determination and the Notch pathway. *Development* 124:2245–2254, 1997.
- [38793] 12345.Moran, J. L.; Johnston, S. H.; Rauskolb, C.; Bhalerao, J.; Bowcock, A. M.; Vogt, T. F.: Genomic structure, mapping, and expression analysis of the mammalian lunatic, manic, and radical fringe genes. *Mammalian Genome* 10: 535–541, 1999.
- [38794] 12346.Van Tine, B. A.; Knops, J.; Shaw, G. M.; May, W. A.: Assignment of human MFNG, manic fringe *Drosophila* homolog, to 22q13.1 using tyramide fluorescence in situ hybridization (T-FISH). *Cytogenet. Cell Genet.* 87:132–133, 1999.
- [38795] 12347.Laufer, E.; Dahn, R.; Orozco, O. E.; Yeo, C.-Y.; Pisenti, J.; Henrique, D.; Abbott, U. K.; Fallon, J. F.; Tabin, C.: Expression of Radical fringe in limb-bud ectoderm reg-

ulates apical ectodermal ridge formation. *Nature* 386:366–373, 1997.

[38796] 12348. Rodriguez–Esteban, C.; Schwabe, J. W. R.; De La Pena, J.; Foys, B.; Eshelman, B.; Izpisua Belmonte, J. C.: Radical fringe position the apical ectodermal ridge at the dorsoventral boundary of the vertebrate limb. *Nature* 386: 360–366, 1997.

[38797] 12349. Hofmann, Y.; Becker, J.; Wright, F.; Avner, E. D.; Mrug, M.; Guay–Woodford, L. M.; Somlo, S.; Zerres, K.; Germino, G. G.; Onuchic, L. F.: Genomic structure of the gene for the human P1 protein (MCM3) and its exclusion as a candidate for autosomal recessive polycystic kidney disease. *Europ. J. Hum. Genet.* 8: 163–166, 2000.

[38798] 12350. Starborg, M.; Brundell, E.; Gell, K.; Larsson, C.; White, I.; Daneholt, B.; Hoog, C.: A murine replication protein accumulates temporarily in the heterochromatic regions of nuclei prior to initiation of DNA replication. *J. Cell Sci.* 108: 927–934, 1995.

[38799] 12351. Budde, S. M. S.; van den Heuvel, L. P. W. J.; Janssen, A. J.; Smeets, R. J. P.; Buskens, C. A. F.; De Meirleir, L.; Van Coster, R.; Baethmann, M.; Voit, T.; Trijbels, J. M. F.; Smeitink, J. A. M.: Combined enzymatic complex I and III deficiency associated with mutations in the nuclear en-

coded NDUF54 gene. Biochem. Biophys. Res. Commun. 275: 63–68,2000.

- [38800] 12352.Papa, S.; Sardanelli, A. M.; Cocco, T.; Speranza, F.; Scacco, S.C.; Technikova–Dobrova, Z.: The nuclear encoded–18 kDa (IP) AQDQsubunit of bovine heart complex I is phosphorylated by the mitochondrialcAMP–dependent protein kinase. FEBS Lett. 379: 299–301, 1996.
- [38801] 12353.Petruzzella, V.; Vergari, R.; Puzziferri, I.; Boffoli, D.; Lamantea,E.; Zeviani, M.; Papa, S.: A nonsense mutation in the NDUF54 geneencoding the 18 kDa (AQDQ) subunit of complex I abolishes assemblyand activity of the complex in a patient with Leigh–like syndrome. Hum.Molec. Genet. 10: 529–535, 2001.
- [38802] 12354.van den Heuvel, L.; Ruitenbeek, W.; Smeets, R.; Gelman–Kohan, Z.;Elpeleg, O.; Loeffen, J.; Trijbels, F.; Mariman, E.; de Bruijn, D.;Smeitink, J.: Demonstration of a new pathogenic mutation in humancomplex I deficiency: a 5–bp duplication in the nuclear gene encodingthe 18–kD (AQDQ) subunit. Am. J. Hum. Genet. 62: 262–268, 1998.
- [38803] 12355.Walker, J. E.; Arizmendi, J. M.; Dupuis, A.; Fearnley, I. M.; Finel,M.; Medd, S. M.; Pilkington, S. J.; Runswick, M. J.; Skehel, J. M.: Sequences of 20 subunits of NADH:ubiquinone oxidoreductase frombovine heart mito–

chondria: application of a novel strategy for sequencing–
proteins using the polymerase chain reaction. *J. Molec.*
Biol. 226:1051–1072, 1992.

- [38804] 12356. Chicheportiche, Y.; Bourdon, P. R.; Xu, H.; Hsu, Y.–M.; Scott, H.; Hession, C.; Garcia, I.; Browning, J. L.: TWEAK, a new secreted ligand in the tumor necrosis factor family that weakly induces apoptosis. *J. Biol. Chem.* 272: 32401–32410, 1997.
- [38805] 12357. Kaptein, A.; Jansen, M.; Dilaver, G.; Kitson, J.; Dash, L.; Wang, E.; Owen, M. J.; Bodmer, J.–L.; Tschopp, J.; Farrow, S. N.: Studies on the interaction between TWEAK and the death receptor WSL–1/TRAMP(DR3). *FEBS Lett.* 485: 135–141, 2000.
- [38806] 12358. Marsters, S. A.; Sheridan, J. P.; Pitti, R. M.; Brush, J.; Goddard, A.; Ashkenazi, A.: Identification of a ligand for the death–domain–containing receptor Apo3. *Curr. Biol.* 8: 525–528, 1998.
- [38807] 12359. Schneider, P.; Schwenzer, R.; Haas, E.; Muhlenbeck, F.; Schubert, G.; Scheurich, P.; Tschopp, J.; Wajant, H.: TWEAK can induce cell death via endogenous TNF and TNF receptor 1. *Europ. J. Immun.* 29:1785–1792, 1999.
- [38808] 12360. Wiley, S. R.; Cassiano, L.; Lofton, T.; Davis–Smith, T.; Winkles, J. A.; Lindner, V.; Liu, H.; Daniel, T. O.; Smith,

C. A.; Fanslow, W. C.: A novel TNF receptor family member binds TWEAK and is implicated in angiogenesis. *Immunity* 15: 837–846, 2001.

[38809] 12361. Hoey, T.; Sun, Y.-L.; Williamson, K.; Xu, X.: Isolation of two new members of the NF-AT gene family and functional characterization of the NF-AT proteins. *Immunity* 2: 461–472, 1995.

[38810] 12362. Hoey, T.; Sun, Y. L.; Williamson, K.; Xu, X.: Isolation of two new members of the NF-AT gene family and functional characterization of the NF-AT proteins. *Immunity* 2: 461–472, 1995.

[38811] 12363. Arany, Z.; Newsome, D.; Oldread, E.; Livingston, D. M.; Eckner, R.: A family of transcriptional adaptor proteins targeted by the E1A oncoprotein. *Nature* 374: 81–84, 1995.

[38812] 12364. de Lecea, L.; Ruiz-Lozano, P.; Danielson, P. E.; Peelle-Kirley, J.; Foye, P. E.; Frankel, W. N.; Sutcliffe, J. G.: Cloning, mRNA expression, and chromosomal mapping of mouse and human prepro cortistatin. *Genomics* 42: 499–506, 1997.

[38813] 12365. Ejleskar, K.; Abel, F.; Sjöberg, R.-M.; Backström, J.; Kogner, P.; Martinsson, T.: Fine mapping of the human prepro cortistatin gene (CORT) to neuroblastoma consensus

deletion region 1p36.3–p36.2, but absence of mutations in primary tumors. *Cytogenet. Cell Genet.* 89:62–66, 2000.

- [38814] 12366. Fukusumi, S.; Kitada, C.; Takekawa, S.; Kizawa, H.; Sakamoto, J.; Miyamoto, M.; Hinuma, S.; Kitano, K.; Fujino, M.: Identification and characterization of a novel human cortistatin-like peptide. *Biochem. Biophys. Res. Commun.* 232: 157–163, 1997.
- [38815] 12367. Dong, C.; Zhu, S.; Wang, T.; Yoon, W.; Li, Z.; Alvarez, R. J.; ten Dijke, P.; White, B.; Wigley, F. M.; Goldschmidt-Clermont, P. J.: Deficient Smad7 expression: a putative molecular defect in scleroderma. *Proc. Nat. Acad. Sci.* 99: 3908–3913, 2002.
- [38816] 12368. Kavsak, P.; Rasmussen, R. K.; Causing, C. G.; Bonni, S.; Zhu, H.; Thomsen, G. H.; Wrana, J. L.: Smad7 binds to Smurf2 to form an E3 ubiquitin ligase that targets the TGF- β receptor for degradation. *Molec. Cell* 6: 1365–1375, 2000.
- [38817] 12369. Lallemand, F.; Mazars, A.; Prunier, C.; Bertrand, F.; Kornprost, M.; Gallea, S.; Roman-Roman, S.; Cherqui, G.; Atfi, A.: Smad7 inhibits the survival nuclear factor kappa-B and potentiates apoptosis in epithelial cells. *Oncogene* 20: 879–884, 2001.
- [38818] 12370. Roijer, E.; Moren, A.; ten Dijke, P.; Stenman, G.: As-

signment of the Smad7 gene (MADH7) to human chromosome 18q21.1 by fluorescence in situ hybridization. *Cytogenet. Cell Genet.* 81: 189–190, 1998.

[38819] 12371. Cravatt, B. F.; Giang, D. K.; Mayfield, S. P.; Boger, D. L.; Lerner, R. A.; Gilula, N. B.: Molecular characterization of an enzyme that degrades neuromodulatory fatty-acid amides. *Nature* 384: 83–87, 1996.

[38820] 12372. Giang, D. K.; Cravatt, B. F.: Molecular characterization of human and mouse fatty acid amide hydrolases. *Proc. Nat. Acad. Sci.* 94: 2238–2242, 1997.

[38821] 12373. Sipe, J. C.; Chiang, K.; Gerber, A. L.; Beutler, E.; Cravatt, B. F.: A missense mutation in human fatty acid amide hydrolase associated with problem drug use. *Proc. Nat. Acad. Sci.* 99: 8394–8399, 2002.

[38822] 12374. Wan, M.; Cravatt, B. F.; Ring, H. Z.; Zhang, X.; Francke, U.: Conserved chromosomal location and genomic structure of human and mouse fatty-acid amide hydrolase genes and evaluation of clasper as a candidate neurological mutation. *Genomics* 54: 408–414, 1998.

[38823] 12375. Falany, C. N.; Johnson, M. R.; Barnes, S.; Diasio, R. B.: Glycine and taurine conjugation of bile acids by a single enzyme: molecular cloning and expression of a human liver bile acid CoA:amino acid N-acyltransferase. *J. Biol.*

Chem. 269: 19375–19379, 1994.

- [38824] 12376. Johnson, M. R.; Barnes, S.; Kwakye, J. B.; Diasio, R. B.: Purification and characterization of bile acid–CoA:amino acid N-acyltransferase from human liver. *J. Biol. Chem.* 266: 10227–10233, 1991.
- [38825] 12377. Baier, M.; Bannert, N.; Werner, A.; Lang, K.; Kurth, R.: Molecular cloning, sequence, expression, and processing of the interleukin 16 precursor. *Proc. Nat. Acad. Sci.* 94: 5273–5277, 1997.
- [38826] 12378. Roh, M. H.; Makarova, O.; Liu, C.-J.; Shin, K.; Lee, S.; Laurinec, S.; Goyal, M.; Wiggins, R.; Margolis, B.: The Maguk protein, Pals1, functions as an adapter, linking mammalian homologues of Crumbs and Discs Lost. *J. Cell Biol.* 157: 161–172, 2002.
- [38827] 12379. Bannert, N.; Baier, M.; Werner, A.; Kurth, R.: Interleukin–16 or not? *Nature* 381: 30 only, 1996.
- [38828] 12380. Bannert, N.; Kurth, R.; Baier, M.: The gene encoding mouse interleukin–16 consists of seven exons and maps to Chromosome 7 D2–D3. *Immunogenetics* 49: 704–706, 1999.
- [38829] 12381. Cruikshank, W.; Center, D. M.: Modulation of lymphocyte migration by human lymphokines. II. Purification of a lymphotactic factor (LCF). *J. Immun.* 128: 2569–2574,

1982.

- [38830] 12382.Cruikshank, W. W.; Center, D. M.; Nisar, N.; Wu, M.; Natke, B.;Theodore, A. C.; Kornfeld, H.: Molecular and functional analysisof a lymphocyte chemoattractant factor: association of biologic functionwith CD4 expression. Proc. Nat. Acad. Sci. 91: 5109–5113, 1994.
- [38831] 12383.Drwinga, H. L.; Toji, L. H.; Kim, C. H.; Greene, A. E.; Mulivor,R. A.: NIGMS human/rodent somatic cell hybrid mapping panels 1 and2. Genomics 16: 311–314, 1993.
- [38832] 12384.Hudson, T. J.; Stein, L. D.; Gerety, S. S.; Ma, J.; Cas–tle, A.B.; Silva, J.; Slonim, D. K.; Baptista, R.; Kruglyak, L.; Xu, S. H.;Hu, X.; Colbert, A. M. E.; and 39 others: An STS–based map of thehuman genome. Science 270: 1945–1954, 1995.
- [38833] 12385.Keane, J.; Nicoll, J.; Kim, S.; Wu, D. M. H.; Cruik–shank, W. W.;Brazer, W.; Natke, B.; Zhang, Y.; Center, D. M.; Kornfeld, H.: Conservationof structure and function between human and murine IL–16. J. Immun. 160:5945–5954, 1998.
- [38834] 12386.Scheuerpflug, C. G.; Lichter, P.; Debatin, K.–M.; Mincheva, A.: Assignment of TRADD to human chromo–some band 16q22 by in situ hybridization. Cytogenet.Cell Genet. 92: 347–348, 2001.

- [38835] 12387.Lennon–Dumenil, A.–M.; Barbouche, M.–R.; Védrenne, J.; Prod'Homme,T.; Bejaoui, M.; Ghariani, S.; Char–ron, D.; Fellous, M.; Dellagi,K.; Alcaide–Loridan, C.: Unco–ordinated HLA–D gene expression in aRFXANK–defective patient with MHC class II deficiency. *J. Immun.* 166:5681–5687, 2001.
- [38836] 12388.Lisowska–GrosPierre, B.; Fondaneche, M. C.; Rols, M. P.; Griscelli,C.; Fischer, A.: Two complementation groups account for most casesof inherited MHC class II deficiency. *Hum. Molec. Genet.* 3: 953–958,1994.
- [38837] 12389.Boll, W.; Wagner, P.; Mantei, N.: Structure of the chromosomal gene and cDNAs coding for lactase–phlorizin hydrolase in humans withadult–type hypolactasia or per–sistence of lactase. *Am. J. Hum. Genet.* 48:889–902, 1991.
- [38838] 12390.Wiszniewski, W.; Fondaneche, M.–C.; Lambert, N.; Masternak, K.;Picard, C.; Notarangelo, L.; Schwartz, K.; Bal, J.; Reith, W.; Alcaide,C.; de Saint Basile, G.; Fischer, A.; Lisowska–GrosPierre, B.: Foundereffect for a 26–bp dele–tion in the RFXANK gene in North African majorhistocom–patibility complex class II–deficient patients belonging to– complementation group B. *Immunogenetics* 51: 261–267, 2000.
- [38839] 12391.Harvey, C. B.; Fox, M. F.; Jeggo, P. A.; Mantei, N.;

Povey, S.;Swallow, D. M.: Regional localization of the lactase-phlorizin hydrolase gene, LCT, to chromosome 2q21. *Ann. Hum. Genet.* 57: 179–185, 1993.

[38840] 12392.Kruse, T. A.; Bolund, L.; Byskov, A.; Sjostrom, H.; Noren, O.;Mantei, N.; Semenza, G.: Mapping of the human lactase-phlorizin hydrolase gene to chromosome 2.

(Abstract) *Cytogenet. Cell Genet.* 51: 1026,1989.

[38841] 12393.Kruse, T. A.; Bolund, L.; Grzeschik, K.-H.; Ropers, H. H.; Sjostrom,H.; Noren, O.; Mantei, N.; Semenza, G.: The human lactase-phlorizinhydrolase gene is located on chromosome 2. *FEBS Lett.* 240: 123–126,1988.

[38842] 12394.Mantei, N.; Villa, M.; Enzler, T.; Wacker, H.; Boll, W.; James,O.; Hunziker, W.; Semenza, G.: Complete primary structure of humanand rabbit lactase-phlorizin hydrolase: implications for biosynthesis,membrane anchoring and evolution of the enzyme. *EMBO J.* 7: 2705–2713,1988.

[38843] 12395.Stec, I.; Wright, T. J.; van Ommen, G.-J. B.; de Boer, P. A. J.;van Haeringen, A.; Moorman, A. F. M.; Altherr, M. R.; den Dunnen,J. T.: WHSC1, a 90 kb SET domain-containing gene, expressed in earlydevelopment and homologous to a Drosophila dysmorphology gene maps inthe Wolf-Hirschhorn syndrome critical region and is fused to IgH int(4;14) multiple myeloma. *Hum. Molec. Genet.* 7:

1071–1082, 1998.

- [38844] 12396.Ame, J.–C.; Apiou, F.; Jacobson, E. L.; Jacobson, M. K.: Assignment of the poly(ADP–ribose) glycohydrolase gene (PARG) to human chromosome 10q11.23 and mouse chromosome 14B by in situ hybridization. *Cytogenet. Cell Genet.* 85: 269–270, 1999.
- [38845] 12397.Lin, W.; Ame, J.–C.; Aboul–Ela, N.; Jacobson, E. L.; Jacobson, M. K.: Isolation and characterization of the cDNA encoding bovine poly(ADP–ribose) glycohydrolase. *J. Biol. Chem.* 272: 11895–11901, 1997.
- [38846] 12398.Brown, S. D.; Twells, R. C. J.; Hey, P. J.; Cox, R. D.; Levy, E.R.; Soderman, A. R.; Metzker, M. L.; Caskey, C. T.; Todd, J. A.; Hess, J. F.: Isolation and characterization of LRP6, a novel member of the low density lipoprotein receptor gene family. *Biochem. Biophys. Res. Commun.* 248: 879–888, 1998.
- [38847] 12399.Mao, B.; Wu, W.; Li, Y.; Hoppe, D.; Stannek, P.; Glinka, A.; Niehrs, C.: LDL–receptor–related protein 6 is a receptor for Dickkopf proteins. *Nature* 411:321–325, 2001.
- [38848] 12400.Pinson, K. I.; Brennan, J.; Monkley, S.; Avery, B. J.; Skarnes, W. C.: An LDL–receptor–related protein mediates Wnt signalling in mice. *Nature* 407: 535–538, 2000.

- [38849] 12401.Tamai, K.; Semenov, M.; Kato, Y.; Spokony, R.; Liu, C.; Katsuyama,Y.; Hess, F.; Saint-Jeannet, J.-P.; He, X.: LDL-receptor-relatedproteins in Wnt signal transduction. Nature 407: 530-535, 2000.
- [38850] 12402.Wehrli, M.; Dougan, S. T.; Caldwell, K.; O'Keefe, L.; Schwartz,S.; Vaizel-Ohayon, D.; Schejter, E.; Tomlinson, A.; DiNardo, S.:Arrow encodes an LDL-receptor-related protein essential for Wingless signalling. Nature 407: 527-530, 2000. Note: Erratum: Nature 410:847 only, 2001.
- [38851] 12403.Vinkemeier, U.; Obermann, W.; Weber, K.; Furst, D. O.: The globularhead domain of titin extends into the center of the sarcomeric M band:cDNA cloning, epitope mapping and immunoelectron microscopy of twotitin-associated proteins. J. Cell Sci. 106: 319-330, 1993.
- [38852] 12404.van der Ven, P. F. M.; Speel, E. J. M.; Albrechts, J. C. M.; Ramaekers,F. C. S.; Hopman, A. H. N.; Furst, D. O.: Assignment of the humangene for endosarcomeric cytoskeletal M-protein (MYOM2) to 8p23.3. Genomics 55:253-255, 1999.
- [38853] 12405.Black, R. A.; Rauch, C. T.; Kozlosky, C. J.; Peschon, J. J.; Slack,J. L.; Wolfson, M. F.; Castner, B. J.; Stocking, K. L.; Reddy, P.;Srinivasan, S.; Nelson, N.; Boiani, N.; Schoo-

ley, K. A.; Gerhart, M.; Davis, R.; Fitzner, J. N.; Johnson, R. S.; Paxton, R. J.; March, C. J.; Cerretti, D. P.: A metalloproteinase disintegrin that releases tumour-necrosis factor-alpha from cells. *Nature* 385: 729-733, 1997.

[38854] 12406. Hirohata, S.; Seldin, M. F.; Apte, S. S.: Chromosomal assignment of two ADAM genes, TACE (ADAM17) and MLTNB (ADAM19), to human chromosomes 2 and 5, respectively, and of MltNb to mouse chromosome 11. *Genomics* 54: 178-179, 1998.

[38855] 12407. Moss, M. L.; Jin, S.-L. C.; Milla, M. E.; Burkhart, W.; Carter, H. L.; Chen, W.-J.; Clay, W. C.; Didsbury, J. R.; Hasler, D.; Hoffman, C. R.; Kost, T. A.; Lambert, M. H.; and 13 others: Cloning of a disintegrin metalloproteinase that processes precursor tumour-necrosis factor-alpha. *Nature* 385: 733-736, 1997.

[38856] 12408. Patel, I. R.; Attur, M. G.; Patel, R. N.; Stuchin, S. A.; Abagyan, R. A.; Abramson, S. B.; Amin, A. R.: TNF-alpha convertase enzyme from human arthritis-affected cartilage: isolation of cDNA by differential display, expression of the active enzyme, and regulation of TNF-alpha. *J. Immun.* 160: 4570-4579, 1998.

[38857] 12409. Peschon, J. J.; Slack, J. L.; Reddy, P.; Stocking, K. L.; Sunnarborg, S. W.; Lee, D. C.; Russell, W. E.; Castner, B. J.;

Johnson, R. S.;Fitzner, J. N.; Boyce, R. W.; Nelson, N.; Kozlosky, C. J.; Wolfson,M. F.; Rauch, C. T.; Cerretti, D. P.; Paxton, R. J.; March, C. J.;Black, R. A.: An essential role for ectodomain shedding in mammalian development. *Science* 282: 1281–1284, 1998.

[38858] 12410.Yamazaki, K.; Mizui, Y.; Sagane, K.; Tanaka, I.: Genetic mapping of mouse tumor necrosis factor- α converting enzyme (Tace) to chromosome 12. *Genomics* 49: 336–337, 1998.

[38859] 12411.Inoue, D.; Reid, M.; Lum, L.; Kratzschmar, J.; Weskamp, G.; Myung,Y. M.; Baron, R.; Blobel, C. P.: Cloning and initial characterization of mouse meltrin beta and analysis of the expression of four metalloprotease-disintegrins in bone cells. *J. Biol. Chem.* 273: 4180–4187, 1998.

[38860] 12412.Iyer, N. V.; Leung, S. W.; Semenza, G. L.: The human hypoxia-inducible factor 1- α gene: HIF1A structure and evolutionary conservation. *Genomics* 52:159–165, 1998.

[38861] 12413.Kline, D. D.; Peng, Y.-J.; Manalo, D. J.; Semenza, G. L.; Prabhakar,N. R.: Defective carotid body function and impaired ventilatory response to chronic hypoxia in mice partially deficient for hypoxia-inducible factor 1- α .

Proc. Nat. Acad. Sci. 99: 821–826, 2002.

- [38862] 12414.Lando, D.; Peet, D. J.; Whelan, D. A.; Gorman, J. J.; Whitelaw, M. L.: Asparagine hydroxylation of the HIF trans-activation domain: a hypoxic switch. Science 295: 858–861, 2002.
- [38863] 12415.Marti, H. H.; Katschinski, D. M.; Wagner, K. F.; Schaffer, L.; Stier, B.; Wenger, R. H.: Isoform-specific expression of hypoxia-inducible factor-1- α during the late stages of mouse spermiogenesis. Molec. Endocr. 16: 234–243, 2002.
- [38864] 12416.Maxwell, P. H.; Wiesener, M. S.; Chang, G.-W.; Clifford, S. C.; Vaux, E. C.; Cockman, M. E.; Wykoff, C. C.; Pugh, C. W.; Maher, E. R.; Ratcliffe, P. J.: The tumour suppressor protein VHL targets hypoxia-inducible factors for oxygen-dependent proteolysis. Nature 399: 271–275, 1999.
- [38865] 12417.Min, J.-H.; Yang, H.; Ivan, M.; Gertler, F.; Kaelin, W. G., Jr.; Pavletich, N. P.: Structure of an HIF-1- α -pVHL complex: hydroxyproline recognition in signaling. Science 296: 1886–1889, 2002.
- [38866] 12418.Semenza, G. L.: HIF-1 and human disease: one highly involved factor. Genes Dev. 14: 1983–1991, 2000.
- [38867] 12419.Semenza, G. L.; Rue, E. A.; Iyer, N. V.; Pang, M. G.;

Kearns, W. G.: Assignment of the hypoxia-inducible factor 1- α gene to a region of conserved synteny on mouse chromosome 12 and human chromosome 14q. *Genomics* 34: 437–439, 1996.

[38868] 12420. Sutter, C. H.; Laughner, E.; Semenza, G. L.: Hypoxia-inducible factor 1- α protein expression is controlled by oxygen-regulated ubiquitination that is disrupted by deletions and missense mutations. *Proc. Nat. Acad. Sci.* 97: 4748–4753, 2000.

[38869] 12421. Wenger, R. H.; Rolfs, A.; Kvietikova, I.; Spielmann, P.; Zimmermann, D. R.; Gassmann, M.: The mouse gene for hypoxia-inducible factor-1- α —genomic organization, expression and characterization of an alternative first exon and 5-prime flanking sequence. *Europ. J. Biochem.* 246: 155–165, 1997.

[38870] 12422. Wenger, R. H.; Rolfs, A.; Marti, H. H.; Guenet, J.-L.; Gassmann, M.: Nucleotide sequence, chromosomal assignment and mRNA expression of mouse hypoxia-inducible factor-1- α . *Biochem. Biophys. Res. Commun.* 223: 54–59, 1996.

[38871] 12423. Wenger, R. H.; Rolfs, A.; Spielmann, P.; Zimmermann, D. R.; Gassmann, M.: Mouse hypoxia-inducible factor-1- α is encoded by two different mRNA isoforms:

expression from a tissue-specific and a housekeeping-type promoter. *Blood* 91: 3471–3480, 1998.

[38872] 12424. Tian, H.; Hammer, R. E.; Matsumoto, A. M.; Russell, D. W.; McKnight, S. L.: The hypoxia-responsive transcription factor EPAS1 is essential for catecholamine homeostasis and protection against heart failure during embryonic development. *Genes Dev.* 12: 3320–3324, 1998.

[38873] 12425. Tian, H.; McKnight, S. L.; Russell, D. W.: Endothelial PAS domain protein 1 (EPAS1), a transcription factor selectively expressed in endothelial cells. *Genes Dev.* 11: 72–82, 1997.

[38874] 12426. Hovanessian, A. G.; Laurent, A. G.; Chebath, J.; Galabru, J.; Robert, N.; Svab, J.: Identification of 69-kd and 100-kd forms of 2–5A synthetase in interferon-treated human cells by specific monoclonal antibodies. *EMBO J.* 6: 1273–1280, 1987.

[38875] 12427. Marie, I.; Galabru, J.; Svab, J.; Hovanessian, A. G.: Preparation and characterization of polyclonal antibodies specific for the 69 and 100 k-dalton forms of human 2–5A synthetase. *Biochem. Biophys. Res. Commun.* 160: 580–587, 1989.

[38876] 12428. Marie, I.; Hovanessian, A. G.: The 69-kDa 2–5A synthetase is composed of two homologous and adjacent

functional domains. J. Biol. Chem. 267:9933–9939, 1992.

[38877] 12429.Kools, P.; Van Imschoot, G.; van Roy, F.: Characterization of three novel human cadherin genes (CDH7, CDH19, and CDH20) clustered on chromosome 18q22–q23 and with high homology to chicken cadherin-7. Genomics 68:283–295, 2000.

[38878] 12430.Dantzig, A. H.; Hoskins, J.; Tabas, L. B.; Bright, S.; Shepard, R. L.; Jenkins, I. L.; Duckworth, D. C.; Sportsman, J. R.; Mackensen, D.; Rosteck, P. R., Jr.; Skatrud, P. L.: Association of intestinal peptide transport with a protein related to the cadherin superfamily. Science 264:430–433, 1994.

[38879] 12431.Shibata, T.; Shimoyama, Y.; Gotoh, M.; Hirohashi, S.: Identification of human cadherin-14, a novel neurally specific type II cadherin, by protein interaction cloning. J. Biol. Chem. 272: 5236–5240, 1997.

[38880] 12432.Barabino, S. M. L.; Hubner, W.; Jenny, A.; Minvielle-Sebastia, L.; Keller, W.: The 30 kDa subunit of mammalian cleavage and polyadenylation specificity factor and its yeast homolog are RNA binding zinc finger proteins. Genes Dev. 11: 1703–1716, 1997.

[38881] 12433.Nemeroff, M. E.; Barabino, S. M. L.; Li, Y.; Keller, W.; Krug, R. M.: Influenza virus NS1 protein interacts with

the cellular 30kDa subunit of CPSF and inhibits 3-prime end formation of cellular pre-mRNAs. *Molec. Cell* 1: 991–1000, 1998.

- [38882] 12434. Hsu, D. R.; Economides, A. N.; Wang, X.; Eimon, P. M.; Harland, R. M.: The *Xenopus* dorsalizing factor gremlin identifies a novel family of secreted proteins that antagonize BMP activities. *Molec. Cell* 1: 673–683, 1998.
- [38883] 12435. Topol, L. Z.; Modi, W. S.; Koochekpour, S.; Blair, D. G.: DRM-Gremlin (CKTSF1B1) maps to human chromosome 15 and is highly expressed in adult and fetal brain. *Cytogenet. Cell Genet.* 89: 79–84, 2000.
- [38884] 12436. Beaconsfield, P.; Rainsbury, R.; Kalton, G.: Glucose-6-phosphate dehydrogenase deficiency and the incidence of cancer. *Oncologia* 19: 11–19, 1965.
- [38885] 12437. Ben-Bassat, J.; Ben-Ishay, D.: Hereditary hemolytic anemia associated with glucose-6-phosphate dehydrogenase deficiency (Mediterranean type). *Israel J. Med. Sci.* 5: 1053–1059, 1969.
- [38886] 12438. Benabadji, M.; Merad, F.; Benmoussa, M.; Tra-buchet, G.; Junien, C.; Dreyfus, J. C.; Kaplan, J. C.: Heterogeneity of glucose-6-phosphate dehydrogenase deficiency in Algeria. *Hum. Genet.* 40: 177–184, 1978.
- [38887] 12439. Benohr, H. C.; Klumpp, F.; Waller, H. D.: Glucose-

6-phosphat-DehydrogenaseTyp Schwaben. Dtsch. Med. Wschr. 96: 1029–1032, 1971.

[38888] 12440.Benohr, H. C.; Waller, H. D.: Eigenschaften der Glucose-6-p-dehydrogenase,Typ Tubingen. Klin. Wschr. 48: 71–74, 1970.

[38889] 12441.Benohr, H. C.; Waller, H. D.; Arnold, H.; Blume, K. G.; Lohr, G. W.: Glucose-6-P-Dehydrogenase Typ Bondensee (eine neue Enzymvariante). Klin. Wschr. 49: 1058–1062, 1971.

[38890] 12442.Beutler, E.: Glucose 6-phosphate dehydrogenase deficiency, a new Indian variant, G6PD Jammu. In: Sen, N. N.; Basu, A. K.: Trends in Haematology. Calcutta: Chatterjea Memorial Committee (pub.) 1975. Pp. 279–283.

[38891] 12443.Beutler, E.: Personal Communication. La Jolla, Calif. 11/12/1990.

[38892] 12444.Beutler, E.: G6PD deficiency. Blood 84: 3613–3636, 1994.

[38893] 12445.Beutler, E.: Glucose-6-phosphate dehydrogenase deficiency. New Eng. J. Med. 324: 169–174, 1991.

[38894] 12446.Beutler, E.: Selectivity of proteases as a basis for tissue distribution of enzymes in hereditary deficiencies. Proc. Nat. Acad. Sci. 80:3767–3768, 1983.

[38895] 12447.Beutler, E.: Glucose-6-phosphate dehydrogenase

deficiency. In: Wintrobe, M. M.: Red Cell Metabolism in Hemolytic Anemia. New York: Plenum Press (pub.) 1978.

[38896] 12448. Beutler, E.: The hemolytic effect of primaquine and related compounds: a review. *Blood* 14: 103–139, 1959.

[38897] 12449. Beutler, E.; Grooms, A. M.; Morgan, S. K.; Trinidad, F.: Chronic severe hemolytic anemia due to G-6-PD Charleston: a new deficiency variant. *J. Pediatr.* 80: 1005–1009, 1972.

[38898] 12450. Beutler, E.; Hartman, K.; Gelbart, T.; Forman, L.: G-6-PD Walter Reed: possible insight into 'structural' NADP in G-6-PD. *Am. J. Hemat.* 23: 25–30, 1986.

[38899] 12451. Beutler, E.; Keller, J. W.; Matsumoto, F.: A new glucose 6-phosphate dehydrogenase (G6PD) variant associated with nonspherocytic hemolytic anemia: G6PD Atlanta. *I.R.C.S.* 4: 479, 1976.

[38900] 12452. Beutler, E.; Kuhl, W.: The NT 1311 polymorphism of G6PD: G6PD Mediterranean mutation may have originated independently in Europe and Asia. *Am. J. Hum. Genet.* 47: 1008–1012, 1990.

[38901] 12453. Beutler, E.; Kuhl, W.: Linkage between a PvuII restriction fragment length polymorphism and G6PD A- (202A/376G): evidence for a single origin of the common G6PD A- mutation. *Hum. Genet.* 85: 9–11, 1990.

- [38902] 12454.Beutler, E.; Kuhl, W.; Gelbart, T.; Forman, L.: DNA sequence abnormalities of human glucose-6-phosphate dehydrogenase variants. J.Biol. Chem. 266: 4145-4150, 1991.
- [38903] 12455.Beutler, E.; Kuhl, W.; Ramirez, E.; Lisker, R.: Some Mexican glucose-6-phosphate dehydrogenase variants revisited. Hum. Genet. 86:371-374, 1991.
- [38904] 12456.Beutler, E.; Kuhl, W.; Saenz, G. F.; Rodriguez, W.: Mutation analysis of glucose-6-phosphate dehydrogenase (G6PD) variants in Costa Rica. Hum. Genet. 87: 462-464, 1991.
- [38905] 12457.Beutler, E.; Mathai, C. K.; Smith, J. E.: Biochemical variants of glucose-6-phosphate dehydrogenase giving rise to congenital nonspherocytic hemolytic disease. Blood 31: 131-150, 1968.
- [38906] 12458.Beutler, E.; Matsumoto, F.: A new glucose 6-phosphate dehydrogenase variant: G6PD (-) Los Angeles. I.R.C.S. 5: 89, 1977.
- [38907] 12459.Beutler, E.; Matsumoto, F.; Daiber, A.: Nonspherocytic hemolytic anemia due to G-6-PD Panama. I.R.C.S. 2: 1389, 1974.
- [38908] 12460.Beutler, E.; Rosen, R.: Nonspherocytic congenital hemolytic anemia due to a new G-6-PD variant: G-6-PD

Alhambra. Pediatrics 45: 230–235,1970.

- [38909] 12461.Beutler, E.; Westwood, B.; Prchal, J. T.; Vaca, G.; Bartsocas,C. S.; Baronciani, L.: New glucose–6–phosphate dehydrogenase mutationsfrom various ethnic groups. Blood 80: 255–256, 1992.
- [38910] 12462.Beutler, E.; Yoshida, A.: Human glucose–6–phosphate dehydrogenasevariants: a supplementary tabulation. Ann. Hum. Genet. 37: 151–156,1973.
- [38911] 12463.Boivin, P.; Galand, C.: Nouvelles variantes de la glucose–6–phosphatedehydrogenase erythrocytaire. Rev. Franc. Etud. Clin. Biol. 13:30–39, 1968.
- [38912] 12464.Botha, M. C.; Dern, R. J.; Mitchell, M.; West, C.; Beutler, E.: G6PD Capetown, a variant of glucose–6–phosphate dehydrogenase. Am.J. Hum. Genet. 21: 547–551, 1969.
- [38913] 12465.Boyer, S. H.; Graham, J. B.: Linkage between the X chromosomeloci for glucose–6–phosphate dehydrogenase electrophoretic variationand hemophilia A. Am. J. Hum. Genet. 17: 320–324, 1965.
- [38914] 12466.Boyer, S. H.; Porter, I. H.; Weilbaecher, R. G.: Electrophoreticheterogeneity of glucose–6–phosphate dehydrogenase and its relationship to enzyme deficiency in man. Proc. Nat. Acad. Sci. 48: 1868–1876,1962.

- [38915] 12467. Busch, D.; Bote, K.: Glucose-6-phosphate-dehydrogenase-Defectin Deutschland. II. Eigenschaften des Enzyms (Typ Freiburg). *Klin. Wschr.* 48: 74-78, 1970.
- [38916] 12468. Calabro, V.; Giacobbe, A.; Vallone, D.; Montanaro, V.; Cascone, A.; Filosa, S.; Battistuzzi, G.: Genetic heterogeneity at the glucose-6-phosphatedehydrogenase locus in southern Italy: a study on a population from the Matera district. *Hum. Genet.* 86: 49-53, 1990.
- [38917] 12469. Cappadoro, M.; Giribaldi, G.; O'Brien, E.; Turrini, F.; Mannu, F.; Ulliers, D.; Simula, G.; Luzzatto, L.; Arese, P.: Early phagocytosis of glucose-6-phosphate dehydrogenase (G6PD)-deficient erythrocytes parasitized by *Plasmodium falciparum* may explain malaria protection in G6PD deficiency. *Blood* 92: 2527-2534, 1998.
- [38918] 12470. Cappellini, M. D.; Sampietro, M.; Toniolo, D.; Carandina, G.; Pittalis, S.; Martinez di Montemuros, F.; Tavazzi, D.; Fiorelli, G.: Biochemical and molecular characterization of a new sporadic glucose-6-phosphatedehydrogenase variant described in Italy: G6PD Modena. *Brit. J. Haemat.* 87: 209-211, 1994.
- [38919] 12471. Carandina, G.; Moretto, E.; Zecchi, G.; Conighi, C.: Glucose-6-phosphate dehydrogenase Ferrara. A new vari-

ant of G6PD identified in Northern Italy. *Acta Haemat.* 56: 116–122, 1976.

[38920] 12472. Carson, P. E.; Flanagan, C. L.; Ickes, C. E.; Alving, A. S.: Enzymatic deficiency in primaquine-sensitive erythrocytes. *Science* 124:484–485, 1956.

[38921] 12473. Poy, F.; Yaffe, M. B.; Sayos, J.; Saxena, K.; Morra, M.; Sumegi, J.; Cantley, L. C.; Terhorst, C.; Eck, M. J.: Crystal structures of the XLP protein SAP reveal a class of SH2 domains with extended, phosphotyrosine-independent sequence recognition. *Molec. Cell* 4:555–561, 1999.

[38922] 12474. Provisor, A. J.; Iaccone, J. J.; Chilcote, R. R.; Neiburger, R. G.; Crussi, F. G.; Baehner, R. L.: Acquired agammaglobulinemia after a life-threatening illness with clinical and laboratory features of infectious mononucleosis in three related male children. *New Eng. J. Med.* 293: 62–65, 1975.

[38923] 12475. Purtilo, D. T.: Pathogenesis and phenotypes of an X-linked recessive lymphoproliferative syndrome. *Lancet* II: 882–885, 1976.

[38924] 12476. Purtilo, D. T.: X-linked lymphoproliferative syndrome: an immunodeficiency disorder with acquired agammaglobulinemia, fatal infectious mononucleosis, or malignant lymphoma. *Arch. Path. Lab. Med.* 105:

119–121, 1981.

- [38925] 12477. Purtilo, D. T.; Bhawan, J.; Hutt, L. M.; De Nicola, L.; Szymanski, I.; Yang, J. P. S.; Boto, W.; Naier, R.; Thorley-Lawson, D.: Epstein-Barrvirus in the X-linked recessive lymphoproliferative syndrome. *Lancet* I:798–801, 1978.
- [38926] 12478. Purtilo, D. T.; Cassel, C. K.; Yang, J. P. S.: Fatal infectious mononucleosis in familial lymphohistiocytosis. (Letter) *New Eng. J. Med.* 201: 736 only, 1974.
- [38927] 12479. Purtilo, D. T.; Cassel, C. K.; Yang, J. P. S.; Harper, R.; Stephenson, S. R.; Landing, B. H.; Vewter, G. F.: X-linked recessive progressive combined variable immunodeficiency (Duncan's disease). *Lancet* I:935–941, 1975.
- [38928] 12480. Purtilo, D. T.; DeFlorio, D., Jr.; Hutt, L. M.; Bhawan, J.; Yang, J. P. S.; Otto, R. L.; Edwards, W.: Variable phenotypic expression of an X-linked recessive lymphoproliferative syndrome. *New Eng. J. Med.* 297: 1077–1081, 1977.
- [38929] 12481. Purtilo, D. T.; Grierson, H. L.: Methods of detection of new families with X-linked lymphoproliferative disease. *Cancer Genet. Cytogenet.* 51: 143–153, 1991.
- [38930] 12482. Purtilo, D. T.; Sakamoto, K.; Barnabei, V.; Seeley, J.; Bechtold, T.; Rogers, G.; Yetz, J.; Harada, S.; the XLP collaborators: Epstein-Barrvirus-induced diseases in boys with the X-linked lymphoproliferative syndrome (XLP): up-

date on studies of the registry. *Am. J. Med.* 73:49–56, 1982.

[38931] 12483. Purtilo, D. T.; Yang, J. P. S.; Allegra, S.; DeFlorio, D.; Hutt, L. M.; Soltani, M.; Vawter, G. F.: Hematopathology and pathogenesis of the X-linked recessive lymphoproliferative syndrome. *Am. J. Med.* 62:225–233, 1977.

[38932] 12484. Sanger, W. G.; Grierson, H. L.; Skare, J.; Wyandt, H.; Pirruccello, S.; Fordyce, R.; Purtilo, D. T.: Partial Xq25 deletion in a family with the X-linked lymphoproliferative disease (XLP). *Cancer Genet. Cytogenet.* 47: 163–169, 1990.

[38933] 12485. Sayos, J.; Wu, C.; Morra, M.; Wang, N.; Zhang, X.; Allen, D.; van Schaik, S.; Notarangelo, L.; Gehat, R.; Roncarolo, M. G.; Oettgen, H.; De Vries, J. E.; Aversall, G.; Terhorst, C.: The X-linked lymphoproliferative-disease gene product SAP regulates signals induced through the co-receptor SLAM. *Nature* 395: 462–469, 1998.

[38934] 12486. Scher, I.: The CBA/N mouse strain: an experimental model illustrating the influence of the X-chromosome on immunity. *Adv. Immun.* 33: 1–71, 1982.

[38935] 12487. Schuster, V.; Kreth, H. W.: X-linked lymphoproliferative disease. In: Ochs, H. D.; Smith, C. I. E.; Puck, J. M. (eds.): *Primary Immunodeficiency Diseases: A Molecular and Genetic Approach*. New York: Oxford University Press

1999. Pp. 222–232.

- [38936] 12488. Seemayer, T. A.; Gross, T. G.; Egeler, R. M.; Pirruccello, S. J.; Davis, D. J.; Kelly, C. M.; Okano, M.; Lanyi, A.; Sumegi, J.: X-linked lymphoproliferative disease: twenty-five years after the discovery. *Pediat. Res.* 38: 471–478, 1995.
- [38937] 12489. Skare, J.; Grierson, H.; Wyandt, H.; Sanger, W.; Milunsky, J.; Purtilo, D.; Sullivan, J.; Milunsky, A.: Genetics of the X-linked lymphoproliferative syndrome. (Abstract) *Am. J. Hum. Genet.* 45 (suppl.): A161 only, 1989.
- [38938] 12490. Skare, J.; Madan, S.; Glaser, J.; Purtilo, D.; Nitowsky, H.; Pulijaal, V.; Milunsky, A.: First prenatal diagnosis of X-linked lymphoproliferative disease. *Am. J. Med. Genet.* 44: 79–81, 1992.
- [38939] 12491. Skare, J.; Milunsky, A.; Byron, K.; Sullivan, J.: The mutation causing X-linked lymphoproliferative syndrome lies in Xq26. (Abstract) *Am. J. Hum. Genet.* 41: A185 only, 1987.
- [38940] 12492. Skare, J.; Wu, B.-L.; Madan, S.; Pulijaal, V.; Purtilo, D.; Haber, D.; Nelson, D.; Sylla, B.; Grierson, H.; Nitowsky, H.; Glaser, J.; Wissink, J.; White, B.; Holden, J.; Housman, D.; Lenoir, G.; Wyandt, H.; Milunsky, A.: Characterization of three overlapping deletions causing X-linked lymphopro-

liferative disease. *Genomics* 16: 254–255,1993.

- [38941] 12493. Skare, J. C.; Grierson, H. L.; Sullivan, J. L.; Nussbaum, R. L.; Purtilo, D. T.; Sylla, B. S.; Lenoir, G. M.; Reilly, D. S.; White, B. N.; Milunsky, A.: Linkage analysis of seven kindreds with the X-linked lymphoproliferative syndrome (XLP) confirms that the XLP locus is near DXS42 and DXS37. *Hum. Genet.* 82: 354–358, 1989.
- [38942] 12494. Skare, J. C.; Milunsky, A.; Byron, K. S.; Sullivan, J. L.: Mapping the X-linked lymphoproliferative syndrome. *Proc. Nat. Acad. Sci.* 84: 2015–2018, 1987.
- [38943] 12495. Skare, J. C.; Sullivan, J. L.; Milunsky, A.: Mapping the mutation causing the X-linked lymphoproliferative syndrome in relation to restriction fragment length polymorphisms on Xq. *Hum. Genet.* 82: 349–353, 1989.
- [38944] 12496. Steinherz, R.; Levy, Y.; Litwin, A.; Nitzan, M.; Friedman, E.; Levin, S.: X-linked lymphoproliferative syndrome: a new kindred with variable phenotypic expression. *Am. J. Dis. Child.* 139: 191–193, 1985.
- [38945] 12497. Nehlin, J. O.; Hara, E.; Kuo, W.-L.; Collins, C.; Campisi, J.: Genomic organization, sequence, and chromosomal localization of the human helix-loop-helix *Id1* gene. *Biochem. Biophys. Res. Commun.* 231: 628–634, 1997.
- [38946] 12498. Loftin, C. D.; Trivedi, D. B.; Tiano, H. F.; Clark, J. A.;

Lee, C. A.; Epstein, J. A.; Morham, S. G.; Breyer, M. D.; Nguyen, M.; Hawkins, B. M.; Goulet, J. L.; Smithies, O.; Koller, B. H.; Langenbach, R.: Failure of ductus arteriosus closure and remodeling in neonatal mice deficient in cyclooxygenase-1 and cyclooxygenase-2. *Proc. Nat. Acad. Sci.* 98: 1059–1064, 2001.

[38947] 12499. Macchia, L.; Di Paola, R.; Guerrese, M.-C.; Chiechi, L. M.; Tursi, A.; Caiaffa, M. F.; Haeggstrom, J. Z.: Expression of prostaglandin endoperoxide H synthase 1 and 2 in human placenta at term. *Biochem. Biophys. Res. Commun.* 233: 496–501, 1997.

[38948] 12500. Morham, S. G.; Langenbach, R.; Loftin, C. D.; Tiano, H. F.; Vouloumanos, N.; Jennette, J. C.; Mahler, J. F.; Kluckman, K. D.; Ledford, A.; Lee, C. A.; Smithies, O.: Prostaglandin synthase 2 gene disruption causes severe renal pathology in the mouse. *Cell* 83: 473–482, 1995.

[38949] 12501. Neufang, G.; Furstenberger, G.; Heidt, M.; Marks, F.; Muller-Decker, K.: Abnormal differentiation of epidermis in transgenic mice constitutively expressing cyclooxygenase-2 in skin. *Proc. Nat. Acad. Sci.* 98: 7629–7634, 2001.

[38950] 12502. O'Banion, M. K.; Sadowski, H. B.; Winn, V.; Young, D. A.: A serum- and glucocorticoid-regulated 4-kilobase

mRNA encodes a cyclooxygenase-related protein. J. Biol. Chem. 266: 23261–23267, 1991.

- [38951] 12503. O'Banion, M. K.; Winn, V. D.; Young, D. A.: cDNA cloning and functional activity of a glucocorticoid-regulated inflammatory cyclooxygenase. Proc. Nat. Acad. Sci. 89: 4888–4892, 1992.
- [38952] 12504. Oshima, M.; Dinchuk, J. E.; Kargman, S. L.; Oshima, H.; Hancock, B.; Kwong, E.; Trzaskos, J. M.; Evans, J. F.; Taketo, M. M.: Suppression of intestinal polyposis in Apc(delta-716) knockout mice by inhibition of cyclooxygenase 2 (COX-2). Cell 87: 803–809, 1996.
- [38953] 12505. Salmenkivi, K.; Haglund, C.; Ristimäki, A.; Arola, J.; Heikkilä, P.: Increased expression of cyclooxygenase-2 in malignant pheochromocytomas. J. Clin. Endocr. Metab. 86: 5615–5619, 2001.
- [38954] 12506. Tay, A.; Squire, J. A.; Goldberg, H.; Skorecki, K.: Assignment of the human prostaglandin-endoperoxide synthase 2 (PTGS2) gene to 1q25 by fluorescence in situ hybridization. Genomics 23: 718–719, 1994.
- [38955] 12507. Swan, S. K.; Rudy, D. W.; Lasseter, K. C.; Ryan, C. F.; Buechel, K. L.; Lambrecht, L. J.; Pinto, M. B.; Dilzer, S. C.; Obrda, O.; Sundblad, K. J.; Gumbs, C. P.; Ebel, D. L.; Quan, H.; Larson, P. J.; Schwartz, J. I.; Musliner, T. A.;

Gertz, B. J.; Brater, D. C.; Yao, S.-L.:Effect of cyclooxygenase-2 inhibition on renal function in elderly persons receiving a low-salt diet: a randomized controlled trial. *Ann.Intern. Med.* 133: 1-9, 2000.

- [38956] 12508.Tazawa, R.; Xu, X.-M.; Wu, K. K.; Wang, L.-H.: Characterization of the genomic structure, chromosomal location and promoter of human prostaglandin H synthase-2 gene. *Biochem. Biophys. Res. Commun.* 203:190-199, 1994.
- [38957] 12509.Tsujii, M.; DuBois, R. N.: Alterations in cellular adhesion and apoptosis in epithelial cells overexpressing prostaglandin endoperoxide synthase 2. *Cell* 83: 493-501, 1995.
- [38958] 12510.Zhou, X.-L.; Lei, Z. M.; Rao, C. V.: Treatment of human endometrial gland epithelial cells with chorionic gonadotropin/luteinizing hormone increases the expression of the cyclooxygenase-2 gene. *J. Clin. Endocr.Metab.* 84: 3364-3377, 1999.
- [38959] 12511.Maho, A.; Bensimon, A.; Vassart, G.; Parmentier, M.: Mapping of the CCXCR1, CX3CR1, CCBP2 and CCR9 genes to the CCR cluster within the 3p21.3 region of the human genome. *Cytogenet. Cell Genet.* 87:265-268, 1999.

- [38960] 12512. Anderson, D. M.; Johnson, L.; Glaccum, M. B.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Valentine, V.; Kirstein, M. N.; Shapiro, D. N.; Morris, S. W.; Grabstein, K.; Cosman, D.: Chromosomal assignment and genomic structure of IL15. *Genomics* 25: 701–706, 1995.
- [38961] 12513. Grabstein, K. H.; Eisenman, J.; Shanebeck, K.; Rauch, C.; Srinivasan, S.; Fung, V.; Beers, C.; Richardson, J.; Schoenborn, M. A.; Ahdieh, M.; Johnson, L.; Alderson, M. R.; Watson, J. D.; Anderson, D. M.; Giri, J. G.: Cloning of a T cell growth factor that interacts with the beta chain of the interleukin–2 receptor. *Science* 264: 965–968, 1994.
- [38962] 12514. Roberts, A. I.; Lee, L.; Schwarz, E.; Groh, V.; Spies, T.; Ebert, E. C.; Jabri, B.: Cutting edge: NKG2D receptors induced by IL–15 costimulate CD28–negative effector CTL in the tissue microenvironment. *J. Immun.* 167: 5527–5530, 2001.
- [38963] 12515. Robinson, P.; Okhuysen, P. C.; Chappell, C. L.; Lewis, D. E.; Shahab, I.; Lahoti, S.; White, A. C., Jr.: Expression of IL–15 and IL–4 in IFN–gamma–independent control of experimental human *Cryptosporidium parvum* infection. *Cytokine* 15: 39–46, 2001.
- [38964] 12516. Chen, X.; Vinkemeier, U.; Zhao, Y.; Jeruzalmi, D.; Darnell, J. E., Jr.; Kuriyan, J.: Crystal structure of a tyrosine

phosphorylatedSTAT-1 dimer bound to DNA. Cell 93: 827-839, 1998.

- [38965] 12517.Copeland, N. G.; Gilbert, D. J.; Schindler, C.; Zhong, Z.; Wen,Z.; Darnell, J. E., Jr.; Mui, A. L.-F.; Miyajima, A.; Quelle, F. W.;Ihle, J. N.; Jenkins, N. A.: Distribution of the mammalian Stat genefamily in mouse chromosomes. Ge-nomics 29: 225-228, 1995.
- [38966] 12518.Darnell, J. E., Jr.; Kerr, I. M.; Stark, G. M.: Jak-STAT pathwaysand transcriptional activation in response to IFNs and other extracellularsignaling proteins. Science 264: 1415-1421, 1994.
- [38967] 12519.Durbin, J. E.; Hackenmiller, R.; Simon, M. C.; Levy, D. E.: Targetedisruption of the mouse Stat1 gene results in compromised innate immunityto viral disease. Cell 84: 443-450, 1996.
- [38968] 12520.Haddad, B.; Pabon-Pena, C. R.; Young, H.; Sun, W. H.: Assignmentof STAT1 to human chromosome 2q32 by FISH and radiation hybrids. Cytogenet.Cell Genet. 83: 58-59, 1998.
- [38969] 12521.Ihle, J. N.: STATs: signal transducers and activators of transcription. Cell 84:331-334, 1996.
- [38970] 12522.Meraz, M. A.; White, J. M.; Sheehan, K. C. F.; Bach, E. A.; Rodig,S. J.; Dighe, A. S.; Kaplan, D. H.; Riley, J. K.;

Greenlund, A. C. Campbell, D.; Carver-Moore, K.; DuBois, R. N.; Clark, R.; Aguet, M.; Schreiber, R. D.: Targeted disruption of the Stat1 gene in mice reveals unexpected physiologic specificity in the JAK-STAT signaling pathway. *Cell* 84:431-442, 1996.

- [38971] 12523. Mowen, K. A.; Tang, J.; Zhu, W.; Schurter, B. T.; Shuai, K.; Herschman, H. R.; David, M.: Arginine methylation of STAT1 modulates IFN- α /beta-induced transcription. *Cell* 104: 731-741, 2001.
- [38972] 12524. Ramana, C. V.; Chatterjee-Kishore, M.; Nguyen, H.; Stark, G. R.: Complex roles of Stat1 in regulating gene expression. *Oncogene* 19:2619-2627, 2000.
- [38973] 12525. Yamamoto, K.; Kobayashi, H.; Arai, A.; Miura, O.; Hirosawa, S.; Miyasaka, N.: cDNA cloning, expression and chromosome mapping of the human STAT4 gene: both STAT4 and STAT1 genes are mapped to 2q32.2-q32.3. *Cytogenet. Cell Genet.* 77: 207-210, 1997.
- [38974] 12526. Ware, R. E.; Howard, T. A.; Kamitani, T.; Chang, H.-M.; Yeh, E. T. H.; Seldin, M. F.: Chromosomal assignment of genes involved in glycosylphosphatidylinositol anchor biosynthesis: implications for the pathogenesis of paroxysmal nocturnal hemoglobinuria. *Blood* 83:3753-3757, 1994.

- [38975] 12527.Watanabe, R.; Inoue, N.; Westfall, B.; Taron, C. H.; Orlean, P.;Takeda, J.; Kinoshita, T.: The first step of glycosylphosphatidylinositolbiosynthesis is mediated by a complex of PIG-A, PIG-H, PIG-C and GPI1. EMBOJ. 17: 877-885, 1998.
- [38976] 12528.Watanabe, R.; Kinoshita, T.; Masaki, R.; Yamamoto, A.; Takeda,J.; Inoue, N.: PIG-A and PIG-H, which participate in glycosylphosphatidylinositolanchor biosynthesis, form a protein complex in the endoplasmic reticulum. J.Biol. Chem. 271: 26868-26875, 1996.
- [38977] 12529.Chernajovsky, Y.; Brown, A.; Clark, J.: Human kinesin light (beta)chain gene: DNA sequence and functional characterization of its promoterand first exon. DNA Cell Biol. 15: 965-974, 1996.
- [38978] 12530.Goedert, M.; Marsh, S.; Carter, N.: Localization of the humankinesin light chain gene (KNS2) to chromosome 14q32.3 by fluorescencein situ hybridization. Genomics 32: 173-175, 1996.
- [38979] 12531.Kamal, A.; Stokin, G. B.; Yang, Z.; Xia, C.; Goldstein, L. S.:Axonal transport of amyloid precursor protein is mediated by directbinding to the kinesin light chain subunit of kinesin-I. Neuron 28:449-459, 2000.
- [38980] 12532.Butzow, R.; Huhtala, M.-L.; Bohn, H.; Virtanen, I.;

Seppala, M.: Purification and characterization of placental protein 5. *Biochem.Biophys. Res. Commun.* 150: 483–490, 1988.

[38981] 12533.Kisiel, W.; Sprecher, C. A.; Foster, D. C.: Evidence that a second human tissue factor pathway inhibitor (TFPI–2) and human placental protein 5 are equivalent. (Letter) *Blood* 84: 4384–4385, 1994.

[38982] 12534.Miyagi, Y.; Yasumitsu, H.; Eki, T.; Miyata, S.; Koshikawa, N.; Hirahara, F.; Aoki, I.; Misugi, K.; Miyazaki, K.: Assignment of the human PP5/TFPI–2 gene to 7q22 by FISH and PCR–based human/rodent cell hybrid mapping panel analysis. *Genomics* 35: 267–268, 1996.

[38983] 12535.Miyagi, Y.; Yasumitsu, H.; Mizushima, H.; Koshikawa, N.; Matsuda, Y.; Itoh, H.; Hori, T.–A.; Aoki, I.; Misugi, K.; Miyazaki, K.: Cloning of the cDNA encoding mouse PP5/TFPI–2 and mapping of the gene to chromosome 6. *DNA and Cell Biology* 15: 947–954, 1996.

[38984] 12536.Siiteri, J. E.; Koistinen, R.; Salem, H. T.; Bohn, H.; Seppala, M.: Placental protein 5 is related to blood coagulation and fibrinolytic systems. *Life Sci.* 30: 1885–1891, 1982.

[38985] 12537.Sprecher, C. A.; Kisiel, W.; Mathewes, S.; Foster, D. C.: Molecular cloning, expression, and partial characteriza–

tion of a second humantissue–factor–pathway inhibitor.

Proc. Nat. Acad. Sci. 91: 3353–3357,1994.

[38986] 12538.Maglott, D. R.; Durkin, A. S.; Lane, S. A.; Callen, D. F.; Feldblyum,T. V.; Nierman, W. C.: The gene for membrane protein E16 (D16S469E)maps to human chromosome 16q24.3 and is expressed in human brain,thymus, and retina. Genomics 23: 303–304, 1994.

[38987] 12539.Kastury, K.; Druck, T.; Huebner, K.; Barletta, C.; Acampora, D.;Simeone, A.; Faiella, A.; Boncinelli, E.: Chromosome locations ofhuman EMX and OTX genes. Genomics 22: 41–45, 1994.

[38988] 12540.Simeone, A.; Acampora, D.; Gulisano, M.; Stornaiuolo, A.; Boncinelli,E.: Nested expression domains of four homeobox genes in developingrostral brain. Nature 358: 687–690, 1992.

[38989] 12541.Bishop, K. M.; Goudreau, G.; O'Leary, D. D. M.: Regulation ofarea identity in the mammalian neocortex by Emx2 and Pax6. Science 288:344–349, 2000.

[38990] 12542.Boncinelli, E.; Gulisano, M.; Spada, F.; Broccoli, V.: Emx andOtx gene expression in the developing mouse brain. Ciba Found. Symp. 193:100–116, 1995.

[38991] 12543.Bosetti, A.; Faiella, A.; Boncinelli, E.; Consalez, G. G.: Linkagemapping of Emx2 to mouse chromosome 19.

Mammalian Genome 8: 71–72,1997.

- [38992] 12544.Noonan, F. C.; Mutch, D. G.; Mallon, M. A.; Good-fellow, P. J.:Characterization of the homeodomain gene EMX2: sequence conservation,expression analysis, and a search for mutations in endometrial cancers. Genomics 76:37–44, 2001.
- [38993] 12545.Acampora, D.; Mazan, S.; Avantaggiato, V.; Barone, P.; Tuorto,F.; Lallemand, Y.; Brulet, P.; Simeone, A.: Epilepsy and brain abnormalitiesin mice lacking the Otx1 gene. Nature Genet. 14: 218–222, 1996.
- [38994] 12546.Boncinelli, E.; Gulisano, M.; Broccoli, V.: Emx and Otx homeoboxgenes in the developing mouse brain. J. Neurobiol. 24: 1356–1366,1993.
- [38995] 12547.Frantz, G. D.; Weimann, J. M.; Levin, M. E.; Mc-Connell, S. K.:Otx1 and Otx2 define layers and regions in developing cerebral cortexand cerebellum. J. Neurosci. 14: 5725–5740, 1994.
- [38996] 12548.Avraham, S.; Jiang, S.; Ota, S.; Fu, Y.; Deng, B.; Dowler, L. L.;White, R. A.; Avraham, H.: Structural and functional studies of theintracellular tyrosine kinase MATK gene and its translated product. J.Biol. Chem. 270: 1833–1842, 1995.
- [38997] 12549.Bennett, B. D.; Cowley, S.; Jiang, S.; London, R.;

Deng, B.; Grabarek, J.; Groopman, J. E.; Goeddel, D. V.; Avraham, H.: Identification and characterization of a novel tyrosine kinase from megakaryocytes. *J. Biol. Chem.* 269: 1068–1074, 1994.

[38998] 12550. Klages, S.; Adam, D.; Class, K.; Fagnoli, J.; Bolen, J. B.; Penhallow, R. C.: Ctk: a protein-tyrosine kinase related to Csk that defines an enzyme family. *Proc. Nat. Acad. Sci.* 91: 2597–2601, 1994.

[38999] 12551. Sakano, S.; Iwama, A.; Inazawa, J.; Ariyama, T.; Ohno, M.; Suda, T.: Molecular cloning of a novel non-receptor tyrosine kinase, HYL (hematopoietic consensus tyrosine-lacking kinase). *Oncogene* 9: 1155–1161, 1994.

[39000] 12552. Zrihan-Licht, S.; Lim, J.; Keydar, I.; Sliwkowski, M. X.; Groopman, J. E.; Avraham, H.: Association of Csk-homologous kinase (CHK) (formerly MATK) with HER-2/ErbB-2 in breast cancer cells. *J. Biol. Chem.* 272: 1856–1863, 1997.

[39001] 12553. Shimizu, S.; Narita, M.; Tsujimoto, Y.: Bcl-2 family proteins regulate the release of apoptogenic cytochrome c by the mitochondrial channel VDAC. *Nature* 399: 483–487, 1999.

[39002] 12554. Nichols, A. F.: Personal Communication. Berkeley, Calif. 10/4/1995.

- [39003] 12555.Campbell, H. D.; Schimansky, T.; Claudianos, C.; Ozsarac, N.; Kasprzak, A. B.; Cotsell, J. N.; Young, I. G.; de Couet, H. G.; Gabor Miklos, G. L.: The *Drosophila melanogaster* flightless-I gene involved in gastrulation and muscle degeneration encodes gelsolin-like and leucine-rich repeat domains and is conserved in *Caenorhabditis elegans* and humans. *Proc. Nat. Acad. Sci.* 90: 11386–11390, 1993.
- [39004] 12556.Harmon, D. L.; Gardner-Medwin, D.; Stirling, J. L.: Two new mutations in a late infantile Tay-Sachs patient are both in exon 1 of the beta-hexosaminidase alpha subunit gene. *J. Med. Genet.* 30: 123–128, 1993.
- [39005] 12557.Buckle, V.; Mondello, C.; Darling, S.; Craig, I. W.; Goodfellow, P. N.: Homologous expressed genes in the human sex chromosome pairing region. *Nature* 317: 739–741, 1985.
- [39006] 12558.Slaugenhaupt, S. A.; Roca, A. L.; Liebert, C. B.; Altherr, M. R.; Gusella, J. F.; Reppert, S. M.: Mapping of the gene for the Mel1a-melatonin receptor to human chromosome 4 (MTNR1A) and mouse chromosome 8 (Mtnr1a). *Genomics* 27:355–357, 1995.
- [39007] 12559.Hattori, Y.; Yamashiro, Y.; Ohba, Y.; Miyaji, T.; Morishita, M.; Yamamoto, K.; Yamamoto, K.; Narai, S.;

Kimura, A.: A new beta-thalassemia mutation (initiation codon ATG-to-GTG) found in the Japanese population. Hemoglobin 15:317-325, 1991.

- [39008] 12560. Holmes, L. B.: Norrie's disease--an X-linked syndrome of retinal malformation, mental retardation and deafness. New Eng. J. Med. 284:367-368, 1971.
- [39009] 12561. Isashiki, Y.; Ohba, N.; Yanagita, T.; Hokita, N.; Doi, N.; Nakagawa, M.; Ozawa, M.; Kuroda, N.: Novel mutation at the initiation codon in the Norrie disease gene in two Japanese families. Hum. Genet. 95:105-108, 1995.
- [39010] 12562. Isashiki, Y.; Ohba, N.; Yanagita, T.; Hokita, N.; Hotta, Y.; Hayakawa, M.; Fujiki, K.; Tanabe, U.: Mutations in the Norrie disease gene: a new mutation in a Japanese family. (Letter) Brit. J. Ophthalmol. 79:703-708, 1995.
- [39011] 12563. Johnson, K.; Mintz-Hittner, H. A.; Conley, Y. P.; Ferrell, R. E.: X-linked exudative vitreoretinopathy caused by an arginine to leucine substitution (R121L) in the Norrie disease protein. Clin. Genet. 50: 113-115, 1996.
- [39012] 12564. Johnston, S. S.; Hanna, J. E.; Nevin, N. C.; Bryars, J. H.: Norrie's disease. Birth Defects Orig. Art. Ser. 18(6): 729-738, 1982.
- [39013] 12565. Katayama, S.; Wohlfeld, M.; Golbus, M. S.: First demonstration of recombination between the gene for

Norrie disease and probe L1.28. Am.J. Med. Genet. 30: 967–970, 1988.

[39014] 12566.Kivlin, J. D.; Sanborn, G. E.; Wright, E.; Cannon, L.; Carey,J.: Further linkage data on Norrie disease. Am. J. Med. Genet. 26:733–736, 1987.

[39015] 12567.Lindsay, S.; Thiselton, D. L.; Bateman, J. B.; Ngo, J. T.; Sparkes,R. S.; Coleman, M.; Davies, K. E.; Bhattacharya, S. S.: Localisationof the gene for Norrie disease to be-tween DXS7 and DXS426 on Xp. Hum.Genet. 88: 349–350, 1992.

[39016] 12568.Meindl, A.; Berger, W.; Meitinger, T.; van de Pol, D.; Achatz,H.; Dorner, C.; Haasemann, M.; Hellebrand, H.; Gal, A.; Cremers, F.;Ropers, H.–H.: Norrie disease is caused by mutations in an extracellularprotein resembling C-terminal globular domain of mucins. Nature Genet. 2:139–143, 1992.

[39017] 12569.Meindl, A.; Lorenz, B.; Achatz, H.; Hellebrand, H.; Schmitz–Valckenberg,P.; Meitinger, T.: Missense muta-tions in the NDP gene in patientswith a less severe course of Norrie disease. Hum. Molec. Genet. 4:489–490, 1995.

[39018] 12570.Meitinger, T.; Meindl, A.; Bork, P.; Rost, B.; Sander, C.; Haasemann,M.; Murken, J.: Molecular modeling of the Norrie disease proteinpredicts a cystine knot growth fac–

tor tertiary structure. *NatureGenet.* 5: 376–380, 1993.

[39019] 12571. Moreira-Filho, C. A.; Neustein, I.: A presumptive new variant of Norrie's disease. *J. Med. Genet.* 16: 125–128, 1979.

[39020] 12572. Nance, W. E.; Hara, S.; Hansen, A.; Elliott, J.; Lewis, M.; Chown, B.: Genetic linkage studies in a Negro kindred with Norrie's disease. *Am.J. Hum. Genet.* 21: 423–429, 1969.

[39021] 12573. Ngo, J.; Spence, M. A.; Cortessis, V.; Bateman, J. B.; Sparkes, R. S.: Duplicate report crossing over in Norrie disease family. (Letter) *Am.J. Med. Genet.* 33: 286, 1989.

[39022] 12574. Ngo, J. T.; Bateman, J. B.; Cortessis, V.; Sparkes, R. S.; Mohandas, T.; Inana, G.; Spence, M. A.: Norrie disease: linkage analysis using a 4.2-kb RFLP detected by a human ornithine aminotransferase cDNA probe. *Genomics* 4: 539–545, 1989.

[39023] 12575. Ngo, J. T.; Spence, M. A.; Cortessis, V.; Sparkes, R. S.; Bateman, J. B.: Recombinational event between Norrie disease and DXS7 loci. *Clin. Genet.* 34: 43–47, 1988.

[39024] 12576. Norrie, G.: Nogle Blindheds årsager: en oversigt. *Hospitalstidende* 76: 141–147, 1933.

[39025] 12577. Ohba, N.; Yamashita, T.: Primary vitreoretinal dysplasia resembling Norrie's disease in a female: association

with X autosome chromosomal translocation. *Brit. J. Ophthalmol.* 70: 64–71, 1986.

[39026] 12578. Phillips, C. I.; Newton, M.; Duvall, J.; Holloway, S.; Levy, A.M.: Probably Norrie's disease due to mutation: two sporadic sibships of two males each, a necropsy of one case, and, given Norrie's disease, a calculation of the gene mutation frequency. *Brit. J. Ophthalmol.* 70:305–313, 1986.

[39027] 12579. Rehm, H. L.; Gutierrez-Espeleta, G. A.; Garcia, R.; Jimenez, G.; Khetarpal, U.; Priest, J. M.; Sims, K. B.; Keats, B. J. B.; Morton, C. C.: Norrie disease gene mutation in a large Costa Rican kindred with a novel phenotype including venous insufficiency. *Hum. Mutat.* 9: 402–408, 1997.

[39028] 12580. Schuback, D. E.; Chen, Z. Y.; Craig, I. W.; Breakefield, X. O.; Sims, K. B.: Mutations in the Norrie disease gene. *Hum. Mutat.* 5:285–292, 1995.

[39029] 12581. Shastry, B. S.: Identification of a recurrent missense mutation in the Norrie disease gene associated with a simplex case of exudative vitreoretinopathy. *Biochem. Biophys. Res. Commun.* 246: 35–38, 1998.

[39030] 12582. Shastry, B. S.; Hejtmancik, J. F.; Plager, D. A.; Hartzler, M.K.; Trese, M. T.: Linkage and candidate gene analysis in X-linked familial exudative vitreoretinopathy. *Genomics* 27: 341–344, 1995.

- [39031] 12583. Shastry, B. S.; Hejtmancik, J. F.; Trese, M. T.: Identification of novel missense mutations in the Norrie disease gene associated with one X-linked and four sporadic cases of familial exudative vitreoretinopathy. *Hum. Mutat.* 9: 396–401, 1997.
- [39032] 12584. Sims, K. B.; Lebo, R. V.; Benson, G.; Shalish, C.; Schuback, D.; Chen, Z. Y.; Bruns, G.; Craig, I. W.; Golbus, M. S.; Breakefield, X. O.: The Norrie disease gene maps to a 150 kb region on chromosome Xp11.3. *Hum. Molec. Genet.* 1: 83–89, 1992.
- [39033] 12585. Sims, K. B.; Ozelius, L.; Corey, T.; Rinehart, W. B.; Liberfarb, R.; Haines, J.; Chen, W. J.; Norio, R.; Sankila, E.; de la Chapelle, A.; Murphy, D. L.; Gusella, J.; Breakefield, X. O.: Norrie disease gene is distinct from the monoamine oxidase genes. *Am. J. Hum. Genet.* 45:424–434, 1989.
- [39034] 12586. Taylor, P. J.; Coates, T.; Newhouse, M. L.: Episkopi blindness: hereditary blindness in a Greek Cypriot family. *Brit. J. Ophthalmol.* 43:340–344, 1959.
- [39035] 12587. Torrente, I.; Mangino, M.; Gennarelli, M.; Novelli, G.; Giannotti, A.; Vadala, P.; Dallapiccola, B.: Two new missense mutations (A105T and C110G) in the norrin gene in two Italian families with Norrie disease and familial exudative vitreoretinopathy. (Letter) *Am. J. Med. Genet.* 72:

242–244, 1997.

- [39036] 12588.Burgoyne, P. S.: Genetic homology and crossing over in the X and Y chromosomes of mammals. *Hum. Genet.* 61: 85–90, 1982.
- [39037] 12589.Cooke, H. J.; Brown, W. R. A.; Rappold, G. A.: Hypervariable telomeric sequences from the human sex chromosomes are pseudoautosomal. *Nature* 317:687–692, 1985.
- [39038] 12590.Darling, S. M.; Banting, G. S.; Pym, B.; Wolfe, J.; Goodfellow, P. N.: Cloning an expressed gene shared by the human sex chromosomes. *Proc. Nat. Acad. Sci.* 83: 135–139, 1986.
- [39039] 12591.Dracopoli, N. C.; Rettig, W. J.; Albino, A. P.; Esposito, D.; Archidiacono, N.; Rocchi, M.; Siniscalco, M.; Old, L. J.: Genes controlling gp25/30 cell–surface molecules map to chromosomes X and Y and escape X–inactivation. *Am. J. Hum. Genet.* 37: 199–207, 1985.
- [39040] 12592.Gelin, C.; Aubrit, F.; Phalipon, A.; Raynal, B.; Cole, S.; Kaczorek, M.; Bernard, A.: The E2 antigen, a 32 kD glycoprotein involved in T–cell adhesion processes, is the MIC2 gene product. *EMBO J.* 8:3253–3259, 1989.
- [39041] 12593.Geller, R. L.; Shapiro, L. J.; Mohandas, T. K.: Fine mapping of the distal short arm of the human X chromo–

some using X/Y translocations. Am.J. Hum. Genet. 38: 884–890, 1986.

- [39042] 12594. Goodfellow, P.; Banting, G.; Sheer, D.; Ropers, H. H.; Caine, A.; Ferguson-Smith, M. A.; Povey, S.; Voss, R.: Genetic evidence that a Y-linked gene in man is homologous to a gene on the X chromosome. Nature 302:346–349, 1983.
- [39043] 12595. Goodfellow, P.; Pym, B.; Mohandas, T.; Shapiro, L. J.: The cell surface antigen locus, MIC2X, escapes X-inactivation. Am. J. Hum. Genet. 36: 777–782, 1984.
- [39044] 12596. Goodfellow, P. J.; Darling, S. M.; Thomas, N. S.; Goodfellow, P. N.: A pseudoautosomal gene in man. Science 234: 740–743, 1986.
- [39045] 12597. Goodfellow, P. N.; Tippet, P.: A human quantitative polymorphism related to Xg blood groups. Nature 289: 404–405, 1981.
- [39046] 12598. Levy, R.; Dilley, J.; Fox, R. I.; Warnke, R.: A human thymus-leukemia antigen defined by hybridoma monoclonal antibodies. Proc. Nat. Acad. Sci. 76: 6552–6556, 1979.
- [39047] 12599. Pettersen, R. D.; Bernard, G.; Olafsen, M. K.; Pourte, M.; Lie, S. O.: CD99 signals caspase-independent T cell death. J. Immun. 166:4931–4942, 2001.

- [39048] 12600.Ropers, H. H.; Zimmer, J.; Strobl, G.; Goodfellow, P.: The MIC2X(12E7) locus maps distally from STS on Xp. (Abstract) Cytogenet.Cell Genet. 40: 736 only, 1985.
- [39049] 12601.Simmler, M.-C.; Rouyer, F.; Vergnaud, G.; Nys-trom-Lahti, M.; Ngo,K. Y.; de la Chapelle, A.; Weissenbach, J.: Pseudoautosomal DNA sequences in the pairing region of the human sex chromosomes. Nature 317:692-697, 1985.
- [39050] 12602.Tippett, P.; Shaw, M.-A.; Green, C. A.; Daniels, G. L.: The 12E7red cell quantitative polymorphism: control by the Y-borne locus,Yg. Ann. Hum. Genet. 50: 339-347, 1986.
- [39051] 12603.Janz, R.; Sudhof, T. C.; Hammer, R. E.; Unni, V.; Siegelbaum, S.A.; Bolshakov, V. Y.: Essential roles in synaptic plasticity for synaptogyrin I and synaptophysin I. Neuron 24: 687-700, 1999.
- [39052] 12604.McMahon, H. T.; Bolshakov, V. Y.; Janz, R.; Hammer, R. E.; Siegelbaum,S. A.; Sudhof, T. C.: Synaptophysin, a major synaptic vesicle protein,is not essential for neurotransmitter release. Proc. Nat. Acad. Sci. 93:4760-4764, 1996.
- [39053] 12605.Brown, C. J.; Sekiguchi, T.; Nishimoto, T.; Willard, H. F.: Regional localization of CCG1 gene which comple-

ments hamster cell cycle mutationBN462 to Xq11–Xq13.

Somat. Cell Molec. Genet. 15: 93–96, 1989.

[39054] 12606.Derry, J. M. J.; Barnard, P. J.: Localization of the Ccg1 geneon the mouse X chromosome. (Abstract) Cyto-genet. Cell Genet. 51:988, 1989.

[39055] 12607.Dikstein, R.; Ruppert, S.; Tjian, R.: TAFII250 is a bi-partiteprotein kinase that phosphorylates the base tran-scription factor RAP74. Cell 84:781–790, 1996.

[39056] 12608.Giles, R. E.; Ruddle, F. H.: X-linkage of a human genetic locusthat corrects the DNA synthesis lesion in tsC1AGOH mouse cells. Genetics 93:975–996, 1979.

[39057] 12609.Hisatake, K.; Hasegawa, S.; Takada, R.; Nakatani, Y.; Horikoshi,M.; Roeder, R. G.: The p250 subunit of native TATA box-binding factorTFIID is the cell-cycle regulatory protein CCG1. Nature 362: 179–181,1993.

[39058] 12610.Jacobson, R. H.; Ladurner, A. G.; King, D. S.; Tjian, R.: Structureand function of a human TAFII250 double bromodomain module. Science 288:1422–1425, 2000.

[39059] 12611.Jha, K. K.; Ozer, H. L.: Genetic studies with a mu-tant mouse cell,ts–2 Balb–3T3, with a temperature–sensi-tive defect in DNA synthesis.(Abstract) Genetics 86: s32–s33, 1977.

[39060] 12612.Jha, K. K.; Siniscalco, M.; Ozer, H. L.: Temperature–

sensitivemutants of Balb-3T3 cells. III. Hybrids between ts2 and other mousemutant cells affected in DNA synthesis and correction of ts2 defectby human X chromosome. Somat. Cell Genet. 6: 603-614, 1980.

[39061] 12613.Almind, K.; Ahlgren, M. G.; Hansen, T.; Urhammer, S. A.; Clausen,J. O.; Pedersen, O.: Discovery of a Met300Val variant in Shc andstudies of its relationship to birth weight and length, impaired insulinsecretion, insulin resistance, and type 2 diabetes mellitus. J. Clin.Endocr. Metab. 84: 2241-2244, 1999.

[39062] 12614.Harun, R. B.; Smith, K. K.; Leek, J. P.; Markham, A. F.; Norris,A.; Morrison, J. F. J.: Characterization of human SHC p66 cDNA andits processed pseudogene mapping to Xq12-q13.1. Genomics 42: 349-352,1997.

[39063] 12615.McGlade, J.; Cheng, A.; Pelicci, G.; Pelicci, P. G.; Pawson, T.: Shc proteins are phosphorylated and regulated by the v-src and v-fpsprotein-tyrosine-kinases. Proc. Nat. Acad. Sci. 89: 8869-8873, 1992.

[39064] 12616.Migliaccio, E.; Giorgio, M.; Mele, S.; Pelicci, G.; Reboldi, P.;Pandolfi, P. P.; Lanfrancone, L.; Pelicci, P. G.: The p66(shc) adaptorprotein controls oxidative stress response and life span in mammals. Nature 402:309-313, 1999.

- [39065] 12617.Nemoto, S.; Finkel, T.: Redox regulation of fork-head proteins through a p66shc-dependent signaling pathway. *Science* 295: 2450–2452, 2002.
- [39066] 12618.Pelicci, G.; Lanfrancone, L.; Grignani, F.; McGlade, J.; Cavallo, F.; Forni, G.; Nicoletti, I.; Grignani, F.; Pawson, T.; Pelicci, P.G.: A novel transforming protein (SHC) with an SH2 domain is implicated in mitogenic signal transduction. *Cell* 70: 93–104, 1992.
- [39067] 12619.Yulug, I. G.; Egan, S. E.; See, C. G.; Fisher, E. M. C.: A human SHC-related sequence maps to chromosome 17, the SHC gene maps to chromosome 1. *Hum. Genet.* 96: 245–248, 1995.
- [39068] 12620.Zhang, L.; Camerini, V.; Bender, T. P.; Ravichandran, K. S.: A nonredundant role for the adapter protein Shc in thymic T cell development. *Nature Immunol.* 3: 749–755, 2002.
- [39069] 12621.Abramovitz, M.; Boie, Y.; Nguyen, T.; Rushmore, T. H.; Bayne, M.A.; Metters, K. M.; Slipetz, D. M.; Grygorczyk, R.: Cloning and expression of a cDNA for the human prostanoid FP receptor. *J. Biol. Chem.* 269:2632–2636, 1994.
- [39070] 12622.Betz, R.; Lagercrantz, J.; Kedra, D.; Dumanski, J. P.; Nordenskjöld, A.: Genomic structure, 5-prime flanking se-

quences, and precise localization in 1p31.1 of the human prostaglandin F receptor gene. *Biochem. Biophys. Res. Commun.* 254: 413–416, 1999.

- [39071] 12623. Sugimoto, Y.; Yamasaki, A.; Segi, E.; Tsuboi, K.; Aze, Y.; Nishimura, T.; Oida, H.; Yoshida, N.; Tanaka, T.; Katsuyama, M.; Hasumoto, K.; Murata, T.; Hirata, M.; Ushikubi, F.; Negishi, M.; Ichikawa, A.; Narumiya, S.: Failure of parturition in mice lacking the prostaglandin F receptor. *Science* 277:681–683, 1997.
- [39072] 12624. Geppert, M.; Khvotchev, M.; Krasnoperov, V.; Goda, Y.; Missler, M.; Hammer, R. E.; Ichtchenko, K.; Petrenko, A. G.; Sudhof, T. C.: Neurexin I- α is a major α -latrotoxin receptor that cooperates in α -latrotoxin action. *J. Biol. Chem.* 273: 1705–1710, 1998.
- [39073] 12625. Ichtchenko, K.; Hata, Y.; Nguyen, T.; Ullrich, B.; Missler, M.; Moomaw, C.; Sudhof, T. C.: Neuroligin 1: a splice site-specific ligand for beta-neurexins. *Cell* 81: 435–443, 1995.
- [39074] 12626. Kleiderlein, J. J.; Nisson, P. E.; Jessee, J.; Li, W.-B.; Becker, K. G.; Derby, M. L.; Ross, C. A.; Margolis, R. L.: CCG repeats in cDNAs from human brain. *Hum. Genet.* 103: 666–673, 1998. Note: Erratum: *Hum. Genet.* 104: 113 only, 1999.

- [39075] 12627.Apte, S. S.; Mattei, M.-G.; Olsen, B. R.: Mapping of human BAXgene to chromosome 19q13.3-q13.4 and isolation of a novel alternatively spliced transcript, BAX-delta. *Genomics* 26: 592-594, 1995.
- [39076] 12628.Niyonsaba, F.; Iwabuchi, K.; Someya, A.; Hirata, M.; Matsuda, H.; Ogawa, H.; Nagaoka, I.: A cathelicidin family of human antibacterial peptide LL-37 induces mast cell chemotaxis. *Immunology* 106: 20-26, 2002.
- [39077] 12629.Margolis, R.: Personal Communication. Baltimore, Md. 3/29/2000.
- [39078] 12630.Missler, M.; Sudhof, T. C.: Neurexins: three genes and 1001 products. *Trends Genet.* 14: 20-26, 1998.
- [39079] 12631.Ikeshima, H.; Imai, S.; Shimoda, K.; Hata, J.; Takano, T.: Expression of a MADS box gene, MEF2D, in neurons of the mouse central nervous system: implication of its binary function in myogenic and neurogenic cell lineages. *Neurosci. Lett.* 200: 117-120, 1995.
- [39080] 12632.Delhase, M.; Hayakawa, M.; Chen, Y.; Karin, M.: Positive and negative regulation of I-kappa-B kinase activity through IKK-beta subunit phosphorylation. *Science* 284:309-312, 1999.
- [39081] 12633.DiDonato, J. A.; Hayakawa, M.; Rothwarf, D. M.; Zandi, E.; Karin, M.: A cytokine-responsive I-kappaB kinase

that activates the transcriptionfactor NF-kappaB. Nature 388: 548-554, 1997.

[39082] 12634.Mercurio, F.; Zhu, H.; Murray, B. W.; Shevchenko, A.; Bennett,B. L.; Li, J.; Young, D. B.; Barbosa, M.; Mann, M.; Manning, A.; Rao,A.: IKK-1 and IKK-2: cytokine-activated I-kappa-B kinases essentialfor NF-kappa-B activation. Science 278: 860-866, 1997.

[39083] 12635.Matsushima, A.; Kaisho, T.; Rennert, P. D.; Nakano, H.; Kurosawa,K.; Uchida, D.; Takeda, K.; Akira, S.; Matsumoto, M.: Essential roleof nuclear factor (NF)-kappa-B-inducing kinase and inhibitor of kappa-B(I-kappa-B) kinase alpha in NF-kappa-B activation through lymphotoxinbeta receptor, but not through tumor necrosis factor receptor I. J.Exp. Med. 193: 631-636, 2001.

[39084] 12636.Brzezinski, A.: Melatonin in humans. New Eng. J. Med. 336: 186-195,1997.

[39085] 12637.Liu, R.-Y.; Zhou, J.-N.; van Heerikhuize, J.; Hofman, M. A.; Swaab,D. F.: Decreased melatonin levels in post-mortem cerebrospinal fluidin relation to aging, Alzheimer's disease, and apolipoprotein E-epsilon-4/4genotype. J. Clin. Endocr. Metab. 84: 323-327, 1999.

- [39086] 12638.Nelson, C. S.; Ikeda, M.; Gompf, H. S.; Robinson, M. L.; Fuchs,N. K.; Yoshioka, T.; Neve, K. A.; Allen, C. N.: Regulation of melatonin1a receptor signaling and traffick- ing by asparagine-124. *Molec. Endocr.* 15:1306-1317, 2001.
- [39087] 12639.Reppert, S. M.; Weaver, D. R.: Melatonin madness. *Cell* 83: 1059-1062,1995.
- [39088] 12640.Reppert, S. M.; Weaver, D. R.; Ebisawa, T.: Cloning and characterizationof a mammalian melatonin receptor that mediates reproductive and circadianresponses. *Neu- ron* 13: 1177-1185, 1994.
- [39089] 12641.Muller-Pillasch, F.; Zimmerhackl, F.; Lacher, U.; Schultz, N.;Hameister, H.; Varga, G.; Friess, H.; Buchler, M.; Adler, G.; Gress,T. M.: Cloning of novel transcripts of the human guanine-nucleotide-exchange factor Mss4: in situ chromosomal mapping and expression in pancreatic- cancer. *Genomics* 46: 389-396, 1997.
- [39090] 12642.Yu, H.; Schreiber, S. L.: Cloning, Zn(2+) binding, and structural characterization of the guanine nucleotide exchange factor human Mss4. *Biochemistry* 34:9103-9110, 1995.
- [39091] 12643.Yabe, D.; Nakamura, T.; Kanazawa, N.; Tashiro, K.; Honjo, T.:Calumenin, a Ca(2+)-binding protein retained in

the endoplasmic reticulum with a novel carboxyl-terminal sequence, HDEF. J. Biol. Chem. 272:18232–18239, 1997.

- [39092] 12644. Yabe, D.; Taniwaki, M.; Nakamura, T.; Kanazawa, N.; Tashiro, K.; Honjo, T.: Human calumenin gene (CALU): cDNA isolation and chromosomal mapping to 7q32. Genomics 49: 331–333, 1998.
- [39093] 12645. Ito, T.; Yang, M.; May, W. S.: RAX, a cellular activator for double-stranded RNA-dependent protein kinase during stress signaling. J. Biol. Chem. 274:15427–15432, 1999.
- [39094] 12646. Patel, R. C.; Sen, G. C.: PACT, a protein activator of the interferon-induced protein kinase, PKR. EMBO J. 17: 4379–4390, 1998.
- [39095] 12647. Scott, A. F.: Personal Communication. Baltimore, Md. 2001.
- [39096] 12648. Shearman, L. P.; Zylka, M. J.; Weaver, D. R.; Kolakowski, L. F., Jr.; Reppert, S. M.: Two period homologs: circadian expression and photic regulation in the suprachiasmatic nuclei. Neuron 19: 1261–1269, 1997.
- [39097] 12649. Toh, K. L.; Jones, C. R.; He, Y.; Eide, E. J.; Hinz, W. A.; Virshup, D. M.; Ptacek, L. J.; Fu, Y.-H.: An hPer2 phosphorylation site mutation in familial advanced sleep phase syndrome. Science 291: 1040–1043, 2001.

- [39098] 12650.Engelender, S.; Wanner, T.; Kleiderlein, J. J.; Wakabayashi, K.;Tsuji, S.; Takahashi, H.; Ashworth, R.; Margolis, R. L.; Ross, C.A.: Organization of the human synphilin-1 gene, a candidate for Parkinson'sdisease. *Mammalian Genome* 11: 763–766, 2000.
- [39099] 12651.Chien, W.; Pei, L.: A novel binding factor facilitates nucleartranslocation and transcriptional activation function of the pituitarytumor–transforming gene product. *J. Biol. Chem.* 275: 19422–19427,2000.
- [39100] 12652.Yaspo, M.–L.; Aaltonen, J.; Horelli–Kuitunen, N.; Peltonen, L.;Lehrach, H.: Cloning of a novel human putative type Ia integral membraneprotein mapping to 21q22.3. *Genomics* 49: 133–136, 1998.
- [39101] 12653.Yaspo, M.–L.; Gellen, L.; Mott, R.; Korn, B.; Nizetic, D.; Poustka,A. M.; Lehrach, H.: Model for a transcript map of human chromosome21: isolation of new coding sequences from exon and enriched cDNAlibraries. *Hum. Molec. Genet.* 4: 1291–1304, 1995.
- [39102] 12654.Ghiso, J. A.; Holton, J.; Miravalle, L.; Calero, M.; Lashley, T.;Vidal, R.; Houlden, H.; Wood, N.; Neubert, T. A.; Rostagno, A.; Plant,G.; Revesz, T.; Frangione, B.: Systemic amyloid deposits in familialBritish dementia. *J. Biol. Chem.* 276: 43909–43914, 2001.

- [39103] 12655. Stromgren, E.; Dalby, A.; Dalby, M.; Ranheim, B.: *Acta Neurol. Scand.* 46 (suppl. 43): 97–98, 1970.
- [39104] 12656. Gandhi, R.; Elble, R. C.; Gruber, A. D.; Schreur, K. D.; Ji, H.-L.; Fuller, C. M.; Pauli, B. U.: Molecular and functional characterization of a calcium-sensitive chloride channel from mouse lung. *J. Biol. Chem.* 273: 32096–32101, 1998.
- [39105] 12657. Gruber, A. D.; Elble, R. C.; Ji, H.-L.; Schreur, K. D.; Fuller, C. M.; Pauli, B. U.: Genomic cloning, molecular characterization, and functional analysis of human CLCA1, the first human member of the family of Ca^{2+} -activated Cl^- channel proteins. *Genomics* 54: 200–214, 1998.
- [39106] 12658. Gruber, A. D.; Schreur, K. D.; Ji, H.-L.; Fuller, C. M.; Pauli, B. U.: Molecular cloning and transmembrane structure of hCLCA2 from human lung, trachea, and mammary gland. *Am. J. Physiol.* 276: C1261–C1270, 1999.
- [39107] 12659. Liao, C.; Wang, X. Y.; Wei, H. Q.; Li, S. Q.; Merghoub, T.; Pandolfi, P. P.; Wolgemuth, D. J.: Altered myelopoiesis and the development of acute myeloid leukemia in transgenic mice overexpressing cyclin A1. *Proc. Nat. Acad. Sci.* 98: 6853–6858, 2001.
- [39108] 12660. Liu, D.; Matzuk, M. M.; Sung, W. K.; Guo, Q.; Wang, P.; Wolgemuth, D. J.: Cyclin A1 is required for meiosis in

the male mouse. *NatureGenet.* 20: 377–380, 1998.

[39109] 12661. Muller, C.; Yang, R.; Beck-von-Peccoz, L.; Idos, G.; Verbeek, W.; Koeffler, H. P.: Cloning of the cyclin A1 genomic structure and characterization of the promoter region: GC boxes are essential for cell cycle-regulated transcription of the cyclin A1 gene. *J. Biol. Chem.* 274: 11220–11228, 1999.

[39110] 12662. Albertsen, H. M.; Smith, S. A.; Mazoyer, S.; Fujimoto, E.; Stevens, J.; Williams, B.; Rodriguez, P.; Cropp, C. S.; Slijepcevic, P.; Carlson, M.; Robertson, M.; Bradley, P.; and 9 others: A physical map and candidate genes in the BRCA1 region on chromosome 17q12–21. *NatureGenet.* 7: 472–479, 1994.

[39111] 12663. Han, H.-J.; Sudo, K.; Inazawa, J.; Nakamura, Y.: Isolation and mapping of a human gene (RABL) encoding a small GTP-binding protein homologous to the Ras-related RAB gene. *Cytogenet. Cell Genet.* 73:137–139, 1996.

[39112] 12664. Csoka, A. B.; Scherer, S. W.; Stern, R.: Expression analysis of six paralogous human hyaluronidase genes clustered on chromosomes 3p21 and 7q31. *Genomics* 60: 356–361, 1999.

[39113] 12665. Heckel, D.; Comtesse, N.; Brass, N.; Blin, N.; Zang, K. D.; Meese, E.: Novel immunogenic antigen homologous

to hyaluronidase in meningioma. *Hum.Molec. Genet.* 7: 1859–1872, 1998.

- [39114] 12666.Dolganov, G. M.; Maser, R. S.; Novikov, A.; Tosto, L.; Chong, S.;Bressan, D. A.; Petrini, J. H. J.: Human Rad50 is physically associatedwith human Mre11: identification of a conserved multiprotein compleximplicated in recombinational DNA repair. *Molec. Cell Biol.* 16:4832–4841, 1996.
- [39115] 12667.Hopfner, K.–P.; Craig, L.; Moncalian, G.; Zinkel, R. A.; Usui,T.; Owen, B. A. L.; Karcher, A.; Henderson, B.; Bodmer, J.–L.; McMurray,C. T.; Carney, J. P.; Petrini, J. H. J.; Tainer, J. A.: The Rad50zinc–hook is a structure joining Mre11 complexes in DNA recombinationand repair. *Nature* 418: 562–566, 2002.
- [39116] 12668.Luo, G.; Yao, M. S.; Bender, C. F.; Mills, M.; Bladl, A. R.; Bradley,A.; Petrini, J. H. J.: Disruption of mRad50 causes embryonic stemcell lethality, abnormal embryonic development, and sensitivity toionizing radiation. *Proc. Nat. Acad. Sci.* 96: 7376–7381, 1999.
- [39117] 12669.Trujillo, K. M.; Yuan, S.–S. F.; Lee, E. Y.–H. P.; Sung, P.: Nucleaseactivities in a complex of human recombination and DNA repair factorsRad50, Mre11, and p95. *J. Biol. Chem.* 273: 21447–21450, 1998.

- [39118] 12670. Clark, J.; Lu, Y. J.; Sidhar, S. K.; Parker, C.; Gill, S.; Smedley, D.; Hamoudi, R.; Linehan, W.; Shipley, J.; Cooper, C.: Fusion of splicing factor genes PSF and NonO (p54nrb) to the TFE3 gene in papillary renal cell carcinoma. *Oncogene* 15: 2233–2239, 1997.
- [39119] 12671. Heimann, P.; El Housni, H.; Ogur, G.; Weterman, M. A. J.; Petty, E. M.; Vassart, G.: Fusion of a novel gene, RCC17, to the TFE3 gene in t(X;17)(p11.2;q25.3)-bearing papillary renal cell carcinomas. *Cancer Res.* 61: 4130–4135, 2001.
- [39120] 12672. Henthorn, P. S.; Stewart, C. C.; Kadesch, T.; Puck, J. M.: The gene encoding human TFE3, a transcription factor that binds the immunoglobulin heavy-chain enhancer, maps to Xp11.22. *Genomics* 11: 374–378, 1991.
- [39121] 12673. Joyama, S.; Ueda, T.; Shimizu, K.; Kudawara, I.; Mano, M.; Funai, H.; Takemura, K.; Yoshikawa, H.: Chromosome rearrangement at 17q25 and Xp11.2 in alveolar soft-part sarcoma: a case report and review of the literature. *Cancer* 86: 1246–1250, 1999.
- [39122] 12674. Ladanyi, M.; Lui, M. Y.; Antonescu, C. R.; Krause-Boehm, A.; Meindl, A.; Argani, P.; Healey, J. H.; Ueda, T.; Yoshikawa, H.; Meloni-Ehrig, A.; Sorensen, P. H. B.; Mertens, F.; Mandahl, N.; van den Berghe, H.; Sciort, R.; Dal

Cin, P.; Bridge, J.: The der(17)t(X;17)(p11;q25) of human alveolar soft part sarcoma fuses the TFE3 transcription factor gene to ASPL, a novel gene at 17q25. *Oncogene* 20: 48–57, 2001.

[39123] 12675. Macchi, P.; Notarangelo, L.; Giliani, S.; Strina, D.; Repetto, M.; Sacco, M. G.; Vezzoni, P.; Villa, A.: The genomic organization of the human transcription factor 3 (TFE3) gene. *Genomics* 28: 491–494, 1995.

[39124] 12676. Shipley, J. M.; Birdsall, S.; Clark, J.; Crew, J.; Gill, S.; Linehan, M.; Gnarr, J.; Gisher, S.; Craig, I. W.; Cooper, C. S.: Mapping the X chromosome breakpoint in two papillary renal cell carcinoma cell lines with a t(X;1)(p11.2;q21.2) and the first report of a female case. *Cytogenet. Cell. Genet.* 71: 280–284, 1995.

[39125] 12677. Sidhar, S. K.; Clark, J.; Gill, S.; Hamoudi, R.; Crew, A. J.; Gwilliam, R.; Ross, M.; Linehan, W. M.; Birdsall, S.; Shipley, J.; Cooper, C. S.: The t(X;1)(p11.2;q21.2) translocation in papillary renal cell carcinoma fuses a novel gene PRCC to the TFE3 transcription factor gene. *Hum. Molec. Genet.* 5: 1333–1338, 1996.

[39126] 12678. Larola, G.; Cuesta, R.; Brewer, G.; Schneider, R. J.: Control of mRNA decay by heat shock–ubiquitin–proteasome pathway. *Science* 284: 499–502, 1999.

- [39127] 12679.Deutsch, D.; Palmon, A.; Dafni, L.; Fisher, L.; Termine, J. D.;Young, M.: Cloning, sequencing, and characterization of tuftelin:a novel acidic enamel protein. *Connect. Tissue Res.* 27: 121 only,1992.
- [39128] 12680.Deiss, L. P.; Feinstein, E.; Berissi, H.; Cohen, O.; Kimchi, A.: Identification of a novel serine/threonine kinase and a novel 15-kDprotein as potential mediators of the gamma interferon-induced celldeath. *Genes Dev.* 9: 15-30, 1995.
- [39129] 12681.Feinstein, E.; Druck, T.; Kastury, K.; Berissi, H.; Goodart, S.A.; Overhauser, J.; Kimchi, A.; Huebner, K.: Assignment of DAP1 andDAPK: genes that positively mediate programmed cell death triggeredby IFN-gamma--to chromosome regions 5p12.2 (sic) and 9q34.1, respectively. *Genomics* 29:305-307, 1995.
- [39130] 12682.Fink, T. M.; Vaesen, M.; Kratzin, H. D.; Lichter, P.; Zimmer, M.: Localization of the gene encoding the putative human HLA class IIassociated protein (PHAPI) to chromosome 15q22.3-q23 by fluorescencein situ hybridization. *Genomics* 29: 309-310, 1995.
- [39131] 12683.Li, M.; Makkinje, A.; Damuni, Z.: Molecular identification ofl-1(PP2A), a novel potent heat-stable inhibitor protein of proteinphosphatase 2A. *Biochemistry* 35:

6998–7002, 1996.

- [39132] 12684. Matilla, A.; Koshy, B. T.; Cummings, C. J.; Isobe, T.; Orr, H. T.; Zoghbi, H. Y.: The cerebellar leucine-rich acidic nuclear protein interacts with ataxin-1. *Nature* 389: 974–978, 1997. Note: Erratum: *Nature* 391: 818 only, 1998.
- [39133] 12685. Sandberg, A. A.; Morgan, R.; McCallister, J. A.; Kaiser-McCaw, B.; Hecht, F.: Acute myeloblastic leukemia (AML) with t(6;9)(p23;q34): a specific subgroup of AML? *Cancer Genet. Cytogenet.* 10: 139–142, 1983.
- [39134] 12686. Vaesen, M.; Barnikol-Watanabe, S.; Gotz, H.; Awni, L. A.; Cole, T.; Zimmermann, B.; Kratzin, H. D.; Hilschmann, N.: Purification and characterization of two putative HLA class II associated proteins: PHAPI and PHAPII. *Biol. Chem. Hoppe-Seyler* 375: 113–126, 1994.
- [39135] 12687. von Lindern, M.; van Baal, S.; Wiegant, J.; Raap, A.; Hagemeijer, A.; Grosveld, G.: Can, a putative oncogene associated with myeloid leukemogenesis may be activated by fusion of its 3-prime half to different genes: characterization of the set gene. *Molec. Cell Biol.* 12: 3346–3355, 1992.
- [39136] 12688. Hughes, K. A.; Hurlstone, A. F. L.; Tobias, E. S.; McFarlane, R.; Black, D. M.: Absence of ST7 mutations in tu-

mor-derived cell lines and tumors. *Nature Genet.* 29: 380–381, 2001.

- [39137] 12689. Thomas, N. A.; Choong, D. Y. H.; Jokubaitis, V. J.; Neville, P. J.; Campbell, I. G.: Mutation of the ST7 tumor suppressor gene on 7q31.1 is rare in breast, ovarian and colorectal cancers. *Nature Genet.* 29: 379–380, 2001.
- [39138] 12690. Zenklusen, J. C.; Conti, C. J.; Green, E. D.: Mutational and functional analyses reveal that ST7 is a highly conserved tumor-suppressor gene on human chromosome 7q31. *Nature Genet.* 27: 392–398, 2001.
- [39139] 12691. Zenklusen, J. C.; Rodriguez, L. V.; LaCava, M.; Wang, Z.; Goldstein, L. S.; Conti, C. J.: Novel susceptibility locus for mouse hepatomas: evidence for a conserved tumor suppressor gene. *Genome Res.* 6: 1070–1076, 1996.
- [39140] 12692. Zenklusen, J. C.; Weitzel, J. N.; Ball, H. G.; Conti, C. J.: Allelic loss at 7q31.1 in human primary ovarian carcinomas suggests the existence of a tumor suppressor gene. *Oncogene* 11: 359–363, 1995.
- [39141] 12693. Lee, P. L.; Gelbart, T.; West, C.; Adams, M.; Blackstone, R.; Beutler, E.: Three genes encoding zinc finger proteins on human chromosome 6p21.3: members of a new subclass of the Kruppel gene family containing the conserved SCAN box domain. *Genomics* 43: 191–201,

1997.

- [39142] 12694.Tirosvoutis, K. N.; Divane, A.; Jones, M.; Affara, N. A.: Characterization of a novel zinc finger gene (ZNF165) mapping to 6p21 that is expressed specifically in testis. *Genomics* 28: 485–490, 1995.
- [39143] 12695.Deutsch, D.; Palmon, A.; Fisher, L. W.; Kolodny, N.; Termine, J.D.; Young, M. F.: Sequencing of bovine enamel-elin (tuftelin), a novel acidic enamel protein. *J. Biol. Chem.* 266: 16021–16028, 1991.
- [39144] 12696.Deutsch, D.; Palmon, A.; Young, M. F.; Selig, S.; Kearns, W. G.; Fisher, L. W.: Mapping of the human tuftelin (TUFT1) gene to chromosome 1 by fluorescence in situ hybridization. (Abstract) *Mammalian Genome* 5:461–462, 1994.
- [39145] 12697.MacDougall, M.; Simmons, D.; Dodds, A.; Knight, C.; Luan, X.; Zeichner-David, M.; Zhang, C.; Ryu, O. H.; Qian, Q.; Simmer, J. P.; Hu, C.-C.: Cloning, characterization, and tissue expression pattern of mouse tuftelin cDNA. *J. Dent. Res.* 77: 1970–1978, 1998.
- [39146] 12698.Dickeson, S. K.; Helmkamp, G. M., Jr.; Yarbrough, L. R.: Sequence of a human cDNA encoding phosphatidylinositol transfer protein and occurrence of a related sequence in widely divergent eukaryotes. *Gene*

142:301–305, 1994.

- [39147] 12699. Dickeson, S. K.; Lim, C. N.; Schuyler, G. T.; Dalton, T. P.; Helmkamp, G. M., Jr.; Yarbrough, L. R.: Isolation and sequence of cDNA clones encoding rat phosphatidylinositol transfer protein. *J. Biol. Chem.* 264:16557–16564, 1989.
- [39148] 12700. Hay, J. C.; Martin, T. F. J.: Phosphatidylinositol transfer protein required for ATP-dependent priming of Ca^{2+} -activated secretion. *Nature* 366:572–575, 1993.
- [39149] 12701. Vihtelic, T. S.; Goebel, M.; Milligan, S.; O'Tousa, J. E.; Hyde, D. R.: Localization of *Drosophila* retinal degeneration B, a membrane-associated phosphatidylinositol transfer protein. *J. Cell Biol.* 122: 1013–1022, 1993.
- [39150] 12702. Wirtz, K. W. A.: Phospholipid transfer proteins. *Annu. Rev. Biochem.* 60:73–99, 1991.
- [39151] 12703. Fitzgibbon, J.; Pilz, A.; Gayther, S.; Appukuttan, B.; Dulai, K. S.; Delhanty, J. D. A.; Helmkamp, G. M., Jr.; Yarbrough, L. R.; Hunt, D. M.: Localization of the gene encoding human phosphatidylinositol transfer protein (PITPN) to 17p13.3: a gene showing homology to the *Drosophila* retinal degeneration B gene (*rdgB*). *Cytogenet. Cell Genet.* 67:205–207, 1994.
- [39152] 12704. Hamilton, B. A.; Smith, D. J.; Mueller, K. L.; Kerre–

brock, A. W.;Bronson, R. T.; van Berkel, V.; Daly, M. J.; Kruglyak, L.; Reeve,M. P.; Nemhauser, J. L.; Hawkins, T. L.; Rubin, E. M.; Lander, E.S.: The vibrator mutation causes neurodegeneration via reduced expressionof PITP-alpha: positional complementation cloning and extragenic suppression. Neuron 18:711-722, 1997.

[39153] 12705.Hara, T.; Yamauchi, M.; Takahashi, E.; Hoshino, M.; Aoki, K.; Ayusawa,D.; Kawakita, M.: The UDP-galactose translocator gene is mapped toband Xp11.23-p11.22 containing the Wiskott-Aldrich syndrome locus. Somat.Cell Molec. Genet. 19: 571-575, 1993.

[39154] 12706.Ishida, N.; Miura, N.; Yoshioka, S.; Kawakita, M.: Molecular cloningand characterization of a novel isoform of the human UDP-galactosetransporter, and of related complementary DNAs belonging to the nucleotide-sugartransporter gene family. J. Biochem. 120: 1074-1078, 1996.

[39155] 12707.Miura, N.; Ishida, N.; Hoshino, M.; Yamauchi, M.; Hara, T.; Ayusawa,D.; Kawakita, M.: Human UDP-galactose translocator: molecular cloningof a complementary DNA that complements the genetic defect of a mutantcell line deficient in UDP-galactose translocator. J. Biochem. 120:236-241, 1996.

- [39156] 12708.Lund, A.; Udd, B.; Juvonen, V.; Andersen, P. M.; Cederquist, K.; Davis, M.; Gellera, C.; Kolmel, C.; Ronnevi, L.-O.; Sperfeld, A.-D.; Sorensen, S.-A.; Tranebjaerg, L.; Van Maldergem, L.; Watanabe, M.; Weber, M.; Yeung, L.; Savontaus, M.-L.: Multiple founder effects in spinal and bulbar muscular atrophy (SBMA, Kennedy disease) around the world. *Europ. J. Hum. Genet.* 9: 431–436, 2001.
- [39157] 12709.Edelhoff, S.; Ayer, D. E.; Zervos, A. S.; Steingrims-son, E.; Jenkins, N. A.; Copeland, N. G.; Eisenman, R. N.; Brent, R.; Distech, C. M.: Mapping of two genes encoding members of a distinct subfamily of MAX interacting proteins: MAD to human chromosome 2 and mouse chromosome 6, and MXI1 to chromosome 10 and mouse chromosome 19. *Oncogene* 9:665–668, 1994.
- [39158] 12710.Ferguson, G. D.; Anagnostaras, S. G.; Silva, A. J.; Herschman, H. R.: Deficits in memory and motor performance in synaptotagmin IV mutant mice. *Proc. Nat. Acad. Sci.* 97: 5598–5603, 2000.
- [39159] 12711.Ferguson, G. D.; Chen, X.-N.; Korenberg, J. R.; Herschman, H. R.: The human synaptotagmin IV gene defines an evolutionary break point between syntenic mouse and human chromosome regions but retains ligand inducibility

and tissue specificity. J. Biol. Chem. 275:
36920–36926, 2000.

- [39160] 12712. Holterhus, P.-M.; Wiebel, J.; Sinnecker, G. H. G.; Bruggenwirth, H. T.; Sippell, W. G.; Brinkmann, A. O.; Kruse, K.; Hiort, O.: Clinical and molecular spectrum of somatic mosaicism in androgen insensitivity syndrome. *Pediatr. Res.* 46: 684–690, 1999.
- [39161] 12713. Foresta, C.; Bertella, A.; Moro, E.; Roverato, A.; Merico, M.; Ferlin, A.: Sertoli cell function in infertile patients with and without microdeletions of the azoospermia factors on the Y chromosome long arm. *J. Clin. Endocr. Metab.* 86: 2414–2419, 2001.
- [39162] 12714. Kobayashi, K.; Mizuno, K.; Hida, A.; Komaki, R.; Tomita, K.; Matsushita, I.; Namiki, M.; Iwamoto, T.; Tamura, S.; Minowada, S.; Nakahori, Y.; Nakagome, Y.: PCR analysis of the Y chromosome long arm in azoospermic patients: evidence for a second locus required for spermatogenesis. *Hum. Molec. Genet.* 3: 1965–1967, 1994.
- [39163] 12715. Agulnik, A. I.; Zharkikh, A.; Boettger-Tong, H.; Bourgeron, T.; McElreavey, K.; Bishop, C. E.: Evolution of the DAZ gene family suggests that Y-linked DAZ plays little, or a limited, role in spermatogenesis but underlines a recent African origin for human populations. *Hum. Molec.*

Genet. 7: 1371–1377, 1998.

[39164] 12716. Anonymous: A missing piece on the Y. (Editorial) Nature Genet. 10:367–368, 1995.

[39165] 12717. Cooke, H. J.; Lee, M.; Kerr, S.; Ruggiu, M.: A murine homologue of the human DAZ gene is autosomal and expressed only in male and female gonads. Hum. Molec. Genet. 5: 513–516, 1996.

[39166] 12718. Foresta, C.; Moro, E.; Garolla, A.; Onisto, M.; Ferlin, A.: Y chromosome microdeletions in cryptorchidism and idiopathic infertility. J. Clin. Endocr. Metab. 84: 3660–3665, 1999.

[39167] 12719. Makova, K. D.; Li, W.-H.: Strong male-driven evolution of DNA sequences in humans and apes. Nature 416: 624–626, 2002.

[39168] 12720. Menke, D. B.; Mutter, G. L.; Page, D. C.: Expression of DAZ, an azoospermia factor candidate, in human spermatogonia. (Letter) Am. J. Hum. Genet. 60: 237–241, 1997.

[39169] 12721. Moro, E.; Ferlin, A.; Yen, P. H.; Franchi, P. G.; Palka, G.; Foresta, C.: Male infertility caused by a de novo partial deletion of the DAZ cluster on the Y chromosome. J. Clin. Endocr. Metab. 85: 4069–4073, 2000.

[39170] 12722. Reijo, R.; Lee, T.-Y.; Salo, P.; Alagappan, R.; Brown,

L. G.; Rosenberg, M.; Rozen, S.; Jaffe, T.; Straus, D.; Hovatta, O.; de laChapelle, A.; Silber, S.; Page, D. C.: Diverse spermatogenic defects in humans caused by Y chromosome deletions encompassing a novel RNA-binding protein gene. *Nature Genet.* 10: 383–393, 1995.

[39171] 12723. Reijo, R.; Seligman, J.; Dinulos, M. B.; Jaffe, T.; Brown, L. G.; Disteche, C. M.; Page, D. C.: Mouse autosomal homolog of DAZ, a candidate male sterility gene in humans, is expressed in male germ cells before and after puberty. *Genomics* 35: 346–352, 1996.

[39172] 12724. Saxena, R.; Brown, L. G.; Hawkins, T.; Alagappan, R. K.; Skaletsky, H.; Reeve, M. P.; Reijo, R.; Rozen, S.; Dinulos, M. B.; Disteche, C. M.; Page, D. C.: The DAZ gene cluster on the human Y chromosome arose from an autosomal gene that was transposed, repeatedly amplified and pruned. *Nature Genet.* 14: 292–299, 1996.

[39173] 12725. Najmabadi, H.; Huang, V.; Yen, P.; Subbarao, M. N.; Bhasin, D.; Banaag, L.; Naseeruddin, S.; de Kretser, D. M.; Baker, H. W. G.; McLachlan, R. I.; Loveland, K. A.; Bhasin, S.: Substantial prevalence of microdeletions of the Y-chromosome in infertile men with idiopathic azoospermia and oligozoospermia detected using a sequence-tagged site-based mapping strategy. *J. Clin. Endocr. Metab.* 81:

1347–1352, 1996.

- [39174] 12726.Wang, G.; Sawai, N.; Kotliarova, S.; Kanazawa, I.; Nukina, N.:Ataxin-3, the MJD1 gene product, interacts with the two human homologsof yeast DNA repair protein RAD23, HHR23A and HHR23B. *Hum. Molec.Genet.* 9: 1795–1803, 2000.
- [39175] 12727.Ng, J. M. Y.; Vrieling, H.; Sugasawa, K.; Ooms, M. P.; Grootegoed,J. A.; Vreeburg, J. T. M.; Visser, P.; Beems, R. B.; Gorgels, T. G.M. F.; Hanaoka, F.; Hoeijmakers, J. H. J.; van der Horst, G. T. J.: Developmental defects and male sterility in mice lacking the ubiquitin-likeDNA repair gene mHR23B. *Molec. Cell. Biol.* 22: 1233–1245, 2002.
- [39176] 12728.Volker, M.; Mone, M. J.; Karmakar, P.; van Hoffen, A.; Schul, W.;Vermeulen, W.; Hoeijmakers, J. H. J.; van Driel, R.; van Zeeland,A. A.; Mullenders, L. H. F.: Sequential assembly of the nucleotideexcision repair factors in vivo. *Molec. Cell* 8: 213–224, 2001.
- [39177] 12729.Abramson, J. S.; Mills, E. L.; Sawyer, M. K.; Regelman, W. R.;Nelson, J. D.; Quie, P. G.: Recurrent infections and delayed separationof the umbilical cord in an infant with abnormal phagocytic cell locomotionand oxidative response during particle phagocytosis. *J. Pediat.* 99:887–894, 1981.

- [39178] 12730.Allende, L. M.; Hernandez, M.; Corell, A.; Garcia-Perez, M. A.;Varela, P.; Moreno, A.; Caragol, I.; Garcia-Martin, F.; Guillen-Perales,J.; Olive, T.; Espanol, T.; Arnaiz-Villena, A.: A novel CD18 genomicdeletion in a patient with severe leucocyte adhesion deficiency: apossible CD2/lymphocyte function-associated antigen-1 functional associationin humans. *Immunology* 99: 440-450, 2000.
- [39179] 12731.Anderson, D. C.; Springer, T. A.: Leukocyte adhesion deficiency:an inherited defect in the Mac-1, LFA-1, and p150,95 glycoproteins. *Annu.Rev. Med.* 38: 175-194, 1987.
- [39180] 12732.Arnaout, M. A.; Dana, N.; Gupta, S. K.; Tenen, D. G.; Fathallah,D. M.: Point mutations impairing cell surface expression of the commonbeta subunit (CD18) in a patient with leukocyte adhesion molecule(Leu-CAM) deficiency. *J. Clin. Invest.* 85: 977-981, 1990.
- [39181] 12733.Arnaout, M. A.; Pitt, J.; Cohen, H. J.; Melamed, J.; Rosen, F.S.; Colten, H. R.: Deficiency of a granulocyte-membrane glycoprotein(gp150) in a boy with recurrent bacterial infections. *New Eng. J.Med.* 306: 693-699, 1982.
- [39182] 12734.Arnaout, M. A.; Spits, H.; Terhorst, C.; Pitt, J.; Todd, R. F.,III: Deficiency of a leukocyte surface glycoprotein (LFA-1) in twopatients with Mo1 deficiency: effects of cell

activation on Mo1/LFA-1 surface expression in normal and deficient leukocytes. *J. Clin. Invest.* 74:1291-1300, 1984.

[39183] 12735. Anderson, D. C.; Schmalstieg, F. C.; Finegold, M. J.; Hughes, B.J.; Rothlein, R.; Miller, L. J.; Kohl, S.; Tosi, M. F.; Jacobs, R.L.; Waldrop, T. C.; Goldman, A. S.; Shearer, W. T.; Springer, T. A.: The severe and moderate phenotypes of heritable Mac-1, LFA-1 deficiency: their quantitative definition and relation to leukocyte dysfunction and clinical features. *J. Infect. Dis.* 152: 668-689, 1985.

[39184] 12736. Breviario, F.; d'Aniello, E. M.; Golay, J.; Peri, G.; Bottazzi, B.; Bairoch, A.; Saccone, S.; Marzella, R.; Predazzi, V.; Rocchi, M.; Della Valle, G.; Dejana, E.; Mantovani, A.; Introna, M.: Interleukin-1-inducible genes in endothelial cells: cloning of a new gene related to C-reactive protein and serum amyloid P component. *J. Biol. Chem.* 267: 22190-22197, 1992.

[39185] 12737. Lee, T. H.; Lee, G. W.; Ziff, E. B.; Vilcek, J.: Isolation and characterization of eight tumor necrosis factor-induced gene sequences from human fibroblasts. *Molec. Cell. Biol.* 10: 1982-1988, 1990.

[39186] 12738. Pallini, R.; Guazzi, G. C.; Cannella, C.; Cacace, M. G.: Cloning and sequence analysis of the human liver rhodanese: comparison with the bovine and chicken enzymes.

Biochem. Biophys. Res. Commun. 180:887–893, 1991.

- [39187] 12739.Greco, A.; Mariani, C.; Miranda, C.; Lupas, A.; Pagliardini, S.; Pomati, M.; Pierotti, M. A.: The DNA rearrangement that generates the TRK-T3 oncogene involves a novel gene on chromosome 3 whose product has a potential coiled-coil domain. *Molec. Cell Biol.* 15: 6118–6127, 1995.
- [39188] 12740.Mencinger, M.; Panagopoulos, I.; Andreasson, P.; Lassen, C.; Mitelman, F.; Aman, P.: Characterization and chromosomal mapping of the human TFG gene involved in thyroid carcinoma. *Genomics* 41: 327–331, 1997.
- [39189] 12741.Kramps, T.; Peter, O.; Brunner, E.; Nellen, D.; Froesch, B.; Chatterjee, S.; Murone, M.; Zullig, S.; Basler, K.: Wnt/Wingless signaling requires BCL9/legless-mediated recruitment of pygopus to the nuclear beta-catenin-TCF complex. *Cell* 109: 47–60, 2002.
- [39190] 12742.Willis, T. G.; Zalcberg, I. R.; Coignet, L. J. A.; Wlodarska, M.; Stul, D. M.; Jadayel, D. M.; Bastard, C.; Treleaven, J. G.; Catovsky, D.; Silva, M. L. M.; Dyer, M. J. S.: Molecular cloning of translocation t(1;14)(q21;q32) defines a novel gene (BCL9) at chromosome 1q21. *Blood* 91:1873–1881, 1998.
- [39191] 12743.Kim, D.-H.; Iijima, H.; Goto, K.; Sakai, J.; Ishii, H.;

Kim, H.-J.; Suzuki, H.; Kondo, H.; Saeki, S.; Yamamoto, T.: Human apolipoprotein E receptor 2: a novel lipoprotein receptor of the low density lipoprotein receptor family predominantly expressed in brain. *J. Biol. Chem.* 271:8373–8380, 1996.

[39192] 12744. Kim, D.-H.; Magoori, K.; Inoue, T. R.; Mao, C. C.; Kim, H.-J.; Suzuki, H.; Fujita, T.; Endo, Y.; Saeki, S.; Yamamoto, T. T.: Exon/intron organization, chromosome localization, alternative splicing, and transcription units of the human apolipoprotein E receptor 2 gene. *J. Biol. Chem.* 272:8498–8504, 1997.

[39193] 12745. Davidson, R. G.; Nitowsky, H. M.; Childs, B.: Demonstration of two populations of cells in the human female heterozygous for glucose-6-phosphate dehydrogenase variants. *Proc. Nat. Acad. Sci.* 50: 481–485, 1963.

[39194] 12746. De Flora, A.; Morelli, A.; Benatti, U.; Giuntini, P.; Ferraris, A. M.; Galiano, S.; Ravazzolo, R.; Gaetani, G. F.: G6PD Napoli and Ferrara II: two new glucose-6-phosphate dehydrogenase variants having similar characteristics but different intracellular lability and specific activity. *Brit. J. Haemat.* 48: 417–423, 1981.

[39195] 12747. Dern, R. J.; McCurdy, P. R.; Yoshida, A.: A new

structural variant of glucose-6-phosphate dehydrogenase with a high production rate (G6PD Hektoen). J. Lab. Clin. Med. 73: 283-290, 1969.

- [39196] 12748. De Vita, G.; Alcalay, M.; Sampietro, M.; Cappelini, M. D.; Fiorelli, G.; Toniolo, D.: Two point mutations are responsible for G6PD polymorphism in Sardinia. Am. J. Hum. Genet. 44: 233-240, 1989.
- [39197] 12749. Du, C.; Xu, Y.; Hua, X.; Wu, Q.; Liu, L.; Wu, M.: Studies on erythrocyte glucose-6-phosphate dehydrogenase variants in Chinese. III: Gd(-) Miaozi-Baisha. Acta Genet. Sinica 11(2): 153-158, 1984.
- [39198] 12750. Du, C.-S.; Hua, X.-Y.; Wu, Q.-L.; Li, C.-Q.; Zheng, J.-F.; Li, H.-L.: Studies on erythrocyte glucose-6-phosphate dehydrogenase variants in Chinese. IV. Gd(-) Gaohe associated with paroxysmal nocturnal hemoglobinuria. Chinese J. Pathophysiol. 1: 12-15, 1985.
- [39199] 12751. Du, C. S.; Xu, Y. K.; Hua, X. Y.; Wu, Q. L.; Liu, L. B.: Glucose-6-phosphate dehydrogenase variants and their frequency in Guangdong, China. Hum. Genet. 80: 385-388, 1988.
- [39200] 12752. Eber, S. W.; Gahr, M.; Schroter, W.: Glucose-6-phosphate dehydrogenase (G6PD) Iserlohn and G6PD Regensburg: two new severe enzyme defects in German

families. Blut 51: 109–115, 1985.

- [39201] 12753. Echard, G.; Gillois, M.:
G6PD--alpha-GAL-PGK--HPRT synteny in the rabbit,
Oryctolagus cuniculus. (Abstract) Cytogenet. Cell Genet.
25:148–149, 1979.
- [39202] 12754. Elizondo, J.; Saenz, G. F.; Paez, C. A.; Ramon, M.;
Garcia, M.; Gutierrez, A.; Estrada, M.: G6PD Puerto Limon:
a new deficient variant of glucose-6-phosphate dehydro-
genase associated with congenital nonsphero-
cytic hemolytic anemia. Hum. Genet. 62: 110–112, 1982.
- [39203] 12755. Engstrom, P. F.; Beutler, E.: G-6-PD Tripler: a
unique variant associated with chronic hemolytic disease.
Blood 36: 10–13, 1970.
- [39204] 12756. Epstein, C. J.: Mammalian oocytes: X-chromosome
activity. Science 163:1078–1079, 1969.
- [39205] 12757. Ermakov, N. V.; Chernyak, N. B.; Tokarev, Y. N.:
Properties of new variant of glucose-6-phosphate dehy-
drogenase (Regar variant): glucose metabolism in erythro-
cytes containing abnormal enzyme. Biokhimiia
48:577–583, 1983.
- [39206] 12758. Estrada, M.; Garcia, M.; Gutierrez, A.; Quintero, I.;
Gonzalez, R.: G6PD Varadero. Vox Sang. 43: 102–104,
1982.

- [39207] 12759.Fairbanks, V. F.; Nepo, A. G.; Beutler, E.; Dickson, E. R.; Honig,G.: Glucose-6-phosphate dehydrogenase variants: reexamination ofG6PD Chicago and Cornell and a new variant (G6PD Pea Ridge) resemblingG6PD Chicago. Blood 55: 216-220, 1980.
- [39208] 12760.Faust, C. J.; Levinson, B.; Gitschier, J.; Herman, G. E.: Extensionof the physical map in the region of the mouse X chromosome homologousto human Xq28 and identification of an exception to conserved linkage. Genomics 13:1289-1295, 1992.
- [39209] 12761.Feldman, R.; Gromisch, D. S.; Luhby, A. L.; Beutler, E.: Congenitalnonspherocytic hemolytic anemia due to glucose-6-phosphate dehydrogenaseEast Harlem: a new deficient variant. J. Pediat. 90: 89-91, 1977.
- [39210] 12762.Fernandez, M.; Fairbanks, V. F.: Glucose-6-phosphate dehydrogenasedeficiency in the Philippines: report of a new variant--G6PD Panay. MayoClin. Proc. 43: 645-660, 1968.
- [39211] 12763.Ferraris, A. M.; Broccia, G.; Meloni, T.; Forteleoni, G.; Gaetani,G. F.: Glucose-6-phosphate dehydrogenase deficiency and incidenceof hematologic malignancy. Am. J. Hum. Genet. 42: 516-520, 1988.100. Filosa, E.: Personal Communication. La Jolla, Calif. 1989.101. Filosa, S.; Cal-

abro, V.; Vallone, D.; Poggi, V.; Mason, P.; Pagnini, D.;
 Alfinito, F.; Rotoli, B.; Martini, G.; Luzzatto, L.; Battis-
 tuzzi, G.: Molecular basis of chronic non-spherocytic
 haemolytic anaemia: a new G6PD variant (393arg-to-his)
 with abnormal K(m) G6P and marked in vivo instability.
 Brit. J. Haemat. 80: 111–116, 1992.102. Filosa, S.; Gia-
 cometti, N.; Wangwei, C.; De Mattia, D.; Pagnini, D.;
 Alfinito, F.; Schettini, F.; Luzzatto, L.; Martini, G.: Somatic-
 cell selection is a major determinant of the blood-cell phe-
 notype in heterozygotes for glucose-6-phosphate dehy-
 drogenase mutations causing severe enzyme deficiency.
 Am. J. Hum. Genet. 59: 887–895, 1996.103. Fiorelli, G.;
 Manoussakis, C.; Sampietro, M.; Pittalis, S.; Guglielmino, C.
 R.; Cappellini, M. D.: Different polymorphic variants of
 glucose-6-phosphate dehydrogenase (G6PD) in Italy. Ann.
 Hum. Genet. 53: 229–236, 1989.104. Fite, E.; Morell, F.;
 Zuazu, J.; Julia, A.; Morera, J.: Leucocyte glucose-
 6-phosphate dehydrogenase deficiency and necrotizing
 pneumonia. Europ. J. Resp. Dis. 64: 150–154, 1983.105.
 Francke, U.; Bakay, B.; Connor, J. D.; Coldwell, J. G.; Ny-
 han, W. L.: Linkage relationships of X-linked enzymes glu-
 cose-6-phosphate dehydrogenase and hypoxanthine gua-
 nine phosphoribosyltransferase. Am. J. Hum. Genet. 26:

512–522, 1974.106. Friedman, M. J.; Trager, W.: The biochemistry of resistance to malaria. *Sci. Am.* 244(3): 154–164, 1981.107. Frigerio, R.; Sole, G.; Olla, N.; Lovicu, M.; Passiu, G.; Carcassi, U.: Cagliari II: a new G-6-PD variant. *Haematologica* 72: 241–243, 1987.108. Fujii, H.; Miwa, S.; Takegawa, S.; Takahashi, K.; Hirono, A.; Takizawa, T.; Morisaki, T.; Kanno, H.; Taguchi, T.; Okamura, J.: Gd(-) Gifu and Gd(-) Fukuoka: two new variants of glucose-6-phosphate dehydrogenase found in Japan. *Hum. Genet.* 66: 276–278, 1984.109. Fujii, H.; Miwa, S.; Tani, K.; Takegawa, S.; Fujinami, N.; Takahashi, K.; Nakayama, S.; Konno, M.; Sato, T.: Glucose 6-phosphate dehydrogenase variants: a unique variant (G6PD Kobe) showed an extremely increased affinity for galactose 6-phosphate and a new variant (G6PD Sapporo) resembling G6PD Pea Ridge. *Hum. Genet.* 58: 405–407, 1981.110. Gaetani, G. F.; Galiano, S.; Melani, C.; Miglino, M.; Forni, G. L.; Napoli, G.; Perrone, L.; Ferraris, A. M.: A new glucose-6-phosphate dehydrogenase variant with congenital nonspherocytic hemolytic anemia (G6PD Genova): biochemical characterization and mosaicism expression in the heterozygote. *Hum. Genet.* 84: 337–340, 1990.111. Gahr, M.; Bornhalm, D.; Schroeter, W.: Bio-

chemische Eigenschafteneiner neuen Variante des Glucose-6-phosphadehydrogenase (G6PD) Mangelsmit Favis-mus: G6PD Bielefeld. *Klin. Wschr.* 55: 379–384, 1977.112.

Gahr, M.; Bornhalm, D.; Schroeter, W.: Haemolytic anemia dueto glucose 6-phosphate dehydrogenase (G6PD) defi-ciency: demonstrationof two new biochemical variants, G6PD Hamm and G6PD Tarsus. *Brit.J. Haemat.* 33: 363–370, 1976.113.

Gahr, M.; Schroeter, W.: Glucose 6-phosphate dehydrogenase (G6PD)Hamburg, a new vari-ant with chronic nonspherocytic hemolytic anemia. *Eu-rop.J. Clin. Invest.* 4: 187–191, 1974.114.

Gahr, M.; Schroeter, W.; Sturzenegger, M.; Bornhalm, D.; Marti,H. R.: Glucose 6-phosphate dehydrogenase (G6PD) deficiency in Switzerland. *Helv.Paediat. Acta* 31: 159–166, 1976.115.

Geerdink, R. A.; Horst, R.; Staal, G. E.: An Iraqi Jewish fam-ilywith a new red cell glucose 6-phosphate dehydroge-nase variant (Gd-Bagdad)and kernicterus. *Israel J. Med. Sci.* 9: 1040–1043, 1973.116.

Gellin, J.; Benne, F.; Renard, C.; Vaiman, M.; Hors-Cayla, M.C.; Gillois, M.: Pig gene mapping: syntenry, attempt to assign thehistocompatibility complex (SLA). (Abstract) *Cytogenet. Cell Genet.* 25:159, 1979.117.

Gomez-Gallego, F.; Garrido-Pertierra, A.; Bautista, J. M.: Structuraldefects underlying protein dys-

function in human glucose-6-phosphatedehydrogenase A- deficiency. *J. Biol. Chem.* 275: 9256-9262, 2000.118.

Gonzalez, R.; Estrada, M.; Garcia, M.; Gutierrez, A.: G6PD Ciudad de la Habana: a new slow variant with deficiency found in a Cuban family. *Hum. Genet.* 55: 133-135, 1980.119.

Gonzalez, R.; Wade, M.; Estrada, M.; Svach, E.; Colombo, B.: G6PD Pinar del Rio: a new variant discovered in a Cuban family. *Biochem. Genet.* 15: 909-913, 1977.120.

Goss, S. J.; Harris, H.: Gene transfer by means of cell fusion. I. Statistical mapping of the human X-chromosome by analysis of radiation induced gene segregation. *J. Cell Sci.* 25: 17-37, 1977.121.

Gourdin, D.; Vergnes, H.; Bouloux, C.; Ruffie, J.; Gherardi, M.: Polymorphism of erythrocyte G6PD in the baboon. *Am. J. Phys. Anthropol.* 37: 281-288, 1972.122.

Gray, G. R.; Stamatoyanopoulos, G.; Naiman, S. C.; Kliman, M. R.; Klebanoff, S. J.; Austin, T.; Yoshida, A.; Robinson, G. C. G.: Neutrophil dysfunction, chronic granulomatous disease, and non-spherocytic haemolytic anaemia caused by complete deficiency of glucose-6-phosphatedehydrogenase. *Lancet* II: 530-534, 1973.123.

Grossman, A.; Ramanathan, K.; Justice, P.; Gordon, J.; Shahidi, N. T.; Hsia, D. Y. Y.: Congenital nonspherocytic hemolytic anemia associated

with erythrocyte G-6-PD deficiency in a Negro family. Pediatrics 37:624-629, 1966.124. Gutierrez, A.; Garcia, M.; Estrada, M.; Quintero, I.; Gonzalez, R.: Glucose-6-phosphate dehydrogenase (G6PD) Guantnamo and G6PDCaujeri: two new glucose-6-phosphate dehydrogenase-deficient variants found in Cuba. Biochem. Genet. 25: 231-238, 1987.125. Hall, K.; Schreeder, M. T.; Prchal, J. T.: G6PD Huntsville: a new glucose-6-phosphate dehydrogenase associated with chronic hemolytic anemia. Hum. Genet. 79: 90-91, 1988.126. Harkonen, M.; Vuopio, P.: Red cell glucose-6-phosphate dehydrogenase deficiency in Finland. Ann. Clin. Res. 6: 187-197, 1974.127. Harley, J. D.; Agar, N. S.; Yoshida, A.: Glucose 6-phosphate dehydrogenase variant Gd(+) Alexandra associated with neonatal jaundice and Gd(-) Camperdown in a young man with lamellar cataracts. J. Lab. Clin. Med. 91: 295-300, 1978.128. Helge, H.; Borner, K.: Kongenitale nicht-sphaerozytare haemolytische Anaemie, Katarakt und Glucose-6-phosphat-dehydrogenase-mangel. Dtsch. Med. Wschr. 91: 1584-1589, 1966.129. Hirono, A.; Beutler, E.: Alternative splicing of human glucose-6-phosphate dehydrogenase messenger RNA in different tissues. J. Clin. Invest. 83:343-346, 1989.130. Hirono, A.;

Beutler, E.: Molecular cloning and nucleotide sequence of cDNA for human glucose-6-phosphate dehydrogenase variant A(-). *Proc. Nat. Acad. Sci.* 85: 3951-3954, 1988.131. Hirono, A.; Fujii, H.; Shima, M.; Miwa, S.: G6PD Nara: a new class 1 glucose-6-phosphate dehydrogenase variant with an eight amino acid deletion. *Blood* 82: 3250-3252, 1993.132. Hirono, A.; Kawate, K.; Honda, A.; Fujii, H.; Miwa, S.: A single mutation 202G-A in the human glucose-6-phosphate dehydrogenase gene (G6PD) can cause acute hemolysis by itself. (Letter) *Blood* 99: 1498 only, 2002.133. Hirono, A.; Kuhl, W.; Gelbart, T.; Forman, L.; Fairbanks, V.F.; Beutler, E.: Identification of the binding domain for NADP(+) of human glucose-6-phosphate dehydrogenase by sequence analysis of mutants. *Proc. Nat. Acad. Sci.* 86: 10015-10017, 1989.134. Hitzeroth, H. W.; Bender, K.: Age-dependency of somatic selection in South African Negro G-6-PD heterozygotes. *Hum. Genet.* 58: 338-343, 1981.135. Honig, G. R.; Habaccon, E.; Vida, L. N.; Matsumoto, F.; Beutler, E.: Three new variants of glucose-6-phosphate dehydrogenase associated with chronic nonspherocytic hemolytic anemia: G6PD Lincoln Park, G6PD Arlington Heights, and G6PD West Town. *Am. J. Hemat.* 6: 353-360, 1979.136. Hook, E. B.;

Stamatoyannopoulos, G.; Yoshida, A.; Motulsky, A.G.: Glucose-6-phosphate dehydrogenase Madrona: a slow electrophoretic glucose-6-phosphate dehydrogenase variant with kinetic characteristics similar to those of normal type. *J. Lab. Clin. Med.* 72: 404-409, 1968. 137.

Hors-Cayla, M. C.; Heuertz, S.; Van Cong, N.; Benne, F.: Cattle gene mapping by somatic cell hybridization. (Abstract) *Cytogenet. Cell Genet.* 25: 165-166, 1979. 138.

Howell, E. B.; Nelson, A. J.; Jones, O. W.: A new G-6-PD variant associated with chronic non-spherocytic haemolytic anaemia in a Negro family. *J. Med. Genet.* 9: 160-164, 1972. 139.

Hutz, M. H.; Yoshida, A.; Salzano, F. M.: Three rare G-6-PD variants from Porto Alegre, Brazil. *Hum. Genet.* 39: 191-197, 1977. 140.

Ishwad, C. S.; Naik, S. N.: A new glucose-6-phosphate dehydrogenase variant (G-6-PD Kalyan) found in a Koli family. *Hum. Genet.* 66: 171-175, 1984. 141.

Johnson, G. J.; Kaplan, M. E.; Beutler, E.: G6PD Long Prairie: a new mutant exhibiting normal sensitivity to inhibition by NADPH and accompanied by non-spherocytic hemolytic anemia. *Blood* 49: 247-251, 1977. 142.

Johnston, P. G.; VandeBerg, J. L.; Sharman, G. B.: Inheritance of erythrocyte glucose 6-phosphate dehydrogenase in the red-necked wallaby, *Macropus rufogriseus* (Desmarest)

consistent with paternal X inactivation. *Biochem. Genet.* 13: 235–242, 1975.143. Junien, C.; Kaplan, J.-C.; Meienhofer, M. C.; Maigret, P.; Sender, A.: G6PD Baudelocque: a new unstable variant characterized in cultured fibroblasts. *Enzyme* 18: 48–59, 1974.144. Kaeda, J. S.; Chhotray, G. P.; Ranjit, M. R.; Bautista, J. M.; Reddy, P. H.; Stevens, D.; Naidu, J. M.; Britt, R. P.; Vulliamy, T.J.; Luzzatto, L.; Mason, P. J.: A new glucose-6-phosphate dehydrogenase variant, G6PD Orissa (44 ala-gly), is the major polymorphic variant in tribal populations in India. *Am. J. Hum. Genet.* 57: 1335–1341, 1995.145. Kageoka, T.; Satoh, C.; Goriki, K.; Fujita, M.; Neriishi, S.; Yamamura, K.; Kaneko, J.; Masunari, N.: Electrophoretic variants of blood proteins in Japanese. IV. Prevalence and enzymologic characteristics of G6PD variants in Hiroshima and Nagasaki. *Hum. Genet.* 70: 101–108, 1985.146. Kahn, A.; Bernard, J.-F.; Cottreau, D.; Mazie, J.; Boivin, P.: Gd(-) Abrami, a deficient G6PD variant with hemizygous expression in blood cells of a woman with primary myelofibrosis. *Humangenetik* 30: 41–46, 1975.147. Kahn, A.; Boivin, P.; Hakim, J.; Lagneau, J.: Heterogeneity of glucose-6-phosphate dehydrogenase erythrocyte deficiency in the black race: kinetic study and description of two new variants Gd(-) Dakar et

Gd(-) Mali. *Nouv. Rev. Franc. Hemat.* 11:741-758, 1971.148. Kahn, A.; Boivin, P.; Lagneau, J.: Phenotypes de la glucose-6-phosphatedehydrogenase erythrocytaire dans la race noire. *Humangenetik* 18:261-270, 1973.149. Kahn, A.; Boulard, M.; Hakim, J.; Schaison, G.; Boivin, P.; Bernard, J.: Anemie hemolytique congenitale non spherocytaire par deficien glucose 6-phosphate dehydrogenase erythrocytaire: description de deux nouvelles variants: Gd(-) Saint Louis (Paris) et Gd(-) Hayem. *Nouv. Rev. Franc. Hemat.* 14: 587-600, 1974.150. Kahn, A.; Dao, C.; Cottreau, D.; Bilski-Pasquier, G.: 'Gd(-)Hotel Dieu': a new G-6PD variant with chronic hemolysis in a Negropatient from Senegal. *Hum. Genet.* 39: 353-358, 1977.151. Kahn, A.; Exters, A.; Habedank, M.: Gd(-) Aachen, a new variant of deficient glucose-6-phosphate dehydrogenase. *Human-genetik* 32:171-180, 1976.152. Kahn, A.; Hakim, J.; Cottreau, D.; Boivin, P.: Gd (-) Matam. An African glucose 6-phosphate dehydrogenase variant with enzyme deficiency. Biochemical and immunological properties in various hemopoietic tissues. *Clin. Chim. Acta* 59: 183-190, 1975.153. Kahn, A.; North, M. L.; Cottreau, D.; Giron, G.; Lang, J. M.: G6PD Vientiane: a new glucose-6-phosphate dehydrogenase variant with increased stability. *Hum.*

Genet. 43: 85–89, 1978.154. Kahn, A.; North, M. L.; Messer, J.; Boivin, P.: G–6PD 'Ankara':a new G–6PD variant with deficiency found in a Turkish family. Humangenetik 27:247–250, 1975.155. Kanno, H.; Huang, I.–Y.; Kan, Y. W.; Yoshida, A.: Two structuralgenes on different chromosomes are required for encoding the majorsubunit of human red cell glucose–6–phosphate dehydrogenase. Cell 58:595–606, 1989.156. Kanno, H.; Takano, T.; Fujii, H.; Tani, K.; Morisaki, T.; Hirono,A.; Kumakawa, T.; Ogura, H.; Takahashi, K.; Tsutsumi, H.; Miwa, S.: A new glucose–6–phosphate variant (G6PD Iwate) associated with congenitalnon–spherocytic hemolytic anemia. Acta Haemat. Jpn. 51: 715–719,1987.157. Kaplan, J. C.; Hanzlickova–Leroux, A.; Nicholas, A. M.; Rosa,R.; Weiler, C.; Lepercq, G.: A new glucose–6–phosphate dehydrogenasevariant (G6PD Port–Royal). Enzyme 12: 25–32, 1971.158. Kaplan, J. C.; Rosa, R.; Seringe, P.; Hoeffel, J. C.: Le polymorphisme genetique de la glucose–6–phosphate deshydrogenase erythrocytairechez l'homme. II. Etude d'une nouvelle variete a activite diminuee:le type 'Kabyle.'. Enzym. Biol. Clin. 8: 332–340, 1967.159. Kaplan, M.; Hammerman, C.; Vreman, H. J.; Stevenson, D. K.; Beutler,E.: Acute hemolysis and severe neonatal hyperbilirubinemia in glucose–

6-phosphatedehydrogenase deficient heterozygotes. J. Pediat. 139: 137–140, 2001.160. Kaplan, M.; Renbaum, P.; Levy-Lahad, E.; Hammerman, C.; Lahad, A. Beutler, E.: Gilbert syndrome and glucose-6-phosphate dehydrogenase deficiency: a dose-dependent genetic interaction crucial to neonatal hyperbilirubinemia. Proc. Nat. Acad. Sci. 94: 12128–12132, 1997.161. Kappas, A.; Drummond, G. S.; Valaes, T.: A single dose of Sn-mesoporphyrin prevents development of severe hyperbilirubinemia in glucose-6-phosphatedehydrogenase-deficient newborns. Pediatrics 108: 25–30, 2001.162. Karadsheh, N. S.; Awidi, A. S.; Tarawneh, M. S.: Two new glucose-6-phosphate dehydrogenase (G6PD) variants associated with hemolytic anemia: G6PD Amman-1 and G6PD Amman-2. Am. J. Hemat. 22: 185–192, 1986.163. Kay, A. C.; Kuhl, W.; Prchal, J.; Beutler, E.: The origin of glucose-6-phosphate-dehydrogenase (G6PD) polymorphisms in African-Americans. Am. J. Hum. Genet. 50: 394–398, 1992.164. Kirkman, H. N.; Kidson, C.; Kennedy, M.: Variants of human glucose-6-phosphatedehydrogenase. Studies of samples from New Guinea. In: Beutler, E.: Hereditary Disorders of Erythrocyte Metabolism. New York: Grune and Stratton (pub.) 1968. Pp. 126–145.165.

Kirkman, H. N.; Lie-Injo, L. E.: Variants of glucose 6-phosphatedehydrogenase in Indonesia. *Nature* 221: 959, 1969.166. Kirkman, H. N.; McCurdy, P. R.; Naiman, J. L.: Functionally abnormal glucose-6-phosphate dehydrogenases. *Cold Spring Harbor Symp. Quant. Biol.* 29: 391-398, 1964.167. Kirkman, H. N.; Ramot, B.; Lee, J. T.: Altered aggregational properties in a genetic variant of human glucose-6-phosphate dehydrogenase. *Biochem. Genet.* 3: 137-150, 1969.168. Kirkman, H. N.; Riley, H. D., Jr.: Congenital nonspherocytic hemolytic anemia. *Am. J. Dis. Child.* 102: 313-320, 1961.169. Kirkman, H. N.; Rosenthal, I. M.; Simon, E. B.; Carson, P. E.; Brinson, A. G.: 'Chicago I' variant of glucose-6-phosphate dehydrogenase in congenital hemolytic disease. *J. Lab. Clin. Med.* 63: 715-725, 1964.170. Kirkman, H. N.; Schettini, E.; Pickard, B. M.: Mediterranean variant of glucose-6-phosphate dehydrogenase. *J. Lab. Clin. Med.* 63: 726-735, 1964.171. Kirkman, H. N.; Simon, E. R.; Pickard, B. M.: Seattle variant of glucose-6-phosphate dehydrogenase. *J. Lab. Clin. Med.* 66: 834-840, 1965.172. Kissin, C.; Cotte, J.: Etude d'un variant de glucose-6-phosphatedeshydrogenase: I B type Constantine. *Enzyme* 11: 277-284, 1970.173. Kitao, T.; Ito, K.; Hattori,

K.; Matsuki, T.; Yoneyama, Y.: G6PD Kanazawa: a new variant of glucose-6-phosphate dehydrogenase associated with congenital nonspherocytic hemolytic anemia. *Acta Haemat.* 68: 131-135, 1982.174. Kojima, H.: Congenital nonspherocytic hemolytic disease (CNHD) due to a G-6-PD variant: G-6-PD Kyoto. *Acta Haemat. Jpn.* 35: 32-38, 1972.175. Koliakos, G.; Kalomenopoulou, M.; Grammatikos, P.; Dimitriadou, A.; Kouzi-Koliakos, K.; Zacharaki, R.; Skaragas, G.; Kokka, A.; Trakatellis, A.: A new glucose 6-phosphate dehydrogenase variant (G6PD Thessaloniki) in a patient with idiopathic myelofibrosis. *Hum. Hered.* 39: 141-149, 1989.176. Krasnopskaya, K. D.; Bochkov, N. P.: Genetic heterogeneity of hereditary enzymopathies. *Vestn. Akad. Med. Nauk. SSSR* 9: 56-64, 1982.177. Krasnopskaya, K. D.; Shatskaya, T. L.; Filippov, I. K.; Annenkov, G. A.; Zakharova, T. V.; Mekhtiev, N. K.; Movsum-Zade, K. M.: Genetic heterogeneity of G6PD deficiency: study of mutant alleles in Sheki district of Azerbaijan. *Genetika* 13: 1455-1461, 1977.178. Kumakawa, T.; Suzuki, S.; Fujii, H.; Miwa, S.: Frequency of glucose 6-phosphate dehydrogenase (G6PD) deficiency in Tokyo and a new variant: G6PD Musashino. *Acta Haemat. Jpn.* 50: 25-28, 1987.179. Kurdi-Haidar, B.; Ma-

son, P. J.; Berrebi, A.; Ankra-Badu, G.; Al-Ali, A.; Oppenheim, A.; Luzzatto, L.: Origin and spread of the glucose-6-phosphate dehydrogenase variant (G6PD-Mediterranean) in the Middle East. *Am. J. Hum. Genet.* 47: 1013-1019, 1990.180. Kwiatkowska, J.; Kacprzak-Bergman, I.: New erythrocyte glucose-6-phosphate dehydrogenase variant. *Acta Haemat.* 46: 188-192, 1971.181. Kwok, C. J.; Martin, A. C. R.; Au, S. W. N.; Lam, V. M. S.: G6PDdb, an integrated database of glucose-6-phosphate dehydrogenase (G6PD) mutations. *Hum. Mutat.* 19: 217-224, 2002.182. Lee, K. T.; Thomas, W. A.; Janakidevi, K.; Kroms, M.; Reiner, J. M.; Borg, K. Y.: Mosaicism in female hybrid hares heterozygous for glucose-6-phosphate dehydrogenase (G-6-PD). I. General properties of a hybrid hare model with special reference to atherogenesis. *Exp. Molec. Path.* 34: 191-201, 1981.183. Lenzerini, L.; Meera Khan, P.; Filippi, G.; Rattazzi, M. C.; Rat, A. K.: Characterization of glucose-6-phosphate dehydrogenase variants. I. Occurrence of a G6PD Seattle-like variant in Sardinia and its interaction with G6PD Mediterranean variant. *Am. J. Hum. Genet.* 21: 142-153, 1969.184. Lisker, R.; Linares, C.; Motulsky, A. G.: Glucose-6-phosphate dehydrogenase Mexico, a new variant

with enzyme deficiency, abnormal mobility and absence of hemolysis. J. Lab. Clin. Med. 29: 788–793, 1972. 185.

Lisker, R.; Perez-Briceno, R.; Beutler, E.: A new glucose-6-phosphate dehydrogenase variant, Gd(-) Tepic, characterized by moderate enzyme deficiency and mild episodes of hemolytic anemia. Hum. Genet. 69:19–21, 1985. 186.

Lisker, R.; Perez-Briceno, R.; Rave, V.; Yoshida, A.: Glucose-6-phosphate dehydrogenase Gd(-) Distrito Federal: nueva variante asociada a deficiencia enzimática moderada y anemia hemolítica ocasional. Rev. Invest. Clin. 33:209–211, 1981. 187.

Lisker, R.; Perez Briceno, R.; Aguilár, L.; Yoshida, A.: A variant glucose-6-phosphate dehydrogenase Gd(-) Chiapas associated with moderate enzyme deficiency and occasional hemolytic anemia. Hum. Genet. 43:81–84, 1978. 188.

Lisker, R.; Perez Briceno, R.; Zavala, C.; Navarrette, J. I.; Wessels, M.; Yoshida, A.: A glucose 6-phosphate dehydrogenase Gd(-) Castilla variant characterized by mild deficiency associated with drug-induced hemolytic anemia. J. Lab. Clin. Med. 90: 754–759, 1977. 189.

Liu, Y.; Phelan, J.; Go, R. C. P.; Prchal, J. F.; Prchal, J. T.: Rapid determination of clonality by detection of two closely-linked X chromosome exonic polymorphisms using allele-specific PCR. J. Clin. Invest. 99: 1984–1990,

1997.190. Long, W. K.; Kirkman, H. N.; Sutton, H. H.: Electrophoretically slow variants of glucose-6-phosphate dehydrogenase from red cells of Negroes. J. Lab. Clin. Med. 65: 81-87, 1965.191. Longo, L.; Vanegas, O. C.; Patel, M.; Rosti, V.; Li, H.; Waka, J.; Merghoub, T.; Pandolfi, P. P.; Notaro, R.; Manova, K.; Luzzatto, L.: Maternally transmitted severe glucose 6-phosphate dehydrogenase deficiency is an embryonic lethal. EMBO J. 21: 4229-4239, 2002.192. Luzzatto, L.: Genetic heterogeneity and pathophysiology of G6PD deficiency. Brit. J. Haemat. 28: 151-156, 1974.193. Luzzatto, L.: Personal Communication. London, England 4/1990.194. Luzzatto, L.; Afolayam, A.: Enzyme properties of different types of human erythrocyte glucose-6-phosphate dehydrogenase with characterization of two new genetic variants. J. Clin. Invest. 47: 1833-1842, 1968.195. Luzzatto, L.; Martini, G.: X-Linked Wiskott-Aldrich syndrome in a girl. (Letter) New Eng. J. Med. 338: 1850-1851, 1998.196. Luzzatto, L.; Usanga, E. A.; Bienze, U.; Esan, G. F. J.; Fusuan, F. A.: Imbalance in X-chromosome expression: evidence for a human X-linked gene affecting growth of hemopoietic cells. Science 205:1418-1420, 1979.197. Luzzatto, L.; Usanga, E. A.; Reddy, S.: Glucose-6-phosphate dehydrogenase deficient

red cells: resistance to infection by malarial parasites. *Science* 164: 839–842, 1969.198. MacDonald, D.; Town, M.; Mason, P.; Vulliamy, T.; Luzzatto, L.; Goff, D. K.: Deficiency in red blood cells. (Letter) *Nature* 350:115, 1991.199. Maeda, M.; Constantoulakis, P.; Chen, C.-S.; Stamatoyannopoulos, G.; Yoshida, A.: Molecular abnormalities of a human glucose-6-phosphate dehydrogenase variant associated with undetectable enzyme activity and immunologically cross-reacting material. *Am. J. Hum. Genet.* 51:386–395, 1992.200. Mallouh, A. A.; Abu-Osba, Y. K.: Bacterial infections in children with glucose-6-phosphate dehydrogenase deficiency. *J. Pediat.* 111:850–852, 1987.201. Mamlok, R. J.; Mamlok, V.; Mills, G. C.; Daeschner, C. W., III; Schmalstieg, F. C.; Anderson, D. C.: Glucose-6-phosphate dehydrogenase deficiency, neutrophil dysfunction and *Chromobacterium violaceum* sepsis. *J. Pediat.* 111: 852–854, 1987.202. Mamlok, R. J.; Mills, G. C.; Goldblum, R. M.; Daeschner, C. W.: Glucose-6-phosphate dehydrogenase Beaumont: a new variant with severe enzyme deficiency and chronic nonspherocytic hemolytic anemia. *Enzyme* 34:15–21, 1985.203. Mandelli, F.; Amadori, S.; DeLaurenzi, A.; Kahn, A.; Isacchi, G.; Papa, G.: Glucose-6-phosphate dehydrogenase Velletri: a new

variant with reduced activity in a patient with congenital non-spherocytic haemolytic anemia. *Acta Haemat.* 57: 121–126, 1977.

204. Marks, P. A.; Banks, J.; Gross, R.: Genetic heterogeneity of glucose-6-phosphate dehydrogenase deficiency. *Nature* 194: 454–456, 1962.

205. Martin, S. K.; Miller, L. H.; Alling, D.; Okoye, V. C.; Esan, G. J. F.; Osunkoya, B. O.; Deane, M.: Severe malaria and glucose-6-phosphate-dehydrogenase deficiency: a reappraisal of the malaria-G6PD hypothesis. *Lancet* I: 524–526, 1979.

206. Martini, G.; Toniolo, D.; Vulliamy, T.; Luzzatto, L.; Dono, R.; Viglietto, G.; Paonessa, G.; D'Urso, M.; Persico, M. G.: Structural analysis of the X-linked gene encoding human glucose 6-phosphate dehydrogenase. *EMBO J.* 5: 1849–1855, 1986.

207. Mason, P. J.: New insights into G6PD deficiency. (Annotation) *Brit. J. Haemat.* 94: 585–591, 1996.

208. Mason, P. J.; Stevens, D. J.; Luzzatto, L.; Brenner, S.; Aparicio, S.: Genomic structure and sequence of the *Fugu rubripes* glucose-6-phosphate dehydrogenase gene (G6PD). *Genomics* 26: 587–591, 1995.

209. McCann, S. R.; Smithwick, A. M.; Temperley, I. J.; Tipton, K.: G6PD (Dublin): chronic non-spherocytic haemolytic anaemia resulting from glucose-6-phosphate dehydrogenase deficiency in an Irish kin-

dred. J. Med. Genet. 17: 191–193, 1980.210. McCurdy, P. R.: Use of genetic linkage for the detection of female carriers of hemophilia. New Eng. J. Med. 285: 218–219, 1971.211. McCurdy, P. R.; Blackwell, R. Q.; Todd, D.; Tso, S. C.; Tuchinda, S.: Further studies on glucose-6-phosphate dehydrogenase deficiency in Chinese subjects. J. Lab. Clin. Med. 75: 788–797, 1970.212. McCurdy, P. R.; Kamel, K.; Selim, O.: Heterogeneity of red cell glucose 6-phosphate dehydrogenase (G6PD) deficiency in Egypt. J. Lab. Clin. Med. 84: 673–680, 1974.213. McCurdy, P. R.; Kirkman, H. N.; Naiman, J. L.; Jim, R. T. S.; Pickard, B. M.: A Chinese variant of glucose-6-phosphate dehydrogenase. J. Lab. Clin. Med. 67: 374–385, 1966.214. McCurdy, P. R.; Mahmood, L.: Red cell glucose-6-phosphate dehydrogenase deficiency in Pakistan. J. Lab. Clin. Med. 76: 943–948, 1970.215. McCurdy, P. R.; Maldonado, N. I.; Dillon, D. E.: Variants of glucose-6-phosphate dehydrogenase (G-6-PD) associated with G-6-PD deficiency in Puerto Ricans. J. Lab. Clin. Med. 82: 432–437, 1973.216. McCurdy, P. R.; Maldonado, N. I.; Dillon, D. E.; Conrad, M. E.: Variants of glucose-6-phosphate dehydrogenase (G-6-PD) associated with G-6-PD deficiency in Puerto Ricans. J. Lab. Clin. Med. 82: 432–437, 1973.217. Meloni, T.;

Carta, F.; Forteleoni, G.; Carta, A.; Ena, F.; Meloni, G. F.: Glucose 6-phosphate dehydrogenase deficiency and cataract of patients in northern Sardinia. *Am. J. Ophthalmol.* 110: 661-664, 1990. 218. Mentzer, W. C., Jr.; Warner, R.; Addiego, J.; Smith, B.; Walter, T.: G6PD San Francisco: a new variant of glucose-6-phosphate dehydrogenase associated with congenital nonspherocytic hemolytic anemia. *Blood* 55:195-198, 1980. 219. Mesbah-Namin, S. A.; Sanati, M. H.; Mowjoodi, A.; Mason, P. J.; Vulliamy, T. J.; Noori-Daloui, M. R.: Three major glucose-6-phosphate dehydrogenase-deficient polymorphic variants identified in Mazandaran state of Iran. *Brit. J. Haematol.* 117: 763-764, 2002. 220. Miller, D. R.; Wollman, M. R.: A new variant of glucose 6-phosphate dehydrogenase deficiency hereditary hemolytic anemia, G6PD Cornell: erythrocyte, leukocyte and platelet studies. *Blood* 44: 277-284, 1974. 221. Mills, G. C.; Alperin, J. B.; Trimmer, K. B.: Studies on variant glucose-6-phosphate dehydrogenase: G6PD Fort Worth. *Biochem. Med.* 13:264-275, 1975. 222. Milner, G.; Delamore, I. W.; Yoshida, A.: G-6-PD Manchester: a new variant associated with chronic nonspherocytic hemolytic anemia. *Blood* 43:271-276, 1974. 223. Miwa, S.; Fujii, H.: Molecular basis of ery-

thoenzymopathies associated with hereditary hemolytic anemia: tabulation of mutant enzymes. *Am. J. Hemat.* 51: 122–132, 1996.224. Miwa, S.; Fujii, H.; Nakashima, K.; Miura, Y.; Yamada, K.; Hagiwara, T.; Fukuda, M.: Three new electrophoretically normal glucose–6-phosphatedehydrogenase variants associated with congenital nonspherocytic hemolytic anemia found in Japan: G6PD Ogikubo, Yokohama, and Akita. *Hum. Genet.* 45:11–17, 1978.225. Miwa, S.; Fujii, H.; Nakatsuji, T.; Ishida, Y.; Oda, E.; Kaneto, A.; Motokawa, M.; Ariga, Y.; Fukuchi, S.; Sasai, S.; Hiraoka, K.; Kashii, H.; Kodama, T.: Four new electrophoretically slow-moving glucose–6-phosphate dehydrogenase variants associated with congenital nonspherocytic hemolytic anemia found in Japan: Gd(–) Kurume, Gd(–) Fukushima, Gd(–) Yamaguchi, and Gd(–) Wakayama. *Am. J. Hemat.* 5:131–138, 1978.226. Miwa, S.; Nakashima, K.; Ono, J.; Fujii, H.; Suzuki, E.: Three glucose 6-phosphate dehydrogenase variants found in Japan. *Hum. Genet.* 36:327–334, 1977.227. Miwa, S.; Ono, J.; Nakashima, K.; Abe, S.; Kageoka, T.; Shinohara, K.; Isobe, J.; Yamaguchi, H.: Two new glucose 6-phosphate dehydrogenase variants associated with congenital nonspherocytic hemolytic anemia found in

Japan: Gd(-) Tokushima and Gd(-) Tokyo. Am. J. Hemat. 1:433-442, 1976.228. Modiano, G.; Battistuzzi, G.; Esan, G. J. F.; Testa, U.; Luzzatto, L.: Genetic heterogeneity of 'normal' human erythrocyte glucose-6-phosphatedehydrogenase: an isoelectrophoretic polymorphism. Proc. Nat. Acad.Sci. 76: 852-856, 1979.229. Mohrenweiser, H. W.; Neel, J. V.: Frequency of thermostabilityvariants: estimation of total 'rare' variant frequency in human populations. Proc.Nat. Acad. Sci. 78: 5729-5733, 1981.230. Morelli, A.; Benatti, U.; Guida, L.; De Flora, A.: G6PD Cagliari:a new low activity glucose 6-phosphate dehydrogenase variant characterizedby enhanced intracellular lability. Hum. Genet. 66: 62-65, 1984.231. Morisaki, T.; Fujii, H.; Takegawa, S.; Tani, K.; Hirono, A.;Takizawa, T.; Takahashi, K.; Shinogi, M.; Teshirogi, T.; Miwa, S.: G6PD Sendagi: a new glucose-6-phosphate dehydrogenase variant associatedwith congenital hemolytic anemia. Hum. Genet. 65: 214-215, 1983.232. Nafa, K.; Reghis, A.; Osmani, N.; Baghli, L.; Benabadji, M.;Kaplan, J.-C.; Vulliamy, T. J.; Luzzatto, L.: G6PD Aures: a new mutation(48 ile-to-thr) causing mild G6PD deficiency is associated with favism. Hum.Molec. Genet. 2: 81-82, 1993.233. Nagel, R. L.; Ranney, H. M.: Genetic epidemiology of

structural mutations of the beta-globin gene. *Semin. Hemat.* 27: 342–359, 1990.234. Nakai, T.; Yoshida, A.: G6PD Heian. A glucose-6-phosphate dehydrogenase variant associated with hemolytic anemia found in Japan. *Clin. Chim. Acta* 51: 199–203, 1974.235. Nakashima, K.; Ono, J.; Abe, S.; Miwa, S.; Yoshida, A.: G6PD Ube: a glucose 6-phosphate dehydrogenase variant found in four unrelated Japanese families. *Am. J. Hum. Genet.* 29: 24–30, 1977.236. Nakatsuji, T.; Miwa, S.: Incidence and characteristics of glucose-6-phosphate dehydrogenase variants in Japan. *Hum. Genet.* 51: 297–305, 1979.237. Nance, W. E.: Turner's syndrome, twinning, and an unusual variant of glucose-6-phosphate dehydrogenase. *Am. J. Hum. Genet.* 16: 380–392, 1964.238. Necheles, T. F.; Snyder, L. M.; Strauss, W.: *Glucose-6-phosphate dehydrogenase* Boston. A new variant associated with congenital nonspherocytic hemolytic disease. *Humangenetik* 13: 218–221, 1971.239. Niazi, G.; Adeyokunu, A.; Westwood, B.; Beutler, E.: G6PD Aures: a rare mutant of G6PD in Saudi Arabia: molecular and clinical presentations. *Saudi Med. J.* 17: 311–314, 1996.240. Ninfali, P.; Baronciani, L.; Bardoni, A.; Bresolin, N.: Muscle expression of glucose-6-phosphate dehydrogenase deficiency in different variants. *Clin. Genet.*

48: 232–237, 1995.241. Notaro, R.; Afolayan, A.; Luz-
 zatto, L.: Human mutations in glucose6–phosphate dehy-
 drogenase reflect evolutionary history. FASEB J.
 14:485–494, 2000.242. Nowicki, L.; Strobel, S.; Martin, H.;
 Koschwitz, U.: Ueber eineneue erythrocytaere glucose
 6–phosphatdehydrogenase Variante, TypFrankfurt. Klin.
 Wschr. 52: 478–484, 1974.243. Nsouly, G. M.; Prchal, J.
 T.: Characterization of a new G6PDvariant and its associ-
 ated oxidative damage. Clin. Res. 29: 829,1981.244.
 O'Brien, S. J.: The extent and character of biochemical ge-
 neticvariation in the domestic cat. J. Hered. 71: 2–8,
 1980.245. Ogura, H.; Morisaki, T.; Tani, K.; Kanno, H.;
 Tsutsumi, H.; Takahashi,K.; Miyamori, T.; Fujii, H.; Miwa,
 S.: A new glucose–6–phosphatedehydrogenase variant
 (G6PD Tsukui) associated with congenital hemolyticane-
 mia. Hum. Genet. 78: 369–371, 1988.246. Ohno, S.: Sex
 Chromosomes and Sex–linked Genes. Berlin andNew York:
 Springer (pub.) 1967.247. Orzalesi, N.; Sorcinelli, R.;
 Guiso, G.: Increased incidenceof cataracts in male subjects
 deficient in glucose–6–phosphate dehydrogenase.
 Arch.Ophthal. 99: 69–70, 1981.248. Othieno–Obel, A.:
 East African variant of glucose–
 6–phosphatedehydrogenase. East Afr. Med. J. 49:

230–234, 1972.249. Pai, G. S.; Sprenkle, J. A.; Do, T. T.; Maren, C. E.; Migeon, B. R.: Localization of loci for hypoxanthine phosphoribosyltransferase and glucose-6-phosphate dehydrogenase and biochemical evidence of nonrandom X chromosome expression from studies of a human X-autosome translocation. *Proc. Nat. Acad. Sci.* 77: 2810–2813, 1980.250. Panich, V.: G6PD variants in Lao-tians. *Hum. Hered.* 24: 285–290, 1974.251. Panich, V.: G6PD Intanon, a new glucose 6-phosphate dehydrogenase variant. *Humangenetik* 21: 203–205, 1974.252. Panich, V.: G6PD characterization in Thailand. *Genetics* 74(suppl.): s208 only, 1973.253. Panich, V.: Glucose-6-phosphate dehydrogenase in Thailand. *Hum. Genet.* 53: 227–228, 1980.254. Panich, V.; Bumrungtrakul, P.; Jitjai, C.; Kamolmatayakul, S.; Khoprasert, B.; Klaisuvan, C.; Kongmuang, U.; Maneechai, P.; Pornpatkul, M.; Ruengrairatanaroje, P.; Surapruk, P.; Viriyayudhakorn, S.: Glucose-6-phosphate dehydrogenase deficiency in South Vietnamese. *Hum. Hered.* 30: 361–364, 1980.255. Panich, V.; Na-Nakorn, S.: G6PD variants in Thailand. *J. Med. Assoc. Thai.* 63: 537–543, 1980.256. Panich, V.; Sungnate, T.: Characterization of glucose 6-phosphate dehydrogenase in Thailand: the occurrence of 6 variants among 50

G6PDdeficient Thai. Humangenetik 18: 39–46, 1973.257. Panich, V.; Sungnate, T.; Na-Nakorn, S.: Acute intravascular hemolysis and renal failure in a new glucose 6-phosphate dehydrogenase variant: G6PD Siriraj. J. Med. Assoc. Thai. 55: 726–731, 1972.258. Panich, V.; Sungnate, T.; Wasi, P.; Na-Nakorn, S.: G-6-PD Mahidol: the most common glucose-6-phosphate dehydrogenase variant in Thailand. J. Med. Assoc. Thai. 55: 576–585, 1972.259. Pawlak, A. L.; Mazurkiewicz, C. A.; Ordynski, J.; Ruzynkova, D.; Horst, A.: G6PD Poznan, variant with severe enzyme deficiency. Humangenetik 28:163–165, 1975.260. Pawlak, A. L.; Zagorski, Z.; Ruzynkova, D.; Horst, A.: Polish variant of glucose-6-phosphate dehydrogenase (G-6-PD Lublin). Humangenetik 10:340–343, 1970.261. Pekrun, A.; Eber, S. W.; Schroter, W.: G6PD Avenches and G6PD Moosburg: biochemical and erythrocyte membrane characterization. Blut 58:11–14, 1989.262. Perroni, L.; Tassara, P.; Baldi, M.; Reali, R.; Scartezzini, P.: G6PD variants detected in Genoa area. In: Weatherall, D. J.; Fiorelli, G.; Gorini, S.: Advances in Red Blood Cell Biology. New York: Raven Press (pub.) 1982. Pp. 409–416.263. Persico, M. G.; Toniolo, D.; Nobile, C.; D'Urso, M.; Luzzatto, L.: cDNA sequences of human glu-

cose 6-phosphate dehydrogenase cloned in pBR322. Nature 294: 778–780, 1981.264. Persico, M. G.; Viglietto, G.; Martini, G.; Toniolo, D.; Paonessa, G.; Moscatelli, C.; Dono, R.; Vulliamy, T.; Luzzatto, L.; D'Urso, M.: Isolation of human glucose-6-phosphate dehydrogenase (G6PD) cDNA-clones: primary structure of the protein and unusual 5-prime non-coding region. Nucleic Acids Res. 14: 2511–2522, 1986.265. Picat, C.; Etiemble, J.; Boivin, P.; Le Prise, P.-Y.: Gd(-)Rennes, a new deficient variant of glucose-6-phosphate dehydrogenase associated with congenital nonspherocytic hemolytic anemia found in France. Hum. Genet. 55: 125–127, 1980.266. Pinto, P. V. C.; Newton, W. A., Jr.; Richardson, K. E.: Evidence for four types of erythrocyte glucose-6-phosphate dehydrogenase from G-6-PD deficient human subjects. J. Clin. Invest. 45: 823–831, 1966.267. Poggi, V.: Personal Communication. London, England 1989.268. Poon, M.-C.; Hall, K.; Scott, C. W.; Prchal, J. T.: G6PD Viangchan: a new glucose 6-phosphate dehydrogenase variant from Laos. Hum. Genet. 78: 98–99, 1988.269. Porter, I. H.; Boyer, S. H.; Watson-Williams, E. J.; Adam, A.; Szeinberg, A.; Siniscalco, M.: Variation of glucose-6-phosphate dehydrogenase in different populations. Lancet I: 895–899, 1964.270.

Porter, I. H.; Schulze, J.; McKusick, V. A.: Genetical linkage between the loci for glucose-6-phosphate dehydrogenase deficiency and colour-blindness in American Negroes. *Ann. Hum. Genet.* 26: 107-122, 1962.271. Prchal, J.; Moreno, H.; Conrad, M.; Vitek, A.: G-6-PD Dothan: a new variant associated with chronic hemolytic anemia. *I.R.C.S.* 7:348, 1979.272. Prchal, J. T.: Personal Communication. Birmingham, Ala. 1985.273. Prchal, J. T.; Crist, W. M.; Malluh, A.; Vitek, A.; Tauxe, W.N.; Carroll, A. J.: A new glucose-6-phosphate dehydrogenase deficient variant in a patient with Chediak-Higashi syndrome. *Blood* 56: 476-480, 1980.274. Prchal, J. T.; Hall, K.; Csepregy, M.; Lilly, M.; Berkow, R.; Scott, C. W.: Two apparent glucose-6-phosphate dehydrogenase variants in normal XY males: G6PD Alabama. *Am. J. Med.* 84: 517-523, 1988.275. Pretsch, W.; Charles, D. J.; Merkle, S.: X-linked glucose-6-phosphate-dehydrogenase deficiency in *Mus musculus*. *Biochem. Genet.* 26: 89-103, 1988.276. Puck, J. M.; Willard, H. F.: X inactivation in females with X-linked disease. *New Eng. J. Med.* 338: 325-327, 1998.277. Ramot, B.; Ben-Bassat, I.; Shchory, M.: New glucose-6-phosphate dehydrogenase variants observed in Israel and their association with congenital nonspherocytic

hemolytic disease. J. Lab. Clin. Med. 74:895–901, 1969.278. Ramot, B.; Brok, F.: A new glucose–6–phosphate dehydrogenasemutant (Tel–Hashomer mutant). Ann. Hum. Genet. 28: 167–172, 1964.279. Rattazzi, M. C.; Corash, L. M.; Van Zzanen, G. E.; Jaffe, E.R.; Piomelli, S.: G6PD deficiency and chronic hemolysis: four newmutants--relationships between clinical syndrome and enzyme kinetics. Blood 38:205–218, 1971.280. Rattazzi, M. C.; Lenzerini, L.; Meera Khan, P.; Luzzatto, L.: Characterization of glucose–6–phosphate dehydrogenase variants.II. G6PD Kephalaria, G6PD Attica, and G6PD 'Seattle–like' found inGreece. Am. J. Hum. Genet. 21: 154–167, 1969.281. Ravindranath, Y.; Beutler, E.: Two new variants of glucose–6–phosphatedehydrogenase associated with hereditary non–spherocytic hemolyticanemia: G6PD Wayne and G6PD Huron. Am. J. Hemat. 24: 357–363, 1987.282. Reys, L.; Manso, C.; Stamatoyannopoulos, G.: Genetic studieson Southeastern Bantu of Mozambique. I. Variants of glucose–6–phosphatedehydrogenase. Am. J. Hum. Genet. 22: 203–215, 1970.283. Rinaldi, A.; Filippi, G.; Siniscalco, M.: Variability of redcell phenotypes between and within individuals in an unbiased sampleof 77 heterozygotes for G6PD deficiency in Sardinia. Am. J.

Hum.Genet. 28: 496–505, 1976.284. Rosenstrauss, M.; Chasin, L. A.: Isolation of mammalian cell mutants deficient in glucose-6-phosphate dehydrogenase activity: linkage to hypoxanthine phosphoribosyl transferase. Proc. Nat. Acad. Sci. 72:493–497, 1975.285. Roth, E. F., Jr.; Raventos-Suarez, C.; Rinaldi, A.; Nagel, R.L.: Glucose-6-phosphate dehydrogenase deficiency inhibits in vitro growth of Plasmodium falciparum. Proc. Nat. Acad. Sci. 80: 298–299, 1983.286. Roychoudhury, A. K.; Nei, M.: Human Polymorphic Genes: World Distribution. New York: Oxford Univ. Press (pub.) 1988.287. Ruwando, C.; Khe, S. C.; Snow, R. W.; Yates, S. N. R.; Kwiatkowski, D.; Gupta, S.; Warn, P.; Alisopp, G. E. M.; Gilbert, S. C.; Peschu, N.; Newbold, C. I.; Greenwood, S. M.; Marsh, K.; Hill, A. V. S.: Natural selection of hemi- and heterozygotes for G6PD deficiency in Africa by resistance to severe malaria. Nature 376: 246–249, 1995.288. Saenz, G. F.; Chaves, M.; Berrantes, A.; Elizondo, J.; Montero, A. G.; Yoshida, A.: A glucose-6-phosphate dehydrogenase variant, Gd(-) Santa-maria found in Costa Rica. Acta Haemat. 72: 37–40, 1984.289. Saidi, N.; Hors-Cayla, M. C.; Van Cong, N.; Benne, F.: Sheep gene mapping by somatic cell hybridization. (Abstract) Cytogenet. Cell Genet. 25: 200, 1979.290.

Samuel, A. P. W.; Saha, N.; Omer, A.; Hoffbrand, A. V.: Quantitative expression of G6PD activity of different phenotypes of G6PD and haemoglobin in a Sudanese population. *Hum. Hered.* 31: 110–115, 1981.291. Sansone, G.; Perroni, L.; Testa, U.; Marenzi, C.; Luzzatto, L.: New genetic variants of glucose 6-phosphate dehydrogenase (G6PD) in Italy. *Ann. Hum. Genet.* 45: 97–104, 1981.292. Sansone, G.; Perroni, L.; Yoshida, A.: Glucose-6-phosphate dehydrogenase variants from Italian subjects associated with severe neonatal jaundice. *Brit. J. Haemat.* 31: 159–165, 1975.293. Sansone, G.; Perroni, L.; Yoshida, A.; Dave, V.: A new glucose 6-phosphate dehydrogenase variant (Gd Trinacria) in two unrelated families of Sicilian ancestry. *Ital. J. Biochem.* 26: 44–50, 1977.294. Shatskaya, T. L.; Krasnopol'skaya, K. D.; Annenkov, G. A.: A description of new mutant forms of erythrocyte glucose-6-phosphate dehydrogenase isolated at the territory of the Soviet Union. *Genetika* 11: 116–122, 1975.295. Shatskaya, T. L.; Krasnopol'skaya, K. D.; Idelson, L. J.: The new form of glucose 6-phosphate dehydrogenase (G6PD 'Kaluga') from erythrocytes of a patient with chronic non-spherocytic hemolytic anemia. *Vopr. Med. Khim.* 22: 764–768, 1976.296. Shatskaya, T. L.; Krasnopol'skaya, K.

D.; Idelson, L. J.: Mutantforms of erythrocyte glucose 6-phosphate dehydrogenase in Ashkenazi: description of two new variants, G6PD Kirovograd and G6PD Zhitomir. Humangenetik 33:175–178, 1976.297. Shatskaya, T. L.; Krasnopol'skaya, K. D.; Tzoneva, M.; Mavrudieva, M.; Toncheva, D.: Variants of erythrocyte glucose-6-phosphate dehydrogenase (G6PD) in Bulgarian populations. Hum. Genet. 54: 115–117, 1980.298. Shatskaya, T. L.; Krasnopol'skaya, K. D.; Zakharova, T. V.: Regularities of distribution of Gd-alleles in Azerbaijan. II. Identification of G6PD mutant forms. Genetika 16: 2217–2225, 1980.299. Shows, T. B.; Brown, J. A.: Human X-linked genes regionally mapped utilizing X-autosome translocations and somatic cell hybrids. Proc. Nat. Acad. Sci. 72: 2125–2129, 1975.300. Shows, T. B.; Brown, J. A.; Chapman, V. M.: Comparative gene mapping of HPRT, G6PD and PGK in man, mouse, and Muntjac deer. Birth Defects Orig. Art. Ser. XII(7): 436–439, 1976.301. Shows, T. B.; Tashian, R. E.; Brewer, G. J.: Erythrocyte glucose-6-phosphate dehydrogenase in Caucasians: new inherited variant. Science 145:1056–1057, 1964.302. Sidi, Y.; Aderka, D.; Brok-Simoni, F.; Benjamin, D.; Ramot, B.; Pinkhas, J.: Viral hepatitis with extreme hyperbilirubine-

mia, massive hemolysis and encephalopathy in a patient with a new G6PD variant. *Israel J. Med. Sci.* 16: 130–133, 1980.303. Siegel, N. H.; Beutler, E.: Hemolytic anemia caused by G-6-PD Carswell, a new variant. *Ann. Intern. Med.* 75: 437–439, 1971.304. Smith, J. E.; Ryer, K.; Wallace, L.: Glucose-6-phosphate dehydrogenase deficiency in a dog. *Enzyme* 21: 379–382, 1976.305. Snyder, L. M.; Necheles, T. F.; Reddy, W. J.: G-6-PD Worcester: a new variant, associated with X-linked optic atrophy. *Am. J. Med.* 49:125–132, 1970.306. Stamatoyannopoulos, G.; Kotsakis, P.; Voigtlander, V.; Akrivakis, A.; Motulsky, A. G.: Electrophoretic diversity of glucose-6-phosphate dehydrogenase among Greeks. *Am. J. Hum. Genet.* 22: 587–596, 1970.307. Stamatoyannopoulos, G.; Voigtlander, V.; Akrivakis, A.: Thessaly variant of glucose-6-phosphate dehydrogenase. *Humangenetik* 9: 23–25, 1970.308. Stamatoyannopoulos, G.; Voigtlander, V.; Kotsakis, P.; Akrivakis, A.: Genetic diversity of the 'Mediterranean' glucose-6-phosphate dehydrogenase deficiency phenotype. *J. Clin. Invest.* 50: 1253–1261, 1971.309. Stamatoyannopoulos, G.; Yoshida, A.; Bacopoulos, C.; Motulsky, A. G.: Athens variant of glucose-6-phosphate dehydrogenase. *Science* 157:831–833,

1967.310. Stevens, D. J.; Wanachiwanawin, W.; Mason, P. J.; Vulliamy, T.J.; Luzzatto, L.: G6PD Canton a common deficient variant in SouthEast Asia caused by a 459 arg-to-leu mutation. *Nucleic Acids Res.* 18:7190, 1990.311. Stocco dos Santos, R. C.; Barretto, O. C. O.; Nonoyama, K.; Castro, N. H. C.; Ferraz, O. P.; Walter-Moura, J.; Vescio, C. C. S.; Becak, W.: X-linked syndrome: mental retardation, hip luxation, and G6PDvariant (Gd(+)) Butantan). *Am. J. Med. Genet.* 39: 133–136, 1991.312. Stockham, S. L.; Harvey, J. W.; Kinden, D. A.: Equine glucose-6-phosphatedehydrogenase deficiency. *Vet. Path.* 31: 518–527, 1994.313. Streiff, F.; Vigneron, C.: Anemie hemolytique chronique pardeficit en glucose 6-phosphate deshydrogenase dans une famille d'origineLorraine. *Nouv. Rev. Franc. Hemat.* 11: 279–290, 1971.314. Takahashi, K.; Fujii, H.; Takegawa, S.; Tani, K.; Hirono, A.; Takizawa, T.; Kawakatsu, T.; Miwa, S.: A new glucose-6-phosphatedehydrogenase variant (G6PD Nagano) associated with congenital hemolyticanemia. *Hum. Genet.* 62: 368–370, 1982.315. Takizawa, T.; Fujii, H.; Takegawa, S.; Takahashi, K.; Hirono, A.; Morisaki, T.; Kanno, H.; Oka, R.; Yoshioka, H.; Miwa, S.: A uniqueelectrophoretic slow-moving glucose 6-phosphate dehydrogenase vari-

ant(G6PD Asahikawa) with a markedly acidic pH optimum. Hum. Genet. 68:70–72, 1984.316. Takizawa, T.; Huang, I.-Y.; Ikuta, T.; Yoshida, A.: Human glucose-6-phosphatedehydrogenase: primary structure and cDNA cloning. Proc. Nat. Acad.Sci. 83: 4157–4161, 1986.317. Takizawa, T.; Yoneyama, Y.; Miwa, S.; Yoshida, A.: A single nucleotide base transition is the basis of the common human glucose-6-phosphatedehydrogenase variant A(+). Genomics 1: 228–231, 1987.318. Takizawa, T.; Yoshida, A.: Molecular abnormality of the common glucose-6-phosphate dehydrogenase variant, G6PD A(+), and restriction-fragment-length polymorphism. (Abstract) Am. J. Hum. Genet. 41: A241, 1987.319. Talalak, P.; Beutler, E.: G-6-PD Bangkok: a new variant found in congenital non-spherocytic hemolytic disease (CNHD). Blood 33:772–776, 1969.320. Tanaka, K. R.; Beutler, E.: Hereditary hemolytic anemia due to glucose-6-phosphate dehydrogenase Torrance: a new variant. J. Lab. Clin. Med. 73: 657–667, 1969.321. Tang, T. K.; Huang, C.-S.; Huang, M. J.; Tam, K.-B.; Yeh, C.-H.; Tang, C.-J. C.: Diverse point mutations result in glucose-6-phosphatedehydrogenase (G6PD) polymorphism in Taiwan. Blood 79: 2135–2140, 1992.322. Testa, U.; Meloni, T.; Lania, A.; Battistuzzi, G.; Cutillo,

S.;Luzzatto, L.: Genetic heterogeneity of glucose-6-phosphate dehydrogenase deficiency in Sardinia. Hum. Genet. 56: 99-105, 1980.323. Thigpen, J. T.; Steinberg, M. H.; Beutler, E.; Gillespie, G.T., Jr.; Dreiling, B. J.; Morrison, F. S.: Glucose-6-phosphate dehydrogenase Jackson, a new variant associated with hemolytic anemia. Acta Haemat. 51:310-314, 1974.324. Tishkoff, S. A.; Varkonyi, R.; Cahinhinan, N.; Abbes, S.; Argyropoulos, G.; Destro-Bisol, G.; Drousiotou, A.; Dangerfield, B.; Lefranc, G.;Loiselet, J.; Piro, A.; Stoneking, M.; Tagarelli, A.; Tagarelli, G.;Touma, E. H.; Williams, S. M.; Clark, A. G.: Haplotype diversity and linkage disequilibrium at human G6PD: recent origin of alleles that confer malarial resistance. Science 293: 455-462, 2001.325. Tokarev, Y. N.; Chernyak, N. B.; Batischev, A. I.; Lanzina, N.V.; Alexeyev, G. A.: Etude des proprietes electrophoretiques et cinetiques de la glucose-6-phosphate deshydrogenase (Gd) d'erythrocytes dans les deficits hereditaires de l'enzyme: description d'une nouvelle variante de glucose-6-phosphate deshydrogenase: la Gd Kremenchug. Nouv.Rev. Franc. Hemat. 20: 557-564, 1978.326. Toncheva, D.: Variants of glucose-6-phosphate dehydrogenase in a Vietnamese population. Hum. Hered. 36:

348–351, 1986.327. Toncheva, D.; Tzoneva, M.: Genetic polymorphism of G6PD in a Bulgarian population. Hum. Genet. 67: 340–342, 1984.328. Toniolo, D.; Martini, G.; Migeon, B. R.; Dono, R.: Expression of the G6PD locus on the human X chromosome is associated with demethylation of three CpG islands within 100 kb of DNA. EMBO J. 7: 401–406, 1988.329. Town, M.; Athanasiou–Metaxa, M.; Luzzatto, L.: Intragenic interspecific complementation of glucose 6–phosphate dehydrogenase in human–hamster cell hybrids. Somat. Cell Molec. Genet. 16: 97–108, 1990.330. Town, M.; Bautista, J. M.; Mason, P. J.; Luzzatto, L.: Both mutations in G6PD A– are necessary to produce the G6PD deficient phenotype. Hum. Molec. Genet. 1: 171–174, 1992.331. Usanga, E. A.; Bienzle, U.; Cancedda, K.; Fasuan, F. A.; Ajayi, O.; Luzzatto, L.: Genetic variants of human erythrocyte glucose 6–phosphate dehydrogenase: new variants in West Africa characterized by column chromatography. Ann. Hum. Genet. 40: 279–286, 1977.332. Vaca, G.; Ibarra, B.; Garcia Cruz, D.; Medina, C.; Romero, F.; Cantu, J. M.; Beutler, E.: G–6–PD Jalisco and G–6–PD Morelia: two new Mexican variants. Hum. Genet. 71: 82–85, 1985.333. Vaca, G.; Ibarra, B.; Romero, F.; Olivares, N.; Cantu, J. M.; Beutler, E.: G–6–PD Guadalajara: a

new mutant associated with chronic nonspherocytic hemolytic anemia. *Hum. Genet.* 61: 175–176, 1982.334.

Ventura, A.; Panizon, F.; Soranzo, M. R.; Veneziano, G.; Sansone, G.; Testa, U.; Luzzatto, L.: Congenital dyserythropoietic anaemia type II associated with a new type of G6PD deficiency (G6PD Gabrovizza). *Acta Haemat.* 71: 227–234, 1984.335.

Vergnes, H.; Gherardi, M.; Bouloux, C.: Erythrocyte glucose-6-phosphate dehydrogenase in the Niokolonko (Malinke of the Niokolo) of the Eastern Senegal: identification of a slow variant with normal activity (Tacoma-like). *Hum. Hered.* 25: 80–87, 1975.336.

Vergnes, H.; Gherardi, M.; Quilici, J. C.; Yoshida, A.; Giaccardy, R.: G6PD Luz-Saint-Sauveur: a new variant with abnormal electrophoretic mobility, mild enzyme deficiency and absence of hemolytic disorders. *I.R.C.S.* 7:14, 1973.337.

Vergnes, H.; Gherardi, M.; Yoshida, A.: G6PD Lozere and Trinacria-like: segregation of two non-hemolytic variants in a French family. *Hum. Genet.* 34: 293–298, 1976.338.

Vergnes, H.; Ribet, A.; Bommelaer, G.; Amadieu, J.; Brun, H.: GD(–) Muret and GD(–) Colomiers, two new variants of glucose-6-phosphate dehydrogenase associated with favism. *Hum. Genet.* 57: 332–334, 1981.339.

Vergnes, H.; Yoshida, A.;

Gourdin, D.; Gherardi, M.; Bierme, R.; Ruffie, J.: Glucose 6-phosphate dehydrogenase Toulouse: a new variant with marked instability and severe deficiency discovered in a family of Mediterranean ancestry. *Acta Haemat.* 51: 240–249, 1974.340. Viglietto, G.; Montanaro, V.; Calabro, V.; Vallone, D.; D'Urso, M.; Persico, M. G.; Battistuzzi, G.: Common glucose-6-phosphate dehydrogenase (G6PD) variants from the Italian population: biochemical and molecular characterization. *Ann. Hum. Genet.* 54: 1–15, 1990.341. Vives-Corrons, J.-L.; Kuhl, W.; Pujades, M. A.; Beutler, E.: Molecular genetics of the glucose-6-phosphate dehydrogenase (G6PD) Mediterranean variant and description of a new G6PD mutant, G6PD Andalus (1361A). *Am. J. Hum. Genet.* 47: 575–579, 1990.342. Vives-Corrons, J. L.; Feliu, E.; Pujades, M. A.; Cardellach, F.; Rozman, C.; Carreras, A.; Jou, J. M.; Vallespi, M. T.; Zuazu, F. J.: Severe glucose-6-phosphate dehydrogenase (G6PD) deficiency associated with chronic hemolytic anemia, granulocyte dysfunction, and increased susceptibility to infections: description of a new molecular variant (G6PD Barcelona). *Blood* 59: 428–434, 1982.343. Vives-Corrons, J. L.; Pujades, A.: Heterogeneity of 'Mediterranean type' glucose-6-phosphate dehydrogenase (G6PD) deficiency in

Spain and description of two new variants associated with favism. Hum. Genet. 60: 216–221, 1982.344. Vives–Corrons, J. L.; Pujades, A.; Curia, M. D.: Caracterización molecular de la glucosa–6–fosfato deshidrogenasa (G6PD) en 24 casos de déficit enzimático y descripción de una nueva variante (G6PD–Bética). Sangre 25:1049–1064, 1980.345. Vives–Corrons, J. L.; Pujades, M. A.; Petit, J.; Colomer, D.; Corbella, M.; Aguilar i Bascompte, J. L.; Merino, A.: Chronic nonspherocytic hemolytic anemia (CNSHA) and glucose 6 phosphate dehydrogenase (G6PD) deficiency in a patient with familial amyloidotic polyneuropathy (FAP): molecular study of a new variant (G6PD Clinic) with markedly acidic pH optimum. Hum. Genet. 81: 161–164, 1989.346. Vulliamy, T.; Beutler, E.; Luzzatto, L.: Variants of glucose–6–phosphate dehydrogenase are due to missense mutations spread throughout the coding region of the gene. Hum. Mutat. 2: 159–167, 1993.347. Vulliamy, T.; Mason, P.; Luzzatto, L.: The molecular basis of glucose–6–phosphate dehydrogenase deficiency. Trends Genet. 8: 138–143, 1992.348. Vulliamy, T.; Rovira, A.; Yusoff, N.; Colomer, D.; Luzzatto, L.; Vives–Corrons, J.–L.: Independent origin of single and double mutations in the human

glucose 6-phosphate dehydrogenase gene. Hum.Mutat. 8: 311–318, 1996.349. Vulliamy, T. J.: Personal Communication. London, England 1989.350. Vulliamy, T. J.; D'Urso, M.; Battistuzzi, G.; Estrada, M.; Foulkes, N. S.; Martini, G.; Calabro, V.; Poggi, V.; Giordano, R.; Town, M.; Luzzatto, L.; Persico, M. G.: Diverse point mutations in the human glucose-6-phosphate dehydrogenase gene cause enzyme deficiency and mild or severe hemolytic anemia. Proc. Nat. Acad. Sci. 85: 5171–5175, 1988.351. Vulliamy, T. J.; Kaeda, J. S.; Ait-Chafa, D.; Mangerini, R.; Roper, D.; Barbot, J.; Mehta, A. B.; Athanassiou-Metaxa, M.; Luzzatto, L.; Mason, P. J.: Clinical and haematological consequences of recurrent G6PD mutations and a single new mutation causing chronic nonspherocytic haemolytic anaemia. Brit. J. Haemat. 101: 670–675, 1998.352. Vulliamy, T. J.; Othman, A.; Town, M.; Nathwani, A.; Falusi, A. G.; Mason, P. J.; Luzzatto, L.: Polymorphic sites in the African population detected by sequence analysis of the glucose-6-phosphate dehydrogenase gene outline the evolution of the variants A and A-. Proc. Nat. Acad. Sci. 88: 8568–8571, 1991.353. Vulliamy, T. J.; Wanachiwanawin, W.; Mason, P. J.; Luzzatto, L.: G6PD Mahidol, a common deficient variant in South East Asia is caused by a

(163)glycine-to-serine mutation. Nucleic Acids Res. 17:5868, 1989.354. Vuopio, P.; Harkonen, M.; Helske, T.; Naeveri, H.: Red cellglucose-6-phosphate dehydrogenase deficiency in Finland: characterization of a new variant with severe enzyme deficiency. Scand. J. Haemat. 15:145-152, 1975.355. Vuopio, P.; Harkonen, M.; Johnsson, P.; Nuuti-nen, M.: Red cellglucose-phosphate dehydrogenase defi-ciency in Finland. Ann. Clin.Res. 5: 168-178, 1973.356. Waitz, R.; Boivin, P.; Oberling, F.; Casenave, J. P.; North, M. L.; Mayer, S.: Variante Gd(-) Strasbourg de la glucose-6-phosphatedehydrogenase. Nouv. Rev. Franc. Hemat. 10: 312-314, 1970.357. Wang, Y. M.; Patterson, J. H.; Van Eys, J.: The potential use of xylitol in glucose-6-phosphate dehydrogenase deficiency anemia. J.Clin. Invest. 50: 1421-1428, 1971.358. Weimer, T. A.; Salzano, F. M.; Hutz, M. H.: Erythrocyte isozymes and hemoglobin types in a southern Brazilian population. J. Hum.Evol. 10: 319-328, 1981.359. Weimer, T. A.; Schuler, L.; Beutler, E.; Salzano, F. M.: Gd(+)Laguna, a new rare glucose-6-phosphate dehydrogenase variant from Brazil. Hum. Genet. 65: 402-404, 1984.360. Weinreich, J.; Busch, D.; Gottstein, U.; Schaefer, J.; Rohr, J.: Ueber zwei neue Faelle von hereditaerer nichtsphaerocytaererhaemolytischer

Anaemie bei glucose-

6-phosphat-dehydrogenase-Defektin einer Nord

Deutschen Familie. Klin. Wschr. 46: 146-149, 1968.361.

Welch, S. G.; McGregor, I. A.; Williams, K.: A new variant of human erythrocyte G6PD occurring at a high frequency amongst the population of two villages in The Gambia,

West Africa. Hum. Genet. 40: 305-309, 1978.362.

Westring, D. W.; Pisciotto, A. V.: Anemia, cataracts, and seizures in patients with glucose-6-phosphate dehydrogenase deficiency. Arch. Intern. Med. 118: 385-390,

1966.363. WHO: Nomenclature of glucose-6-phosphate dehydrogenase in man. Bull. WHO 36: 319-322, 1967.

Note: See Also: Canad. Med. Assoc. J. 97: 422-424,

1967.364. WHO: Scientific group on the standardization of procedures for the study of glucose-6-phosphate dehydrogenase. WHO Techn. Rep. (pub.) Ser. No. 366:

1967.365. Wilson, W. W.: Congenital hemolytic anemia due to a deficiency of glucose 6-phosphate dehydrogenase. Rocky Mt. Med. J. 73: 160-162, 1976.366.

Witt, I.; Yoshioka, S.: Biochemical characterization of a glucose-

6-phosphatedehydrogenase variant with favism: G-6-PD Zaehringen. Klin. Wschr. 50: 205-209, 1972.367.

Wong, P. W. K.; Shih, L.-Y.; Hsia, D. Y. Y.: Characterization of glu-

cose-6-phosphate dehydrogenase among Chinese. *Nature* 208:1323-1324, 1965.368. Yermakov, N.; Tokarev, J.; Chernjak, N.; Schoenian, G.; Grieger, M.; Guckler, G.; Jacobasch, G.; Mahmudova, M.; Bahramov, S.: Newstable mutant Gd(-) variants: G6PD Tashkent and G6PD Nucus: molecularbasis of hereditary enzyme deficiency. *Acta Biol. Med. Ger.* 40:559-562, 1981.369. Yoshida, A.: Personal Communication. Duarte, Calif. 2/26/1996.370. Yoshida, A.: Amino acid substitution (histidine to tyrosine)in a glucose-6-phosphatate dehydrogenase variant (G6PD Hek-toen) associatedwith over-production. *J. Mol. Biol.* 52: 483-490, 1970.371. Yoshida, A.: A single amino acid substitution (asparagine toaspartic acid) between normal (B plus) and the common Negro variant(A plus) of human glucose-6-phosphate dehydrogenase. *Proc. Nat. Acad.Sci.* 57: 835-840, 1967.372. Yoshida, A.: Human glucose-6-phosphate dehydrogenase: purificationand characteri-zation of Negro type variant (A+) and comparison with-normal enzyme (B+). *Biochem. Genet.* 1: 81-99, 1967.373. Yoshida, A.; Baur, E. W.; Motulsky, A. G.: A Philippino glucose-6-phosphatedehydrogenase variant (G6PD Union) with enzyme deficiency and alteredsubstrate specificity. *Blood* 35: 506-513, 1970.374. Yoshida, A.;

Beutler, E.: Human glucose-6-phosphate dehydrogenase-variants: a supplementary tabulation. *Ann. Hum. Genet.* 41: 347-355, 1978.375. Yoshida, A.; Beutler, E.; Motulsky, A. G.: Table of human glucose-6-phosphatedehydrogenase variants. *Bull. WHO* 45: 243-253, 1971.376. Yoshida, A.; Stamatoyannopoulos, G.; Motulsky, A. G.: Negrovariant of glucose-6-phosphate dehydrogenase deficiency (A-) in man. *Science* 155:97-99, 1967.377. Yoshida, A.; Takizawa, T.: The same extra FokI cleavage site exists in glucose-6-phosphate dehydrogenase variants A(+) and A(-). *Am.J. Hum. Genet.* 43: 131-133, 1988.378. Yoshida, A.; Takizawa, T.; Prchal, J. T.: RFLP of the X chromosome-linked glucose-6-phosphate dehydrogenase locus in blacks. *Am. J. Hum. Genet.* 42:872-876, 1988.379. Zinkham, W. H.: A deficiency of glucose-6-phosphate dehydrogenase activity in lens from individuals with primaquine-sensitive erythrocytes. *Bull. Johns Hopkins Hosp.* 109: 206-216, 1961.380. Zuo, L.; Chen, E.; Du, C. S.; Chang, C. N.; Chiu, D. T. Y.: Genetic study of Chinese G6PD variants by direct PCR sequencing. (Abstract) *Blood* 76(suppl. 1): 51A, 1990.

[39212] 12764. Campuzano, V.; Montermini, L.; Molto, M. D.; Pi-

anese, L.; Cossee, M.; Cavalcanti, F.; Monros, E.; Rodius, F.; Duclos, F.; Monticelli, A.; Zara, F.; Canizares, J.; Koutnikova, H.; Bidichandani, S. I.; Gellera, C.; Brice, A.; Trouillas, P.; De Michele, G.; Filla, A.; De Frutos, R.; Palau, F.; Patel, P. I.; Di Donato, S.; Mandel, J.-L.; Coccozza, S.; Koenig, M.; Pandolfo, M.: Friedreich's ataxia: autosomal recessive disease caused by an intronic GAA triplet repeat expansion. *Science* 271: 1423–1427, 1996.

[39213] 12765. Ilyin, G. P.; Rialland, M.; Pigeon, C.; Guguen-Guilouzo, C.: cDNA cloning and expression analysis of new members of the mammalian F-box protein family. *Genomics* 67: 40–47, 2000.

[39214] 12766. Inase, N.; Fushimi, K.; Ishibashi, K.; Uchida, S.; Ichioka, M.; Sasaki, S.; Marumo, F.: Isolation of human aquaporin 3 gene. *J. Biol. Chem.* 270: 17913–17916, 1995.

[39215] 12767. Ishibashi, K.; Sasaki, S.; Saito, F.; Ikeuchi, T.; Marumo, F.: Structure and chromosomal localization of a human water channel (AQP3) gene. *Genomics* 27: 352–354, 1995. Note: Erratum: *Genomics* 30: 633 only, 1995.

[39216] 12768. Ma, T.; Hara, M.; Sougrat, R.; Verbavatz, J.-M.; Verkman, A. S.: Impaired stratum corneum hydration in

mice lacking epidermal waterchannel aquaporin-3. *J. Biol. Chem.* 277: 17147–17153, 2002.

- [39217] 12769. Ishibashi, K.; Sasaki, S.; Fushimi, K.; Uchida, S.; Kuwahara, M.; Saito, H.; Furukawa, T.; Nakajima, K.; Yamaguchi, Y.; Gojobori, T.; Marumo, F.: Molecular cloning and expression of a member of the aquaporin family with permeability to glycerol and urea in addition to water expressed at the basolateral membrane of kidney collecting duct cells. *Proc. Nat. Acad. Sci.* 91: 6269–6273, 1994.
- [39218] 12770. Ma, T.; Song, Y.; Yang, B.; Gillespie, A.; Carlson, E. J.; Epstein, C. J.; Verkman, A. S.: Nephrogenic diabetes insipidus in mice lacking aquaporin-3 water channels. *Proc. Nat. Acad. Sci.* 97: 4386–4391, 2000.
- [39219] 12771. Mulders, S. M.; Olde Weghuis, D.; van Boxtel, J. A. F.; Geurts van Kessel, A.; Echevarria, M.; van Os, C. H.; Deen, P. M. T.: Localization of the human gene for aquaporin 3 (AQP3) to chromosome 9, region p21–p12, using fluorescent in situ hybridization. *Cytogenet. Cell Genet.* 72:303–305, 1996.
- [39220] 12772. Sougrat, R.; Morand, M.; Gondran, C.; Barre, P.; Gobin, R.; Bonte, F.; Dumas, M.; Verbavatz, J.-M.: Functional expression of AQP3 in human skin epidermis and reconstructed epidermis. *J. Invest. Derm.* 118:678–685,

2002.

- [39221] 12773.Andersson, P.; McGuire, J.; Rubio, C.; Gradin, K.; Whitelaw, M.L.; Pettersson, S.; Hanberg, A.; Poellinger, L.: A constitutivelyactive dioxin/aryl hydrocarbon receptor induces stomach tumors. *Proc.Nat. Acad. Sci.* 99: 9990–9995, 2002.
- [39222] 12774.Ema, M.; Matsushita, N.; Sogawa, K.; Ariyama, T.; Inazawa, J.;Nemoto, T.; Ota, M.; Oshimura, M.; Fujii–Kuriyama, Y.: Human arylhydrocarbonreceptor: functional expression and chromosomal assignment to 7p21. *J.Biochem.* 116: 845–851, 1994.
- [39223] 12775.Le Beau, M. M.; Carver, L. A.; Espinosa, R., III; Schmidt, J. V.;Bradfield, C. A.: Chromosomal localization of the human AHR locusencoding the structural gene for the Ah receptor to 7p21–p15. *Cytogenet.Cell Genet.* 66: 172–176, 1994.
- [39224] 12776.Micka, J.; Milatovich, A.; Menon, A.; Grabowski, G. A.; Puga, A.;Nebert, D. W.: Human Ah receptor (AHR) gene: localization to 7p15and suggestive correlation of polymorphism with CYP1A1 inducibility. *Pharmacogenetics* 7:95–101, 1997.
- [39225] 12777.Shimizu, Y.; Nakatsuru, Y.; Ichinose, M.; Takahashi, Y.; Kume,H.; Mimura, J.; Fujii–Kuriyama, Y.; Ishikawa, T.:

Benzo[a]pyrenecarcinogenicity is lost in mice lacking the aryl hydrocarbon receptor. *Proc.Nat. Acad. Sci.* 97: 779–782, 2000.

- [39226] 12778.Adams, M. D.; Kelley, J. M.; Gocayne, J. D.; Dubnick, M.; Polymeropoulos, M. H.; Xiao, H.; Merril, C. R.; Wu, A.; Olde, B.; Moreno, R. F.; Kerlavage, A. R.; McCombie, W. R.; Venter, J. C.: Complementary DNA sequencing: expressed sequence tags and human genome project. *Science* 252: 1651–1656, 1991.
- [39227] 12779.Polvi, A.; Armstrong, E.; Lai, C.; Lemke, G.; Huebner, K.; Spritz, R. A.; Guida, L. C.; Nicholls, R. D.; Alitalo, K.: The human TYRO3 gene and pseudogene are located in chromosome 15q14–q25. *Gene* 134:289–293, 1993.
- [39228] 12780.Chen, P.; Hao, W.; Rife, L.; Wang, X. P.; Shen, D.; Chen, J.; Ogden, T.; Van Boemel, G. B.; Wu, L.; Yang, M.; Fong, H. K. W.: A photic visual cycle of rhodopsin regeneration is dependent on Rgr. *Nature Genet.* 28: 256–260, 2001.
- [39229] 12781.Chen, X.-N.; Korenberg, J. R.; Jiang, M.; Shen, D.; Fong, H. K.W.: Localization of the human RGR opsin gene to chromosome 10q23. *Hum.Genet.* 97: 720–722, 1996.
- [39230] 12782.Jiang, M.; Pandey, S.; Fong, H. K. W.: An opsin homologue in the retina and pigment epithelium. *Invest.*

Ophthalmol. Visual Sci. 34:3669–3678, 1993.

- [39231] 12783. Morimura, H.; Saindelle–Ribeaudéau, F.; Berson, E. L.; Dryja, T.P.: Mutations in RGR, encoding a light-sensitive opsin homologue, in patients with retinitis pigmentosa. (Letter) Nature Genet. 23:393–394, 1999.
- [39232] 12784. Shen, D.; Jiang, M.; Hao, W.; Tao, L.; Salazar, M.; Fong, H. K.W.: A human opsin-related gene that encodes a retinaldehyde-binding protein. Biochemistry 33: 13117–13125, 1994.
- [39233] 12785. Asano, H.; Ishida, A.; Hasegawa, M.; Ono, T.; Yoshida, M. C.; Taniguchi, M.; Kanno, M.: The mouse Mel-18 'RING-finger' gene: genomic organization, promoter analysis and chromosomal assignment. DNA Seq. 3: 369–377, 1993.
- [39234] 12786. Ishida, A.; Asano, H.; Hasegawa, M.; Koseki, H.; Ono, T.; Yoshida, M. C.; Taniguchi, M.; Kanno, M.: Cloning and chromosome mapping of the human Mel-18 gene which encodes a DNA-binding protein with a new 'RING-finger' motif. Gene 129: 249–255, 1993.
- [39235] 12787. Tagawa, M.; Sakamoto, T.; Shigemoto, K.; Matsubara, H.; Tamura, Y.; Ito, T.; Nakamura, I.; Okitsu, A.; Imai, K.; Taniguchi, M.: Expression of novel DNA-binding protein with zinc finger structure in various tumor cells. J.

Biol. Chem. 265: 20021–20026, 1990.

- [39236] 12788.Hara, E.; Yamaguchi, T.; Nojima, H.; Ide, T.; Campisi, J.; Okayama,H.; Oda, K.: Id-related genes encoding helix-loop-helix proteinsare required for G1 progression and are repressed in senescent humanfibroblasts. J. Biol. Chem. 269: 2139–2145, 1994.
- [39237] 12789.Mathew, S.; Chen, W.; Murty, V. V. V. S.; Benezra, R.; Chaganti,R. S. K.: Chromosomal assignment of human ID1 and ID2 genes. Genomics 30:385–387, 1995.
- [39238] 12790.Chen, M. X.; McPartlin, A. E.; Brown, L.; Chen, Y. H.; Barker,H. M.; Cohen, P. T. W.: A novel human protein serine/threonine phosphatase,which possesses four tetratricopeptide repeat motifs and localizesto the nucleus. EMBO J. 13: 4278–4290, 1994.
- [39239] 12791.Xu, X.; Lagercrantz, J.; Zickert, P.; Bajalica-Lagercrantz, S.;Zetterberg, A.: Chromosomal localization and 5-prime sequence ofthe human protein serine/threonine phosphatase 5-prime gene. Biochem.Biophys. Res. Commun. 218: 514–517, 1996.
- [39240] 12792.Yong, W. H.; Ueki, K.; Chou, D.; Reeves, S. A.; von Deimling, A.;Gusella, J. F.; Mohrenweiser, H. W.; Buckler, A. J.; Louis, D. N.: Cloning of a highly conserved human protein serine-threonine phosphatasegene from the

glioma candidate region on chromosome 19q13.3. *Genomics* 29:533–536, 1995.

[39241] 12793. Breitbart, R. E.; Liang, C.; Smoot, L. B.; Laheru, D. A.; Mahdavi, V.; Nadal-Ginard, B.: A fourth human MEF2 transcription factor, hMEF2D, is an early marker of the myogenic lineage. *Development* 118: 1095–1106, 1993.

[39242] 12794. Hobson, G. M.; Krahe, R.; Garcia, E.; Siciliano, M. J.; Funanage, V. L.: Regional chromosomal assignments for four members of the MADS domain transcription enhancer factor 2 (MEF2) gene family to human chromosomes 15q26, 19p12, 5q14, and 1q12–q23. *Genomics* 29: 704–711, 1995.

[39243] 12795. Mao, Z.; Bonni, A.; Xia, F.; Nadal-Vicens, M.; Greenberg, M. E.: Neuronal activity-dependent cell survival mediated by transcription factor MEF2. *Science* 286: 785–790, 1999.

[39244] 12796. Martin, J. F.; Miano, J. M.; Hustad, C. M.; Copeland, N. G.; Jenkins, N. A.; Olson, E. N.: A Mef2 gene that generates a muscle-specific isoform via alternative mRNA splicing. *Molec. Cell. Biol.* 14: 1647–1656, 1994.

[39245] 12797. Pollock, R.; Treisman, R.: Human SRF-related proteins: DNA-binding properties and potential regulatory targets. *Genes Dev.* 5: 2327–2341, 1991.

- [39246] 12798.Yu, Y.-T.; Breitbart, R. E.; Smoot, L. B.; Lee, Y.; Mahdavi, V.;Nadal-Ginard, B.: Human myocyte-specific enhancer factor 2 comprisesa group of tissue-restricted MADS box transcription factors. *GenesDev.* 6: 1783-1798, 1992.
- [39247] 12799.Youn, H.-D.; Sun, L.; Prywes, R.; Liu, J. O.: Apoptosis of T cellsmediated by Ca(2+)-induced release of the transcription factor MEF2. *Science* 286:790-793, 1999.
- [39248] 12800.Krainc, D.; Haas, M.; Ward, D. C.; Lipton, S. A.; Bruns, G.; Leifer,D.: Assignment of human myocyte-specific enhancer binding factor2C (hMEF2C) to human chromosome 5q14 and evidence that MEF2C is evolutionarily-conserved. *Genomics* 29: 809-811, 1995.
- [39249] 12801.Leifer, D.; Krainc, D.; Yu, Y.-T.; McDermott, J.; Breitbart, R.E.; Heng, J.; Neve, R. L.; Kosofsky, B.; Nadal-Ginard, B.; Lipton,S. A.: MEF2C, a MADS/MEF2-family transcription factor expressed ina laminar distribution in cerebral cortex. *Proc. Nat. Acad. Sci.* 90:1546-1550, 1993.
- [39250] 12802.Greengard, P.; Valtorta, F.; Czernik, A. J.; Benfenati, F.: Synapticvesicle phosphoproteins and regulation of synaptic function. *Science* 259:780-785, 1993.
- [39251] 12803.Li, L.; Chin, L.-S.; Greengard, P.; Copeland, N. G.;

Gilbert, D.J.; Jenkins, N. A.: Localization of the synapsin II (SYN2) gene to human chromosome 3 and mouse chromosome 6. *Genomics* 28: 365–366, 1995.

[39252] 12804. McCright, B.; Rivers, A. M.; Audlin, S.; Virshup, D. M.: The B56 family of protein phosphatase 2A (PP2A) regulatory subunits encodes differentiation-induced phosphoproteins that target PP2A to both nucleus and cytoplasm. *J. Biol. Chem.* 271: 22081–22089, 1996.

[39253] 12805. Van Hoof, C.; Aly, M. S.; Garcia, A.; Cayla, X.; Cassiman, J. J.; Merlevede, W.; Goris, J.: Structure and chromosomal localization of the human gene of the phosphotyrosyl phosphatase activator (PTPA) of protein phosphatase 2A. *Genomics* 28: 261–272, 1995.

[39254] 12806. Andre, E.; Becker–Andre, M.: Expression of an N-terminally truncated form of human focal adhesion kinase in brain. *Biochem. Biophys. Res. Commun.* 190: 140–147, 1993.

[39255] 12807. Fiedorek, F. T., Jr.; Kay, E. S.: Mapping of the focal adhesion kinase (Fadk) gene to mouse chromosome 15 and human chromosome 8. *Mammalian Genome* 6: 123–126, 1995.

[39256] 12808. Polte, T. R.; Hanks, S. K.: Interaction between focal adhesion kinase and Crk-associated tyrosine kinase sub-

strate p130-Cas. Proc.Nat. Acad. Sci. 92: 10678–10682, 1995.

[39257] 12809.Schaller, M. D.; Borgman, C.; Cobb, B. S.; Vines, R. R.; Reynolds,A. B.; Parsons, J. T.: pp125(FAK), a structurally distinctive protein-tyrosinekinase associated with focal adhesions. Proc. Nat. Acad. Sci. 89:5192–5196, 1992.

[39258] 12810.Arnemann, J.; Epplen, J. T.; Cooke, H. J.; Sauer-
mann, U.; Engel,W.; Schmidtke, J.: A human Y-
chromosomal DNA sequence expressed in testicular tissue.
Nucleic Acids Res. 15: 8713–8724, 1987.

[39259] 12811.Arnemann, J.; Jakubiczka, S.; Thuring, S.;
Schmidtke, J.: Cloning and sequence analysis of a human
Y-chromosome-derived, testicular cDNA, TSPY. Genomics
11: 108–114, 1991.

[39260] 12812.Guttenbach, M.; Muller, U.; Schmid, M.: A human
moderately repeated Y-specific DNA sequence is evolu-
tionarily conserved in the Y chromosome of the great apes.
Genomics 13: 363–367, 1992.

[39261] 12813.Jakubiczka, S.; Schnieders, F.; Schmidtke, J.: A
bovine homologue of the human TSPY gene. Genomics 17:
732–735, 1993.

[39262] 12814.Lau, Y.-F. C.: Sex chromosome genetics '99: go-

nadoblastoma, testicular and prostate cancers, and the TSPY gene. *Am. J. Hum. Genet.* 64:921–927, 1999.

- [39263] 12815. Manz, E.; Schnieders, F.; Brechlin, A. M.; Schmidtke, J.: TSPY-related sequences represent a micro-heterogeneous gene family organized as constitutive elements in DYZ5 tandem repeat units on the human Y chromosome. *Genomics* 17:726–731, 1993.
- [39264] 12816. Mazeyrat, S.; Mitchell, M. J.: Rodent Y chromosome TSPY gene is functional in rat and non-functional in mouse. *Hum. Molec. Genet.* 7:557–562, 1998.
- [39265] 12817. Schnieders, F.; Dork, T.; Arnemann, J.; Vogel, T.; Werner, M.; Schmidtke, J.: Testis-specific protein, Y-encoded (TSPY) expression in testicular tissues. *Hum. Molec. Genet.* 5: 1801–1807, 1996.
- [39266] 12818. Vogel, T.; Boettger-Tong, H.; Nanda, I.; Dechend, F.; Agulnik, A. I.; Bishop, C. E.; Schmid, M.; Schmidtke, J.: A murine TSPY. *Chromosome Res.* 6: 35–40, 1998.
- [39267] 12819. Zhang, J. S.; Yang-Feng, T. L.; Muller, U.; Mohandas, T. K.; deJong, P. J.; Lau, Y.-F. C.: Molecular isolation and characterization of an expressed gene from the human Y-chromosome. *Hum. Molec. Genet.* 1:717–726, 1992.
- [39268] 12820. Donnelly, P.; Tavaré, S.; Balding, D. J.; Griffiths, R. C.: Estimating the age of the common ancestor of men

from the ZFY intron.(Letter) Science 272:1357–1359, 1996.

[39269] 12821.Dorit, R. L.; Akashi, H.; Gilbert, W.: Absence of polymorphism at the ZFY locus on the human Y chromosome. Science 268: 1183–1185,1995.

[39270] 12822.Dorit, R. L.; Akashi, H.; Gilbert, W.: Estimating the age of the common ancestor of men from the ZFY intron.(Letter) Science 272:1361–1362, 1996.

[39271] 12823.Fu, Y.-X.; Li, W.-H.: Estimating the age of the common ancestor of men from the ZFY intron.(Letter) Science 272: 1356–1357, 1996.

[39272] 12824.Mahaffey, C. L.; Bayleran, J. K.; Yeh, G. Y.; Lee, T. C.; Page,D. C.; Simpson, E. M.: Intron/exon structure confirms that mouse Zfy1 and Zfy2 are members of the ZFY gene family. Genomics 41: 123–127,1997.

[39273] 12825.Palmer, M. S.; Berta, P.; Sinclair, A. H.; Pym, B.; Goodfellow,P. N.: Comparison of human ZFY and ZFX transcripts. Proc. Nat. Acad.Sci. 87: 1681–1685, 1990.

[39274] 12826.Rogers, J.; Samollow, P. B.; Comuzzie, A. G.: Estimating the age of the common ancestor of men from the ZFY intron.(Letter) Science 272:1360–1361, 1996.

[39275] 12827.Sinclair, A. H.; Foster, J. W.; Spencer, J. A.; Page, D. C.; Palmer,M.; Goodfellow, P. N.; Graves, J. A. M.: Se-

quences homologous to ZFY, a candidate human sex-determining gene, are autosomal in marsupials. *Nature* 336:780–783, 1988.

- [39276] 12828. Slattery, J. P.; Sanner-Wachter, L.; O'Brien, S. J.: Novel geneconversion between X–Y homologues located in the nonrecombining region of the Y chromosome in Felidae (mammalia). *Proc. Nat. Acad. Sci.* 97:5307–5312, 2000.
- [39277] 12829. Weiss, G.; von Haeseler: Estimating the age of the common ancestor of men from the ZFY intron. (Letter) *Science* 272: 1359–1360, 1996.
- [39278] 12830. Zambrowicz, B. P.; Findley, S. D.; Simpson, E. M.; Page, D. C.; Palmiter, R. D.: Characterization of the murine Zfy1 and Zfy2 promoters. *Genomics* 24:406–408, 1994.
- [39279] 12831. Mach, B.; Steimle, V.; Martinez-Soria, E.; Reith, W.: Regulation of MHC class II genes: lessons from a disease. *Annu. Rev. Immun.* 14:301–331, 1996.
- [39280] 12832. Reith, W.: Personal Communication. Geneva, Switzerland 5/30/1997.
- [39281] 12833. Scholl, T.; Mahanta, S. K.; Strominger, J. L.: Specific complex formation between the type II bare lymphocyte syndrome-associated transactivators CIITA and RFX5. *Proc. Nat. Acad. Sci.* 94: 6330–6334, 1997.

- [39282] 12834.Emery, P.; Durand, B.; Mach, B.; Reith, W.: RFX proteins, a novel family of DNA binding proteins conserved in the eukaryotic kingdom. *Nucleic Acids Res.* 24: 803–807, 1996.
- [39283] 12835.Pugliati, L.; Reith, W.; Fey, S.; Mach, B.: Mapping the RF-X gene, encoding a DNA-binding protein controlling HLA class II gene expression, to 19p13. (Abstract) *Cytogenet. Cell Genet.* 51: 1061 only, 1989.
- [39284] 12836.Mizuta, M.; Inagaki, N.; Nemoto, Y.; Matsukura, S.; Takahashi, M.; Seino, S.: Synaptotagmin III is a novel isoform of rat synaptotagmin expressed in endocrine and neuronal cells. *J. Biol. Chem.* 269: 11675–11678, 1994.
- [39285] 12837.Li, X.-J.; Wang, D.-Y.; Zhu, Y.; Guo, R.-J.; Wang, X.-D.; Lubomir, K.; Mukai, K.; Sasaki, H.; Yoshida, H.; Oka, T.; Machinami, R.; Shinmura, K.; Tanaka, M.; Sugimura, H.: Mxi1 mutations in human neurofibrosarcomas. *Jpn. J. Cancer Res.* 90: 740–746, 1999.
- [39286] 12838.Prochownik, E. V.; Grove, L. E.; Deubler, D.; Zhu, X. L.; Stephenson, R. A.; Rohr, L. R.; Yin, X.; Brothman, A. R.: Commonly occurring loss and mutation of the MXI1 gene in prostate cancer. *Genes Chromosomes Cancer* 22: 295–304, 1998.
- [39287] 12839.Schreiber-Agus, N.; Meng, Y.; Hoang, T.; Hou, H.,

Jr.; Chen, K.;Greenberg, R.; Cordon-Cardo, C.; Lee, H.-W.; DePinho, R. A.: Role of Mxi1 in ageing organ systems and the regulation of normal and neoplastic growth. *Nature* 393: 483–487, 1998.

[39288] 12840. Shapiro, D. N.; Valentine, V.; Eagle, L.; Yin, X.; Morris, S. W.; Prochownik, E. V.: Assignment of the human MAD and MXI1 genes to chromosomes 2p12–p13 and 10q24–q25. *Genomics* 23: 282–285, 1994.

[39289] 12841. Wechsler, D. S.; Hawkins, A. L.; Li, X.; Jabs, E. W.; Griffin, C. A.; Dang, C. V.: Localization of the human Mxi1 transcription factor gene (MXI1) to chromosome 10q24–q25. *Genomics* 21: 669–672, 1994.

[39290] 12842. Wechsler, D. S.; Shelly, C. A.; Dang, C. V.: Genomic organization of human MXI1, a putative tumor suppressor gene. *Genomics* 32: 466–470, 1996.

[39291] 12843. Zervos, A. S.; Gyuris, J.; Brent, R.: Mxi1, a protein that specifically interacts with Max to bind Myc–Max recognition sites. *Cell* 72: 223–232, 1993. Note: Erratum: *Cell* 79: 389 only, 1994.

[39292] 12844. Boie, Y.; Rushmore, T. H.; Darmon-Goodwin, A.; Grygorczyk, R.; Slipetz, D. M.; Metters, K. M.; Abramovitz, M.: Cloning and expression of a cDNA for the human prostanoid IP receptor. *J. Biol. Chem.* 269: 12173–12178,

1994.

- [39293] 12845. Ishikawa, T.; Tamai, Y.; Rochelle, J. M.; Hirata, M.; Namba, T.; Sugimoto, Y.; Ichikawa, A.; Narumiya, S.; Taketo, M. M.; Seldin, M.F.: Mapping of the genes encoding mouse prostaglandin D, E, and F and prostacyclin receptors. *Genomics* 32: 285–288, 1996.
- [39294] 12846. Murata, T.; Ushikubi, F.; Matsuoka, T.; Hirata, M.; Yamasaki, A.; Sugimoto, Y.; Ichikawa, A.; Aze, Y.; Tanaka, T.; Yoshida, N.; Ueno, A.; Oh-ishi, S.; Narumiya, S.: Altered pain perception and inflammatory response in mice lacking prostacyclin receptor. *Nature* 388: 678–682, 1997.
- [39295] 12847. Ogawa, Y.; Tanaka, I.; Inoue, M.; Yoshitake, Y.; Isse, N.; Nakagawa, O.; Usui, T.; Itoh, H.; Yoshimasa, T.; Narumiya, S.; Nakao, K.: Structural organization and chromosomal assignment of the human prostacyclin receptor gene. *Genomics* 27: 142–148, 1995.
- [39296] 12848. Hoffman, I.; Balling, R.: Chromosomal localization of the murine cadherin-11. *Mammalian Genome* 6: 304 only, 1995.
- [39297] 12849. Okazaki, M.; Takeshita, S.; Kawai, S.; Kikuno, R.; Tsujimura, A.; Kudo, A.; Amann, E.: Molecular cloning and characterization of OB-cadherin, a new member of cadherin family expressed in osteoblasts. *J. Biol. Chem.* 269:

12092–12098, 1994.

- [39298] 12850.Tanihara, H.; Sano, K.; Heimark, R. L.; St. John, T.; Suzuki, S.: Cloning of five human cadherins clarifies characteristic features of cadherin extracellular domain and provides further evidence for two structurally different types of cadherin. *Cell Adhes. Commun.* 2:15–26, 1994.
- [39299] 12851.Christie, P. T.; Curley, A.; Nesbit, M. A.; Chapman, C.; Genet, S.; Harper, P. S.; Keeling, S. L.; Wilkie, A. O. M.; Winter, R. M.; Thakker, R. V.: Mutational analysis in X-linked spondyloepiphyseal dysplasia tarda. *J. Clin. Endocr. Metab.* 86: 3233–3236, 2001.
- [39300] 12852.Fiedler, J.; Bittner, M.; Puhl, W.; Brenner, R. E.: Mutations in the X-linked spondyloepiphyseal dysplasia tarda (SEDL) coding sequence are not a common cause of early primary osteoarthritis in men. (Letter) *Clin. Genet.* 62: 94–95, 2002.
- [39301] 12853.Gecz, J.; Hillman, M. A.; Gedeon, A. K.; Cox, T. C.; Baker, E.; Mulley, J. C.: Gene structure and expression study of the SEDL gene for spondyloepiphyseal dysplasia tarda. *Genomics* 69: 242–251, 2000.
- [39302] 12854.Gedeon, A. K.; Colley, A.; Jamieson, R.; Thompson, E. M.; Rogers, J.; Sillence, D.; Tiller, G. E.; Mulley, J. C.; Gecz, J.: Identification of the gene (SEDL) causing X-linked

spondyloepiphyseal dysplasia tarda. *Nature Genet.* 22: 400–404, 1999.

- [39303] 12855. Gedeon, A. K.; Tiller, G. E.; Le Merrer, M.; Heuertz, S.; Tranebjaerg, L.; Chitayat, D.; Robertson, S.; Glass, I. A.; Savarirayan, R.; Cole, W. G.; Rimoin, D. L.; Kousseff, B. G.; Ohashi, H.; Zabel, B.; Munnich, A.; Gecz, J.; Mulley, J. C.: The molecular basis of X-linked spondyloepiphyseal dysplasia tarda. *Am. J. Hum. Genet.* 68: 1386–1397, 2001.
- [39304] 12856. Grunebaum, E.; Arpaia, E.; MacKenzie, J. J.; Fitzpatrick, J.; Ray, P. N.; Roifman, C. M.: A missense mutation in the SEDL gene results in delayed onset of X linked spondyloepiphyseal dysplasia in a large pedigree. (Letter) *J. Med. Genet.* 38: 409–411, 2001.
- [39305] 12857. Mumm, S.; Christie, P. T.; Finnegan, P.; Jones, J.; Dixon, P. H.; Pannett, A. A. J.; Harding, B.; Gottesman, G. S.; Thakker, R. V.; Whyte, M. P.: A five-base pair deletion in the sedlin gene causes spondyloepiphyseal dysplasia tarda in a six-generation Arkansas kindred. *J. Clin. Endocr. Metab.* 85: 3343–3347, 2000.
- [39306] 12858. Mumm, S.; Zhang, X.; Vacca, M.; D'Esposito, M.; Whyte, M. P.: The sedlin gene for spondyloepiphyseal dysplasia tarda escapes X-inactivation and contains a non-canonical splice site. *Gene* 273: 285–293, 2001.

- [39307] 12859.Takahashi, T.; Takahashi, I.; Tsuchida, S.; Oyama, K.; Komatsu,M.; Saito, H.; Takada, G.: An SEDL gene mutation in a Japanese kindred of X-linked spondyloepiphyseal dysplasia tarda. (Letter) Clin. Genet. 61:319–320, 2002.
- [39308] 12860.Tiller, G. E.; Hannig, V. L.; Dozier, D.; Carrel, L.; Trevarthen,K. C.; Wilcox, W. R.; Mundlos, S.; Haines, J. L.; Gedeon, A. K.; Gecz,J.: A recurrent RNA-splicing mutation in the SEDL gene causes X-linked spondyloepiphyseal dysplasia tarda. Am. J. Hum. Genet. 68: 1398–1407,2001.
- [39309] 12861.Whyte, M. P.; Gottesman, G. S.; Eddy, M. C.; McAlister, W. H.: X-linked recessive spondyloepiphyseal dysplasia tarda: clinical and radiographic evolution in a 6-generation kindred and review of the literature. Medicine 78: 9–25, 1999.
- [39310] 12862.Becker, M. A.; Heidler, S. A.; Bell, G. I.; Seino, S.; Le Beau,M. M.; Westbrook, C. A.; Neuman, W.; Shapiro, L. J.; Mohandas, T.K.; Roessler, B. J.; Palella, T. D.: Cloning of cDNAs for human phosphoribosylpyrophosphatesynthetases 1 and 2 and X chromosome localization of PRPS1 and PRPS2 genes. Genomics 8: 555–561, 1990.
- [39311] 12863.Buchner, G.; Montini, E.; Andolfi, G.; Quaderi, N.; Cainarca, S.;Messali, S.; Bassi, M. T.; Ballabio, A.; Meroni, G.; Franco, B.:MID2, a homologue of the Opitz syndrome

gene MID1: similarities insubcellular localization and differences in expression during development. Hum.Molec. Genet. 8: 1397–1407, 1999.

- [39312] 12864.Braverman, N.; Lin, P.; Moebius, F. F.; Obie, C.; Moser, A.; Glossmann,H.; Wilcox, W. R.; Rimoin, D. L.; Smith, M.; Kratz, L.; Kelley, R.I.; Valle, D.: Mutations in the gene encoding 3-beta-hydroxysteroid-delta(8),delta(7)-isomerasecause X-linked dominant Conradi-Hunermann syndrome. Nature Genet. 22:291–294, 1999.
- [39313] 12865.Cho, S. Y.; Kim, J. H.; Paik, Y. K.: Cholesterol biosynthesisfrom lanosterol: differential inhibition of sterol delta 8-isomeraseand other lanosterol-converting enzymes by tamoxifen. Molec. Cells 8:233–239, 1998.
- [39314] 12866.Clayton, P. T.; Kalter, D. C.; Atherton, D. J.; Besley, G. T.;Broadhead, D. M.: Peroxisomal enzyme deficiency in X-linked dominantConradi-Hunermann syndrome. J. Inherit. Metab. Dis. 12: 358–360,1989.
- [39315] 12867.Derry, J. M. J.; Gormally, E.; Means, G. D.; Zhao, W.; Meindl,A.; Kelley, R. I.; Boyd, Y.; Herman, G. E.: Mutations in a delta(8)-delta(7)sterol isomerase in the tattered mouse and X-linked dominant chondrodysplasiapunctata. Nature Genet. 22: 286–290, 1999.

- [39316] 12868.Grange, D. K.; Kratz, L. E.; Braverman, N. E.; Kelley, R. I.:CHILD syndrome caused by deficiency of 3-beta-hydroxysteroid-delta-8,delta-7-isomerase. *Am.J. Med. Genet.* 90: 328-335, 2000.
- [39317] 12869.Hanner, M.; Moebius, F. F.; Weber, F.; Grabner, M.; Striessnig,J.; Glossmann, H.: Phenylalkylamine Ca(2+) antagonist binding protein:molecular cloning, tissue distribution, and heterologous expression. *J.Biol. Chem.* 270: 7551-7557, 1995.
- [39318] 12870.Has, C.; Bruckner-Tuderman, L.; Muller, D.; Floeth, M.; Folkers,E.; Donnai, D.; Traupe, H.: The Conradi-Hunermann-Happle syndrome(CDPX2) and emopamil binding protein: novel mutations, and somaticand gonadal mosaicism. *Hum. Molec. Genet.* 9: 1951-1955, 2000.
- [39319] 12871.Holmes, R. D.; Wilson, G. N.; Hajra, A. K.: Peroxisomal enzymedeficiency in the Conradi-Hunerman (sic) form of chondrodysplasiapunctata. *New Eng. J. Med. (Letter)* 316: 1608 only, 1987.
- [39320] 12872.Ikegawa, S.; Ohashi, H.; Ogata, T.; Honda, A.; Tsukahara, M.; Kubo,T.; Kimizuka, M.; Shimode, M.; Hasegawa, T.; Nishimura, G.; Nakamura,Y.: Novel and recurrent EBP mutations in X-linked dominant chondrodysplasiapunctata. *Am. J. Med. Genet.* 94: 300-305, 2000.

- [39321] 12873.Kelley, R. I.; Wilcox, W. G.; Smith, M.; Kratz, L. E.; Moser,A.; Rimoin, D. S.: Abnormal sterol metabolism in patients with Conradi–Hunermann–Happlesyndrome and sporadic lethal chondrodysplasia punctata. *Am. J. Med.Genet.* 83: 213–219, 1999.
- [39322] 12874.Aksoy, S.; Brandriff, B. F.; Ward, A.; Little, P. F. R.; Weinshilboum,R. M.: Human nicotinamide N–methyltransferase gene: molecular cloning,structural characterization and chromosomal localization. *Genomics* 29:555–561, 1995.
- [39323] 12875.Aksoy, S.; Szumlanski, C. L.; Weinshilboum, R. M.: Human livernicotinamide N–methyltransferase: cDNA cloning, expression, and biochemicalcharacterization. *J. Biol. Chem.* 269: 14835–14840, 1994.
- [39324] 12876.Bennett, B. D.; Wang, Z.; Kuang, W.–J.; Wang, A.; Groopman, J.E.; Goeddel, D. V.; Scadden, D. T.: Cloning and characterizationof HTK, a novel transmembrane tyrosine kinase of the EPH subfamily. *J.Biol. Chem.* 269: 14211–14218, 1994.
- [39325] 12877.Berclaz, G.; Andres, A.–C.; Albrecht, D.; Dreher, E.; Ziemiecki,A.; Gusterson, B. A.; Crompton, M. R.: Expression of the receptorprotein tyrosine kinase myk–1/htk in normal and malignant mammaryepithelium. *Biochem. Bio–*

phys. Res. Commun. 226: 869–875, 1996.

- [39326] 12878. Gerety, S. S.; Wang, H. U.; Chen, Z.-F.; Anderson, D. J.: Symmetrical mutant phenotypes of the receptor EphB4 and its specific transmembrane ligand ephrin-B2 in cardiovascular development. *Molec. Cell* 4: 403–414, 1999.
- [39327] 12879. Moynihan, T. P.; Ardley, H. C.; Leek, J. P.; Thompson, J.; Brindle, N. S.; Markham, A. F.; Robinson, P. A.: Characterization of a human ubiquitin-conjugating enzyme gene UBE2L3. *Mammalian Genome* 7: 520–525, 1996.
- [39328] 12880. Flanagan, J. R.; Becker, K. G.; Ennist, D. L.; Gleason, S. L.; Driggers, P. H.; Levi, B.-Z.; Appella, E.; Ozato, K.: Cloning of a negative transcription factor that binds to the upstream conserved region of Moloney murine leukemia virus. *Molec. Cell. Biol.* 12:38–44, 1992.
- [39329] 12881. Hariharan, N.; Kelley, D. E.; Perry, R. P.: Delta, a transcription factor that binds to downstream elements in several polymerase II promoters, is a functionally versatile zinc finger protein. *Proc. Nat. Acad. Sci.* 88: 9799–9803, 1991.
- [39330] 12882. Oei, S. L.; Shi, Y.: Transcription factor Yin Yang 1 stimulates poly(ADP-ribosyl)ation and DNA repair.

Biochem. Biophys. Res. Commun. 284:450–454, 2001.

- [39331] 12883.Park, K.; Atchison, M. L.: Isolation of a candidate repressor/activator,NF-E1 (YY-1, delta), that binds to the immunoglobulin kappa 3-primeenhancer and the immunoglobulin heavy-chain micro-E1 site.. Proc.Nat. Acad. Sci. 88: 9804–9808, 1991.
- [39332] 12884.Shi, Y.; Seto, E.; Chang, L.-S.; Shenk, T.: Transcriptional repressionby YY1, a human GLI-Kruppel-related protein, and relief of repressionby adenovirus E1A protein. Cell 67: 377–388, 1991.
- [39333] 12885.Yao, Y.-L.; Dupont, B. R.; Ghosh, S.; Fang, Y.; Leach, R. J.; Seto,E.: Cloning, chromosomal localization and promoter analysis of thehuman transcription factor YY1. Nucleic Acids Res. 26: 3776–3783,1998.
- [39334] 12886.Zhu, W.; Lossie, A. C.; Camper, S. A.; Gumucio, D. L.: Chromosomallocalization of the transcription factor YY1 in the mouse and human. MammalianGenome 5: 234–236, 1994.
- [39335] 12887.Hakimi, M.-A.; Bochar, D. A.; Schmiesing, J. A.; Dong, Y.; Barak,O. G.; Speicher, D. W.; Yokomori, K.; Shiekhattar, R.: A chromatinremodelling complex that loads cohesin onto human chromosomes. Nature 418:994–998, 2002.

- [39336] 12888. Muchardt, C.; Reyes, J. C.; Bourachot, B.; Leguoy, E.; Yaniv, M.: The hbrm and BRG-1 proteins, components of the human SNF/SWI complex, are phosphorylated and excluded from the condensed chromosomes during mitosis. *EMBO J.* 15: 3394–3402, 1996.
- [39337] 12889. Muchardt, C.; Yaniv, M.: A human homologue of *Saccharomyces cerevisiae* SNF2/SWI2 and *Drosophila* brm genes potentiates transcriptional activation by the glucocorticoid receptor. *EMBO J.* 12: 4279–4290, 1993.
- [39338] 12890. Muchardt, C.; Yaniv, M.; Mattei, M.-G.: Assignment of HBRM, the human homolog of *S. cerevisiae* SNF2/SWI2 and *Drosophila* brm genes, to chromosome region 9p23–p24, by in situ hybridization. *Mammalian Genome* 5: 241–243, 1994.
- [39339] 12891. Nomura, N.; Miyajima, N.; Sazuka, T.; Tanaka, A.; Kawarabayashi, Y.; Sato, S.; Nagase, T.; Seki, N.; Ishikawa, K.; Tabata, S.: Prediction of the coding sequences of unidentified human genes. I. The coding sequences of 40 new genes (KIAA0001–KIAA0040) deduced by analysis of randomly sampled cDNA clones from human immature myeloid cell line KG-1. *DNA Res.* 1: 27–35, 1994.
- [39340] 12892. Kikuno, R.; Nagase, T.; Ishikawa, K.; Hirosawa, M.; Miyajima, N.; Tanaka, A.; Kotani, H.; Nomura, N.; Ohara,

O.: Prediction of the coding sequences of unidentified human genes. XIV. The complete sequences of 100 new cDNA clones from brain which code for large proteins in vitro. DNA Res. 6: 197–205, 1999.

[39341] 12893. Polsky, D.; Young, A. Z.; Busam, K. J.; Alani, R. M.: The transcriptional repressor of p16/Ink4a, Id1, is up-regulated in early melanomas. Cancer Res. 61: 6008–6011, 2001.

[39342] 12894. Walker, J. L.; Dixon, J.; Fenton, C. R.; Hungerford, J.; Lynch, S. A.; Stenhouses, S. A. R.; Christian, A.; Craig, I. W.: Two new mutations in exon 3 of the NDP gene: S73X and S101F associated with severe and less severe ocular phenotype, respectively. Hum. Mutat. 9: 53–56, 1997.

[39343] 12895. Lee, D. K.; Horikoshi, M.; Roeder, R. G.: Interaction of TFIID in the minor groove of the TATA element. Cell 67: 1241–1250, 1991.

[39344] 12896. Wilson, R. B.; Kiledjian, M.; Shen, C.-P.; Benezra, R.; Zwollo, P.; Dymecki, S. M.; Desiderio, S. V.; Kadesch, T.: Repression of immunoglobulin enhancers by the helix-loop-helix protein Id: implications for B-lymphoid-cell development. Molec. Cell. Biol. 11: 6185–6191, 1991.

[39345] 12897. Battaglia, G.; Princivale, A.; Forti, F.; Lizier, C.; Zeviani, M.: Expression of the SMN gene, the spinal muscular

atrophy determining gene, in the mammalian central nervous system. *Hum. Molec. Genet.* 6:1961–1971, 1997.

[39346] 12898. Burglen, L.; Lefebvre, S.; Clermont, O.; Burlet, P.; Viollet, L.; Cruaud, C.; Munnich, A.; Melki, J.: Structure and organization of the human survival motor neurone (SMN) gene. *Genomics* 32: 479–482, 1996.

[39347] 12899. Burglen, L.; Spiegel, R.; Ignatius, J.; Cobben, J. M.; Landrieu, P.; Lefebvre, S.; Munnich, A.; Melki, J.: SMN gene deletion in variant of infantile spinal muscular atrophy. (Letter) *Lancet* 346: 316–317, 1995.

[39348] 12900. Bussaglia, E.; Clermont, O.; Tizzano, E.; Lefebvre, S.; Burglen, L.; Cruaud, C.; Urtizberea, J. A.; Colomer, J.; Munnich, A.; Baiget, M.; Melki, J.: A frame-shift deletion in the survival motor neurone gene in Spanish spinal muscular atrophy patients. *Nature Genet.* 11:335–337, 1995.

[39349] 12901. Callebaut, I.; Mornon, J. P.: The human EBNA-2 coactivator p100: multidomain organization and relationship to the staphylococcal nuclease fold and to the tudor protein involved in *Drosophila melanogaster* development. *Biochem. J.* 321: 125–132, 1997.

[39350] 12902. Campbell, L.; Daniels, R. J.; Dubowitz, V.; Davies, K. E.: Maternal mosaicism for a second mutational event in a type I spinal muscular atrophy family. *Am. J. Hum. Genet.*

63: 37–44, 1998.

- [39351] 12903.Campbell, L.; Hunter, K. M. D.; Mohaghegh, P.; Tinsley, J. M.;Brasch, M. A.; Davies, K. E.: Direct interaction of Smn with dp103,a putative RNA helicase: a role for Smn in transcription regulation? Hum.Molec. Genet. 9: 1093–1100, 2000.
- [39352] 12904.Chen, K.–L.; Wang, Y. L.; Rennert, H.; Joshi, I.; Mills, J. K.;Leonard, D. G. B.; Wilson, R. B.: Duplications and de novo deletionsof the SMNt gene demonstrated by fluorescence–based carrier testingfor spinal muscular atrophy. Am. J. Med. Genet. 85: 463–469, 1999.
- [39353] 12905.Warburg, M.: Norrie's disease: a new hereditary bilateral pseudotumourof the retina. Acta Ophthal. (Copenh) 39: 757–772, 1961.
- [39354] 12906.Warburg, M.: Norrie's disease (atrofia bulborum hereditaria). ActaOphthal. 41: 134–146, 1963.
- [39355] 12907.Warburg, M.: Norrie's disease, a congenital progressive oculo–acoustico–cerebraldegeneration. Acta Ophthal. 89 (suppl.): 1–147, 1966.
- [39356] 12908.Warburg, M.; Hauge, M.; Sanger, R.: Norrie's disease and theXg blood group system: linkage data. Acta Genet. Statist. Med. 15:103–115, 1965.
- [39357] 12909.Whitnall, S. E.; Norman, R. M.: Microphthalmia and

the visual pathways: a case associated with blindness and imbecility, and sex-linked. *Brit.J. Ophthal.* 24: 229–244, 1940.

- [39358] 12910. Wilson, W. M. G.: Congenital blindness (pseudoglioma) occurring as a sex-linked developmental anomaly. *Canad. Med. Assoc. J.* 60:580–584, 1949.
- [39359] 12911. Wolff, G.; Mayerova, A.; Wienker, T. F.; Atalianis, P.; Ioannou, P.; Warburg, M.: Clinical reinvestigation and linkage analysis in the family with Episkopi blindness (Norrie disease). *J. Med. Genet.* 29:816–819, 1992.
- [39360] 12912. Wong, F.; Goldberg, M. F.; Hao, Y.: Identification of a nonsense mutation at codon 128 of the Norrie's disease gene in a male infant. *Arch. Ophthal.* 111: 1553–1557, 1993.
- [39361] 12913. Woodruff, G.; Newbury-Ecob, R.; Plaha, D. S.; Young, I. D.: Manifesting heterozygosity in Norrie's disease?. *Brit. J. Ophthal.* 77: 813–814, 1993.
- [39362] 12914. Zhu, D.; Antonarakis, S. E.; Schmeckpeper, B. J.; Diergaarde, P. J.; Greb, A. E.; Maumenee, I. H.: Microdeletion in the X-chromosome and prenatal diagnosis in a family with Norrie disease. *Am. J. Med. Genet.* 33: 485–488, 1989.
- [39363] 12915. Avner, P.; Bucan, M.; Arnaud, D.; Lehrach, H.; Rapp,

U.: A-raf oncogene localizes on mouse X chromosome to region some 10–17 centimorgans proximal to hypoxanthine phosphoribosyltransferase gene. *Somat. Cell Molec. Genet.* 13: 267–272, 1987.

[39364] 12916. Beck, T. W.; Huleihel, M.; Gunnell, M.; Bonner, T. I.; Rapp, U. R.: The complete coding sequence of the human A-raf-1 oncogene and transforming activity of a human A-raf carrying retrovirus. *Nucleic Acids Res.* 15: 595–609, 1987.

[39365] 12917. Huebner, K.; ar-Rushdi, A.; Griffin, C. A.; Isobe, M.; Kozak, C.; Emanuel, B. S.; Nagarajan, L.; Cleveland, J. L.; Bonner, T. I.; Goldsborough, M. D.; Croce, C. M.; Rapp, U.: Actively transcribed genes in the raf oncogene group, located on the X chromosome in mouse and human. *Proc. Nat. Acad. Sci.* 83: 3934–3938, 1986.

[39366] 12918. Lee, J.-E.; Beck, T. W.; Brennscheidt, U.; DeGennaro, L. J.; Rapp, U. R.: The complete sequence and promoter activity of the human A-raf-1 gene (ARAF1). *Genomics* 20: 43–55, 1994.

[39367] 12919. Mark, G. E.; Seeley, T. W.; Shows, T. B.; Mountz, J. D.: Pks, a raf-related sequence in humans. *Proc. Nat. Acad. Sci.* 83: 6312–6316, 1986.

[39368] 12920. Popescu, N. C.; Mark, G. E.: Localization of the pKs

gene, a related sequence on human chromosomes X and 7. *Oncogene* 4: 517–519, 1989.

[39369] 12921. Dowdy, S. F.; Fasching, C. L.; Araujo, D.; Lai, K.-M.; Livanos, E.; Weissman, B. E.; Stanbridge, E. J.: Suppression of tumorigenicity in Wilms' tumor by the p15.5–p14 region of chromosome 11. *Science* 254:293–295, 1991.

[39370] 12922. Dowdy, S. F.; Lai, K.-M.; Weissman, B. E.; Matsui, Y.; Hogan, B. L. M.; Stanbridge, E. J.: The isolation and characterization of a novel cDNA demonstrating an altered mRNA level in nontumorigenic Wilms' microcell hybrid cells. *Nucleic Acids Res.* 19: 5763–5769, 1991.

[39371] 12923. Farmer, A. A.; Loftus, T. M.; Mills, A. A.; Sato, K. Y.; Neill, J. D.; Tron, T.; Yang, M.; Trumpower, B. L.; Stanbridge, E. J.: Extreme evolutionary conservation of QM, a novel c-Jun associated transcription factor. *Hum. Molec. Genet.* 3: 723–728, 1994.

[39372] 12924. Kaneko, K.; Kobayashi, H.; Onodera, O.; Miyatake, T.; Tsuji, S.: Genomic organization of a cDNA (QM) demonstrating an altered mRNA level in nontumorigenic Wilms' microcell hybrid cells and its localization to Xq28. *Hum. Molec. Genet.* 1: 529–533, 1992.

[39373] 12925. Korn, B.; Sedlacek, Z.; Manca, A.; Kioschis, P.; Konnecki, D.; Lehrach, H.; Poustka, A.: A strategy for the se-

lection of transcribed sequences in the Xq28 region. Hum. Molec. Genet. 1: 235–242, 1992.

[39374] 12926. van den Ouweland, A. M. W.; Verdijk, M.; Mannens, M. M. A. M.; van Oost, B. A.: The QM gene is X-linked and therefore not involved in suppression of tumorigenesis in Wilms' tumor. Hum. Molec. Genet. 90:144–146, 1992.

[39375] 12927. Weissman, B. E.; Saxon, P. J.; Pasquale, S. R.; Jones, G. R.; Geiser, A. G.; Stanbridge, E. J.: Introduction of a normal human chromosome 11 into a Wilms' tumor cell line controls its tumorigenic expression. Science 236:175–180, 1987.

[39376] 12928. Nishimoto, T.; Sekiguchi, T.; Kai, R.; Yamashita, K.; Takahashi, T.; Sekiguchi, M.: Large-scale selection and analysis of temperature-sensitive mutants for cell reproduction from BHK cells. Somat. Cell Genet. 8:811–812, 1982.

[39377] 12929. O'Brien, T.; Tjian, R.: Functional analysis of the human TAFII250 N-terminal kinase domain. Molec. Cell 1: 905–911, 1998.

[39378] 12930. Ruppert, S.; Wang, E. H.; Tjian, R.: Cloning and expression of human TAF(II)250: a TBP-associated factor implicated in cell-cycle regulation. Nature 362: 175–179, 1993.

- [39379] 12931.Schwartz, H. E.; Holmes, S.; Meiss, H. K.: Assignment of temperature-sensitive mutations of BHK cells to the X-chromosome. (Abstract) J. Cell Biol. 75:393A, 1977.
- [39380] 12932.Schwartz, H. E.; Moser, G. C.; Holmes, S.; Meiss, H. K.: Assignment of temperature-sensitive mutations of BHK cells to the X-chromosome. Somat.Cell Genet. 5: 217-224, 1979.
- [39381] 12933.Sekiguchi, T.; Miyata, T.; Nishimoto, T.: Molecular cloning of the cDNA of human X chromosomal gene (CCG1) which complements the temperature-sensitive G(1) mutants, tsBN462 and ts13, of the BHK cell line. EMBO J. 7: 1683-1687, 1988.
- [39382] 12934.Sekiguchi, T.; Nohiro, Y.; Nakamura, Y.; Hisamoto, N.; Nishimoto, T.: The human CCG1 gene, essential for progression of the G1 phase, encodes a 210-kilodalton nuclear DNA-binding protein. Molec. Cell.Biol. 11: 3317-3325, 1991.
- [39383] 12935.Sekiguchi, T.; Yoshida, M. C.; Sekiguchi, M.; Nishimoto, T.: Isolation of a human X chromosome-linked gene essential for progression from G1 to S phase of the cell cycle. Exp. Cell Res. 169: 395-407, 1987.
- [39384] 12936.Simchen, G.: Cell cycle mutants. Annu. Rev. Genet. 12: 161-191, 1978.

- [39385] 12937.Slater, M. L.; Ozer, H. L.: Temperature-sensitive mutants of Balb-3T3 cells: description of a mutant affected in cellular and polyomavirus DNA synthesis. *Cell* 7: 289-295, 1976.
- [39386] 12938.Starr, D. B.; Hawley, D. K.: TFIIID binds in the minor groove of the TATA box. *Cell* 67: 1231-1240, 1991.
- [39387] 12939.Abel, A.; Walcott, J.; Woods, J.; Duda, J.; Merry, D. E.: Expression of expanded repeat androgen receptor produces neurologic disease in transgenic mice. *Hum. Molec. Genet.* 10: 107-116, 2001.
- [39388] 12940.Bailey, C. K.; Andriola, I. F. M.; Kampinga, H. H.; Merry, D. E.: Molecular chaperones enhance the degradation of expanded polyglutamine repeat androgen receptor in a cellular model of spinal and bulbar muscular atrophy. *Hum. Molec. Genet.* 11: 515-523, 2002.
- [39389] 12941.Batch, J. A.; Williams, D. M.; Davies, H. R.; Brown, B. D.; Evans, B. A. J.; Hughes, I. A.; Patterson, M. N.: Androgen receptor gene mutations identified by SSCP in fourteen subjects with androgen insensitivity syndrome. *Hum. Molec. Genet.* 1: 497-503, 1992.
- [39390] 12942.Beitel, L. K.; Kazemi-Esfarjani, P.; Kaufman, M.; Lumbroso, R.; DiGeorge, A. M.; Killinger, D. W.; Trifiro, M. A.; Pinsky, L.: Substitution of arginine-839 by cysteine or

histidine in the androgen receptor causes different receptor phenotypes in cultured cells and coordinated degrees of clinical androgen resistance. *J. Clin. Invest.* 94: 546–554, 1994.

- [39391] 12943. Belsham, D. D.; Pereira, F.; Greenberg, C. R.; Liao, S.; Wrogemann, K.: Leu676-to-pro mutation of the androgen receptor causes complete androgen insensitivity syndrome in a large Hutterite kindred. *Hum. Mutat.* 5: 28–33, 1995.
- [39392] 12944. Bevan, C. L.; Brown, B. B.; Davies, H. R.; Evans, B. A. J.; Hughes, I. A.; Patterson, M. N.: Functional analysis of six androgen receptor mutations identified in patients with partial androgen insensitivity syndrome. *Hum. Molec. Genet.* 5: 265–273, 1996.
- [39393] 12945. Biancalana, V.; Serville, F.; Pommier, J.; Julien, J.; Hanauer, A.; Mandel, J. L.: Moderate instability of the trinucleotide repeat in spinobulbar muscular atrophy. *Hum. Molec. Genet.* 1: 255–258, 1992.
- [39394] 12946. Boehmer, A. L. M.; Brinkmann, A. O.; Niermeijer, M. F.; Bakker, L.; Halley, D. J. J.; Drop, S. L. S.: Germ-line and somatic mosaicism in the androgen insensitivity syndrome: implications for genetic counseling. (Letter) *Am. J. Hum. Genet.* 60: 1003–1006, 1997.

- [39395] 12947.Boehmer, A. L. M.; Brinkmann, A. O.; Nijman, R. M.; Verleun-Mooijman,M. C. T.; de Ruiter, P.; Niermeijer, M. F.; Drop, S. L. S.: Phenotypicvariation in a family with partial androgen insensitivity syndromeexplained by differences in 5-alpha dihydrotestosterone availability. *J.Clin. Endocr. Metab.* 86: 1240–1246, 2001.
- [39396] 12948.Bouvattier, C.; Carel, J.-C.; Lecoindre, C.; David, A.; Sultan,C.; Bertrand, A.-M., Morel, Y.; Chaussain, J.-L.: Postnatal changesof T, LH, and FSH in 46,XY infants with mutations in the AR gene. *J.Clin. Endocr. Metab.* 87: 29–32, 2002.
- [39397] 12949.Brown, C. J.; Goss, S. J.; Lubahn, D. B.; Joseph, D. R.; Wilson,E. M.; French, F. S.; Willard, H. F.: Androgen receptor locus onthe human X chromosome: regional localization to Xq11–12 and descriptionof a DNA polymorphism. *Am. J. Hum. Genet.* 44: 264–269, 1989.
- [39398] 12950.Brown, T. R.; Lubahn, D. B.; Wilson, E. M.; Joseph, D. R.; French,F. S.; Migeon, C. J.: Deletion of the steroid-binding domain of thehuman androgen receptor gene in one family with complete androgeninsensitivity syndrome: evidence for further genetic heterogeneityin this syndrome. *Proc. Nat. Acad. Sci.* 85: 8151–8155, 1988.
- [39399] 12951.Bruggenwirth, H. T.; Boehmer, A. L. M.; Ramnarain,

S.; Verleun-Mooijman, M. C. T.; Satijn, D. P. E.; Trapman, J.; Grootegeed, J. A.; Brinkmann, A. O.: Molecular analysis of the androgen-receptor gene in a family with receptor-positive partial androgen insensitivity: an unusual type of intronic mutation. *Am. J. Hum. Genet.* 61: 1067–1077, 1997.

[39400] 12952. Buchanan, G.; Yang, M.; Harris, J. M.; Nahm, H. S.; Han, G.; Moore, N.; Bentel, J. M.; Matusik, R. J.; Horsfall, D. J.; Marshall, V. R.; Greenberg, N. M.; Tilley, W. D.: Mutations at the boundary of the hinge and ligand binding domain of the androgen receptor confer increased transactivation function. *Molec. Endocr.* 15: 46–56, 2001.

[39401] 12953. Butler, R.; Leigh, P. N.; McPhaul, M. J.; Gallo, J.-M.: Truncated forms of the androgen receptor are associated with polyglutamine expansion in X-linked spinal and bulbar muscular atrophy. *Hum. Molec. Genet.* 7: 121–127, 1998.

[39402] 12954. Muller, G.; Schempp, W.: Mapping the human ZFX locus to Xp21.3 by in situ hybridization. *Hum. Genet.* 82: 82–84, 1989.

[39403] 12955. Palmer, M. S.; Sinclair, A. H.; Berta, P.; Ellis, N. A.; Goodfellow, P. N.; Abbas, N. E.; Fellous, M.: Genetic evidence that ZFY is not the testis-determining factor. *Nature*

342: 937–939, 1989.

- [39404] 12956. Masutani, C.; Sugasawa, K.; Yanagisawa, J.; Sonoyama, T.; Ui, M.; Enomoto, T.; Takio, K.; Tanaka, K.; van der Spek, P. J.; Bootsma, D.; Hoeijmakers, J. H. J.; Hanaoka, F.: Purification and cloning of a nucleotide excision repair complex involving the xeroderma pigmentosum group C protein and a human homologue of yeast RAD23. *EMBO J.* 13:1831–1843, 1994.
- [39405] 12957. Covert, D. D.; Le, T. T.; McAndrew, P. E.; Strasswimmer, J.; Crawford, T. O.; Mendell, J. R.; Coulson, S. E.; Androphy, E. J.; Prior, T. W.; Burghes, A. H. M.: The survival motor neuron protein in spinal muscular atrophy. *Hum. Molec. Genet.* 6: 1205–1214, 1997.
- [39406] 12958. Campbell, H. D.; Fountain, S.; McLennan, I. S.; Berven, L. A.; Crouch, M. F.; Davy, D. A.; Hooper, J. A.; Waterford, K.; Chen, K.-S.; Lupski, J. R.; Ledermann, B.; Young, I. G.; Matthaei, K. I.: Flii, a gelsolin-related cytoskeletal regulator essential for early mammalian embryonic development. *Molec. Cell. Biol.* 22: 3518–3526, 2002.
- [39407] 12959. Campbell, H. D.; Fountain, S.; Young, I. G.; Claudianos, C.; Hoheisel, J. D.; Chen, K.-S.; Lupski, J. R.: Genomic structure, evolution, and expression of human FLII, a gel-

solin and leucine-rich-repeat family member: overlap with LLGL. *Genomics* 42: 46–54, 1997.

- [39408] 12960. Connelly, M. A.; Zhang, H.; Kieleczawa, J.; Anderson, C. W.: The promoters for human DNA-PK(cs) (PRKDC) and MCM4: divergently transcribed genes located at chromosome 8 band q11. *Genomics* 47: 71–83, 1998.
- [39409] 12961. Aksoy, I. A.; Wood, T. C.; Weinshilboum, R.: Human liver estrogensulfotransferase: identification by cDNA cloning and expression. *Biochem. Biophys. Res. Commun.* 200: 1621–1629, 1994.
- [39410] 12962. Bernier, F.; Leblanc, G.; Labrie, F.; Luu-The, V.: Structure of human estrogen and aryl sulfotransferase gene: two mRNA species issued from a single gene. *J. Biol. Chem.* 269: 28200–28205, 1994.
- [39411] 12963. Her, C.; Aksoy, I. A.; Kimura, S.; Brandriff, B. F.; Wasmuth, J. J.; Weinshilboum, R. M.: Human estrogen sulfotransferase gene (STE): cloning, structure, and chromosomal localization. *Genomics* 29: 16–23, 1995.
- [39412] 12964. Weinshilboum, R.: Personal Communication. Rochester, Minn. 10/9/1995.
- [39413] 12965. Weinshilboum, R. M.; Otterness, D. M.; Aksoy, I. A.; Wood, T. C.; Her, C.; Raftogianis, R. B.: Sulfotransferase molecular biology: cDNAs and genes. *FASEB J.* 11: 3–14,

1997.

- [39414] 12966. Bartley, T. D.; Bogenberger, J.; Hunt, P.; Li, Y.-S.; Lu, H. S.; Martin, F.; Chang, M.-S.; Samal, B.; Nichol, J. L.; Swift, S.; Johnson, M. J.; Hsu, R.-Y.; and 41 others: Identification and cloning of a megakaryocyte growth and development factor that is a ligand for the cytokine receptor Mpl. *Cell* 77: 1117–1124, 1994.
- [39415] 12967. Chang, M.; McNinch, J.; Basu, R.; Shutter, J.; Hsu, R.; Perkins, C.; Mar, V.; Suggs, S.; Welcher, A.; Li, L.; Lu, H.; Bartley, T.; Hunt, P.; Martin, F.; Samal, B.; Bogenberger, J.: Cloning and characterization of the human megakaryocyte growth and development factor (MGDF) gene. *J. Biol. Chem.* 270: 511–514, 1995.
- [39416] 12968. Chang, M.-S.; Hsu, R.-Y.; McNinch, J.; Copeland, N. G.; Jenkins, N. A.: The gene for murine megakaryocyte growth and development factor (thrombopoietin, Thpo) is located on mouse chromosome 16. *Genomics* 26:636–637, 1995.
- [39417] 12969. de Sauvage, F. J.; Hass, P. E.; Spencer, S. D.; Malloy, B. E.; Gurney, A. L.; Spencer, S. A.; Darbonne, W. C.; Henzel, W. J.; Wong, S. C.; Kuang, W.-J.; Oles, K. J.; Hultgren, B.; Solberg, L. A., Jr.; Goeddel, D. V.; Eaton, D. L.: Stimulation of megakaryocytopoiesis and thrombopoiesis by the

c-Mpl ligand. *Nature* 369: 533–538, 1994.

- [39418] 12970. Farese, A. M.; Hunt, P.; Grab, L. B.; MacVittie, T. J.: Combined administration of recombinant human megakaryocyte growth and development factor and granulocyte colony-stimulating factor enhances multilineage hematopoietic reconstitution in nonhuman primates after radiation-induced marrow aplasia. *J. Clin. Invest.* 97: 2145–2151, 1996.
- [39419] 12971. Foster, D. C.; Sprecher, C. A.; Grant, F. J.; Kramer, J. M.; Kuijper, J. L.; Holly, R. D.; Whitmore, T. E.; Heipel, M. D.; Bell, L. A.; Ching, A. F. T.; McGrane, V.; Hart, C.; O'Hara, P. J.; Lok, S.: Human thrombopoietin: gene structure, cDNA sequence, expression, and chromosomal localization. *Proc. Nat. Acad. Sci.* 91: 13023–13027, 1994.
- [39420] 12972. Ghilardi, N.; Wiestner, A.; Kikuchi, M.; Ohsaka, A.; Skoda, R. C.: Hereditary thrombocythaemia in a Japanese family is caused by a novel point mutation in the thrombopoietin gene. *Brit. J. Haemat.* 107: 310–316, 1999.
- [39421] 12973. Kaushansky, K.; Lok, S.; Holly, R. D.; Broudy, V. C.; Lin, N.; Bailey, M. C.; Forstrom, J. W.; Buddle, M. M.; Oort, P. J.; Hagen, F. S.; Roth, G. J.; Papayannopoulou, T.; Foster, D. C.: Promotion of megakaryocyte progenitor expansion and differentiation by the c-Mpl ligand thrombopoietin.

Nature 369: 568–571, 1994.

[39422] 12974.Kondo, T.; Okabe, M.; Sanada, M.; Kurosawa, M.; Suzuki, S.; Kobayashi,M.; Hosokawa, M.; Asaka, M.: Famil-
ial essential thrombocythemia associatedwith one–base
deletion in the 5–prime–untranslated region of the throm-
bopoietingene. Blood 92: 1091–1096, 1998.

[39423] 12975.Li, J.; Yang, C.; Xia, Y.; Bertino, A.; Glaspy, J.;
Roberts, M.;Kuter, D. J.: Thrombocytopenia caused by the
development of antibodyesto thrombopoietin. Blood 98:
3241–3248, 2001.

[39424] 12976.Lok, S.; Kaushansky, K.; Holly, R. D.; Kuijper, J. L.;
Lofton–Day,C. E.; Oort, P. J.; Grant, F. J.; Heipel, M. D.;
Burkhead, S. K.;Kramer, J. M.; Bell, L. A.; Sprecher, C. A.;
Blumberg, H.; Johnson,R.; Prunkard, D.; Ching, A. F. T.;
Mathewes, S. L.; Bailey, M. C.;Forstrom, J. W.; Buddle, M.
M.; Osborn, S. G.; Evans, S. J.; Sheppard,P. O.; Presnell, S.
R.; O'Hara, P. J.; Hagen, F. S.; Roth, G. J.;Foster, D. C.:
Cloning and expression of murine thrombopoietin cD-
NAand stimulation of platelet production in vivo. Nature
369: 565–568,1994.

[39425] 12977.Metcalf, D.: Thrombopoietin: at last. Nature 369:
519–520,1994.

[39426] 12978.Methia, N.; Louache, F.; Vainchenker, W.; Wendling,

F.: Oligodeoxynucleotides antisense to the proto-oncogene c-mpl specifically inhibit in vitro megakaryocytopoiesis. *Blood* 82: 1395–1401, 1993.

[39427] 12979. Ratajczak, M. Z.; Ratajczak, J.; Marlicz, W.; Pletcher, C. H., Jr.; Machalinski, B.; Moore, J.; Hung, H.; Gewirtz, A. M.: Recombinant human thrombopoietin (TPO) stimulates erythropoiesis by inhibiting erythroid progenitor cell apoptosis. *Brit. J. Haemat.* 98: 8–17, 1997.

[39428] 12980. Sohma, Y.; Akahori, H.; Seki, N.; Hori, T.; Ogami, K.; Kato, T.; Shimada, Y.; Kawamura, K.; Miyazaki, H.: Molecular cloning and chromosomal localization of the human thrombopoietin gene. *FEBS Lett.* 353: 57–61, 1994.

[39429] 12981. Wendling, F.; Maraskovsky, E.; Debili, N.; Florindo, C.; Teepe, M.; Titeux, M.; Methia, N.; Breton-Gorius, J.; Cosman, D.; Vainchenker, W.: c-Mpl ligand is a humoral regulator of megakaryocytopoiesis. *Nature* 369: 571–574, 1994.

[39430] 12982. Dualan, R.; Brody, T.; Keeney, S.; Nichols, A. F.; Admon, A.; Linn, S.: Chromosomal localization and cDNA cloning of the genes (DDB1 and DDB2) for the p127 and p48 subunits of a human damage-specific DNA binding protein. *Genomics* 29: 62–69, 1995.

[39431] 12983. Koken, M. H. M.; Reynolds, P.; Jaspers-Dekker, I.;

Prakash, L.;Prakash, S.; Bootsma, D.; Hoeijmakers, J. H. J.:
Structural and functional conservation of two human ho-
mologs of the yeast DNA repair gene RAD6. Proc.Nat.
Acad. Sci. 88: 8865–8869, 1991.

[39432] 12984.Knoll, A.; Schunkert, H.; Reichwald, K.; Danser, A.
H. J.; Bauer,D.; Platzer, M.; Stein, G.; Rosenthal, A.: Human
renin binding protein:complete genomic sequence and as-
sociation of an intronic T/C polymorphismwith the
prorenin level in males. Hum. Molec. Genet. 6:
1527–1534,1997.

[39433] 12985.Laan, M.; Paabo, S.: Mapping genes by drift-
generated linkage disequilibrium. (Letter) Am. J. Hum.
Genet. 63: 654–656, 1998.

[39434] 12986.Takahashi, S.; Inoue, H.; Miyake, Y.: The human
gene for renin-bindingprotein. J. Biol. Chem. 267:
13007–13013, 1992.

[39435] 12987.Terwilliger, J. D.; Zollner, S.; Laan, M.; Paabo, S.:
Mappinggenes through the use of linkage disequilibrium
generated by geneticdrift: 'drift mapping' in small popula-
tions with no demographic expansion. Hum.Hered. 48:
138–154, 1998.

[39436] 12988.van den Ouweland, A. M. W.; Verdijk, M.; Kiochis,
P.; Poustka,A.; van Oost, B. A.: The human renin-binding

protein gene (RENBP) maps in Xq28. Genomics 21: 279–281, 1994.

[39437] 12989. Warren, S. T.; Knight, S. J. L.; Peters, J. F.; Stayton, C. L.; Consalez, G. G.; Zhang, F.: Isolation of the human chromosomal band Xq28 within somatic cell hybrids by fragile X site breakage. Proc. Nat. Acad. Sci. 87:

3856–3860, 1990.

[39438] 12990. Calvo, R. M.; Asuncion, M.; Sancho, J.; San Millan, J. L.; Escobar–Morreale, H. F.: The role of the CAG repeat polymorphism in the androgen receptor gene and of skewed X–chromosome inactivation, in the pathogenesis of hirsutism. J. Clin. Endocr. Metab. 85: 1735–1740, 2000.

[39439] 12991. Gao, X.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Gridley, T.: Assignment of the murine Notch2 and Notch3 genes to chromosomes 3 and 17. Genomics 49: 160–161, 1998.

[39440] 12992. Larsson, C.; Lardelli, M.; White, I.; Lendahl, U.: The human NOTCH1, 2, and 3 genes are located at chromosome positions 9q34, 1p13–p11, and 19p13.2–p13.1 in regions of neoplasia–associated translocation. Genomics 24: 253–258, 1994.

[39441] 12993. Dichgans, M.; Herzog, J.; Gasser, T.: NOTCH3 mutation involving three cysteine residues in a family with

typical CADASIL. *Neurology* 57:1714–1717, 2001.

[39442] 12994.Joutel, A.; Corpechot, C.; Ducros, A.; Vahedi, K.; Chabriat, H.;Mouton, P.; Alamowitch, S.; Domenga, V.; Cecillon, M.; Marechal,E.; Maciazek, J.; Vayssiere, C.; Cruaud, C.; Cabanis, E.–A.; Ruchoux,M. M.; Weissenbach, J.; Bach, J. F.; Bousser, M. G.; Tournier–Lasserre,E.: Notch3 mutations in CADASIL, a hereditary adult–onset condition–causing stroke and dementia. *Nature* 383: 707–710, 1996.

[39443] 12995.Joutel, A.; Dodick, D. D.; Parisi, J. E.; Cecillon, M.; Tournier–Lasserre,E.; Bousser, M. G.: De novo mutation in the Notch3 gene causing CADASIL. *Ann.Neurol.* 47: 388–391, 2000.

[39444] 12996.Joutel, A.; Vahedi, K.; Corpechot, C.; Troesch, A.; Chabriat, H.;Vayssiere, C.; Cruaud, C.; Maciazek, J.; Weissenbach, J.; Bousser,M.–G.; Bach, J.–F.; Tournier–Lasserre, E.: Strong clusteringandstereotyped nature of Notch3 mutations in CADASIL patients. *Lancet* 350:1511–1515, 1997.

[39445] 12997.Rebay, I.; Fehon, R. G.; Artavanis–Tsakonas, S.: Specific truncationsof Drosophila Notch define dominant activated and dominant negativeforms of the receptor. *Cell* 74: 319–329, 1993.

- [39446] 12998.Deed, R. W.; Hirose, T.; Mitchell, E. L. D.; Santibanez-Koref, M. F.; Norton, J. D.: Structural organisation and chromosomal mapping of the human Id-3 gene. *Gene* 151: 309–314, 1994.
- [39447] 12999.Ellmeier, W.; Aguzzi, A.; Kleiner, E.; Kurzbauer, R.; Weith, A.: Mutually exclusive expression of a helix–loop–helix gene and N-myc in human neuroblastomas and in normal development. *EMBO J.* 11: 2563–2571, 1992.
- [39448] 13000.Kee, B. L.; Rivera, R. R.; Murre, C.: Id3 inhibits B lymphocyte progenitor growth and survival in response to TGF- β . *Nature Immun.* 2:242–247, 2001.
- [39449] 13001.Lyden, D.; Young, A. Z.; Zagzag, D.; Yan, W.; Gerald, W.; O'Reilly, R.; Bader, B. L.; Hynes, R. O.; Zhuang, Y.; Manova, K.; Benezra, R.: Id1 and Id3 are required for neurogenesis, angiogenesis and vascularization of tumour xenografts. *Nature* 401: 670–677, 1999.
- [39450] 13002.Pan, L.; Sato, S.; Frederick, J. P.; Sun, X.-H.; Zhuang, Y.: Impaired immune responses and B-cell proliferation in mice lacking the Id3 gene. *Molec. Cell. Biol.* 19: 5969–5980, 1999.
- [39451] 13003.Yeh, K.; Lim, R. W.: Genomic organization and promoter analysis of the murine Id3 gene. *Gene* 254: 163–171, 2000.

- [39452] 13004.Braun, A.; Kammerer, S.; Weissenhorn, W.; Weiss, E. H.; Cleve,H.: Sequence of a putative human housekeeping gene (HK33) localizedon chromosome 1. *Gene* 146: 291–295, 1994.
- [39453] 13005.Gotte, K.; Girzalsky, W.; Linkert, M.; Baumgart, E.; Kammerer,S.; Kunau, W.–H.; Erdmann, R.: Pex19p, a far–nesylated protein essentialfor peroxisome biogenesis. *Molec. Cell. Biol.* 18: 616–628, 1998.
- [39454] 13006.James, G. L.; Goldstein, J. L.; Pathak, R. K.; Ander–son, R. G.W.; Brown, M. S.: PxF, a prenylated protein of peroxisomes *J. Biol.Chem.* 269: 14182–14190, 1994.
- [39455] 13007.Kammerer, S.; Arnold, N.; Gutensohn, W.; Mewes, H.–W.; Kunau, W.–H.;Hofler, G.; Roscher, A. A.; Braun, A.: Genomic organization and molecularcharacterization of a gene encoding HsPXF, a human peroxisomal farnesylated–protein. *Genomics* 45: 200–210, 1997.
- [39456] 13008.Caskey, C. T.; Pizzuti, A.; Fu, Y.–H.; Fenwick, R. G., Jr.; Nelson,D. L.: Triplet repeat mutations in human dis–ease. *Science* 256:784–789, 1992.
- [39457] 13009.Chang, B.; Zheng, S. L.; Hawkins, G. A.; Isaacs, S. D.; Wiley,K. E.; Turner, A.; Carpten, J. D.; Bleecker, E. R.; Walsh, P. C.;Trent, J. M.; Meyers, D. A.; Isaacs, W. B.; Xu, J.: Polymorphic GGC repeats in the androgen receptor gene

are associated with hereditary and sporadic prostate cancer risk. *Hum. Genet.* 110: 122–129, 2002.

- [39458] 13010. Chang, C.; Kokontis, J.; Liao, S.: Molecular cloning of human and rat complementary DNA encoding androgen receptors. *Science* 240:324–326, 1988.
- [39459] 13011. Choong, C. S.; Kempainen, J. A.; Zhou, Z.-X.; Wilson, E. M.: Reduced androgen receptor gene expression with first exon CAG repeat expansion. *Molec. Endocr.* 10: 1527–1535, 1996.
- [39460] 13012. Choong, C. S.; Quigley, C. A.; French, F. S.; Wilson, E. M.: A novel missense mutation in the amino-terminal domain of the human androgen receptor gene in a family with partial androgen insensitivity syndrome causes reduced efficiency of protein translation. *J. Clin. Invest.* 98: 1423–1431, 1996.
- [39461] 13013. Chu, J.; Zhang, R.; Zhao, Z.; Zou, W.; Han, Y.; Qi, Q.; Zhang, H.; Wang, J.-C.; Tao, S.; Liu, X.; Luo, Z.: Male fertility is compatible with an Arg840Cys substitution in the AR in a large Chinese family with divergent phenotypes of AR insensitivity syndrome. *J. Clin. Endocr. Metab.* 87: 347–351, 2002.
- [39462] 13014. Coetzee, G. A.; Ross, R. K.: Re: Prostate cancer and the androgen receptor. (Letter) *J. Nat. Cancer Inst.* 86:

872–873, 1994.

- [39463] 13015. Correa–Cerro, L.; Wöhr, G.; Haussler, J.; Berthoin, P.; Drelon, E.; Mangin, P.; Fournier, G.; Cussenot, O.; Kraus, P.; Just, W.; Paiss, T.; Cantu, J. M.; Vogel, W.: (CAG)_nCAA and GGN repeats in the human androgen receptor gene are not associated with prostate cancer in a French–German population. *Europ. J. Hum. Genet.* 7: 347–362, 1999.
- [39464] 13016. Dowsing, A. T.; Yong, E. L.; Clark, M.; McLachlan, R. I.; de Kretser, D. M.; Trounson, A. O.: Linkage between male infertility and trinucleotide repeat expansion in the androgen–receptor gene. *Lancet* 354: 640–643, 1999.
- [39465] 13017. Elo, J. P.; Kvist, L.; Leinonen, K.; Isomaa, V.; Henttu, P.; Lukkarinen, O.; Vihko, P.: Mutated human androgen receptor gene detected in a prostatic cancer patient is also activated by estradiol. *J. Clin. Endocr. Metab.* 80: 3494–3500, 1995.
- [39466] 13018. Gaddipati, J. P.; McLeod, D. G.; Heidenberg, H. B.; Sesterhenn, I. A.; Finger, M. J.; Moul, J. W.; Srivastava, S.: Frequent detection of codon 877 mutation in the androgen receptor gene in advanced prostate cancers. *Cancer Res.* 54: 2861–2864, 1994.
- [39467] 13019. Gehring, U.; Tomkins, G. M.: Characterization of a hormone receptor defect in the androgen–insensitivity

mutant. *Cell* 3: 59–64, 1974.

- [39468] 13020. Gingrich, J. R.; Barrios, R. J.; Kattan, M. W.; Nahm, H. S.; Finegold, M. J.; Greenberg, N. M.: Androgen-independent prostate cancer progression in the TRAMP model. *Cancer Res.* 57: 4687–4691, 1997.
- [39469] 13021. Gingrich, J. R.; Greenberg, N. M.: A transgenic mouse prostate cancer model. *Toxicol. Path.* 24: 502–504, 1996.
- [39470] 13022. Giovannucci, E.; Stampfer, M. J.; Krithivas, K.; Brown, M.; Brufsky, A.; Talcott, J.; Hennekens, C. H.; Kantoff, P. W.: The CAG repeat within the androgen receptor gene and its relationship to prostate cancer. *Proc. Nat. Acad. Sci.* 94: 3320–3323, 1997.
- [39471] 13023. Gottlieb, B.; Beitel, L. K.; Trifiro, M. A.: Variable expressivity and mutation databases: the androgen receptor gene mutations database. *Hum. Mutat.* 17: 382–388, 2001.
- [39472] 13024. Gottlieb, B.; Trifiro, M.; Lumbroso, R.; Pinsky, L.: The androgen receptor gene mutations database. *Nucleic Acids Res.* 25: 158–162, 1997.
- [39473] 13025. Gottlieb, B.; Trifiro, M.; Lumbroso, R.; Vasiliou, D. M.; Pinsky, L.: The androgen receptor gene mutations database. *Nucleic Acids Res.* 24: 151–154, 1996.

- [39474] 13026.Griffin, J. E.: Androgen resistance--the clinical and molecular spectrum. *New Eng. J. Med.* 326: 611-618, 1992.
- [39475] 13027.Grino, P. B.; Isidro-Gutierrez, R. F.; Griffin, J. E.; Wilson, J. D.: Androgen resistance associated with a qualitative abnormality of the androgen receptor and responsive to high dose androgen therapy. *J.Clin. Endocr. Metab.* 68: 578-584, 1989.
- [39476] 13028.Hardy, D. O.; Scher, H. I.; Bogenreider, T.; Sabbatini, P.; Zhang, Z.-F.; Nanus, D. M.; Catterall, J. F.: Androgen receptor CAG repeat lengths in prostate cancer: correlation with age of onset. *J. Clin. Endocr. Metab.* 81: 4400-4405, 1996.
- [39477] 13029.Hellwinkel, O. J.-C.; Holterhus, P.-M.; Struve, D.; Marschke, C.; Homburg, N.; Hiort, O.: A unique exonic splicing mutation in the human androgen receptor gene indicates a physiologic relevance of regular androgen receptor transcript variants. *J. Clin. Endocr. Metab.* 86: 2569-2575, 2001.
- [39478] 13030.Hickey, T.; Chandy, A.; Norman, R. J.: The androgen receptor CAG repeat polymorphism and X-chromosome inactivation in Australian Caucasian women with infertility related to polycystic ovary syndrome. *J.Clin.*

Endocr. Metab. 87: 161–165, 2002.

- [39479] 13031.Hiort, O.; Sinnecker, G. H. G.; Holterhus, P.–M.; Nitsche, E.M.; Kruse, K.: Inherited and de novo androgen receptor gene mutations: investigation of single–case families. J. Pediat. 132: 939–943, 1998.
- [39480] 13032.Holterhus, P.–M.; Bruggenwirth, H. T.; Hiort, O.; Kleinkauf–Houcken, A.; Kruse, K.; Sinnecker, G. H. G.; Brinkmann, A. O.: Mosaicism due to a somatic mutation of the androgen receptor gene determines phenotype in androgen insensitivity syndrome. J. Clin. Endocr. Metab. 82: 3584–3589, 1997.
- [39481] 13033.Shibuya, H.; Yamaguchi, K.; Shirakabe, K.; Tonegawa, A.; Gotoh, Y.; Ueno, N.; Irie, K.; Nishida, E.; Matsumoto, K.: TAB1: an activator of the TAK1 MAPKKK in TGF–beta signal transduction. Science 272: 1179–1182, 1996.
- [39482] 13034.Boyd, J. M.; Subramanian, T.; Schaeper, U.; La Regina, M.; Bayley, S.; Chinnadurai, G.: A region in the C–terminus of adenovirus 2/5E1a protein is required for association with a cellular phosphoprotein and important for the negative modulation of T24–ras mediated transformation, tumorigenesis and metastasis. EMBO J. 12: 469–478, 1993.

- [39483] 13035.Furusawa, T.; Moribe, H.; Kondoh, H.; Higashi, Y.: Identification of CtBP1 and CtBP2 as corepressors of zinc finger-homeodomain factor delta-EF1. *Molec. Cell. Biol.* 19: 8581–8590, 1999.
- [39484] 13036.Katsanis, N.; Fisher, E. M. C.: A novel C-terminal binding protein (CTBP2) is closely related to CTBP1, an adenovirus E1A-binding protein, and maps to human chromosome 21q21.3. *Genomics* 47: 294–299, 1998.
- [39485] 13037.Schaeper, U.; Boyd, J. M.; Verma, S.; Uhlmann, E.; Subramanian, T.; Chinnadurai, G.: Molecular cloning and characterization of a cellular phosphoprotein that interacts with a conserved C-terminal domain of adenovirus E1A involved in negative modulation of oncogenic transformation. *Proc. Nat. Acad. Sci.* 92: 10467–10471, 1995.
- [39486] 13038.Sewalt, R. G. A. B.; Gunster, M. J.; van der Vlag, J.; Satijn, D. P. E.; Otte, A. P.: C-terminal binding protein is a transcriptional repressor that interacts with a specific class of vertebrate polycomb proteins. *Molec. Cell. Biol.* 19: 777–787, 1999.
- [39487] 13039.Zhang, Q.; Piston, D. W.; Goodman, R. H.: Regulation of corepressor function by nuclear NADH. *Science* 295: 1895–1897, 2002.
- [39488] 13040.Watanabe, T.; Inoue, S.; Hiroi, H.; Orimo, A.;

Kawashima, H.; Muramatsu, M.: Isolation of estrogen-responsive genes with a CpG island library. *Molec. Cell. Biol.* 18: 442–449, 1998.

[39489] 13041. Adra, C. N.; Kobayashi, H.; Rowley, J. D.; Lim, B.: Assignment of the human GDID4 gene, a GDP/GTP-exchange regulator, to chromosome 12p12.3. *Genomics* 24: 188–190, 1994.

[39490] 13042. Leffers, H.; Nielsen, M. S.; Andersen, A. H.; Honore, B.; Madsen, P.; Vandekerckhove, J.; Celis, J. E.: Identification of two human Rho GDP dissociation inhibitor proteins whose overexpression leads to disruption of the actin cytoskeleton. *Exp. Cell Res.* 209: 165–174, 1993.

[39491] 13043. Lelias, J.-M.; Adra, C. N.; Wulf, G. M.; Guillemot, J.-C.; Khagad, M.; Caput, D.; Lim, B.: cDNA cloning of a human mRNA preferentially expressed in hematopoietic cells and with homology to a GDP-dissociation inhibitor for the rho GTP-binding proteins. *Proc. Nat. Acad. Sci.* 90: 1479–1483, 1993.

[39492] 13044. Scherle, P.; Behrens, T.; Staudt, L. M.: Ly-GDI, a GDP-dissociation inhibitor of the RhoA GTP-binding protein, is expressed preferentially in lymphocytes. *Proc. Nat. Acad. Sci.* 90: 7568–7572, 1993.

[39493] 13045. Ishikawa, K.; Nagase, T.; Suyama, M.; Miyajima, N.;

Tanaka, A.;Kotani, H.; Nomura, N.; Ohara, O.: Prediction of coding sequencesof unidentified human genes. X. The complete sequences of 100 newcDNA clones from brain which can code for large proteins in vitro. DNARes. 5: 169–176, 1998.

[39494] 13046.Waldegger, S.; Erdel, M.; Nagl, U. O.; Barth, P.; Raber, G.; Steuer,S.; Utermann, G.; Paulmichl, M.; Lang, F.: Genomic organization andchromosomal localization of the human SGK protein kinase gene. Genomics 51:299–302, 1998.

[39495] 13047.Webster, M. K.; Goya, L.; Ge, Y.; Maiyar, A. C.; Firestone, G.L.: Characterization of sgk, a novel member of the serine/threonineprotein kinase gene family which is transcriptionally induced by glucocorticoidsand serum. Molec. Cell. Biol. 13: 2031–2040, 1993.

[39496] 13048.Aoki, K.; Ishida, R.; Kasai, M.: Isolation and characterizationof a cDNA encoding a translin-like protein, TRAX. FEBS Lett. 401:109–112, 1997.

[39497] 13049.Meng, G.; Aoki, K.; Tokura, K.; Nakahara, K.; Inazawa, J.; Kasai,M.: Genomic structure and chromosomal localization of the gene encodingTRAX, a translin-as-sociated factor X. J. Hum. Genet. 45: 305–308,2000.

[39498] 13050.Eudy, J. D.; Yao, S.; Weston, M. D.; Ma-Edmonds,

M.; Talmadge, C. B.; Cheng, J. J.; Kimberling, W. J.; Sumegi, J.: Isolation of a gene encoding a novel member of the nuclear receptor superfamily from the critical region of Usher syndrome type IIa at 1q41. *Genomics* 50:382–384, 1998.

[39499] 13051. Greschik, H.; Wurtz, J.-M.; Sanglier, S.; Bourguet, W.; van Dorsselaer, A.; Moras, D.; Renaud, J.-P.: Structural and functional evidence for ligand-independent transcriptional activation by the estrogen-related receptor 3. *Molec. Cell* 9: 303–313, 2002.

[39500] 13052. Matsuyoshi, N.; Imamura, S.: Multiple cadherins are expressed in human fibroblasts. *Biochem. Biophys. Res. Commun.* 235: 355–358, 1997.

[39501] 13053. Heron, L.; Virsolvy, A.; Peyrollier, K.; Gribble, F. M.; Le Cam, A.; Ashcroft, F. M.; Bataille, D.: Human alpha-endosulfine, a possible regulator of sulfonylurea-sensitive K(ATP) channel: molecular cloning, expression and biological properties. *Proc. Nat. Acad. Sci.* 95:8387–8391, 1998.

[39502] 13054. Scott, A. F.: Personal Communication. Baltimore, Md. 1/11/2001.

[39503] 13055. Bertilsson, G.; Heidrich, J.; Svensson, K.; Asman, M.; Jendeberg, L.; Sydow-Backman, M.; Ohlsson, R.; Postlind, H.; Blomquist, P.; Berkenstam, A.: Identification of a human nuclear receptor defines a new signaling pathway

for CYP3A induction. *Proc. Nat. Acad. Sci.* 95:
12208–12213, 1998.

[39504] 13056. Blumberg, B.; Sabbagh, W., Jr.; Juguilon, H.; Bolado, J., Jr.; van Meter, C. M.; Ong, E. S.; Evans, R. M.: SXR, a novel steroid and xenobiotic–sensing nuclear receptor. *Genes Dev.* 12: 3195–3205, 1998.

[39505] 13057. Watkins, R. E.; Wisely, G. B.; Moore, L. B.; Collins, J. L.; Lambert, M. H.; Williams, S. P.; Willson, T. M.; Kliewer, S. A.; Redinbo, M. R.: The human nuclear xenobiotic receptor PXR: structural determinants of directed promiscuity. *Science* 292: 2329–2333, 2001.

[39506] 13058. Grenet, J.; Valentine, V.; Kitson, J.; Li, H.; Farrow, S. N.; Kidd, V. J.: Duplication of the DR3 gene on human chromosome 1p36 and its deletion in human neuroblastoma. *Genomics* 49: 385–393, 1998.

[39507] 13059. Adato, A.; Raskin, L.; Petit, C.; Bonne–Tamir, B.: Deafness heterogeneity in a Druze isolate from the Middle East: novel OTOF and PDS mutations, low prevalence of GJB2 35delG mutation and indication for a new DFNB locus. *Europ. J. Hum. Genet.* 8: 437–442, 2000.

[39508] 13060. Yasunaga, S.; Grati, M.; Chardenoux, S.; Smith, T. N.; Friedman, T. B.; Lalwani, A. K.; Wilcox, E. R.; Petit, C.: OTOF encodes multiple long and short isoforms: genetic

evidence that the long ones underlierecessive deafness
DFNB9. *Am. J. Hum. Genet.* 67: 591–600, 2000.

[39509] 13061.Yasunaga, S.; Petit, C.: Physical map of the region
surroundingthe OTOFERLIN locus on chromosome
2p22–p23. *Genomics* 66: 110–112,2000.

[39510] 13062.Niederreither, K.; Subbarayan, V.; Dolle, P.; Cham-
bon, P.: Embryonicretinoic acid synthesis is essential for
early mouse post-implantationdevelopment. *Nature*
Genet. 21: 444–448, 1999.

[39511] 13063.Ono, Y.; Fukuhara, N.; Yoshie, O.: TAL1 and LIM-
only proteinssynergistically induce retinaldehyde dehy-
drogenase 2 expression inT-cell acute lymphoblastic
leukemia by acting as cofactors for GATA3. *Molec.Cell.*
Biol. 18: 6939–6950, 1998.

[39512] 13064.Perlmann, T.: Retinoid metabolism: a balancing act.
Nature Genet. 31:7–9, 2002. Note: Erratum: *Nature Genet.*
31: 221 only, 2002.

[39513] 13065.Wang, X.; Penzes, P.; Napoli, J. L.: Cloning of a
cDNA encodingan aldehyde dehydrogenase and its ex-
pression in *Escherichia coli*:recognition of retinal as sub-
strate. *J. Biol. Chem.* 271: 16288–16293,1996.

[39514] 13066.Zhao, D.; McCaffery, P.; Ivins, K. J.; Neve, R. L.;
Hogan, P.;Chin, W. W.; Drager, U. C.: Molecular identifica-

tion of a major retinoic-acid-synthesizing enzyme, a retinaldehyde-specific dehydrogenase. *Europ. J. Biochem.* 240:15–22, 1996.

[39515] 13067. McGwire, G. B.; Tan, F.; Michel, B.; Rehli, M.; Skidgel, R. A.: Identification of a membrane-bound carboxypeptidase as the mammalian homolog of duck gp180, a hepatitis B virus-binding protein. *LifeSci.* 60: 715–724, 1997.

[39516] 13068. Riley, D. A.; Tan, F.; Miletich, D. J.; Skidgel, R. A.: Chromosomal localization of the genes for human carboxypeptidase D (CPD) and the active 50-kilodalton subunit of human carboxypeptidase N (CPN1). *Genomics* 50:105–108, 1998.

[39517] 13069. Tan, F.; Rehli, M.; Krause, S. W.; Skidgel, R. A.: Sequence of human carboxypeptidase D reveals it to be a member of the regulatory carboxypeptidase family with three tandem active site domains. *Biochem. J.* 327: 81–87, 1997.

[39518] 13070. Gebhard, W.; Schube, M.; Eulitz, M.: cDNA cloning and complete primary structure of the small, active subunit of human carboxypeptidase N (kininase 1). *Eur. J. Biochem.* 178: 603–607, 1989.

[39519] 13071. Skidgel, R. A.; Bennett, C. D.; Schilling, J. W.; Tan,

F.; Weerasinghe,D. K.; Erdos, E. G.: Amino acid sequence of the N-terminus and selected tryptic peptides of the active subunit of human plasma carboxypeptidaseN: comparison with other carboxypeptidases. *Biochem. Biophys. Res. Commun.* 154: 1323–1329, 1988.

- [39520] 13072. Rajadhyaksha, A. Riviere, M.; Van Vooren, P.; Szpirer, J.; Szpirer, C.; Babin, J.; Bina, M.: Assignment of AR1, transcription factor 20 (TCF20), to human chromosome 22q13.3 with somatic cell hybrids and in situ hybridization. *Cytogenet. Cell Genet.* 81: 176–177, 1998.
- [39521] 13073. Sanz, L.; Moscat, J.; Diaz-Meco, M. T.: Molecular characterization of a novel transcription factor that controls stromelysin expression. *Molec. Cell. Biol.* 15: 3164–3170, 1995.
- [39522] 13074. Ding, H.; Descheemaeker, K.; Marynen, P.; Nelles, L.; Carvalho, T.; Carmo-Fonseca, M.; Collen, D.; Belayew, A.: Characterization of a helicase-like transcription factor involved in the expression of the human plasminogen activator inhibitor-1 gene. *DNA Cell Biol.* 15: 429–442, 1996.
- [39523] 13075. Lin, Y.; Sheridan, P. L.; Jones, K. A.; Evans, G. A.: The HIP116SNF2/SWI2-related transcription factor gene (SNF2L3) is located on human chromosome 3q25.1–q26.1 *Genomics* 27: 381–382, 1995.

- [39524] 13076.Moinova, H. R.; Chen, W.-D.; Shen, L.; Smiraglia, D.; Olechnowicz,J.; Ravi, L.; Kasturi, L.; Myeroff, L.; Plass, C.; Parsons, R.; Minna,J.; Willson, J. K. V.; Green, S. B.; Issa, J.-P.; Markowitz, S. D.: HLTF gene silencing in human colon cancer. *Proc. Nat. Acad. Sci.* 99:4562–4567, 2002.
- [39525] 13077.Sheridan, P. L.; Schorpp, M.; Voz, M. L.; Jones, K. A.: Cloning of an SNF2/SWI2-related protein that binds specifically to the SPH motifs of the SV40 enhancer and to the HIV-1 promoter. *J. Biol. Chem.* 270:4575–4587, 1995.
- [39526] 13078.Ambros, P. F.; Schmid, J.; Rumpler, S.; Binder, B. R.; de Martin,R.: Localization of the human I-kappa-B kinase-beta (IKBKB) to chromosome8p11.2 by fluorescence in situ hybridization and radiation hybridmapping. *Genomics* 54: 575–576, 1998.
- [39527] 13079.Pasparakis, M.; Courtois, G.; Hafner, M.; Schmidt-Suppran, M.;Nenci, A.; Toksoy, A.; Krampert, M.; Goebeler, M.; Gillitzer, R.;Israel, A.; Krieg, T.; Rajewsky, K.; Haase, I.: TNF-mediated inflammatory skin disease in mice with epidermis-specific deletion of IKK2. *Nature* 417:861–866, 2002.
- [39528] 13080.Rossi, A.; Kapahi, P.; Natoli, G.; Takahashi, T.; Chen, Y.; Karin,M.; Santoro, M. G.: Anti-inflammatory cyclopentenone prostaglandins are direct inhibitors of I-

kappa-B kinase. *Nature* 403: 103–108,2000.

- [39529] 13081.Shindo, M.; Nakano, H.; Sakon, S.; Yagita, H.; Mihara, M.; Okumura,K.: Assignment of I-kappa-B kinase beta (IKBKB) to human chromosomeband 8p12–p11 by in situ hybridization. *Cytogenet. Cell Genet.* 82:32–33, 1998.
- [39530] 13082.Tang, G.; Yang, J.; Minemoto, Y.; Lin, A.: Blocking caspase-3-mediatedproteolysis of IKK-beta suppresses TNF-alpha-induced apoptosis. *Molec.Cell* 8: 1005–1016, 2001.
- [39531] 13083.Woronicz, J. D.; Gao, X.; Cao, Z.; Rothe, M.; Goeddel, D. V.:IkappaB kinase-beta: NF-kappa-B activation and complex formation withIkappaB kinase-alpha and NIK. *Science* 278: 866–869, 1997.
- [39532] 13084.Yin, M.-J.; Yamamoto, Y.; Gaynor, R. B.: The anti-inflammatoryagents aspirin and salicylate inhibit the activity of I-kappa-B kinase-beta. *Nature* 396:77–80, 1998.
- [39533] 13085.Zandi, E.; Rothwarf, D. M.; Delhase, M.; Hayakawa, M.; Karin,M.: The IkappaB kinase complex (IKK) contains two kinase subunits,IKKalpha and IKKbeta, necessary for IkappaB phosphorylation and NF-kappa-Bactivation. *Cell* 91: 243–252, 1997.
- [39534] 13086.Aoki, M.; Hamada, F.; Sugimoto, T.; Sumida, S.; Akiyama, T.; Toyoshima,K.: The human cot proto-

oncogene encodes two protein serine/threonine kinases with different transforming activities by alternative initiation of translation. *J. Biol. Chem.* 268: 22723–22732, 1993.

- [39535] 13087. Kitson, J.; Raven, T.; Jiang, Y.-P.; Goeddel, D. V.; Giles, K. M.; Pun, K.-T.; Grinham, C. J.; Brown, R.; Farrow, S. N.: A death-domain-containing receptor that mediates apoptosis. *Nature* 384: 372–375, 1996.
- [39536] 13088. Marsters, S. A.; Sheridan, J. P.; Donahue, C. J.; Pitti, R. M.; Gray, C. L.; Goddard, A. D.; Bauer, K. D.; Ashkenazi, A.: Apo-3, a new member of the tumor necrosis factor receptor family, contains a death domain and activates apoptosis and NF- κ B. *Curr. Biol.* 6: 1669–1676, 1996.
- [39537] 13089. Screaton, G. R.; Xu, X.-N.; Olsen, A. L.; Cowper, A. E.; Tan, R.; McMichael, A. J.; Bell, J. I.: LARD: a new lymphoid-specific death domain containing receptor regulated by alternative pre-mRNA splicing. *Proc. Nat. Acad. Sci.* 94: 4615–4619, 1997.
- [39538] 13090. Wang, E. C. Y.; Thern, A.; Denzel, A.; Kitson, J.; Farrow, S. N.; Owen, M. J.: DR3 regulates negative selection during thymocyte development. *Molec. Cell. Biol.* 21: 3451–3461, 2001.

- [39539] 13091. Bataille, V.; Hiles, R.; Bishop, J. A. N.: Retinoblastoma, melanoma and the atypical mole syndrome. *Brit. J. Derm.* 132: 134–138, 1995.
- [39540] 13092. Chilosi, M.; Doglioni, C.; Yan, Z.; Lestani, M.; Menestrina, F.; Sorio, C.; Benedetti, A.; Vinante, F.; Pizzolo, G.; Inghirami, G.: Differential expression of cyclin-dependent kinase 6 in cortical thymocytes and T-cell lymphoblastic lymphoma/leukemia. *Am. J. Pathol.* 152:209–217, 1998.
- [39541] 13093. Costello, J. F.; Plass, C.; Arap, W.; Chapman, V. M.; Held, W. A.; Berger, M. S.; Su Huang, H. J.; Cavenee, W. K.: Cyclin-dependent kinase 6 (cdk6) amplification in human gliomas identified using two-dimensional separation of genomic DNA. *Cancer Res.* 57: 1250–1254, 1997.
- [39542] 13094. Guan, K.-L.; Jenkins, C. W.; Li, Y.; Nichols, M. A.; Wu, X.; O'Keefe, C. L.; Matera, A. G.; Xiong, Y.: Growth suppression by p18, a p16(INK4/MTS1)- and p14(INK4B/MTS2)-related CDK6 inhibitor, correlates with wild-type pRb function. *Genes Dev.* 8: 2939–2952, 1994.
- [39543] 13095. Lien, H.-C.; Lin, C.-W.; Huang, P.-H.; Chang, M.-L.; Hsu, S.-M.: Expression of cyclin-dependent kinase 6 (cdk6) and frequent loss of CD44 in nasal-nasopharyngeal NK/T-cell lymphomas: comparison with CD56-negative peripheral T-cell lymphomas. *Lab. Invest.* 80:

893–900,2000.

- [39544] 13096.Meyerson, M.; Harlow, E.: Identification of G1 kinase activityfor cdk6, a novel cyclin D partner. *Molec. Cell. Biol.* 14: 2077–2086,1994.
- [39545] 13097.Shennan, M. G.; Badin, A.–C.; Walsh, S.; Summers, A.; From, L.;McKenzie, M.; Goldstein, A. M.; Tucker, M. A.; Hogg, D.; Lassam, N.: Lack of germline CDK6 mutations in familial melanoma. *Oncogene* 19:1849–1852, 2000.
- [39546] 13098.Traboulsi, E. I.; Zimmerman, L. E.; Manz, H. J.: Cutaneous malignantmelanoma in survivors of heritable retinoblastoma. *Arch. Ophthal.* 106:1059–1061, 1988.
- [39547] 13099.Blais, A.; Labrie, Y.; Pouliot, F.; Lachance, Y.; Labrie, C.:Structure of the gene encoding the human cyclin–dependent kinase inhibitorp18 and mutational analysis in breast cancer. *Biochem. Biophys. Res.Commun.* 247: 146–153, 1998.
- [39548] 13100.Lapointe, J.; Lachance, Y.; Labrie, Y.; Labrie, C.: A p18 mutantdefective in CDK6 binding in human breast cancer cells. *Cancer Res.* 56:4586–4589, 1996.
- [39549] 13101.Bontemps, Y.; Maquart, F.–X.; , Wegrowski, Y.: Human UDP–glucosedehydrogenase gene: complete cloning and transcription start mapping. *Biochem.Biophys. Res. Commun.* 275: 981–985, 2000.

- [39550] 13102.Hempel, J.; Perozich, J.; Romovacek, H.; Hinich, A.; Kuo, I.; Feingold, D. S.: UDP-glucose dehydrogenase from bovine liver: primary structure and relationship to other dehydrogenases. *Protein Sci.* 3: 1074–1080, 1994.
- [39551] 13103.Marcu, O.; Stathakis, D. G.; Marsh, J. L.: Assignment of the UGDH locus encoding UDP-glucose dehydrogenase to human chromosome band 4p15.1 by radiation hybrid mapping. *Cytogenet. Cell Genet.* 86: 244–245, 1999.
- [39552] 13104.Spicer, A. P.; Kaback, L. A.; Smith, T. J.; Seldin, M. F.: Molecular cloning and characterization of the human and mouse UDP-glucose dehydrogenase genes. *J. Biol. Chem.* 273: 25117–25124, 1998.
- [39553] 13105.Walsh, E. C.; Stainier, D. Y. R.: UDP-glucose dehydrogenase required for cardiac valve formation in zebrafish. *Science* 293: 1670–1674, 2001.
- [39554] 13106.Bernard, M.; Sanseau, P.; Henry, C.; Couturier, A.; Prigent, C.: Cloning of STK13, a third human protein kinase related to *Drosophila* Aurora and budding yeast Ipl1 that maps on chromosome 19q13.3-ter. *Genomics* 53:406–409, 1998.
- [39555] 13107.Tseng, T.-C.; Chen, S.-H.; Hsu, Y.-P. P.; Tang, T. K.: Protein kinase profile of sperm and eggs: cloning and

characterization of two novel testis-specific protein kinases (AIE1, AIE2) related to yeast and fly chromosome segregation regulators. *DNA Cell Biol.* 17:823–833, 1998.

- [39556] 13108. Becker, W.; Weber, Y.; Wetzel, K.; Eirmbter, K.; Tejedor, F. J.; Joost, H.-G.: Sequence characteristics, sub-cellular localization, and substrate specificity of DYRK-related kinases, a novel family of dual specificity protein kinases. *J. Biol. Chem.* 273: 25893–25902, 1998.
- [39557] 13109. Hsu, H.; Xiong, J.; Goeddel, D. V.: The TNF receptor 1-associated protein TRADD signals cell death and NF- κ B activation. *Cell* 81:495–504, 1995.
- [39558] 13110. Pan, M.-G.; Xiong, J.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Goeddel, D. V.: Sequence, genomic organization, and chromosomal localization of the mouse TRADD gene. *J. Inflamm.* 46: 168–175, 1996.
- [39559] 13111. Bao, J.; Zervos, A. S.: Isolation and characterization of Nmi, a novel partner of Myc proteins. *Oncogene* 12: 2171–2176, 1996.
- [39560] 13112. Zhu, M.; John, S.; Berg, M.; Leonard, W. J.: Functional association of Nmi with Stat5 and Stat1 in IL-2- and IFN- γ -mediated signaling. *Cell* 96:121–130, 1999.
- [39561] 13113. Phillips, N. J.; Zeigler, M. R.; Deaven, L. L.: A cDNA from the ovarian cancer critical region of deletion on chro-

mosome 17p13.3. *CancerLett.* 102: 85–90, 1996.

[39562] 13114.Schultz, D. C.; Vanderveer, L.; Berman, D. B.; Hamilton, T. C.;Wong, A. J.; Godwin, A. K.: Identification of two candidate tumorsuppressor genes on chromosome 17p13.3. *Cancer Res.* 56: 1997–2002,1996.

[39563] 13115.Kirchhausen, T.; Davis, A. C.; Frucht, S.; O'Brine Greco, B.; Payne,G. S.; Tubb, B.: AP17 and AP19, the mammalian small chains of theclathrin-associated protein complexes show homology to Yap17p, theirputative homolog in yeast. *J. Biol. Chem.* 266: 11153–11157, 1991.

[39564] 13116.Peyrard, M.; Parveneh, S.; Lagercrantz, S.; Ekman, M.; Fransson,I.; Sahlen, S.; Dumanski, J. P.: Cloning, expression pattern, andchromosomal assignment to 16q23 of the human gamma-adaptin gene (ADTG). *Genomics* 50:275–280, 1998.

[39565] 13117.Takatsu, H.; Sakurai, M.; Shin, H.–W.; Murakami, K.; Nakayama,K.: Identification and characterization of novel clathrin adaptor-relatedproteins. *J. Biol. Chem.* 273: 24693–24700, 1998.

[39566] 13118.Albarosa, R.; Colombo, B. M.; Roz, L.; Magnani, I.; Pollo, B.;Cirenei, N.; Giani, C.; Conti, A. M.; DiDonato, S.; Finocchiaro, G.: Deletion mapping of gliomas suggests the presence of two small regionsfor candidate tumor–

suppressor genes in a 17-cM interval on chromosome 10q.
Am. J. Hum. Genet. 58: 1260–1267, 1996.

- [39567] 13119. Maier, D.; Zhang, Z.; Taylor, E.; Hamou, M.-F.; Gratzl, O.; VanMeir, E. G.; Scott, R. J.; Merlo, A.: Somatic deletion mapping on chromosome 10 and sequence analysis of PTEN/MMAC1 point to the 10q25–26 region as the primary target in low-grade and high-grade gliomas. Oncogene 16:3331–3335, 1998.
- [39568] 13120. Rasheed, B. K.; McLendon, R. E.; Friedman, H. S.; Friedman, A. H.; Fuchs, H. E.; Bigner, D. D.; Bigner, S. H.: Chromosome 10 deletion mapping in human gliomas: a common deletion region in 10q25. Oncogene 10:2243–2246, 1995.
- [39569] 13121. Andre, E.; Conquet, F.; Steinmayr, M.; Stratton, S. C.; Porciatti, V.; Becker-Andre, M.: Disruption of retinoid-related orphan receptor beta changes circadian behavior, causes retinal degeneration and leads to vacillans phenotype in mice. EMBO J. 17: 3867–3877, 1998.
- [39570] 13122. Paravicini, G.; Steinmayr, M.; Andre, E.; Becker-Andre, M.: The metastasis suppressor candidate nucleotide diphosphate kinase NM23 specifically interacts with members of the ROR/RZR nuclear orphan receptor subfamily. Biochem. Biophys. Res. Commun. 227: 82–87, 1996.

- [39571] 13123.Sirlin, J. L.: Vacillans, a neurological mutant in the house mouse linked with brown. *J. Genet.* 54: 42–48, 1956.
- [39572] 13124.Vidal–Puig, A.; Solanes, G.; Grujic, D.; Flier, J. S.; Lowell, B. B.: UCP3: an uncoupling protein homologue expressed preferentially and abundantly in skeletal muscle and brown adipose tissue. *Biochem. Biophys. Res. Commun.* 235: 79–82, 1997.
- [39573] 13125.Vidal–Puig, A. J.; Grujic, D.; Zhang, C.–Y.; Hagen, T.; Boss, O.; Ido, Y.; Szczepanik, A.; Wade, J.; Mootha, V.; Cortright, R.; Muoio, D. M.; Lowell, B. B.: Energy metabolism in uncoupling protein3 gene knockout mice. *J. Biol. Chem.* 275: 16258–16266, 2000.
- [39574] 13126.Lovering, R.; Hanson, I. M.; Borden, K. L. B.; Martin, S.; O'Reilly, N. J.; Evan, G. I.; Rahman, D.; Pappin, D. J. C.; Trowsdale, J.; Freemont, P. S.: Identification and preliminary characterization of a protein motif related to the zinc finger. *Proc. Nat. Acad. Sci.* 90: 2112–2116, 1993.
- [39575] 13127.Bourdi, M.; Demady, D.; Martin, J. L.; Jabbour, S. K.; Martin, B. M.; George, J. W.; Pohl, L. R.: cDNA cloning and baculovirus expression of the human liver endoplasmic reticulum P58: characterization as a protein disulfide isomerase isoform, but not as a protease or acarnitine acyl–

transferase. Arch. Biochem. Biophys. 323: 397–403, 1995.

[39576] 13128. Briquet–Laugier, V.; Xia, Y.–R.; Rooke, K.; Mehra–bian, M.; Lysis, A. J.; Doolittle, M. H.: Mapping of three members of the mouse protein disulfide isomerase family. Mammalian Genome 9: 176–177, 1998.

[39577] 13129. Hirano, N.; Shibasaki, F.; Sakai, R.; Tanaka, T.; Nishida, J.; Yazaki, Y.; Takenawa, T.; Hirai, H.: Molecular cloning of the human glucose–regulated protein ERp57/GRP58, a thiol–dependent reductase: identification of its secretory form and inducible expression by the oncogenic transformation. Europ. J. Biochem. 234: 336–342, 1995.

[39578] 13130. Koivunen, P.; Helaakoski, T.; Annunen, P.; Veijola, J.; Raisanen, S.; Pihlajaniemi, T.; Kivirikko, K. I.: ERp60 does not substitute for protein disulphide isomerase as the beta–subunit of prolyl 4–hydroxylase. Biochem. J. 316: 599–605, 1996.

[39579] 13131. Koivunen, P.; Horelli–Kuitunen, N.; Helaakoski, T.; Karvonen, P.; Jaakkola, M.; Palotie, A.; Kivirikko, K. I.: Structures of the human gene for the protein disulfide isomerase–related polypeptide ERp60 and a processed gene and assignment of these genes to 15q15 and 1q21. Genomics 42: 397–404, 1997.

- [39580] 13132.Oliver, J. D.; van der Wal, F. J.; Bulleid, N. J.; High, S.: Interaction of the thiol-dependent reductase ERp57 with nascent glycoproteins. *Science* 275:86–88, 1997.
- [39581] 13133.Didsbury, J.; Weber, R. F.; Bokoch, G. M.; Evans, T.; Snyderman, R.: rac, a novel ras-related family of proteins that are botulinum toxin substrates. *J. Biol. Chem.* 264: 16378–16382, 1989.
- [39582] 13134.Palmiter, R. D.; Cole, T. B.; Quaife, C. J.; Findley, S. D.: ZnT3, a putative transporter of zinc into synaptic vesicles. *Proc. Nat. Acad. Sci.* 93: 14934–14939, 1996.
- [39583] 13135.Palmiter, R. D.; Findley, S. D.: Cloning and functional characterization of a mammalian zinc transporter that confers resistance to zinc. *EMBOJ.* 14: 639–649, 1995.
- [39584] 13136.Vallee, B. L.; Falchuk, K. H.: The biochemical basis of zinc physiology. *Physiol. Rev.* 73: 79–118, 1993.
- [39585] 13137.Berthelsen, J.; Viggiano, L.; Schulz, H.; Ferretti, E.; Consalez, G. G.; Rocchi, M.; Blasi, F.: PKNOX1, a gene encoding PREP1, a new regulator of Pbx activity, maps on human chromosome 21q22.3 and murine chromosome 17B/C. *Genomics* 47: 323–324, 1998.
- [39586] 13138.Chen, H.; Rossier, C.; Nakamura, Y.; Lynn, A.; Chakravarti, A.; Antonarakis, S. E.: Cloning of a novel

homeobox-containing gene, PKNOX1, and mapping to human chromosome 21q22.3. *Genomics* 41: 193–200, 1997.

- [39587] 13139. Morita, K.; Furuse, M.; Fujimoto, K.; Tsukita, S.: Claudin multigenefamily encoding four-transmembrane domain protein components of tightjunction strands. *Proc. Nat. Acad. Sci.* 96: 511–516, 1999.
- [39588] 13140. Chiang, P.-W.; Fogel, E.; Jackson, C. L.; Lieuallen, K.; Lennon, G.; Qu, X.; Wang, S.-Q.; Kurnit, D. M.: Isolation, sequencing, and mapping of the human homologue of the yeast transcription factor, SPT5. *Genomics* 38: 421–424, 1996.
- [39589] 13141. Chiang, P.-W.; Stubbs, L.; Zhang, L.; Kurnit, D. M.: Isolation of murine SPT5 homologue: completion of the isolation and characterization of human and murine homologues of yeast chromatin structural protein complex SPT4, SPT5, and SPT6. *Genomics* 47: 426–428, 1998.
- [39590] 13142. Grover, J.; Chen, X.-N.; Korenberg, J. R.; Roughley, P. J.: The structure and chromosome location of the human chondroadherin gene (CHAD). *Genomics* 45: 379–385, 1997.
- [39591] 13143. Larsson, T.; Sommarin, Y.; Paulsson, M.; Antons-son, P.; Hedbom, E.; Wendel, M.; Heinegard, D.: Cartilage matrix proteins: a basic 36-kDa protein with a restricted

distribution to cartilage and bone. J.Biol. Chem. 266: 20428–20433, 1991.

[39592] 13144.Tribioli, C.; Frasch, M.; Lufkin, T.: Bapx1: an evolutionary conservedhomologue of the Drosophila bagpipe homeobox gene is expressed insplanchnic mesoderm and the embryonic skeleton. Mech. Dev. 65: 145–162,1997.

[39593] 13145.Tribioli, C.; Lufkin, T.: Molecular cloning, chromosomal mappingand developmental expression of BAPX1, a novel human homeobox–containinggene homologous to Drosophila bagpipe. Gene 203: 225–233, 1997.

[39594] 13146.Yoshiura, K.–I.; Murray, J. C.: Sequence and chromosomal assignmentof human BAPX1, a bagpipe–related gene, to 4p16.1: a candidate genefor skeletal dysplasia. Genomics 45: 425–428, 1997.

[39595] 13147.de Coo, R. F. M.; Buddiger, P.; Smeets, H. J. M.; van Oost, B.A.: Molecular cloning and characterization of the human mitochondrialNADH:oxidoreductase 10–kDa gene (NDUFV3). Genomics 45: 434–437,1997.

[39596] 13148.Fritzler, M. J.; Lung, C.–C.; Hamel, J. C.; Griffith, K. J.; Chan,E. K. L.: Molecular characterization of golgin–245, a novel Golgicomplex protein containing a granin signature. J. Biol. Chem. 270:31262–31268, 1995.

[39597] 13149.Chen, L.–S. K.; Lo, C. F.; Numann, R.; Cuddy, M.:

Characterization of the human and rat phospholemman (PLM) cDNAs and localization of the human PLM gene to chromosome 19q13.1. *Genomics* 41: 435–443, 1997.

- [39598] 13150. Bianchi, M. C.; Tosetti, M.; Fornai, F.; Alessandri, M. G.; Cipriani, P.; De Vito, G.; Canapicchi, R.: Reversible brain creatine deficiency in two sisters with normal blood creatine level. *Ann. Neurol.* 47: 511–513, 2000.
- [39599] 13151. Humm, A.; Fritsche, E.; Mann, K.; Gohl, M.; Huber, R.: Recombinant expression and isolation of human L-arginine:glycine amidinotransferase and identification of its active-site cysteine residue. *Biochem. J.* 322: 771–776, 1997.
- [39600] 13152. Humm, A.; Huber, R.; Mann, K.: The amino acid sequences of human and pig L-arginine:glycine amidinotransferase. *FEBS Lett.* 339: 101–107, 1994.
- [39601] 13153. Item, C. B.; Stockler-Ipsiroglu, S.; Stromberger, C.; Muhl, A.; Alessandri, M. G.; Bianchi, M. C.; Tosetti, M.; Fornai, F.; Cioni, G.: Arginine:glycine amidinotransferase deficiency: the third inborn error of creatine metabolism in humans. *Am. J. Hum. Genet.* 69: 1127–1133, 2001.
- [39602] 13154. Yasuda, H.; Shima, N.; Nakagawa, N.; Yamaguchi, K.; Kinosaki, M.; Mochizuki, S.; Tomoyasu, A.; Yano, K.; Goto, M.; Murakami, A.; Tsuda, E.; Morinaga, T.; Higashio,

K.; Udagawa, N.; Takahashi, N.; Suda, T.: Osteoclast differentiation factor is a ligand for osteoprotegerin/osteoclastogenesis-inhibitory factor and is identical to TRANCE/RANKL. *Proc. Nat. Acad. Sci.* 95:3597–3602, 1998.

[39603] 13155. Bey, F.; Silva Pereira, I.; Coux, O.; Viegas-Pequignot, E.; RecillasTarga, F.; Nothwang, H. G.; Dutrillaux, B.; Scherrer, K.: The prosomal RNA-binding protein p27K is a member of the alpha-type human prosomal gene family. *Molec. Gen. Genet.* 237: 193–205, 1993.

[39604] 13156. Heard, D. J.; Norby, P. L.; Holloway, J.; Vissing, H.: Human ERR-gamma, a third member of the estrogen receptor-related receptor (ERR) subfamily of orphan nuclear receptors: tissue-specific isoforms are expressed during development in the adult. *Molec. Endocr.* 14: 382–392, 2000.

[39605] 13157. Hong, H.; Yang, L.; Stallcup, M. R.: Hormone-independent transcriptional activation and coactivator binding by novel orphan nuclear receptor ERR3. *J. Biol. Chem.* 274: 22618–22626, 1999.

[39606] 13158. Fisher, D. A.; Smith, J. F.; Pillar, J. S.; St. Denis, S. H.; Cheng, J. B.: Isolation and characterization of PDE9A, a novel human cGMP-specific phosphodiesterase. *J. Biol. Chem.* 273: 15559–15564, 1998.

- [39607] 13159. Guipponi, M.; Scott, H. S.; Kudoh, J.; Kawasaki, K.; Shibuya, K.; Shintani, A.; Asakawa, S.; Chen, H.; Lalioti, M. D.; Rossier, C.; Minoshima, S.; Shimizu, N.; Antonarakis, S. E.: Identification and characterization of a novel cyclic nucleotide phosphodiesterase gene (PDE9A) that maps to 21q22.3: alternative splicing of mRNA transcripts, genomic structure and sequence. *Hum. Genet.* 103: 386–392, 1998.
- [39608] 13160. Soderling, S. H.; Bayuga, S. J.; Beavo, J. A.: Identification and characterization of a novel family of cyclic nucleotide phosphodiesterases. *J. Biol. Chem.* 273: 15553–15558, 1998.
- [39609] 13161. Ishibashi, K.; Kuwahara, M.; Gu, Y.; Kageyama, Y.; Tohsaka, A.; Suzuki, F.; Marumo, F.; Sasaki, S.: Cloning and functional expression of a new water channel abundantly expressed in the testis permeable to water, glycerol, and urea. *J. Biol. Chem.* 272: 20782–20786, 1997.
- [39610] 13162. Ishibashi, K.; Yamauchi, K.; Kageyama, Y.; Saito-Ohara, F.; Ikeuchi, T.; Marumo, F.; Sasaki, S.: Molecular characterization of human aquaporin-7 gene and its chromosomal mapping. *Biochim. Biophys. Acta* 1399: 62–66, 1998.
- [39611] 13163. Kuriyama, H.; Kawamoto, S.; Ishida, N.; Ohno, I.;

Mita, S.; Matsuzawa, Y.; Matsubara, K.; Okubo, K.: Molecular cloning and expression of a novel human aquaporin from adipose tissue with glycerol permeability.

Biochem. Biophys. Res. Commun. 241: 53–58, 1997.

[39612] 13164. Billin, A. N.; Eilers, A. L.; Queva, C.; Ayer, D. E.:

Mlx, a novel Max-like BHLHZip protein that interacts with the Max network of transcription factors. J. Biol. Chem. 274: 36344–36350, 1999.

[39613] 13165. Bjerknes, M.; Cheng, H.: TCFL4: a gene at 17q21.1

encoding a putative basic helix–loop–helix leucine–zipper transcription factor. Gene 181:7–11, 1996.

[39614] 13166. Cairo, S.; Merla, G.; Urbinati, F.; Ballabio, A.; Rey-

mond, A.: WBSCR14, a gene mapping to the Williams–Beuren syndrome deleted region, is a new member of the Mlx transcription factor network. Hum. Molec. Genet. 10: 617–627, 2001.

[39615] 13167. Fukuoka, S.-I.; Freedman, S. D.; Scheele, G. A.: A

single gene encodes membrane-bound and free forms of GP-2, the major glycoprotein in pancreatic secretory (zymogen) granule membranes. Proc. Nat. Acad. Sci. 88: 2898–2902, 1991.

[39616] 13168. Fukuoka, S.-I.; Freedman, S. D.; Yu, H.; Sukhatme,

V. P.; Scheele, G. A.: GP-2/THP gene family encodes self-

binding glycosylphosphatidylinositol-anchored proteins in apical secretory compartments of pancreas and kidney. Proc.Nat. Acad. Sci. 89: 1189–1193, 1992.

- [39617] 13169.Fukuoka, S.-I.; Suzuki, M.; Okabayashi, K.; Takahashi, E.: Assignment of pancreatic zymogen granule membrane protein GP2 (GP2) to human chromosome band 9q21.11 to q21.2 by in situ hybridization. Cytogenet.Cell Genet. 79: 231–232, 1997.
- [39618] 13170.Kooy, J.; Toh, B.-H.; Pettitt, J. M.; Erlich, R.; Gleeson, P. A.: Human autoantibodies as reagents to conserved Golgi components: characterization of a peripheral, 230-kDa compartment-specific Golgi protein. J. Biol. Chem. 267: 20255–20263, 1992.
- [39619] 13171.Snow, B. E.; Antonio, L.; Suggs, S.; Gutstein, H. B.; Siderovski, D. P.: Molecular cloning and expression analysis of rat Rgs12 and Rgs14. Biochem. Biophys. Res. Commun. 233: 770–777, 1997.
- [39620] 13172.Kimple, R. J.; Kimple, M. E.; Betts, L.; Sondek, J.; Siderovski, D. P.: Structural determinants for GoLoco-induced inhibition of nucleotide release by G-alpha subunits. Nature 416: 878–881, 2002.
- [39621] 13173.Takesono, A.; et al; et al: Receptor-independent activators of heterotrimeric G-protein signalling pathways.

J. Biol. Chem. 274:33202–33205, 1999.

[39622] 13174.Traver, S.; Bidot, C.; Spassky, N.; Baltauss, T.; de Tand, M.–F.;Thomas, J.–L.; Zalc, B.; Janoueix–Lerosey, I.; de Gunzburg, J.: RGS14is a novel Rap effector that preferentially regulates the GTPase activityof G–alpha–0.

Biochem. J. 350: 19–29, 2000.

[39623] 13175.Buckbinder, L.; Velasco–Miguel, S.; Chen, Y.; Xu, N.; Talbott,R.; Gelbert, L.; Gao, J.; Seizinger, B. R.; Gutkind, J. S.; Kley,N.: The p53 tumor suppressor targets a novel regulator of G proteinsignaling. Proc. Nat. Acad. Sci. 94: 7868–7872, 1997.

[39624] 13176.Chen, C.–K.; Wieland, T.; Simon, M. I.: RGS–r, a retinal specificRGS protein, binds an intermediate conformation of transducin andenhances recycling. Proc. Nat. Acad. Sci. 93: 12885–12889, 1996.

[39625] 13177.Snow, B. E.; Antonio, L.; Suggs, S.; Siderovski, D. P.: Cloningof a retinally abundant regulator of G–protein signaling (RGS–r/RGS16):genomic structure and chromosomal localization of the human gene. Gene 206:247–253, 1998.

[39626] 13178.Zheng, B.; Chen, D.; Farquhar, M. G.: MIR16, a putative membraneglycerophosphodiester phosphodiesterase, interacts with RGS16. Proc.Nat. Acad. Sci. 97:

3999–4004, 2000.

- [39627] 13179. LeBoeuf, R. C.; Caldwell, M.; Guo, Y.; Metz, C.; Davitz, M. A.; Olson, L. K.; Deeg, M. A.: Mouse glycosylphosphatidylinositol-specific phospholipase D (Gpld1) characterization. *Mammalian Genome* 9: 710–714, 1998.
- [39628] 13180. Scallan, B. J.; Fung, W.-J. C.; Tsang, T. C.; Li, S.; Kado-Fong, H.; Huang, K.-S.; Kochan, J. P.: Primary structure and functional activity of a phosphatidylinositol-glycan-specific phospholipase D. *Science* 252:446–448, 1991.
- [39629] 13181. Schofield, J. N.; Rademacher, T. W.: Structure and expression of the human glycosylphosphatidylinositol phospholipase D1 (GPLD1) gene. *Biochim. Biophys. Acta* 1494: 189–194, 2000.
- [39630] 13182. Tsang, T. C.; Fung, W.-J.; Levine, J.; Metz, C. N.; Davitz, M. A.; Burns, D. K.; Huang, K.-S.; Kochan, J. P.: Isolation and expression of two human glycosylphosphatidylinositol phospholipase D (GPI-PLD) cDNAs. (Abstract) *FASEB J. (supp.)* 6: A1922 only, 1992.
- [39631] 13183. Schmitz, F.; Konigstorfer, A.; Sudhof, T. C.: RIBEYE, a component of synaptic ribbons: a protein's journey through evolution provides insight into synaptic ribbon function. *Neuron* 28: 857–872, 2000.

- [39632] 13184. Bergelson, J. M.; Cunningham, J. A.; Droguett, G.; Kurt-Jones, E. A.; Krithivas, A.; Hong, J. S.; Horwitz, M. S.; Crowell, R. L.; Finberg, R. W.: Isolation of a common receptor for coxsackie B viruses and adenoviruses 2 and 5. *Science* 275: 1320–1323, 1997.
- [39633] 13185. Bowles, K. R.; Gibson, J.; Wu, J.; Shaffer, L. G.; Towbin, J. A.; Bowles, N. E.: Genomic organization and chromosomal localization of the human Coxsackievirus B-adenovirus receptor gene. *Hum. Genet.* 105:354–359, 1999.
- [39634] 13186. Carson, S. D.; Chapman, N. N.; Tracy, S. M.: Purification of the putative coxsackievirus B receptor from HeLa cells. *Biochem. Biophys. Res. Commun.* 233: 325–328, 1997.
- [39635] 13187. Griffin, L. D.; Kearney, D.; Ni, J.; Jaffe, R.; Fricker, F. J.; Webber, S.; Demmler, G.; Gelb, B. D.; Towbin, J. A.: Analysis of formalin-fixed and frozen myocardial autopsy samples for viral genome in childhood myocarditis and dilated cardiomyopathy with endocardial fibroelastosis using polymerase chain reaction (PCR). *Cardiovasc. Path.* 4: 3–11, 1995.
- [39636] 13188. Martin, A. B.; Webber, S.; Fricker, F. J.; Jaffe, R.; Demmler, G.; Kearney, D.; Zhang, Y.-H.; Bodurtha, J.; Gelb,

B.; Ni, J.; Bricker, J. T.; Towbin, J. A.: Acute myocarditis: rapid diagnosis by PCR in children. *Circulation* 90: 330–339, 1994.

[39637] 13189. Pauschinger, M.; Bowles, N. E.; Fuentes–Garcia, F. J.; Pham, V.; Kuhl, U.; Schwimmbeck, P. L.; Schultheiss, H.–P.; Towbin, J. A.: Detection of adenoviral genome in the myocardium of adult patients with idiopathic left ventricular dysfunction. *Circulation* 99: 1348–1354, 1999.

[39638] 13190. Tomko, R. P.; Xu, R.; Philipson, L.: HCAR and MCAR: the human and mouse receptors for subgroup C adenoviruses and group B coxsackieviruses. *Proc. Nat. Acad. Sci.* 94: 3352–3356, 1997.

[39639] 13191. Margolis, R. L.; Abraham, M. R.; Gatchell, S. B.; Li, S.–H.; Kidwai, A. S.; Breschel, T. S.; Stine, O. C.; Callahan, C.; McInnis, M. G.; Ross, C. A.: cDNAs with long CAG trinucleotide repeats from human brain. *Hum. Genet.* 100: 114–122, 1997.

[39640] 13192. Dale, M.; Hammond, D. W.; Cox, A.; Nicklin, M. J. H.: The human gene encoding the interleukin–1 receptor accessory protein (IL1RAP) maps to chromosome 3q28 by fluorescence in situ hybridization and radiation hybrid mapping. *Genomics* 47: 325–326, 1998.

[39641] 13193. Huang, J.; Gao, X.; Li, S.; Cao, Z.: Recruitment of

IRAK to the interleukin 1 receptor complex requires interleukin 1 receptor accessory protein. *Proc. Nat. Acad. Sci.* 94: 12829–12832, 1997.

[39642] 13194. Wesche, H.; Korbherr, C.; Kracht, M.; Falk, W.; Resch, K.; Martin, M. U.: The interleukin–1 receptor accessory protein (IL–1RAcP) is essential for IL–1–induced activation of interleukin–1 receptor–associated kinase (IRAK) and stress–activated protein kinases (SAP kinases). *J. Biol. Chem.* 272: 7727–7731, 1997.

[39643] 13195. Saha, P.; Chen, J.; Thome, K. C.; Lawlis, S. J.; Hou, Z.–H.; Hendricks, M.; Parvin, J. D.; Dutta, A.: Human CDC6/Cdc18 associates with Orc1 and cyclin–cdk and is selectively eliminated from the nucleus at the onset of S phase. *Molec. Cell. Biol.* 18: 2758–2767, 1998.

[39644] 13196. Williams, R. S.; Shohet, R. V.; Stillman, B.: A human protein related to yeast Cdc6p. *Proc. Nat. Acad. Sci.* 94: 142–147, 1997.

[39645] 13197. Yan, Z.; DeGregori, J.; Shohet, R.; Leone, G.; Stillman, B.; Nevins, J. R.; Williams, R. S.: Cdc6 is regulated by E2F and is essential for DNA replication in mammalian cells. *Proc. Nat. Acad. Sci.* 95: 3603–3608, 1998.

[39646] 13198. Brais, B.; Bouchard, J.–P.; Xie, Y.–G.; Rochefort, D. L.; Chretien, N.; Tome, F. M. S.; Lafreniere, R. G.; Rom–

mens, J. M.; Uyama, E.; Nohira, O.; Blumen, S.; Korczyn, A. D.; Heutink, P.; Mathieu, J.; Duranceau, A.; Codere, F.; Fardeau, M.; Rouleau, G. A.: Short GCGexpansions in the PABP2 gene cause oculopharyngeal muscular dystrophy. *Nature Genet.* 18: 164–167, 1998. Note: Erratum: *Nature Genet.* 19: 404 only, 1998.

[39647] 13199. Calado, A.; Tome, F. M. S.; Brais, B.; Rouleau, G. A.; Kuhn, U.; Wahle, E.; Carmo-Fonseca, M.: Nuclear inclusions in oculopharyngeal muscular dystrophy consist of poly(A) binding protein 2 aggregates which sequester poly(A) RNA. *Hum. Molec. Genet.* 9: 2321–2328, 2000.

[39648] 13200. Fan, X.; Dion, P.; Laganier, J.; Brais, B.; Rouleau, G. A.: Oligomerization of polyalanine expanded PABPN1 facilitates nuclear protein aggregation that is associated with cell death. *Hum. Molec. Genet.* 10: 2341–2351, 2001.

[39649] 13201. Ikeda, S.; Shiva, N.; Ikeda, A.; Smith, R. S.; Nusinowitz, S.; Yan, G.; Lin, T. R.; Chu, S.; Heckenlively, J. R.; North, M. A.; Naggert, J. K.; Nishina, P. M.; Duyao, M. P.: Retinal degeneration but not obesity is observed in null mutants of the tubby-like protein 1 gene. *Hum. Molec. Genet.* 9: 155–163, 2000.

[39650] 13202. North, M. A.; Naggert, J. K.; Yan, Y.; Noben-Trauth, K.; Nishina, P. M.: Molecular characterization of TUB,

TULP1, and TULP2, members of the novel tubby gene family and their possible relation to ocular diseases. *Proc. Nat. Acad. Sci.* 94: 3128–3133, 1997.

- [39651] 13203. Paloma, E.; Hjelmqvist, L.; Bayes, M.; Garcia-Sandoval, B.; Ayuso, C.; Balcells, S.; Gonzalez-Duarte, R.: Novel mutations in the TULP1 gene causing autosomal recessive retinitis pigmentosa. *Invest. Ophthalmol. Vis. Sci.* 41: 656–659, 2000.
- [39652] 13204. Aoki, N.; Ishii, T.; Ohira, S.; Yamaguchi, Y.; Negi, M.; Adachi, T.; Nakamura, R.; Matsuda, T.: Stage specific expression of milkfat globule membrane glycoproteins in mouse mammary gland: comparison of MFG-E8, butyrophilin, and CD36 with a major milk protein, beta-casein. *Biochim. Biophys. Acta* 1334: 182–190, 1997.
- [39653] 13205. Collins, C.; Nehlin, J. O.; Stubbs, J. D.; Kowbel, D.; Kuo, W.-L.; Parry, G.: Mapping of a newly discovered human gene homologous to the apoptosis associated-murine mammary protein, MFG-E8, to chromosome 15q25. *Genomics* 39: 117–118, 1997.
- [39654] 13206. Haggqvist, B.; Naslund, J.; Sletten, K.; Westermarck, G. T.; Mucchiano, G.; Tjernberg, L. O.; Nordstedt, C.; Engstrom, U.; Westermarck, P.: Medin: an integral fragment of aortic smooth muscle cell-produced lactadherin forms the

most common human amyloid. Proc. Nat. Acad.Sci. 96: 8669–8674, 1999.

[39655] 13207.Hanayama, R.; Tanaka, M.; Miwa, K.; Shinohara, A.; Iwamatsu, A.;Nagata, S.: Identification of a factor that links apoptotic cellsto phagocytes. Nature 417: 182–187, 2002.

[39656] 13208.Larocca, D.; Peterson, J. A.; Urrea, R.; Kuniyoshi, J.; Bistrain,A. M.; Ceriani, R. L.: A M(r) 46,000 human milk fat globule proteinthat is highly expressed in human breast tumors contains factor VIII–likedomains. Cancer Res. 51: 4994–4998, 1991.

[39657] 13209.Stubbs, J. D.; Lekutis, C.; Singer, K. L.; Bui, A.; Yuzuki, D.;Srinivasan, U.; Parry, G.: cDNA cloning of a mouse mammary epithelialcell surface protein reveals the existence of epidermal growth factor–likedomains linked to factor VIII–like sequences. Proc. Nat. Acad. Sci. 87:8417–8421, 1990.

[39658] 13210.Zhang, J.; Kuehl, P.; Green, E. D.; Touchman, J. W.; Watkins, P.B.; Daly, A.; Hall, S. D.; Maurel, P.; Relling, M.; Brimer, C.; Yasuda,K.; Wrighton, S. A.; Hancock, M.; Kim, R. B.; Strom, S.; Thummel,K.; Russell, C. G.; Hudson, J. R., Jr.; Schuetz, E. G.; Boguski, M.S.: The human pregnane X receptor: genomic structure and identificationand func–

tional characterization of natural allelic variants. Pharmacogenetics 11:555–572, 2001.

- [39659] 13211.Su, L.-K.; Qi, Y.: Characterization of human MAPRE genes and their proteins. Genomics 71: 143–149, 2001.
- [39660] 13212.Arai, T.; Akiyama, Y.; Okabe, S.; Ando, M.; Endo, M.; Yuasa, Y.: Genomic structure of the human Smad3 gene and its infrequent alterations in colorectal cancers. Cancer Lett. 122: 157–163, 1998.
- [39661] 13213.Zhang, Y.; Feng, X.-H.; Wu, R.-Y.; Derynck, R.: Receptor-associated Mad homologues synergize as effectors of the TGF-beta response. Nature 383:168–172, 1996.
- [39662] 13214.Zhu, Y.; Richardson, J. A.; Parada, L. F.; Graff, J. M.: Smad3 mutant mice develop metastatic colorectal cancer. Cell 94: 703–714, 1998.
- [39663] 13215.Bruno, E.; Horrigan, S. K.; Van Den Berg, D.; Rozler, E.; Fitting, P. R.; Moss, S. T.; Westbrook, C.; Hoffman, R.: The Smad5 gene is involved in the intracellular signaling pathways that mediate the inhibitory effects of transforming growth factor-beta on human hematopoiesis. Blood 91:1917–1923, 1998.
- [39664] 13216.Gemma, A.; Hagiwara, K.; Vincent, F.; Ke, Y.; Hancock, A. R.; Nagashima, M.; Bennett, W. P.; Harris, C. C.: hSmad5 gene, a human hSmad family member: its full

length cDNA, genomic structure, promoter region and mutation analysis in human tumors. *Oncogene* 16: 951–956, 1998.

[39665] 13217. Cole, A. M.; Kim, Y.-H.; Tahk, S.; Hong, T.; Weis, P.; Waring, A. J.; Ganz, T.: Calcitermin, a novel antimicrobial peptide isolated from human airway secretions. *FEBS Lett.* 504: 5–10, 2001.

[39666] 13218. Guignard, F.; Mauel, J.; Markert, M.: Identification and characterization of a novel human neutrophil protein related to the S100 family. *Biochem. J.* 309: 395–401, 1995.

[39667] 13219. Ilg, E. C.; Troxler, H.; Burgisser, D. M.; Kuster, T.; Markert, M.; Guignard, F.; Hunziker, P.; Birchler, N.; Heizmann, C. W.: Amino acid sequence determination of human S100A12 (P6, Calgranulin C, CGRP, CAAF1) by tandem mass spectrometry. *Biochem. Biophys. Res. Commun.* 225: 146–150, 1996.

[39668] 13220. Marti, T.; Erttmann, K. D.; Gallin, M. Y.: Host–parasite interaction in human onchocerciasis: identification and sequence analysis of a novel human calgranulin. *Biochem. Biophys. Res. Commun.* 221: 454–458, 1996.

[39669] 13221. Wicki, R.; Marenholz, I.; Mischke, D.; Schafer, B. W.; Heizmann, C. W.: Characterization of the human S100A12

(calgranulin C, p6, CAAF1, CGRP) gene, a new member of the S100 gene cluster on chromosome 1q21. *Cell Calcium* 20: 459–464, 1996.

[39670] 13222. Tan, K. B.; Harrop, J.; Reddy, M.; Young, P.; Terrett, J.; Emery, J.; Moore, G.; Truneh, A.: Characterization of a novel TNF-like ligand and recently described TNF ligand and TNF receptor superfamily genes and their constitutive and inducible expression in hematopoietic and non-hematopoietic cells. *Gene* 204: 35–46, 1997.

[39671] 13223. Bonini, J. A.; Martin, S. K.; Dralyuk, F.; Roe, M. W.; Philipson, L. H.; Steiner, D. F.: Cloning, expression, and chromosomal mapping of a novel human CC-chemokine receptor (CCR10) that displays high-affinity binding for MCP-1 and MCP-3. *DNA Cell Biol.* 16: 1249–1256, 1997.

[39672] 13224. Nibbs, R. J. B.; Wylie, S. M.; Yang, J.; Landau, N. R.; Graham, G. J.: Cloning and characterization of a novel promiscuous human beta-chemokine receptor D6. *J. Biol. Chem.* 272: 32078–32083, 1997.

[39673] 13225. Zieger, B.; Tran, H.; Hainmann, I.; Wunderle, D.; Zgaga-Griesz, A.; Blaser, S.; Ware, J.: Characterization and expression analysis of two human septin genes, PNUTL1 and PNUTL2. *Gene* 261: 197–203, 2000.

[39674] 13226. Brandt, S.; Jentsch, T. J.: ClC-6 and ClC-7 are two

novel broadly expressed members of the CLC chloride channel family. FEBS Lett. 377:15–20, 1995.

- [39675] 13227. Eggermont, J.; Buyse, G.; Voets, T.; Tytgat, J.; De Smedt, H.; Droogmans, G.: Alternative splicing of CLC-6 (a member of the CLC chloride-channel family) transcripts generates three truncated isoforms one of which, CLC-6c, is kidney-specific. Biochem. J. 325: 269–276, 1997.
- [39676] 13228. Cleiren, E.; Benichou, O.; Van Hul, E.; Gram, J.; Bollerslav, J.; Singer, F. R.; Beaverson, K.; Aledo, A.; Whyte, M. P.; Yoneyama, T.; deVernejou, M.-C.; Van Hul, W.: Albers-Schonberg disease (autosomal dominant osteopetrosis, type II) results from mutations in the CLCN7 chloride channel gene. Hum. Molec. Genet. 10: 2861–2867, 2001.
- [39677] 13229. Hedblom, E.; Kirkness, E. F.: A novel class of GABA-A receptor subunit in tissues of the reproductive system. J. Biol. Chem. 272: 15346–15350, 1997.
- [39678] 13230. Caloca, M. J.; Garcia-Bermejo, M. L.; Blumberg, P. M.; Lewin, N. E.; Kremmer, E.; Mischak, H.; Wang, S.; Nacro, K.; Bienfait, B.; Marquez, V. E.; Kazanietz, M. G.: Beta-2-chimaerin is a novel target for diacylglycerol: binding properties and changes in subcellular localization mediated by ligand binding to its C1 domain. Proc. Nat. Acad. Sci. 96: 11854–11859, 1999.

- [39679] 13231. Leung, T.; How, B.-E.; Manser, E.; Lim, L.: Cerebellar beta-2-chimaerin, a GTPase-activating protein for p21 Ras-related Rac is specifically expressed in granule cells and has a unique N-terminal SH2 domain J. Biol. Chem. 269: 12888-12892, 1994.
- [39680] 13232. Leung, T.; How, B.-E.; Manser, E.; Lim, L.: Germ cell beta-chimaerin, a new GTPase-activating protein for p21rac, is specifically expressed during the acrosomal assembly stage in rat testis. J. Biol. Chem. 268:3813-3816, 1993.
- [39681] 13233. Yuan, S.; Miller, D. W.; Barnett, G. H.; Hahn, J. F.; Williams, B. R.: Identification and characterization of human beta 2-chimaerin: association with malignant transformation in astrocytoma. Cancer Res. 55: 3456-3461, 1995.
- [39682] 13234. Chung, E.; Hanukoglu, A.; Rees, M.; Thompson, R.; Dillon, M.; Hanukoglu, I.; Bistrizter, T.; Kuhnle, U.; Seckl, J.; Gardiner, R. M.: Exclusion of the locus for autosomal recessive pseudohypoaldosteronism type 1 from the mineralocorticoid receptor gene region on human chromosome 4q by linkage analysis. J. Clin. Endocr. Metab. 80: 3341-3345, 1995.
- [39683] 13235. Nagase, T.; Ishikawa, K.; Suyama, M.; Kikuno, R.;

Hirosawa, M.;Miyajima, N.; Tanaka, A.; Kotani, H.; No-
mura, N.; Ohara, O.: Predictionof the coding sequences of
unidentified human genes. XIII. The complete sequences of
100 new cDNA clones from brain which code for large
proteinsin vitro. DNA Res. 6: 63–70, 1999.

[39684] 13236.Philibert, R. A.; Winfield, S. L.; Sandhu, H. K.; Mar-
tin, B. M.;Ginns, E. I.: The structure and expression of the
human neuroligin–3gene. Gene 246: 303–310, 2000.

[39685] 13237.Arico, M.; Imashuku, S.; Clementi, R.; Hibi, S.; Ter-
amura, T.;Danesino, C.; Haber, D. A.; Nichols, K. E.:
Hemophagocytic lymphohistiocytosis due to germline mu-
tations in SH2D1A, the X–linked lymphoproliferativedis-
ease gene. Blood 97: 1131–1133, 2001.

[39686] 13238.Arkwright, P. D.; Makin, G.; Will, A. M.; Ayres, M.;
Gokhale, D.A.; Fergusson, W. D.; Taylor, G. M.: X linked
lymphoproliferativedisease in a United Kingdom family.
Arch. Dis. Child. 79: 52–55, 1998.

[39687] 13239.Bar, R. S.; DeLor, C. J.; Clausen, K. P.; Hurtubise, P.;
Henle, W.; Hewetson, J. F.: Fatal infectious mononucleosis
in a family. NewEng. J. Med. 290: 363–367, 1974.

[39688] 13240.Benoit, L.; Wang, X.; Pabst, H. F.; Dutz, J.; Tan, R.:
Cuttingedge: defective NK cell activation in X–linked lym-
phoproliferativedisease. J. Immun. 165: 3549–3553,

2000.

- [39689] 13241.Brandau, O.; Schuster, V.; Weiss, M.; Hellebrand, H.; Fink, F.M.; Kreczy, A.; Friedrich, W.; Strahm, B.; Niemeyer, C.; Belohradsky,B. H.; Meindl, A.: Epstein–Barr virus–negative boys with non–Hodgkinlymphoma are mutated in the SH2D1A gene, as are patients with X–linkedlymphoproliferative disease (XLP). Hum. Molec. Genet. 8: 2407–2413,1999.
- [39690] 13242.Coffey, A. J.; Brooksbank, R. A.; Brandau, O.; Oohashi, T.; Howell,G. R.; Bye, J. M.; Cahn, A. P.; Durham, J.; Heath, P.; Wray, P.; Pavitt,R.; Wilkinson, J.; and 31 others: Host response to EBV infectionin X–linked lymphoproliferative disease results from mutations inan SH2–domain encoding gene. Nature Genet. 20: 129–135, 1998.
- [39691] 13243.Czar, M. J.; Kersh, E. N.; Mijares, L. A.; Lanier, G.; Lewis, J.;Yap, G.; Chen, A.; Sher, A.; Duckett, C. S.; Ahmed, R.; Schwartzberg,P. L.: Altered lymphocyte responses and cytokine production in micedeficient in the X–linked lymphoproliferative disease gene SH2D1A/DSHP/SAP. Proc.Nat. Acad. Sci. 98: 7449–7454, 2001.
- [39692] 13244.Dutz, J. P.; Benoit, L.; Wang, X.; Demetrick, D. J.; Junker, A.;de Sa, D.; Tan, R.: Lymphocytic vasculitis in X–

linked lymphoproliferative disease. *Blood* 97: 95–100, 2001.

- [39693] 13245. Grierson, H. L.; Skare, J.; Church, J.; Silberman, T.; Davis, J.R.; Kobrinsky, N.; McGregor, R.; Israels, S.; McCarty, J.; Andrews, L. G.; Blecha, T.; Erdman, S.; Obringer, A.; Scharnhorst, D.; Purtilo, D. T.: Evaluation of families wherein a single male manifests a phenotype of X-linked lymphoproliferative disease (XLP). *Am. J. Med. Genet.* 47:458–463, 1993.
- [39694] 13246. Hambleton, G.; Cottom, D. G.: Familial lymphoma. *Proc. Roy. Soc. Med.* 62: 1095 only, 1969.
- [39695] 13247. Hamilton, J. K.; Paquin, L. A.; Sullivan, J. L.; Maurer, H. S.; Cruzei, F. G.; Provisor, A. J.; Steuber, C. P.; Hawkins, E.; Yawn, D.; Cornet, J.; Clausen, K.; Finkelstein, G. Z.; Landing, B.; Grunnet, M.; Purtilo, D. T.: X-linked lymphoproliferative syndrome registry report. *J. Pediat.* 96: 669–673, 1980.
- [39696] 13248. Harris, A.; Docherty, Z.: X-linked lymphoproliferative disease: a karyotype analysis. *Cytogenet. Cell Genet.* 47: 92–94, 1988.
- [39697] 13249. Harris, A.; Lenoir, G. M.; Lankester, S. A.: X-linked lymphoproliferative disease: linkage studies using DNA probes. *Clin. Genet.* 33: 162–168, 1988.

- [39698] 13250.Hayoz, D.; Lenoir, G. M.; Nicole, A.; Pugin, P.; Regamey, C.:X-linked lymphoproliferative syndrome: identification of a large family in Switzerland. *Am. J. Med.* 84: 529–534, 1988.
- [39699] 13251.Klein, G.; Klein, E.: Sinking surveillance's flagship. *Nature* 395:441–445, 1998.
- [39700] 13252.Levine, P. H.; Kamaraju, L. S.; Connelly, R. R.; Bernard, C. W.; Dorfman, R. F.; Magrath, I.; Easton, J. M.: The American Burkitt's Lymphoma Registry: eight years' experience. *Cancer* 49: 1016–1022, 1982.
- [39701] 13253.Loeffel, S.; Chang, C.–H.; Heyn, R.; Harada, S.; Lipscomb, H.; Sinangil, F.; Volsky, D. J.; McClain, K.; Ochs, H.; Purtilo, D. T.: Necrotizing lymphoid vasculitis in X-linked lymphoproliferative syndrome. *Arch. Path. Lab. Med.* 109: 546–550, 1985.
- [39702] 13254.Lyon, M. F.; Loutit, J. F.: X-linked factor in acquired immunodeficiency syndrome?. (Letter) *Lancet* I: 768 only, 1983.
- [39703] 13255.Mulley, J. C.; Turner, A. M.; Gedeon, A. K.; Berdoukas, V. A.; Huang, T. H. M.; Ledbetter, D. H.; Grier-son, H.; Purtilo, D. T.:X-linked lymphoproliferative disease: prenatal detection of an unaffected histocompatible male. *Clin. Genet.* 42: 76–79, 1992.

- [39704] 13256.Eva, A.; Aaronson, S. A.: Isolation of a new human oncogene from a diffuse B-cell lymphoma. *Nature* 316: 273–275, 1985.
- [39705] 13257.Anderson, S. R.; Warburg, M.: Norrie's disease. *Arch. Ophthalmol.* 66:614–618, 1961.
- [39706] 13258.Bergen, A. A. B.; Wapenaar, M. C.; Schuurman, E. J. M.; Diergaarde, P. J.; Lerach, H.; Monaco, A. P.; Bakker, E.; Bleeker-Wagemakers, E. M.; van Ommen, G. J. B.: Detection of a new submicroscopic Norrie disease deletion interval with a novel DNA probe isolated by differential Alu PCR fingerprint cloning. *Cytogenet. Cell Genet.* 62: 231–235, 1993.
- [39707] 13259.Berger, W.; Meindl, A.; van de Pol, T. J. R.; Cremers, F. P. M.; Ropers, H. H.; Doerner, C.; Monaco, A.; Bergen, A. A. B.; Lebo, R.; Warburg, M.; Zergollern, L.; Lorenz, B.; Gal, A.; Bleeker-Wagemakers, E. M.; Meitinger, T.: Isolation of a candidate gene for Norrie disease by positional cloning. *Nature Genet.* 1: 199–203, 1992.
- [39708] 13260.Berger, W.; van de Pol, D.; Bachner, D.; Oerlemans, F.; Winkens, H.; Hameister, H.; Wieringa, B.; Hendriks, W.; Ropers, H. H.: An animal model for Norrie disease (ND): gene targeting of the mouse ND gene. *Hum. Molec. Genet.* 5: 51–59, 1996.

- [39709] 13261.Eva, A.; Vecchio, G.; Rao, C. D.; Tronick, S. R.; Aaronson, S.A.: The predicted DBL oncogene product defines a distinct class of transforming proteins. *Proc. Nat. Acad. Sci.* 85: 2061–2065, 1988.
- [39710] 13262.Galland, F.; Stefanova, M.; Lafage, M.; Birnbaum, D.: Localization of the 5-prime end of the MCF2 oncogene to human chromosome 15q15–q23. *Cytogenet. Cell Genet.* 60: 114–116, 1992.
- [39711] 13263.Grant, S. G.; Mattei, M.–G.; Galland, F.; Stephenson, D. A.; Keitz, B. T.; Birnbaum, D.; Chapman, V. M.: Localization of the mouse Mcf–2(Dbl) protooncogene within a conserved linkage group on the mouse X chromosome. *Cytogenet. Cell Genet.* 54: 175–181, 1990.
- [39712] 13264.Nguyen, C.; Pontarotti, P.; Birnbaum, D.; Chimini, G.; Rey, J.A.; Mattei, J.–F.; Jordan, B. R.: Large scale physical mapping in the q27 region of the human X chromosome: the coagulation factor IX gene and the mcf.2 transforming sequence are separated by at most 270 kilobase pairs and are surrounded by several 'HTF islands.' *EMBO J.* 6: 3285–3289, 1987.
- [39713] 13265.Nguyen, C.; Poustka, A.–M.; Djabali, M.; Roux, D.; Mattei, J.–F.; Lehrach, H.; Jordan, B. R.: Large-scale mapping and chromosome jumping in the q27 region of the

human X chromosome. *Genomics* 5: 298–303, 1989.

[39714] 13266. Noguchi, T.; Mattei, M.-G.; Oberle, I.; Planche, J.; Imbert, J.; Pelassy, C.; Birg, F.; Birnbaum, D.: Localization of the mcf.2 transforming sequence to the X chromosome. *EMBO J.* 6: 1301–1307, 1987.

[39715] 13267. Ron, D.; Tronick, S. R.; Aaronson, S. A.; Eva, A.: Molecular cloning and characterization of the human DBL proto-oncogene: evidence that its overexpression is sufficient to transform NIH/3T3 cells. *EMBO J.* 7: 2465–2473, 1988.

[39716] 13268. Srivastava, S. K.; Wheelock, R. H. P.; Aaronson, S. A.; Eva, A.: Identification of the protein encoded by the human diffuse B-cell lymphoma (dbl) oncogene. *Proc. Nat. Acad. Sci.* 83: 8868–8872, 1986.

[39717] 13269. Tronick, S. R.; McBride, O. W.; Popescu, N. C.; Eva, A.: Chromosomal localization of DBL oncogene sequences. *Genomics* 5: 546–553, 1989.

[39718] 13270. Giovane, A.; Sobieszczuk, P.; Mignon, C.; Mattei, M.-G.; Wasylyk, B.: Locations of the ets subfamily members net, elk1, and sap1 (ELK3, ELK1, and ELK4) on three homologous regions of the mouse and human genomes. *Genomics* 29: 769–772, 1995.

[39719] 13271. Janz, M.; Lehmann, U.; Olde Weghuis, D.; de Leeuw,

B.; Geurts vanKessel, A.; Gilgenkrantz, S.; Hipskind, R. A.; Nordheim, A.: Refined mapping of the human Ets-related gene Elk-1 to Xp11.2-p11.4, distal to the OATL1 region. Hum. Genet. 94: 442-444, 1994.

[39720] 13272. Rao, V. N.; Huebner, K.; Isobe, M.; ar-Rushdi, A.; Croce, C. M.; Reddy, E. S. P.: Elk, tissue-specific ets-related genes on chromosomes X and 14 near translocation breakpoints. Science 244: 66-70, 1989.

[39721] 13273. Tamai, Y.; Taketo, M.; Nozaki, M.; Seldin, M. F.: Mouse Elk oncogene maps to chromosome X and a novel Elk oncogene (Elk3) maps to chromosome 10. Genomics 26: 414-416, 1995.

[39722] 13274. Yamauchi, T.; Toko, M.; Suga, M.; Hatakeyama, T.; Isobe, M.: Structural organization of the human Elk1 gene and its processed pseudogene Elk2. DNA Res. 6: 21-27, 1999.

[39723] 13275. Bermingham, J. R., Jr.; Scherer, S. S.; O'Connell, S.; Arroyo, E.; Kalla, K. A.; Powell, F. L.; Rosenfeld, M. G.: Tst-1/Oct-6/SCI Pre-regulates a unique step in peripheral myelination and is required for normal respiration. Genes Dev. 10: 1751-1762, 1996.

[39724] 13276. Jaegle, M.; Mandemakers, W.; Broos, L.; Zwart, R.; Karis, A.; Visser, P.; Grosveld, F.; Meijer, D.: The POU factor

Oct-6 and Schwann cell differentiation. Science 273: 507-510, 1996.

[39725] 13277. Sumiyama, K.; Washio-Watanabe, K.; Ono, T.; Yoshida, M. C.; Hayakawa, T.; Ueda, S.: Human class III POU genes, POU3F1 and POU3F3, map to chromosomes 1p34.1 and 3p14.1. Mammalian Genome 9: 180-181, 1998.

[39726] 13278. Costache, M.; Apoil, P.-A.; Cailleau, A.; Elmgren, A.; Larson, G.; Henry, S.; Blancher, A.; Iordachescu, D.; Oriol, R.; Mollicone, R.: Evolution of fucosyltransferase genes in vertebrates. J. Biol. Chem. 272: 29721-29728, 1997.

[39727] 13279. Yanagidani, S.; Uozumi, N.; Ihara, Y.; Miyoshi, E.; Yamaguchi, N.; Taniguchi, N.: Purification and cDNA cloning of GDP-L-Fuc:N-acetyl-beta-D-glucosaminide:alpha-1-6 fucosyltransferase (alpha-1-6 FucT) from human gastric cancer MKN45 cells. J. Biochem. 121: 626-632, 1997.

[39728] 13280. Bekri, S.; Adelaide, J.; Merscher, S.; Grosgeorge, J.; Caroli-Bosc, F.; Perucca-Lostanlen, D.; Kelley, P. M.; Pebusque, M.-J.; Theillet, C.; Birnbaum, D.; Gaudray, P.: Detailed map of a region commonly amplified at 11q13-q14

in human breast carcinoma. *Cytogenet. CellGenet.* 79: 125–131, 1997.

[39729] 13281. Goodwin, R. G.; Alderson, M. R.; Smith, C. A.; Armitage, R. J.; VandenBos, T.; Jerzy, R.; Tough, T. W.; Schoenborn, M. A.; Davis-Smith, T.; Hennen, K.; Falk, B.; Cosman, D.; Baker, E.; Sutherland, G. R.; Grabstein, K. H.; Farrah, T.; Giri, J. G.; Beckmann, M. P.: Molecular and biological characterization of a ligand for CD27 defines a new family of cytokines with homology to tumor necrosis factor. *Cell* 73:447–456, 1993.

[39730] 13282. Wright, R. M.; Vaitaitis, G. M.; Wilson, C. M.; Repine, T. B.; Terada, L. S.; Repine, J. E.: cDNA cloning, characterization, and tissue-specific expression of human xanthine dehydrogenase/xanthine oxidase. *Proc. Nat. Acad. Sci.* 90: 10690–10694, 1993.

[39731] 13283. Kawabata, M.; Saeki, K.: Multiple alternative transcripts of the human homologue of the mouse TRAD/R51H3/RAD51D gene, a member of the Rec A/RAD51 gene family. *Biochem. Biophys. Res. Commun.* 257: 156–162, 1999.

[39732] 13284. Pittman, D. L.; Weinberg, L. R.; Schimenti, J. C.: Identification, characterization, and genetic mapping of Rad51d, a new mouse and human RAD51/RecA-related

gene. *Genomics* 49: 103–111, 1998.

- [39733] 13285. Bell, B.; Scheer, E.; Tora, L.: Identification of hTAFII80–delta links apoptotic signaling pathways to transcription factor TFIID function. *Molec. Cell* 8: 591–600, 2001.
- [39734] 13286. Hisatake, K.; Ohta, T.; Takada, R.; Guermah, M.; Horikoshi, M.; Nakatani, Y.; Roeder, R. G.: Evolutionary conservation of human TATA-binding–polypeptide-associated factors TAFII31 and TAFII80 and interactions of TAFII80 with other TAFs and with general transcription factors. *Proc. Nat. Acad. Sci.* 92: 8195–8199, 1995.
- [39735] 13287. Weinzierl, R. O. J.; Ruppert, S.; Dynlacht, B. D.; Tanese, N.; Tjian, R.: Cloning and expression of *Drosophila* TAFII60 and human TAFII70 reveal conserved interactions with other subunits of TFIID. *EMBO J.* 12: 5303–5309, 1993.
- [39736] 13288. Tsai, K. J.; Chen, S. K.; Ma, Y. L.; Hsu, W. L.; Lee, E. H. Y.: sgk, a primary glucocorticoid-induced gene, facilitates memory consolidation of spatial learning in rats. *Proc. Nat. Acad. Sci.* 99: 3990–3995, 2002.
- [39737] 13289. Waldegger, S.; Barth, P.; Raber, G.; Lang, F.: Cloning and characterization of a putative human serine/

threonine protein kinase transcriptionally modified during anisotonic and isotonic alterations of cell volume.

Proc.Nat. Acad. Sci. 94: 4440–4445, 1997.

- [39738] 13290. Mackay, D. J. G.; Coupe, A.-M.; Shield, J. P. H.; Storr, J. N.P.; Temple, I. K.; Robinson, D. O.: Relaxation of imprinted expression of ZAC and HYMAI in a patient with transient neonatal diabetes mellitus. Hum.Genet. 110: 139–144, 2002.
- [39739] 13291. Spengler, D.; Villalba, M.; Hoffmann, A.; Pantaloni, C.; Houssami, S.; Bockaert, J.; Journot, L.: Regulation of apoptosis and cell cycle arrest by Zac1, a novel zinc finger protein expressed in the pituitary gland and the brain. EMBO J. 16: 2814–2825, 1997.
- [39740] 13292. Temple, I. K.; James, R. S.; Crolla, J. A.; Sitch, F. L.; Jacobs, P. A.; Howell, W. M.; Betts, P.; Baum, J. D.; Shield, J. P. H.: An imprinted gene(s) for diabetes? (Letter) Nature Genet. 9: 110–112, 1995.
- [39741] 13293. Varrault, A.; Ciani, E.; Apiou, F.; Bilanges, B.; Hoffmann, A.; Pantaloni, C.; Bockaert, J.; Spengler, D.; Journot, L.: hZAC encodes a zinc finger protein with antiproliferative properties and maps to a chromosomal region frequently lost in cancer. Proc. Nat. Acad. Sci. 95: 8835–8840, 1998.

- [39742] 13294.Jiang, Y.-W.; Veschambre, P.; Erdjument-Bromage, H.; Tempst, P.;Conaway, J. W.; Conaway, R. C.; Kornberg, R. D.: Mammalian mediatorof transcriptional regulation and its possible role as an end-pointof signal transduction pathways. *Proc. Nat. Acad. Sci.* 95: 8538–8543,1998.
- [39743] 13295.Adams, M. D.; Soares, M. B.; Kerlavage, A. R.; Fields, C.; Venter,J. C.: Rapid cDNA sequencing (expressed sequence tags) from a directionallycloned human infant brain cDNA library. *Nature Genet.* 4: 373–380,1993.
- [39744] 13296.Dai, K.-S.; Liew, C.-C.: A novel human striated muscle RING zincfinger protein, SMRZ, interacts with SMT3b via its RING domain. *J.Biol. Chem.* 276: 23992–23999, 2001.
- [39745] 13297.Abdollahi, A.; Godwin, A. K.; Miller, P. D.; Getts, L. A.; Schultz,D. C.; Taguchi, T.; Testa, J. R.; Hamilton, T. C.: Identificationof a gene containing zinc–finger motifs based on lost expression inmalignantly transformed rat ovarian surface epithelial cells. *CancerRes.* 57: 2029–2034, 1997.
- [39746] 13298.Abdollahi, A.; Roberts, D.; Godwin, A. K.; Schultz, D. C.; Sonoda,G.; Testa, J. R.; Hamilton, T. C.: Identifica–tion of a zinc–fingergene at 6q25: a chromosomal region implicated in development of manysolid tumors. *Onco–*

gene 14: 1973–1979, 1997.

- [39747] 13299.Kamiya, M.; Judson, H.; Okazaki, Y.; Kusakabe, M.; Muramatsu, M.;Takada, S.; Takagi, N.; Arima, T.; Wake, N.; Kamimura, K.; Satomura,K.; Hermann, R.; Bonthron, D. T.; Hayashizaki, Y.: The cell cyclecontrol gene ZAC/PLAGL1 is imprinted: a strong candidate gene fortransient neonatal diabetes. Hum. Molec. Genet. 9: 453–460, 2000.
- [39748] 13300.Ide, H.; Katoh, M.; Sasaki, H.; Yoshida, T.; Aoki, K.; Nawa, Y.;Osada, Y.; Sugimura, T.; Terada, M.: Cloning of human bone morphogeneticprotein type 1B receptor (BMPR–1B) and its expression in prostatecancer in comparison with other BMPRs. Oncogene 14: 1377–1382, 1997.
- [39749] 13301.ten Dijke, P.; Yamashita, H.; Ichijo, H.; Franzen, P.; Laiho, M.;Miyazono, K.; Heldin, C.–H.: Characterization of type I receptorsfor transforming growth factor–beta and activin. Science 264: 101–104,1994.
- [39750] 13302.Blixt, A.; Mahlapuu, M.; Bjursell, C.; Darnfors, C.; Johannesson,T.; Enerback, S.; Carlsson, P.: The two–exon gene of the human forkheadtranscription factor FREAC–2 (FKHL6) is located at 6p25.3. Genomics 53:387–390, 1998.
- [39751] 13303.Best, J. L.; Presky, D. H.; Swerlick, R. A.; Burns, D.

K.; Chu,W.: Cloning of a full-length cDNA sequence encoding a cdc2-related protein kinase from human endothelial cells. *Biochem. Biophys. Res. Commun.* 208: 562–568, 1995.

[39752] 13304. Garriga, J.; Mayol, X.; Grana, X.: The CDC2-related kinase PITALRE is the catalytic subunit of active multimeric protein complexes. *Biochem. J.* 319: 293–298, 1996.

[39753] 13305. Grana, X.; De Luca, A.; Sang, N.; Fu, Y.; Claudio, P. P.; Rosenblatt, J.; Morgan, D. O.; Giordano, A.: PITALRE, a nuclear CDC2-related protein kinase that phosphorylates the retinoblastoma protein in vitro. *Proc. Nat. Acad. Sci.* 91: 3834–3838, 1994.

[39754] 13306. Liu, H.; Rice, A. P.: Genomic organization and characterization of promoter function of the human CDK9 gene. *Gene* 252: 51–59, 2000.

[39755] 13307. Triepels, R.; van den Heuvel, L.; Loeffen, J.; Smeets, R.; Trijbels, F.; Smeitink, J.: The nuclear-encoded human NADH:ubiquinone oxidoreductase NDUF8 subunit: cDNA cloning, chromosomal localization, tissue distribution, and mutation detection in complex-I-deficient patients. *Hum. Genet.* 103:557–563, 1998.

[39756] 13308. Bai, C.; Connolly, B.; Metzker, M. L.; Hilliard, C. A.; Liu, X.; Sandig, V.; Soderman, A.; Galloway, S. M.; Liu, Q.;

Austin, C. P.; Caskey, C. T.: Overexpression of M68/DCR3 in human gastrointestinal tract tumors independent of gene amplification and its location in a four-gene cluster. *Proc. Nat. Acad. Sci.* 97: 1230–1235, 2000.

[39757] 13309. Pitti, R. M.; Marsters, S. A.; Lawrence, D. A.; Roy, M.; Kischkel, F. C.; Dowd, P.; Huang, A.; Donahue, C. J.; Sherwood, S. W.; Baldwin, D. T.; Godowski, P. J.; Wood, W. I.; Gurney, A. L.; Hillan, K. J.; Cohen, R. L.; Goddard, A. D.; Botstein, D.; Ashkenazi, A.: Genomic amplification of a decoy receptor for Fas ligand in lung and colon cancer. *Nature* 396: 699–703, 1998.

[39758] 13310. Ringstad, N.; Nemoto, Y.; De Camilli, P.: The SH3p4/Sh3p8/SH3p13 protein family: binding partners for synaptojanin and dynamin via a Grb2-like Src homology 3 domain. *Proc. Nat. Acad. Sci.* 94: 8569–8574, 1997.

[39759] 13311. Sittler, A.; Walter, S.; Wedemeyer, N.; Hasenbank, R.; Scherzinger, E.; Eickhoff, H.; Bates, G. P.; Lehrach, H.; Wanker, E. E.: SH3GL3 associates with the huntington exon 1 protein and promotes the formation of polyglutamine-containing protein aggregates. *Molec. Cell* 2: 427–436, 1998.

[39760] 13312. Sparks, A. B.; Hoffman, N. G.; McConnell, S. J.; Fowlkes, D. M.; Kay, B. K.: Cloning of ligand targets: systematic isolation of SH3 domain-containing proteins. *Nature*

ture Biotech. 14: 741–744, 1996.

[39761] 13313.Chinnaiyan, A. M.; O'Rourke, K.; Yu, G.–L.; Lyons, R. H.; Garg,M.; Duan, D. R.; Xing, L.; Gentz, R.; Ni, J.; Dixit, V. M.: Signaltransduction by DR3, a death domain–containing receptor related toTNFR–1 and CD95. Science 274: 990–992, 1996.

[39762] 13314.Fracchiolla, N. S.; Colombo, G.; Finelli, P.; Maiolo, A. T.; Neri,A.: EHT, a new member of the MTG8/ETO gene family, maps on 20q11region and is deleted in acute myeloid leukemias. (Letter) Blood 92:3481–3484, 1998.

[39763] 13315.Kitabayashi, I.; Ida, K.; Morohoshi, F.; Yokoyama, A.; Mitsuhashi,N.; Shimizu, K.; Nomura, N.; Hayashi, Y.; Ohki, M.: The AML1–MTG8leukemic fusion protein forms a complex with a novel member of theMTG8(ETO/CDR) family, MTGR1. Molec. Cell. Biol. 18: 846–858, 1998.

[39764] 13316.Nelson, H.; Mandiyan, S.; Noumi, T.; Moriyama, Y.; Miedel, M. C.;Nelson, N.: Molecular cloning of cDNA encoding the C subunit of H(+)-ATPasefrom bovine chromaffin granules. J. Biol. Chem. 265: 20390–20393,1990.

[39765] 13317.van Hille, B.; Vanek, M.; Richener, H.; Green, J. R.; Bilbe, G.: Cloning and tissue distribution of subunits C, D, and E of the humanvacuolar H(+)-ATPase. Biochem. Biophys. Res. Commun. 197: 15–21,1993.

- [39766] 13318. Fujiwara, T.; Suzuki, M.; Tanigami, A.; Ikenoue, T.; Omata, M.; Chiba, T.; Tanaka, K.: The BTRC gene, encoding a human F-box/WD40-repeat protein, maps to chromosome 10q24-q25. *Genomics* 58: 104-105, 1999.
- [39767] 13319. Margottin, F.; Bour, S. P.; Durand, H.; Selig, L.; Benichou, S.; Richard, V.; Thomas, D.; Strebel, K.; Benarous, R.: A novel human WD protein, h-beta TrCp, that interacts with HIV-1 Vpu connects CD4 to the ER degradation pathway through an F-box motif. *Molec. Cell* 1:565-574, 1998.

- [39768] 13320.Fakruddin, J. M.; Chaganti, R. S. K.; Murty, V. V. V. S.: Lackof BCL10 mutations in germ cell tumors and B cell lymphomas. *Cell* 97:683–688, 1999.
- [39769] 13321.Isaacson, P. G.; Spencer, J.: The biology of low grade MALT lymphoma. *J.Clin. Path.* 48: 395–397, 1995.
- [39770] 13322.Ruland, J.; Duncan, G. S.; Elia, A.; del Barco Bar- rantes, I.; Nguyen,L.; Plyte, S.; Millar, D. G.; Bouchard, D.; Wakeham, A.; Ohashi, P.S.; Mak, T. W.: Bcl10 is a positive regulator of antigen receptor–inducedactivation of NF– kappa–B and neural tube closure. *Cell* 104: 33–42,2001.
- [39771] 13323.Thome, M.; Martinon, F.; Hofmann, K.; Rubio, V.; Steiner, V.; Schneider,P.; Mattmann, C.; Tschopp, J.: Equine herpesvirus–2 E10 gene product,but not its cellular homologue, activates NF–kappa–B transcriptionfactor and c–Jun N–terminal kinase. *J. Biol. Chem.* 274: 9962–9968,1999.
- [39772] 13324.Willis, T. G.; Jadayel, D. M.; Du, M.–Q.; Peng, H.; Perry, A. R.;Abdul–Rauf, M.; Price, H.; Karran, L.; Majeko– dunmi, O.; Wlodarska,I.; Pan, L.; Crook, T.; Hamoudi, R.; Isaacson, P. G.; Dyer, M. J.S.: Bcl10 is involved in t(1;14)(p22;q32) of MALT B cell lymphomaand mutated in multiple tumor types *Cell* 96: 35–45, 1999.
- [39773] 13325.Wotherspoon, A. C.; Pan, L. X.; Diss, T. C.; Isaac–

son, P. G.:Cytogenetic study of B-cell lymphoma of mucosa-associated lymphoid tissue. *Cancer Genet. Cytogenet.* 58: 35–38, 1992.

- [39774] 13326. Anderson, P.; Nagler-Anderson, C.; O'Brien, C.; Levine, H.; Watkins, S.; Slayter, H. S.; Blue, M. L.; Schlossman, S. F.: A monoclonal antibody reactive with a 15-kDa cytoplasmic granule-associated protein defines a subpopulation of CD8⁺ T lymphocytes. *J. Immun.* 144: 574–582, 1990.
- [39775] 13327. Forch, P.; Puig, O.; Kedersha, N.; Martinez, C.; Granneman, S.; Seraphin, B.; Anderson, P.; Valcarcel, J.: The apoptosis-promoting factor TIA-1 is a regulator of alternative pre-mRNA splicing. *Molec. Cell* 6: 1089–1098, 2000.
- [39776] 13328. Kawakami, A.; Tian, Q.; Streuli, M.; Poe, M.; Edelhoff, S.; Disteche, C. M.; Anderson, P.: Intron-exon organization and chromosomal localization of the human TIA-1 gene. *J. Immun.* 152: 4937–4945, 1994.
- [39777] 13329. Tian, Q.; Streuli, M.; Saito, H.; Schlossman, S. F.; Anderson, P.: A polyadenylate binding protein localized to the granules of cytolytic lymphocytes induces DNA fragmentation in target cells. *Cell* 67:629–639, 1991.
- [39778] 13330. Zheng, B.; Albrecht, U.; Kaasik, K.; Sage, M.; Lu, W.;

Vaishnav,S.; Li, Q.; Sun, Z. S.; Eichele, G.; Bradley, A.; Lee, C. C.: Nonredundant roles of the mPer1 and mPer2 genes in the mammalian circadian clock. *Cell* 105:683–694, 2001.

[39779] 13331.Cheng, J.–F.; Boyartchuk, V.; Zhu, Y.: Isolation and mapping of human chromosome 21 cDNA: progress in constructing a chromosome 21 expression map. *Genomics* 23: 75–84, 1994.

[39780] 13332.Imamura, Y.; Kubota, R.; Wang, Y.; Asakawa, S.; Kudoh, J.; Mashima,Y.; Oguchi, Y.; Shimizu, N.: Human retina–specific amine oxidase(RAO): cDNA cloning, tissue expression, and chromosomal mapping. *Genomics* 40:277–283, 1997.

[39781] 13333.Imamura, Y.; Noda, S.; Mashima, Y.; Kudoh, J.; Oguchi, Y.; Shimizu,N.: Human retina–specific amine oxidase: genomic structure of the gene (AOC2), alternatively spliced variant, and mRNA expression in retina. *Genomics* 51: 293–298, 1998.

[39782] 13334.Sirotkin, H.; O'Donnell, H.; DasGupta, R.; Halford, S.; St. Jore,B.; Puech, A.; Parimoo, S.; Morrow, B.; Skoultschi, A.; Weissman, S.M.; Scambler, P.; Kucherlapati, R.: Identification of a new human catenin gene family member (ARVCF) from the region deleted in velo–car–

dio-facialsyndrome. Genomics 41: 75–83, 1997.

[39783] 13335.Andrews, N. C.: Mining copper transport genes. Proc. Nat. Acad.Sci. 98: 6543–6545, 2001.

[39784] 13336.Boulton, J.; Strickson, A. J.; Jabs, E. W.; Cheng, J.-F.; Fidler,C.; Wainscoat, J. S.: Physical mapping of the human ATX1 homologue(HAH1) to the critical region of the 5q- syndrome within 5q32, and immediately adjacent to the SPARC gene. Hum. Genet. 106: 127–129,2000.

[39785] 13337.Hamza, I.; Faisst, A.; Prohaska, J.; Chen, J.; Gruss, P.; Gitlin,J. D.: The metallochaperone Atox1 plays a critical role in perinatalcopper homeostasis. Proc. Nat. Acad. Sci. 98: 6848–6852, 2001.

[39786] 13338.Klomp, L. W. J.; Lin, S.-J.; Yuan, D. S.; Klausner, R. D.; Culotta,V. C.; Gitlin, J. D.: Identification and functional expression ofHAH1, a novel human gene involved in copper homeostasis. J. Biol.Chem. 272: 9221–9226, 1997.

[39787] 13339.Castro, A. M.; Snyder, L. M.: G6PD San Jose: a new variant characterized by NADPH inhibition studies. Humangenetik 21: 361–363, 1974.

[39788] 13340.Cayanis, E.; Gomperts, E. D.; Balinsky, D.; Disler, P.; Myers,A.: G6PD Hillbrow: a new variant of glucose-6-phosphate dehydrogenase associated with drug-induced haemolytic anaemia. Brit. J. Haemat. 30:343–350,

1975.

- [39789] 13341. Cayanis, E.; Lane, A. B.; Jenkins, T.; Nurse, G. T.; Balinsky, D.: Glucose-6-phosphate dehydrogenase Porbandar: a new slow variant with slightly reduced activity in a South African family of Indian descent. *Biochem. Genet.* 15: 765-773, 1977.
- [39790] 13342. Cederbaum, A. I.; Beutler, E.: Nonspherocytic hemolytic anemia due to G6PD Grand Prairie. *I.R.C.S.* 3: 579, 1975.
- [39791] 13343. Chan, T. K.; Todd, D.: Characteristics and distribution of glucose-6-phosphate dehydrogenase-deficient variants in South China. *Am. J. Hum. Genet.* 24: 475-484, 1972.
- [39792] 13344. Chan, T. K.; Todd, D.; Lai, M. C. S.: Glucose 6-phosphate dehydrogenase: identity of erythrocyte and leukocyte enzyme with report of a new variant in Chinese. *Biochem. Genet.* 6: 119-124, 1972.
- [39793] 13345. Chang, J.-G.; Chiou, S.-S.; Perng, L.-I.; Chen, T.-C.; Liu, T.-C.; Lee, L.-S.; Chen, P.-H.; Tang, T. K.: Molecular characterization of glucose-6-phosphate dehydrogenase (G6PD) deficiency by natural and amplification created restriction sites: five mutations account for most G6PD deficiency cases in Taiwan. *Blood* 80: 1079-1082, 1992.

- [39794] 13346.Chen, E. Y.; Cheng, A.; Lee, A.; Kuang, W.-J.; Hillier, L.; Green,P.; Schlessinger, D.; Ciccodicola, A.; D'Urso, M.: Sequence of humanglucose-6-phosphate dehydrogenase cloned in plasmids and a yeast artificialchromosome. *Genomics* 10: 792-800, 1991.
- [39795] 13347.Chen, H.-L.; Huang, M.-J.; Huang, C.-S.; Tang, T. K.: G6PD NanKang(517 T-to-C; 173 phe-to-leu): a new Chinese G6PD variant associatedwith neonatal jaundice. *Hum. Hered.* 46: 201-204, 1996.
- [39796] 13348.Chernyak, N. B.; Batischev, A. I.; Lanzina, N. V.; Tokarev, Y.N.; Alexeyev, G. A.: Electrophoretic and kinetic properties of glucose-6-phosphatedehydrogenase from erythrocytes or patients with hemolytic anemia,related to deficiency of the enzyme activity. *Vopr. Med. Khim.* 23:166-171, 1977.
- [39797] 13349.Childs, B.; Zinkham, W.; Browne, E. A.; Kimbro, E. L.; Torbert,J. V.: A genetic study of a defect in glutathione metabolism of theerythrocytes. *Bull. Johns Hopkins Hosp.* 102: 21-37, 1958.
- [39798] 13350.Chiu, D. T. Y.; Zuo, L.; Chao, L.; Chen, E.; Louie, E.; Lubin,B.; Liu, T. Z.; Du, C. S.: Molecular characterization of glucose-6-phosphatedehydrogenase (G6PD) deficiency in patients of Chinese descent andidentification of new base

substitutions in the human G6PD gene. Blood
81:2150–2154, 1993.

[39799] 13351.Chockkalingam, K.; Board, P. G.: Further evidence
for heterogeneity of glucose–6–phosphate dehydrogenase
deficiency in Papua New Guinea. Hum.Genet. 56:
209–212, 1980.

[39800] 13352.Chockkalingam, K.; Board, P. G.; Breguet, G.: Glu–
cose–6–phosphatedehydrogenase variants of Bali Island
(Indonesia). Hum. Genet. 60:60–62, 1982.

[39801] 13353.Chockkalingam, K.; Board, P. G.; Nurse, G. T.: Glu–
cose–6–phosphatedehydrogenase deficiency in Papua New
Guinea: the description of 13 new variants. Hum. Genet.
60: 189–192, 1982.

[39802] 13354.Chuanshu, D.; Yankang, X.; Lin, W. Q. R.; Xiaoyun,
H.: Studies on erythrocyte glucose–6–phosphate dehydro–
genase variants in Chinese.I. Gd(–) Lizu–Baisha. Acta
Acad. Med. Zhong Shan 2: 649–658, 1981.

[39803] 13355.Cocco, P.; Todde, P.; Fornera, S.; Manca, M. B.;
Manca, P. Sias, A. R.: Mortality in a cohort of men express–
ing the glucose–6–phosphatedehydrogenase deficiency.
Blood 91: 706–709, 1998.

[39804] 13356.Cohn, J.; Carter, N.; Warburg, M.: Glucose–
6–phosphate dehydrogenase deficiency in a native Danish

family: a new variant. Scand. J. Haemat. 23:403–406, 1979.

[39805] 13357.Colonna–Romano, S.; Iolascon, A.; Lippo, S.; Pinto, L.; Cutillo,S.; Battistuzzi, G.: Genetic heterogeneity at the glucose–6–phosphatedehydrogenase locus in southern Italy: a study on the population of Naples. Hum. Genet. 69: 228–232, 1985.

[39806] 13358.Cooper, D. W.; Johnston, P. G.; Murtagh, C. E.; Sharman, G. B.;Vandeberg, J. L.; Poole, W. E.: Sex–linked isozymes and sex–chromosomeevolution and inactivation in kangaroos.In: Markeit, C. L.: Isozymes.Developmental Biology. New York: Academic Press (pub.) III: 1975.Pp. 559–573.

[39807] 13359.Cooper, M. R.; Dechatelet, L. R.; McCall, C. E.; Lavia, M. F.;Spurr, C. L.; Baehner, R. L.: Complete defi–ciency of leukocyte glucose–6–phosphatedehydrogenase with defective bactericidal activity. J. Clin. Invest. 51:769–778, 1972.

[39808] 13360.Corash, L.; Spielberg, S.; Bartsocas, C.; Boxer, L.; Steinherz,R.; Sheetz, M.; Egan, M.; Schlessleman, J.; Schul–man, J. D.: Reducedchronic hemolysis during high–dose vitamin E administration in Mediterranean–typeglu–cose–6–phosphate dehydrogenase deficiency. New Eng. J.

Med. 303:416–420, 1980.

- [39809] 13361. Corcoran, C. M.; Calabro, V.; Tamagnini, G.; Town, M.; Haidar, B.; Vulliamy, T. J.; Mason, P. J.; Luzzatto, L.: Molecular heterogeneity underlying the G6PD Mediterranean phenotype. *Hum. Genet.* 88: 688–690, 1992.
- [39810] 13362. Costa, E.; Cabeda, J. M.; Vieira, E.; Pinto, R.; Pereira, S. A.; Ferraz, L.; Santos, R.; Barbot, J.: Glucose-6-phosphate dehydrogenase Aveiro: a de novo mutation associated with chronic nonspherocytic hemolytic anemia. *Blood* 95: 1499–1501, 2000.
- [39811] 13363. Crookston, J. H.; Yoshida, A.; Lin, M.; Booser, D. J.: G6PD Toronto. *Biochem. Genet.* 8: 259–265, 1973.
- [39812] 13364. Csepregy, M.; Hall, M. K.; Berkow, R. L.; Jackson, S.; Prchal, J. T.: Characterization of a new G6PD variant: G6PD Titusville. *Am. J. Med. Sci.* 297: 114–117, 1989.
- [39813] 13365. Csepregy, M.; Yeilding, A.; Lilly, M.; Hall, K.; Scott, C. W.; Prchal, J. T.: Characterization of a new glucose-6-phosphate dehydrogenase variant: G6PD Central City. *Am. J. Hemat.* 28: 61–62, 1988.
- [39814] 13366. D'Urso, M.; Luzzatto, L.; Perroni, L.; Ciccodicola, A.; Gentile, G.; Peluso, I.; Persico, M. G.; Pizzella, T.; Toniolo, D.; Vulliamy, T. J.: An extensive search for RFLP in the human glucose-6-phosphate dehydrogenase locus has re-

vealed a silent mutation in the coding sequence. *Am.J. Hum. Genet.* 42: 735–741, 1988.

- [39815] 13367. Basse, F.; Stout, J. G.; Sims, P. J.; Wiedmer, T.: Isolation of an erythrocyte membrane protein that mediates Ca^{2+} -dependent transbilayer movement of phospholipid. *J. Biol. Chem.* 271: 17205–17210, 1996.
- [39816] 13368. Garrett, R. M.; Johnson, J. L.; Graf, T. N.; Feigenbaum, A.; Rajagopalan, K. V.: Human sulfite oxidase R160Q: identification of the mutation in a sulfite oxidase-deficient patient and expression and characterization of the mutant enzyme. *Proc. Nat. Acad. Sci.* 95: 6394–6398, 1998.
- [39817] 13369. Kisker, C.; Schindelin, H.; Pacheco, A.; Wehbi, W. A.; Garrett, R. M.; Rajagopalan, K. V.; Enemark, J. H.; Rees, D. C.: Molecular basis of sulfite oxidase deficiency from the structure of sulfite oxidase. *Cell* 91: 973–983, 1997.
- [39818] 13370. Corral, J.; Forster, A.; Thompson, S.; Lampert, F.; Kaneko, Y.; Slater, R.; Kroes, W. G.; van der Schoot, C. E.; Ludwig, W.-D.; Karpas, A.; Pocock, C.; Cotter, F.; Rabbitts, T. H.: Acute leukemias of different lineages have similar MLL gene fusions encoding related chimeric proteins resulting from chromosomal translocation. *Proc. Nat. Acad. Sci.* 90: 8538–8542, 1993.

- [39819] 13371. Parry, P.; Wei, Y.; Evans, G.: Cloning and characterization of the t(X;11) breakpoint from a leukemic cell line identify a new member of the forkhead gene family. *Genes Chromosomes Cancer* 11: 79–84, 1994.
- [39820] 13372. Peters, U.; Haberhausen, G.; Kostrzewa, M.; Nolte, D.; Muller, U.: AFX1 and p54(nrb): fine mapping, genomic structure, and exclusion as candidate genes of X-linked dystonia parkinsonism. *Hum. Genet.* 100:569–572, 1997.
- [39821] 13373. Bohme, B.; VandenBos, T.; Cerretti, D. P.; Park, L. S.; Holtrich, U.; Rubsamen-Waigmann, H.; Strebhardt, K.: Cell-cell adhesion mediated by binding of membrane-anchored ligand LERK-2 to the EPH-related receptor human embryonal kinase 2 promotes tyrosine kinase activity. *J. Biol. Chem.* 271: 24747–24752, 1996.
- [39822] 13374. Fletcher, F. A.; Huebner, K.; Shaffer, L. G.; Fairweather, N. D.; Monaco, A. P.; Muller, U.; Druck, T.; Simoneaux, D. K.; Chelly, J.; Belmont, J. W.; Beckmann, M. P.; Lyman, S. D.: Assignment of the gene (EPLG2) encoding a high-affinity binding protein for the receptor tyrosine kinase Elk to a 200-kilobase pair region in human chromosome Xq12. *Genomics* 25: 334–335, 1995.
- [39823] 13375. Fletcher, F. A.; Renshaw, B.; Hollingsworth, T.; Baum, P.; Lyman, S. D.; Jenkins, N. A.; Gilbert, D. J.;

Copeland, N. G.; Davison, B.L.: Genomic organization and chromosomal localization of mouse *Eplg2*, a gene encoding a binding protein for the receptor tyrosine kinase *Elk*. *Genomics* 24: 127–132, 1994.

[39824] 13376. Palmer, A.; Zimmer, M.; Erdmann, K. S.; Eulenburg, V.; Porthin, A.; Heumann, R.; Deutsch, U.; Klein, R.: EphrinB phosphorylation and reverse signaling: regulation by Src kinases and PTP-BL phosphatase. *Molec. Cell* 9: 725–737, 2002.

[39825] 13377. Bourke, E.; Frindt, G.; Flynn, P.; Schreiner, G. E.: Primary hyperoxaluria with normal alpha-ketoglutarate:glyoxylate carboligase activity: treatment with isocarboxazid. *Ann. Intern. Med.* 76: 279–284, 1972.

[39826] 13378. Danpure, C. J.: Personal Communication. Middlesex, England 6/16/1988.

[39827] 13379. Danpure, C. J.: Primary hyperoxaluria type 1 and peroxisome-to-mitochondrion mistargeting of alanine:glyoxylate aminotransferase. *Biochimie* 75:309–315, 1993.

[39828] 13380. Danpure, C. J.; Jennings, P. R.: Peroxisomal alanine:glyoxylate aminotransferase deficiency in primary hyperoxaluria type I. *FEBS Lett.* 201: 20–24, 1986.

[39829] 13381. Danpure, C. J.; Jennings, P. R.; Watts, R. W. E.: En-

zymological diagnosis of primary hyperoxaluria type 1 by measurement of hepatic alanine:glyoxylate aminotransferase activity. *Lancet* I: 289–291, 1987.

[39830] 13382. Brunelli, S.; Faiella, A.; Capra, V.; Nigro, V.; Simone, A.; Cama, A.; Boncinelli, E.: Germline mutation in the homeobox gene EMX2 in patients with severe schizencephaly. *Nature Genet.* 12: 94–96, 1996.

[39831] 13383. Rulten, S.; Thorpe, J.; Kay, J.: Identification of eukaryotic parvulin homologues: a new subfamily of peptidylprolyl cis–trans isomerases. *Biochem. Biophys. Res. Commun.* 259: 557–562, 1999.

[39832] 13384. Barton, D. E.; Yang–Feng, T. L.; Francke, U.: The human tyrosine aminotransferase gene mapped to the long arm of chromosome 16 (region 16q22–q24) by somatic cell hybrid analysis and in situ hybridization. *Hum. Genet.* 72: 221–224, 1986.

[39833] 13385. Beinfang, D. C.; Kuwabara, T.; Pueschel, S. M.: The Richner–Hanhart syndrome: report of a case with associated tyrosinemia. *Arch. Ophthalmol.* 94: 1133–1137, 1976.

[39834] 13386. Bohnert, A.; Anton–Lamprecht, I.: Richner–Hanhart's syndrome: ultrastructural abnormalities of epidermal keratinization indicating a causal relationship to high intracellular tyrosine levels. *J. Invest. Derm.* 79: 68–74,

1982.

- [39835] 13387. Buist, N.: Phenylketonuria and related problems. In: Nyhan, W.L.: Amino Acid Metabolism and Genetic Variation. New York: McGraw-Hill(pub.) 1967. Pp. 117 only.
- [39836] 13388. Chitayat, D.; Balbul, A.; Hani, V.; Mamer, O. A.; Clow, C.; Scriver, C. R.: Hereditary tyrosinaemia type II in a consanguineous Ashkenazijewish family: intrafamilial variation in phenotype; absence of parental phenotype effects on the fetus. J. Inherit. Metab. Dis. 15: 198-203, 1992.
- [39837] 13389. Crovato, F.; Desirello, G.; Gatti, R.; Babbini, N.; Rebora, A.: Richner-Hanhart syndrome spares a plantar autograft. Arch. Derm. 121:539-540, 1985.
- [39838] 13390. Fellman, J. H.; Vanbellinchen, P. J.; Jones, R. T.; Koler, R. D.: Soluble and mitochondrial forms of tyrosine aminotransferase: relationship to human tyrosinemia. Biochemistry 8: 615-622, 1969.
- [39839] 13391. Garibaldi, L. R.; Siliato, F.; De Martini, I.; Scarsi, M. R.; Romano, C.: Oculocutaneous tyrosinosis: report of two cases in the same family. Helv. Paediat. Acta 32: 173-180, 1977.
- [39840] 13392. Goldsmith, L. A.; Kang, E. S.; Bienfang, D. C.; Jimbow, K.; Gerald, P. S.; Baden, H. P.: Tyrosinemia with plan-

tar and palmar keratosis and keratitis. J. Pediat. 83: 798–805, 1973.

- [39841] 13393. Hanhart, E.: Neue Sonderformen von Keratosis palmo-plantaris, u.a. eine regelmaessig-dominante mit systematisierten Lipomen, ferner 2 einfach-rezessive mit Schwachsinn und z.T. mit Hornhautveraenderung des Auges (Ektodermatosyndrom). Dermatologica 94: 286–308, 1947.
- [39842] 13394. Hunziker, N.: Richner-Hanhart syndrome and tyrosinemia type II. Dermatologica 160:180–189, 1980.
- [39843] 13395. Kennaway, N. G.; Buist, N. R. M.: Metabolic studies in a patient with hepatic cytosol tyrosine aminotransferase deficiency. Pediat. Res. 5: 287–297, 1971.
- [39844] 13396. Muller, G.; Scherer, G.; Zentgraf, H.; Ruppert, S.; Herrmann, B.; Lehrach, H.; Schutz, G.: Isolation, characterization and chromosomal mapping of the mouse tyrosine aminotransferase gene. J. Molec. Biol. 184:367–373, 1985.
- [39845] 13397. Natt, E.; Kao, F.-T.; Rettenmeier, R.; Scherer, G.: Assignment of the human tyrosine aminotransferase gene to chromosome 16. Hum. Genet. 72: 225–228, 1986.
- [39846] 13398. Huang, L.; Gitschier, J.: A novel gene involved in zinc transport is deficient in the lethal milk mouse. Nature

Genet. 17: 292–297,1997.

[39847] 13399.McKusick, V. A.: On the X Chromosome of Man.
Washington: Am.Inst. Biol. Sci. (pub.) 1964.

[39848] 13400.Ichinose, H.; Ohye, T.; Suzuki, T.; Sumi-Ichinose,
C.; Nomura,T.; Hagino, Y.; Nagatsu, T.: Molecular cloning
of the human Nurr1gene: characterization of the human
gene and cDNAs. Gene 230: 233–239,1999.

[39849] 13401.Law, S. W.; Conneely, O. M.; DeMayo, F. J.;
O'Malley, B. W.: Identificationof a new brain-specific tran-
scription factor, NURR1. Molec. Endocr. 6:2129–2135,
1992.

[39850] 13402.Mages, H. W.; Rilke, O.; Bravo, R.; Senger, G.;
Kroczeck, R. A.: NOT, a human immediate-early response
gene closely related to thesteroid/thyroid hormone recep-
tor NAK1/TR3. Molec. Endocr. 8: 1583–1591,1994.

[39851] 13403.McEvoy, A. N.; Murphy, E. A.; Ponnio, T.; Conneely,
O. M.; Bresnihan,B.; FitzGerald, O.; Murphy, E. P.: Activa-
tion of nuclear orphan receptorNURR1 transcription by
NF-kappaB and cyclic adenosine
5-prime-monophosphateresponse element-binding pro-
tein in rheumatoid arthritis synovialtissue. J. Immun. 168:
2979–2987, 2002.

[39852] 13404.Okabe, T.; Takayanagi, R.; Imasaki, K.; Haji, M.;

Nawata, H.; Watanabe, T.: cDNA cloning of a NGFI-B/nur77-related transcription factor from an apoptotic human T cell line. *J. Immun.* 154: 3871–3879, 1995.

[39853] 13405. Torii, T.; Kwarai, T.; Nakamura, S.; Kawakami, H.: Organization of the human orphan nuclear receptor Nurr1 gene. *Gene* 230: 225–232, 1999.

[39854] 13406. Xu, P.-Y.; Liang, R.; Jankovic, J.; Hunter, C.; Zeng, Y.-X.; Ashizawa, T.; Lai, D.; Le, W.-D.: Association of homozygous 7048G/7049 variant in the intron six of Nurr1 gene with Parkinson's disease. *Neurology* 58:881–884, 2002.

[39855] 13407. Hanahan, D.: Signaling vascular morphogenesis and maintenance. *Science* 277:48–50, 1997.

[39856] 13408. Wong, S. M. E.; Lowe, A. W.: Sequence of the cDNA encoding human GP-2, the major membrane protein in the secretory granule of the exocrine pancreas. *Gene* 171: 311–312, 1996.

[39857] 13409. Yamamura, T.; Hitomi, J.; Nagasaki, K.; Suzuki, M.; Takahashi, E.; Saito, S.; Tsukada, T.; Yamaguchi, K.: Human CAAF1 gene—molecular cloning, gene structure, and chromosome mapping. *Biochem. Biophys. Res. Commun.* 221: 356–360, 1996.

[39858] 13410. Miyoshi, J.; Higashi, T.; Mukai, H.; Ohuchi, T.;

Kakunaga, T.:Structure and transforming potential of the human cot oncogene encodinga putative protein kinase. Molec. Cell. Biol. 11: 4088–4096, 1991.

[39859] 13411.Ohara, R.; Miyoshi, J.; Aoki, M.; Toyoshima, K.: The murine cotproto–oncogene: genome structure and tissue–specific expression. Jpn.J. Cancer Res. 84: 518–525, 1993.

[39860] 13412.Gould, D. B.; Walter, M. A.: Cloning, characteriza–tion, localization,and mutational screening of the human BARX1 gene. Genomics 68: 336–342,2000.

[39861] 13413.Tissier–Seta, J. P.; Mucchielli, M. L.; Mark, M.; Mattei, M. G.;Goridis, C.; Brunet, J. F.: Barx1, a new mouse homeodomain transcriptionfactor expressed in cranio–fa–cial ectomesenchyme and the stomach. Mech.Dev. 51: 3–15, 1995.

[39862] 13414.Ishihara, H.; Shibasaki, Y.; Kizuki, N.; Wada, T.; Yazaki, Y.;Asano, T.; Oka, Y.: Type I phosphatidylinositol–4–phosphate 5–kinases:cloning of the third isoform and deletion/substitution analysis ofmembers of this novel lipid kinase family. J. Biol. Chem. 273: 8741–8748,1998.

[39863] 13415.Besset, S.; Vincourt, J.–B.; Amalric, F.; Girard, J.–P.: Nuclearlocalization of PAPS synthetase 1: a sulfate activa–tion pathway inthe nucleus of eukaryotic cells. FASEB J. 14: 345–354, 2000.

- [39864] 13416. Faiyaz ul Haque, M.; King, L. M.; Krakow, D.; Cantor, R. M.; Rusiniak, M. E.; Swank, R. T.; Superti-Furga, A.; Haque, S.; Abbas, H.; Ahmad, W.; Ahmad, M.; Cohn, D. H.: Mutations in orthologous genes in human spondyloepimetaphyseal dysplasia and the brachymorphic mouse. *Nature Genet.* 20: 157–162, 1998.
- [39865] 13417. Geller, D. H.; Henry, J. G.; Belch, J.; Schwartz, N. B.: Co-purification and characterization of ATP-sulfurylase and adenosine-5-prime-phosphosulfate kinase from rat chondrosarcoma. *J. Biol. Chem.* 262: 7374–7382, 1987.
- [39866] 13418. Girard, J.-P.; Baekkevold, E. S.; Amalric, F.: Sulfation in high endothelial venules: cloning and expression of the human PAPS synthetase. *FASEB J.* 12: 603–612, 1998.
- [39867] 13419. Lyle, S.; Stanczak, J.; Ng, K.; Schwartz, N. B.: Rat chondrosarcoma ATP sulfurylase and adenosine 5-prime-phosphosulfate kinase reside on a single bifunctional protein. *Biochemistry* 33: 5920–5925, 1994.
- [39868] 13420. Venkatachalam, K. V.; Akita, H.; Strott, C. A.: Molecular cloning, expression, and characterization of human bifunctional 3-prime-phosphoadenosine 5-prime-phosphosulfate synthase and its functional domains. *J. Biol. Chem.* 273: 19311–19320, 1998.

- [39869] 13421.Park, Y. C.; Ye, H.; Hsia, C.; Segal, D.; Rich, R. L.; Liou, H.-C.;Myszka, D. G.; Wu, H.: A novel mechanism of TRAF signaling revealedby structural and functional analyses of the TRADD–TRAF2 interaction. *Cell* 101:777–787, 2000.
- [39870] 13422.Asao, H.; Sasaki, Y.; Arita, T.; Tanaka, N.; Endo, K.; Kasai, H.;Takeshita, T; Endo, Y.; Fujita, T.; Sugamura, K.: Hrs is associatedwith STAM, a signal–transducing adaptor molecule: its suppressiveeffect on cytokine–induced cell growth. *J. Biol. Chem.* 272: 32785–32791,1997.
- [39871] 13423.Takeshita, T.; Arita, T.; Asao, H.; Tanaka, N.; Higuchi, M.; Kuroda,H.; Kaneko, K.; Munakata, H.; Endo, Y.; Fujita, T.; Sugamura, K.:Cloning of a novel signal–transducing adaptor molecule containingan SH3 domain and ITAM. *Biochem. Biophys. Res. Commun.* 225: 1035–1039,1996.
- [39872] 13424.Everett, R. D.; Meredith, M.; Orr, A.; Cross, A.; Kathoria, M.;Parkinson, J.: A novel ubiquitin–specific protease is dynamicallyassociated with the PML nuclear domain and binds to a herpesvirusregulatory protein. *EMBO J.* 16: 1519–1530, 1997. Note: Corrected republication of *EMBO J.*:16: 566–577, 1997.
- [39873] 13425.Li, M.; Chen, D.; Shiloh, A.; Luo, J.; Nikolaev, A. Y.;

Qin, J.;Gu, W.: Deubiquitination of p53 by HAUSP is an important pathwayfor p53 stabilization. *Nature* 416: 648–653, 2002.

- [39874] 13426.Robinson, P. A.; Lomonte, P.; Leek, J. P.; Markham, A. F.; Everett,R. D.: Assignment of herpesvirus–associated ubiquitin–specific proteasegene HAUSP to human chromosome band 16p13.3 by in situ hybridization. *Cytogenet.Cell Genet.* 83: 100 only, 1998.
- [39875] 13427.Zhou, G.; Bao, Z. Q.; Dixon, J. E.: Components of a new humanprotein kinase signal transduction pathway. *J. Biol. Chem.* 270:12665–12669, 1995.
- [39876] 13428.Lee, J.–D.; Ulevitch, R. J.; Han, J.: Primary structure of BMK1:a new mammalian MAP kinase. *Biochem. Biophys. Res. Commun.* 213:715–724, 1995.
- [39877] 13429.Purandare, S. M.; Lee, J.–D.; Patel, P. I.: Assignment of bigMAP kinase (PRKM7) to human chromosome 17 band p11.2 with somaticcell hybrids. *Cytogenet. Cell Genet.* 83: 258–259, 1998.
- [39878] 13430.Regan, C. P.; Li, W.; Boucher, D. M.; Spatz, S.; Su, M. S.; Kuida,K.: Erk5 null mice display multiple extraembryonic vascular and embryoniccardiovascular defects. *Proc. Nat. Acad. Sci.* 99: 9248–9253, 2002.
- [39879] 13431.Barlow, G. M.; Chen, X.–N.; Shi, Z. Y.; Lyons, G. E.;

Kurnit, D.M.; Celle, L.; Spinner, N. B.; Zackai, E.; Pettenati, M. J.; Van Riper, A. J.; Vekemans, M. J.; Mjaatvedt, C. H.; Korenberg, J. R.: Downsyndrome congenital heart disease: a narrowed region and a candidate gene. *Genet. Med.* 3: 91–101, 2001.

[39880] 13432. Schmucker, D.; Clemens, J. C.; Shu, H.; Worby, C. A.; Xiao, J.; Muda, M.; Dixon, J. E.; Zipursky, S. L.: *Drosophila* Dscam is an axon guidance receptor exhibiting extraordinary molecular diversity. *Cell* 101:671–684, 2000.

[39881] 13433. Yamakawa, K.; Huo, Y.-K.; Haendel, M. A.; Hubert, R.; Chen, X.-N.; Lyons, G. E.; Korenberg, J. R.: DSCAM: a novel member of the immunoglobulin superfamily maps in a Down syndrome region and is involved in the development of the nervous system. *Hum. Molec. Genet.* 7: 227–237, 1998.

[39882] 13434. Zhu, J.; Petersen, S.; Tessarollo, L.; Nussenzweig, A.: Targeted disruption of the Nijmegen breakage syndrome gene NBS1 leads to early embryonic lethality in mice. *Curr. Biol.* 11: 105–109, 2001.

[39883] 13435. Cooper, P. R.; Smilnich, N. J.; Day, C. D.; Nowak, N. J.; Reid, L. H.; Pearsall, R. S.; Reece, M.; Prawitt, D.; Landers, J.; Housman, D. E.; Winterpacht, A.; Zabel, B. U.; Pel-

letier, J.; Weissman, B.E.; Shows, T. B.; Higgins, M. J.: Divergently transcribed overlapping genes expressed in liver and kidney and located in the 11p15.5 imprinted domain. *Genomics* 49: 38–51, 1998.

- [39884] 13436. Kershaw, D. B.; Beck, S. G.; Wharram, B. L.; Wiggins, J. E.; Goyal, M.; Thomas, P. E.; Wiggins, R. C.: Molecular cloning and characterization of human podocalyxin-like protein: orthologous relationship to rabbit PCLP1 and rat podocalyxin. *J. Biol. Chem.* 272: 15708–15714, 1997.
- [39885] 13437. Kershaw, D. B.; Wiggins, J. E.; Wharram, B. L.; Wiggins, R. C.: Assignment of the human podocalyxin-like protein (PODXL) gene to 7q32–q33. *Genomics* 45: 239–240, 1997.
- [39886] 13438. Chan, K. K.; Tsui, S. K. W.; Lee, S. M. Y.; Luk, S. C. W.; Liew, C. C.; Fung, K. P.; Waye, M. M. Y.; Lee, C. Y.: Molecular cloning and characterization of FHL2, a novel LIM domain protein preferentially expressed in human heart. *Gene* 210: 345–350, 1998.
- [39887] 13439. Genini, M.; Schwalbe, P.; Scholl, F. A.; Remppis, A.; Mattei, M.-G.; Schafer, B. W.: Subtractive cloning and characterization of DRAL, a novel LIM-domain protein down-regulated in rhabdomyosarcoma. *Dev. Cell Biol.* 16: 433–442, 1997.

- [39888] 13440.Claverie–Martin, F.; Wang, M.; Cohen, S. N.: ARD–1 cDNA from humancells encodes a site–specific single–strand endoribonuclease thatfunctionally resembles Es–cherichia coli RNase E. J. Biol. Chem. 272:13823–13828, 1997.
- [39889] 13441.Van Eynde, A.; Wera, S.; Beullens, M.; Torrekens, S.; Van Leuven,F.; Stalmans, W.; Bollen, M.: Molecular cloning of NIPP–1, a nuclearinhibitor of protein phosphatase–1, reveals homology with polypeptidesinvolved in RNA pro–cessing. J. Biol. Chem. 270: 28068–28074, 1995.
- [39890] 13442.Tekin, M.; Dogu, F.; Tacyildiz, N.; Akar, E.; Ikin–ciogullari,A.; Ogur, G.; Yavuz, G.; Babacan, E.; Akar, N.: 657del5 mutationin the NBS1 gene is associated with Ni–jmegen breakage syndrome ina Turkish family. Clin. Genet. 62: 84–88, 2002.
- [39891] 13443.Varon, R.; Reis, A.; Henze, G.; Einsiedel, H. G.; Sperling, K.;Seeger, K.: Mutations in the Nijmegen break–age syndrome gene (NBS1)in childhood acute lymphoblas–tic leukemia (ALL). Cancer Res. 61:3570–3572, 2001.
- [39892] 13444.Wilda, M.; Demuth, I.; Concannon, P.; Sperling, K.; Hameister,H.: Expression pattern of the Nijmegen break–age syndrome gene, Nbs1,during murine development. Hum. Molec. Genet. 9: 1739–1744, 2000.

- [39893] 13445.Wu, X.; Ranganathan, V.; Weisman, D. S.; Heine, W. F.; Ciccone,D. N.; O'Neill, T. B.; Crick, K. E.; Pierce, K. A.; Lane, W. S.; Rathbun,G.; Livingston, D. M.; Weaver, D. T.: ATM phosphorylation of Nijmegenbreakage syndrome protein is required in a DNA damage response. *Nature* 405:477–482, 2000.
- [39894] 13446.Zhao, S.; Weng, Y.–C.; Yuan, S.–S. F.; Lin, Y.–T.; Hsu, H.–C.;Lin, S.–C. J.; Gerbino, E.; Song, M.; Zdzienicka, M. Z.; Gatti, R.A.; Shay, J. W.; Ziv, Y.; Shiloh, Y.; Lee, E. Y.–H. P.: Functionallink between ataxia–telangiectasia and Nijmegen breakage syndromegene products. *Nature* 405: 473–477, 2000.
- [39895] 13447.Mankodi, A.; Urbinati, C. R.; Yuan, Q.–P.; Moxley, R. T.; Sansone,V.; Krym, M.; Henderson, D.; Schalling, M.; Swanson, M. S.; Thornton,C. A.: Muscleblind localizes to nuclear foci of aberrant RNA in myotonicdystrophy types 1 and 2. *Hum. Molec. Genet.* 10: 2165–2170, 2001.
- [39896] 13448.Hittner, H. M.; Kretzer, F. L.; Antoszyk, J. H.; Ferrell, R. E.;Mehta, R. S.: Variable expressivity of autosomal dominant anteriorsegment mesenchymal dysgenesis in six generations. *Am. J. Ophthal.* 93:57–70, 1982.
- [39897] 13449.Semina, E. V.; Ferrell, R. E.; Mintz–Hittner, H. A.; Bitoun, P.;Alward, W. L. M.; Reiter, R. S.; Funkhauser, C.;

Daack-Hirsch, S.;Murray, J. C.: A novel homeobox gene PITX3 is mutated in familieswith autosomal-dominant cataracts and ASMD. Nature Genet. 19: 167–170,1998.

[39898] 13450.Semina, E. V.; Murray, J. C.; Reiter, R.; Hrstka, R. F.; Graw,J.: Deletion in the promoter region and altered expression of Pitx3homeobox gene in aphakia mice. Hum. Molec. Genet. 9: 1575–1585,2000.

[39899] 13451.Semina, E. V.; Reiter, R. S.; Murray, J. C.: Isolation of a newhomeobox gene belonging to the Pitx/Rieg family: expression duringlens development and mapping to the aphakia region on mouse chromosome19. Hum. Molec. Genet. 6: 2109–2116, 1997.

[39900] 13452.Kent, J.; Lee, M.; Schedl, A.; Boyle, S.; Fantes, J.; Powell, M.;Rushmere, N.; Abbott, C.; van Heyningen, V.; Bickmore, W. A.: Thereticulocalbin gene maps to the WAGR region in human and to the Smalleye Harwell deletion in mouse. Genomics 42: 260–267, 1997.

[39901] 13453.Ozawa, M.: Cloning of a human homologue of mouse reticulocalbinreveals conservation of structural domains in the novel endoplasmicreticulum resident Ca²⁺-binding protein with multiple EF-hand motifs. J.Biochem. 117: 1113–1119, 1995.

[39902] 13454.Ozawa, M.; Muramatsu, T.: Reticulocalbin, a novel

endoplasmicreticulum resident Ca^{2+} -binding protein with multiple EF-hand motifs and a carboxyl-terminal HDEL sequence. J. Biol. Chem. 268: 699–705, 1993.

- [39903] 13455. Luo, G.; Leroy, E.; Kozak, C. A.; Polymeropoulos, M. H.; Horowitz, R.: Mapping of the gene (NRAP) encoding N-RAP in the mouse and human genomes. Genomics 45: 229–232, 1997.
- [39904] 13456. Luo, G.; Zhang, J. Q.; Nguyen, T.-P.; Herrera, A. H.; Paterson, B.; Horowitz, R.: Complete cDNA sequence and tissue localization of N-RAP, a novel nebulin-related protein of striated muscle. Cell Motil. Cytoskeleton 38: 75–90, 1997.
- [39905] 13457. Ichikawa, S.; Ozawa, K.; Hirabayashi, Y.: Assignment of a UDP-glucose:ceramideglucosyltransferase gene (Ugcg) to mouse chromosome band 4B3 by insitu hybridization. Cytogenet. Cell Genet. 83: 14–15, 1998.
- [39906] 13458. Ichikawa, S.; Ozawa, K.; Hirabayashi, Y.: Assignment of a UDP-glucose:ceramideglucosyltransferase gene (UGCG) to human chromosome band 9q31 by insitu hybridization. Cytogenet. Cell Genet. 79: 233–234, 1997.
- [39907] 13459. Ichikawa, S.; Sakiyama, H.; Suzuki, G.; Hidari, K. I.-P. J.; Hirabayashi, Y.: Expression cloning of a cDNA for human ceramide glucosyltransferase that catalyzes the

first glycosylation step of glycosphingolipid synthesis.

Proc.Nat. Acad. Sci. 93: 4638–4643, 1996.

- [39908] 13460.Watanabe, R.; Wu, K.; Paul, P.; Marks, D. L.; Kobayashi, T.; Pittelkow,M. R.; Pagano, R. E.: Up-regulation of glucosylceramide synthaseexpression and activity during human keratinocyte differentiation. J.Biol. Chem. 273: 9651–9655, 1998.
- [39909] 13461.Strahm, B.; Rittweiler, K.; Duffner, U.; Brandau, O.; Orłowska-Volk,M.; Karajannis, M. A.; zur Stadt, U.; Tie-mann, M.; Reiter, A.; Brandis,M.; Meindl, A.; Niemeyer, C. M.: Recurrent B-cell non-Hodgkin's lymphomain two brothers with X-linked lymphoproliferative disease with-out evidence for Epstein-Barr virus infection. Brit. J. Haemat. 108:377–382, 2000.
- [39910] 13462.Sylla, B. S.; Murphy, K.; Cahir-McFarland, E.; Lane, W. S.; Mosialos,G.; Kieff, E.: The X-linked lymphoproliferative syndrome gene productSH2D1A associates with p62(dok) (Dok1) and activates NF-kappa-beta. Proc.Nat. Acad. Sci. 97: 7470–7475, 2000.
- [39911] 13463.Sylla, B. S.; Wang, Q.; Hayoz, D.; Lathrop, G. M.; Lenoir, G.M.: Multipoint linkage mapping of the Xq25–q26 region in a familyaffected by the X-linked lymphoproliferative syndrome. Clin. Genet. 36:359–462, 1989.

- [39912] 13464. Tangye, S. G.; Lazetic, S.; Woollatt, E.; Sutherland, G. R.; Lanier, L. L.; Phillips, J. H.: Cutting edge: human 2B4, an activating NK cell receptor, recruits the protein tyrosine phosphatase SHP-2 and the adaptor signaling protein SAP. *J. Immun.* 162: 6981–6985, 1999.
- [39913] 13465. Tangye, S. G.; Phillips, J. H.; Lanier, L. L.; Nichols, K. E.: Cutting edge: functional requirement for SAP in 2B4-mediated activation of human natural killer cells as revealed by the X-linked lymphoproliferative syndrome. *J. Immun.* 165: 2932–2936, 2000.
- [39914] 13466. Thorley-Lawson, D. A.; Schooley, R. T.; Bhan, A. K.; Nadler, L. M.: Epstein-Barr virus superinduces a new human B cell differentiation antigen (B-LAST 1) expressed on transformed lymphoblasts. *Cell* 30:415–425, 1982.
- [39915] 13467. Vowels, M. R.; Lam-Po-Tang, R.; Berdoukas, V.; Ford, D.; Thierry, D.; Purtilo, D.; Gluckman, E.: Correction of X-linked lymphoproliferative disease by transplantation of cord-blood stem cells. *New Eng. J. Med.* 329: 1623–1625, 1993.
- [39916] 13468. Williams, L. L.; Rooney, C. M.; Conley, M. E.; Brenner, M. K.; Krance, R. A.; Heslop, H. E.: Correction of Duncan's syndrome by allogeneic bone marrow transplantation. *Lancet* 342: 587–588, 1993.

- [39917] 13469.Wu, C.; Nguyen, K. B.; Pien, G. C.; Wang, N.; Gullo, C.; Duncan,H.; Sosa, M. R.; Edwards, M. J.; Borrow, P.; Satoskar, A. R.; Sharpe,A. H.; Biron, C. A.; Terhorst, C.: SAP controls T cell responsesto virus and terminal differ-entiation of T(H)2 cells. *Nature Immun.* 2:410–414, 2001.
- [39918] 13470.Wyandt, H. E.; Skare, J. C.; Grierson, H. L.; Purtilo, D. T.;Milunsky, A.: Detection of a chromosomal deletion of Xq25 in an affectedmale with X-linked lymphoproliferative disease. (Abstract) *Am. J.Hum. Genet.* 45 (suppl.): A108 only, 1989.
- [39919] 13471.Yin, L.; Ferrand, V.; Lavoue, M.–F.; Hayoz, D.; Philippe, N.;Souillet, G.; Seri, M.; Giacchino, R.; Castag-nola, E.; Hodgson, S.;Sylla, B. S.; Romeo, G.: SH2D1A mu-tation analysis for diagnosis ofXLP in typical and atypical patients. *Hum. Genet.* 105: 501–505,1999.
- [39920] 13472.Yin, L.; Tocco, T.; Pauly, S.; Lenoir, G. M.; Romeo, G.: Absenceof SH2D1A point mutation in 62 Burkitts lym-phoma cell lines. *Am.J. Hum. Genet.* 65 (suppl. 1868): A331 only, 1999.
- [39921] 13473.Laxminarayan, K. M.; Chan, B. K.; Tetaz, T.; Bird, P. I.; Mitchell,C. A.: Characterization of a cDNA encoding the 43–kDa membrane–associatedinositol–polyphosphate 5–phosphatase. *J. Biol. Chem.* 269: 17305–17310,1994.

- [39922] 13474.Fink, J. K.; Jones, S. M.; Esposito, C.; Wilkowski, J.: Humanmicrotubule-associated protein 1a (MAP1A) gene: genomic organization,cDNA sequence, and developmen-tal-and tissue-specific expression. *Genomics* 35:577-585, 1996.
- [39923] 13475.Asker, C.; Steinitz, M.; Andersson, K.; Sumegi, J.; Klein, G.;Ingvarsson, S.: Nucleotide sequence of the rat Bmyc gene.. *Oncogene* 4:1523-1527, 1989.
- [39924] 13476.Ingvarsson, S.; Sundaresan, S.; Jin, P.; Francke, U.; Asker, C.;Sumegi, J.; Klein, G.; Sejersen, T.: Chromosome localization andexpression pattern of Lmyc and Bmyc in murine embryonal carcinomacells. *Oncogene* 3: 679-685, 1988.
- [39925] 13477.Kojima, T.; Inazawa, J.; Takamatsu, J.; Rosenberg, R. D.; Saito,H.: Human ryudocan core protein: molecular cloning and characterizationof the cDNA, and chromoso-mal localization of the gene. *Biochem. Biophys.Res. Com-mun.* 190: 814-822, 1993.
- [39926] 13478.Kojima, T.; Shworak, N. W.; Rosenberg, R. D.: Molecular cloningand expression of two distinct cDNA-encoding heparan sulfate proteoglycancore proteins from a rat endothelial cell line. *J. Biol. Chem.* 267:4870-4877, 1992.

- [39927] 13479.Yu, H.; Humphries, D. E.; Watkins, M.; Karlinsky, J. B.: Molecularcloning of the human ryudocan promoter. Biochem. Biophys. Res. Commun. 212:1139–1144, 1995.
- [39928] 13480.McBride, O. W.; Yi, H. F.; Srivastava, M.: The human cytochrome b561 gene (CYB561) is located at 17q11–qter. Genomics 21: 662–663,1994.
- [39929] 13481.Srivastava, M.: Genomic structure and expression of the humangene encoding cytochrome b(561), an integral protein of the cromaffingranule membrane. J. Biol. Chem. 270: 22714–22720, 1995.
- [39930] 13482.Albarosa, R.; DiDonato, S.; Finocchiaro, G.: Redefinition of thecoding sequence of the MXI1 gene and identification of a polymorphicrepeat in the 3–prime non–coding region that allows the detectionof loss of heterozygosity of chromosome 10q25 in glioblastomas. Hum.Genet. 95: 709–711, 1995.
- [39931] 13483.Mitchell, C. A.; Speed, C. J.; Nicholl, J.; Sutherland, G. R.:Chromosomal mapping of the gene (INPP5A) encoding the 43–kDa membrane–associatedinositol polyphosphate 5–phosphatase to 10q26.3 by fluorescence insitu hybridization. Genomics 31: 139–140, 1996.
- [39932] 13484.Sugasawa, K.; Ng, J. M. Y.; Masutani, C.; Iwai, S.; van der Spek,P. J.; Eker, A. P. M.; Hanaoka, F.; Bootsma,

D.; Hoeijmakers, J. H.J.: Xeroderma pigmentosum group C protein complex is the initiator of global genome nucleotide excision repair. *Molec. Cell* 2: 223–232, 1998.

[39933] 13485. van der Spek, P. J.; Smit, E. M. E.; Beverloo, H. B.; Sugawara, K.; Masutani, C.; Hanaoka, F.; Hoeijmakers, J. H. J.; Hagemmeijer, A.: Chromosomal localization of three repair genes: the xeroderma pigmentosum group C gene and two human homologs of yeast RAD23. *Genomics* 23:651–658, 1994.

[39934] 13486. van der Spek, P. J.; Visser, C. E.; Hanaoka, F.; Smit, B.; Hagemmeijer, A.; Bootsma, D.; Hoeijmakers, J. H. J.: Cloning, comparative mapping, and RNA expression of the mouse homologues of the *Saccharomyces cerevisiae* nucleotide excision repair gene RAD23. *Genomics* 31: 20–27, 1996.

[39935] 13487. Netik, A.; Forss-Petter, S.; Holzinger, A.; Molzer, B.; Unterrainer, G.; Berger, J.: Adrenoleukodystrophy-related protein can compensate functionally for adrenoleukodystrophy protein deficiency (X-ALD): implications for therapy. *Hum. Molec. Genet.* 8: 907–913, 1999.

[39936] 13488. Rajantie, I.; Ekman, N.; Iljin, K.; Arighi, E.; Gunji, Y.; Kaukonen, J.; Palotie, A.; Dewerchin, M.; Carmeliet, P.; Alitalo, K.: Bmx tyrosine kinase has a redundant function

downstream of angiopoietin and vascular endothelial growth factor receptors in arterial endothelium.

Molec.Cell. Biol. 21: 4647–4655, 2001.

- [39937] 13489. Tamagnone, L.; Lahtinen, I.; Mustonen, T.; Virtaneva, K.; Francis, F.; Muscatelli, F.; Alitalo, R.; Edvard Smith, C. I.; Larsson, C.; Alitalo, K.: BMX, a novel nonreceptor tyrosine kinase gene of the BTK/ITK/TEC/TK family located in chromosome Xp22.2. *Oncogene* 9:3683–3688, 1994.
- [39938] 13490. Dinulos, M. B.; Bassi, M. T.; Rugarli, E. I.; Chapman, V.; Ballabio, A.; Distèche, C. M.: A new region of conservation is defined between human and mouse X chromosomes. *Genomics* 35: 244–247, 1996.
- [39939] 13491. Schiaffino, M. V.; Bassi, M. T.; Rugarli, E. I.; Renieri, A.; Galli, L.; Ballabio, A.: Cloning of a human homologue of the *Xenopus laevis* APX gene from the ocular albinism type 1 critical region. *Hum. Molec. Genet.* 4: 373–382, 1995.
- [39940] 13492. Bachner, D.; Sedlacek, Z.; Korn, B.; Hameister, H.; Poustka, A.: Expression patterns of two human genes coding for different rabGDP-dissociation inhibitors (GDIs), extremely conserved proteins involved in cellular transport. *Hum. Molec. Genet.* 4: 701–708, 1995.
- [39941] 13493. Black, G. C. M.; Perveen, R.; Bonshek, R.; Cahill, M.;

Clayton-Smith, J.; Lloyd, I. C.; McLeod, D.: Coats' disease of the retina (unilateral retinal telangiectasis) caused by somatic mutation in the NDP gene: a role for norrin in retinal angiogenesis. *Hum. Molec. Genet.* 8:2031–2035, 1999.

[39942] 13494. Mao, M.; Fu, G.; Wu, J.-S.; Zhang, Q.-H.; Zhou, J.; Kan, L.-X.; Huang, Q.-H.; He, K.-L.; Gu, B.-W.; Han, Z.-G.; Shen, Y.; Gu, J.; Yu, Y.-P.; Xu, S.-H.; Wang, Y.-X.; Chen, S.-J.; Chen, Z.: Identification of genes expressed in human CD34+ hematopoietic stem/progenitor cells by expressed sequence tags and efficient full-length cDNA cloning. *Proc. Nat. Acad. Sci.* 95: 8175–8180, 1998.

[39943] 13495. Chen, Y.-T.; Gure, A. O.; Tsang, S.; Stockert, E.; Jager, E.; Knuth, A.; Old, L. J.: Identification of multiple cancer/testis antigens by allogeneic antibody screening of a melanoma cell line library. *Proc. Nat. Acad. Sci.* 95: 6919–6923, 1998.

[39944] 13496. Lucas, S.; De Smet, C.; Arden, K. C.; Viars, C. S.; Lethe, B.; Lurquin, C.; Boon, T.: Identification of a new MAGE gene with tumor-specific expression by representational difference analysis. *Cancer Res.* 58:743–752, 1998.

[39945] 13497. Pearce, W. G.; Sanger, R.; Race, R. R.: Ocular albinism and Xg. *Lancet* I:1282–1283, 1968.

- [39946] 13498.Rosenberg, T.; Schwartz, M.: X-linked ocular albinism: prevalence and mutations--a national study. *Eur. J. Hum. Genet.* 6: 570-577, 1998.
- [39947] 13499.Schiaffino, M. V.; Bassi, M. T.; Galli, L.; Renieri, A.; Bruttini, M.; De Nigris, F.; Bergen, A. A. B.; Charles, S. J.; Yates, J. R.W., Meindl, A.; Lewis, R. A.; King, R. A.; Ballabio, A.: Analysis of the OA1 gene reveals mutations in only one-third of patients with X-linked ocular albinism. *Hum. Molec. Genet.* 4: 2319-2325, 1995.
- [39948] 13500.Schiaffino, M. V.; d'Addio, M.; Alloni, A.; Baschirotto, C.; Valetti, C.; Cortese, K.; Puri, C.; Bassi, M. T.; Colla, C.; De Luca, M.; Tacchetti, C.; Ballabio, A.: Ocular albinism: evidence for a defect in an intracellular signal transduction system. *Nature Genet.* 23: 108-112, 1999.
- [39949] 13501.Schnur, R. E.; Gao, M.; Wick, P. A.; Keller, M.; Benke, P. J.; Edwards, M. J.; Grix, A. W.; Hockey, A.; Jung, J. H.; Kidd, K. K.; Kistenmacher, M.; Levin, A. V.; and 11 others: OA1 mutations and deletions in X-linked ocular albinism. *Am. J. Hum. Genet.* 62: 800-809, 1998.
- [39950] 13502.Schnur, R. E.; Trask, B. J.; van den Engh, G.; Punnett, H. H.; Kistenmacher, M.; Tomeo, M. A.; Naidu, R. E.; Nussbaum, R. L.: An Xp22 microdeletion associated with ocular albinism and ichthyosis: approximation of break-

points and estimation of deletion size by using cloned DNA probes and flow cytometry. *Am. J. Hum. Genet.* 45: 706–720, 1989.

- [39951] 13503. Schnur, R. E.; Wick, P. A.; Bailey, C.; Rebbeck, T.; Weleber, R. G.; Wagstaff, J.; Grix, A. W.; Pagon, R. A.; Hockey, A.; Edwards, M. J.: Phenotypic variability in X-linked ocular albinism: relationship to linkage genotypes. *Am. J. Hum. Genet.* 55: 484–496, 1994.
- [39952] 13504. Vogt, A.: *Die Iris: Albinismus solum bulbi. Atlas Spalt-Lampen-Mikroskopie*(pub.) (3rd ed.): 1942.
- [39953] 13505. Waardenburg, P. J.; Van den Bosch, J.: X-chromosomal ocular albinism in Dutch family. *Ann. Hum. Genet.* 21: 101–122, 1956.
- [39954] 13506. Zhang, Y.; McMahon, R.; Charles, S. J.; Green, J. S.; Moore, A. T.; Barton, D. E.; Yates, J. R. W.: Genetic mapping of the Kallmann syndrome and X linked ocular albinism gene loci. *J. Med. Genet.* 30: 923–925, 1993.
- [39955] 13507. Shapira, E.; Ben-Yoseph, Y.; Eyal, G.; Russell, A.: Enzymatically inactive red cell carbonic anhydrase B in a family with renal tubular acidosis. *J. Clin. Invest.* 53: 59–63, 1974.
- [39956] 13508. Cianfriglia, M.; Miggiano, V. C.; Meo, T.; Muller, H. J.; Muller, E.; Battistuzzi, G.: Evidence for synteny between

the rabbit geneloci coding for HPRT, PGK and G6PD in mouse-rabbit somatic cell hybrids.(Abstract) Cytogenet. Cell Genet. 25: 142, 1979.

- [39957] 13509.Holterhus, P. M.; Sinnecker, G. H. G.; Wollmann, H. A.; Struve,D.; Homburg, N.; Kruse, K.; Hiort, O.: Expression of two functionallydifferent androgen receptors in a patient with androgen insensitivity. Europ.J. Pediat. 158: 702-706, 1999.
- [39958] 13510.Shimmin, L. C.; Chang, B.-H.; Li, W.-H.: Male-driven evolutionof DNA sequences. Nature 362: 745-747, 1993.
- [39959] 13511.Warburg, M.: Personal Communication. Copenhagen, Denmark 1966.
- [39960] 13512.Imasaki, K.; Okabe, T.; Murakami, H.; Tanaka, Y.; Haji, M.; Takayanagi,R.; Nawata, H.: Androgen insensitivity syndrome due to new mutationsin the DNA-binding domain of the androgen receptor. Molec. Cell.Endocr. 120: 15-24, 1996.
- [39961] 13513.Jakubiczka, S.; Nedel, S.; Werder, E. A.; Schleiermacher, E.;Theile, U.; Wolff, G.; Wieacker, P.: Mutations of the androgen receptorgene in patients with complete androgen insensitivity. Hum. Mutat. 9:57-61, 1997.
- [39962] 13514.Kang, H.-Y.; Yeh, S.; Fujimoto, N.; Chang, C.:

Cloning and characterization of human prostate coactivator ARA54, a novel protein that associates with the androgen receptor. *J. Biol. Chem.* 274: 8570–8576, 1999.

- [39963] 13515. Kazemi-Esfarjani, P.; Beitel, L. K.; Trifiro, M.; Kaufman, M.; Rennie, P.; Sheppard, P.; Matusik, R.; Pinsky, L.: Substitution of valine-865 by methionine or leucine in the human androgen receptor causes complete or partial androgen insensitivity, respectively with distinct androgen receptor phenotypes. *Molec. Endocr.* 7: 37–46, 1993.
- [39964] 13516. Kittles, R. A.; Young, D.; Weinrich, S.; Hudson, J.; Argyropoulos, G.; Ukoli, F.; Adams-Campbell, L.; Dunston, G. M.: Extent of linkage disequilibrium between the androgen receptor gene CAG and GGC repeats in human populations: implications for prostate cancer risk. *Hum. Genet.* 109: 253–261, 2001.
- [39965] 13517. Klocker, H.; Kaspar, F.; Eberle, J.; Uberreiter, S.; Radmayr, C.; Bartsch, G.: Point mutation in the DNA binding domain of the androgen receptor in two families with Reifenshtein syndrome. *Am. J. Med. Genet.* 50: 1318–1327, 1992.
- [39966] 13518. Knoke, I.; Allera, A.; Wieacker, P.: Significance of the CAG repeat length in the androgen receptor gene (AR) for the transactivation function of an M780I mutant AR.

Hum. Genet. 104: 257–261, 1999.

- [39967] 13519.Kobayashi, Y.; Miwa, S.; Merry, D. E.; Kume, A.; Mei, L.; Doyu,M.; Sobue, G.: Caspase–3 cleaves the expanded androgen receptor proteinof spinal and bulbar muscular atrophy in a polyglutamine repeat length–dependentmanner. Biochem. Biophys. Res. Commun. 252: 145–150, 1998.
- [39968] 13520.Koivisto, P. A.; Schleutker, J.; Helin, H.; Ehren–van Eekelen,C.; Kallioniemi, O.–P.; Trapman, J.: Androgen receptor gene alterationsand chromosomal gains and losses in prostate carcinomas appearingduring finasteride treatment for benign prostatic hyperplasia. Clin.Cancer Res. 5: 3578–3582, 1999.
- [39969] 13521.Kooy, R. F.; Reyniers, E.; Storm, K.; Vits, L.; van Velzen, D.;de Ruiter, P. E.; Brinkmann, A. O.; de Paepe, A.; Willems, P. J.:CAG repeat contraction in the androgen receptor gene in three brotherswith mental retardation. Am. J. Med. Genet. 85: 209–213, 1999.
- [39970] 13522.La Spada, A.; Fischbeck, K. H.: Androgen receptor gene defectin X–linked spinal and bulbar muscular atrophy. (Abstract) Am. J.Hum. Genet. 49 (suppl.): 20, 1991.
- [39971] 13523.Lewis, M.; Kaita, H.; Giblett, E. R.; Anderson, J.; Philipps,S.; Steinberg, A. G.; McAlpine, P. J.: Multiplicity of

genetic polymorphisms of blood in the Schmiedeleut Hut-
terites. *Am. J. Med. Genet.* 22:477–485, 1985.

- [39972] 13524.Lim, H. N.; Chen, H.; McBride, S.; Dunning, A. M.;
Nixon, R. M.; Hughes, I. A.; Hawkins, J. R.: Longer polyglu-
tamine tracts in the androgen receptor are associated with
moderate to severe undermasculinized genitalia in XY
males. *Hum. Molec. Genet.* 9: 829–834, 2000.
- [39973] 13525.Lobaccaro, J.-M.; Lumbroso, S.; Belon, C.; Galtier-
Dereure, F.; Bringer, J.; Lesimple, T.; Namer, M.; Cutuli, B.
F.; Pujol, H.; Sultan, C.: Androgen receptor gene mutation
in male breast cancer. *Hum. Molec. Genet.* 2: 1799–1802,
1993.
- [39974] 13526.Lubahn, D. B.; Brown, T. R.; Simental, J. A.; Higgs,
H. N.; Migeon, C. J.; Wilson, E. M.; French, F. S.: Sequence
of the intron/exon junctions of the coding region of the
human androgen receptor gene and identification of a
point mutation in a family with complete androgen insen-
sitivity. *Proc. Nat. Acad. Sci.* 86: 9534–9538, 1989.
- [39975] 13527.Lubahn, D. B.; Joseph, D. R.; Sar, M.; Tan, J.; Higgs,
H. N.; Larson, R. E.; French, F. S.; Wilson, E. M.: The human
androgen receptor: complementary deoxyribonucleic acid
cloning, sequence analysis and gene expression in
prostate. *Molec. Endocr.* 2: 1265–1275, 1988.

- [39976] 13528.Lubahn, D. B.; Joseph, D. R.; Sullivan, P. M.; Willard, H. F.; French, F. S.; Wilson, E. M.: Cloning of human androgen receptor complementary DNA and localization to the X chromosome. *Science* 240:327–330, 1988.
- [39977] 13529.Lumbroso, S.; Lobaccaro, J. M.; Georget, V.; Leger, J.; Poujol, N.; Terouanne, B.; Evain-Brion, D.; Czernichow, P.; Sultan, C.: A novel substitution (leu707-to-arg) in exon 4 of the androgen receptor gene causes complete androgen resistance. *J. Clin. Endocr. Metab.* 81:1984–1988, 1996.
- [39978] 13530.Stuppia, L.; Calabrese, G.; Franchi, P. G.; Mingarelli, R.; Gatta, V.; Palka, G.; Dallapiccola, B.: Widening of a Y-chromosome interval–6 deletion transmitted from a father to his infertile son accounts for an oligozoospermia critical region distal to the RBM1 and DAZ genes. (Letter) *Am. J. Hum. Genet.* 59: 1393–1395, 1996.
- [39979] 13531.Brown, G. M.; Furlong, R. A.; Sargent, C. A.; Erickson, R. P.; Longepied, G.; Mitchell, M.; Jones, M. H.; Hargreave, T. B.; Cooke, H. J.; Affara, N. A.: Characterisation of the coding sequence and fine mapping of the human DF-*FRY* gene and comparative expression analysis and mapping to the Sxr-b interval of the mouse Y chromosome of the *Dffry* gene. *Hum. Molec. Genet.* 7: 97–107, 1998.

- [39980] 13532.Ferlin, A.; Moro, E.; Garolla, A.; Foresta, C.: Human male infertility and Y chromosome deletions: role of the AZF-candidate genes DAZ, RBM and DFFRY. *Hum. Reprod.* 14: 1710–1716, 1999.
- [39981] 13533.Foresta, C.; Ferlin, A.; Moro, E.: Deletion and expression analysis of AZFa genes on the human Y chromosome revealed a major role for DBY in male infertility. *Hum. Molec. Genet.* 9: 1161–1169, 2000.
- [39982] 13534.Sargent, C. A.; Boucher, C. A.; Kirsch, S.; Brown, G.; Weiss, B.; Trundley, A.; Burgoyne, P.; Saut, N.; Durand, C.; Levy, N.; Terriou, P.; Hargreave, T.; Cooke, H.; Mitchell, M.; Rappold, G. A.; Affara, N. A.: The critical region of overlap defining the AZFa male infertility interval of proximal Yq contains three transcribed sequences. *J. Med. Genet.* 36: 670–677, 1999.
- [39983] 13535.Shen, P.; Wang, F.; Underhill, P. A.; Franco, C.; Yang, W.-H.; Roxas, A.; Sung, R.; Lin, A. A.; Hyman, R. W.; Vollrath, D.; Davis, R. W.; Cavalli-Sforza, L. L.; Oefner, P. J.: Population genetic implications from sequence variation in four Y chromosome genes. *Proc. Nat. Acad. Sci.* 97: 7354–7359, 2000.
- [39984] 13536.Sun, C.; Skaletsky, H.; Birren, B.; Devon, K.; Tang, Z.; Silber, S.; Oates, R.; Page, D. C.: An azoospermic man

with a de novo pointmutation in the Y-chromosomal gene USP9Y. Nature Genet. 23: 429–432,1999.

[39985] 13537.Thomson, R.; Pritchard, J. K.; Shen, P.; Oefner, P. J.; Feldman,M. W.: Recent common ancestry of human Y chromosomes: evidence fromDNA sequence data. Proc. Nat. Acad. Sci. 97: 7360–7365, 2000.

[39986] 13538.Schiebel, K.; Winkelmann, M.; Mertz, A.; Xu, X.; Page, D. C.; Weil,D.; Petit, C.; Rappold,, G. A.: Abnormal XY interchange between anovel isolated protein kinase gene, PRKY, and its homologue, PRKX,accounts for one third of all (Y+)XX males and (Y-)XY females. Hum.Molec. Genet. 6: 1985–1989, 1997.

[39987] 13539.Greenfield, A.; Scott, D.; Pennisi, D.; Ehrmann, I.; Ellis, P.;Cooper, L.; Simpson, E.; Koopman, P.: An H-YDb epitope is encodedby a novel mouse Y chromosome gene. Nature Genet. 14: 474–478, 1996.

[39988] 13540.Scott, D.; Addey, C.; Ellis, P; James, E.; Mitchell, M. J.; Saut,N.; Jurcevic, S.; Simpson, E.: Dendritic cells permit identificationof genes encoding MHC class II–restricted epitopes of transplantationantigens. Immunity 12: 711–720, 2000.

[39989] 13541.Wu, G.; Chai, J.; Suber, T. L.; Wu, J.–W.; Du, C.; Wang, X.; Shi,Y.: Structural basis of IAP recognition by

Smac/DIABLO. *Nature* 408:1008–1012, 2000.

- [39990] 13542. Macke, J. P.; Hu, N.; Hu, S.; Bailey, M.; King, V. L.; Brown, T.; Hamer, D.; Nathans, J.: Sequence variation in the androgen receptor gene is not a common determinant of male sexual orientation. *Am. J. Hum. Genet.* 53: 844–852, 1993.
- [39991] 13543. Yang, R.-B.; Fulle, H.-J.; Garbers, D. L.: Chromosomal localization and genomic organization of genes encoding guanylyl cyclase receptors expressed in olfactory sensory neurons and retina. *Genomics* 31:367–372, 1996.
- [39992] 13544. Nakamura, H.; Izumoto, Y.; Kambe, H.; Kuroda, T.; Mori, T.; Kawamura, K.; Yamamoto, H.; Kishimoto, T.: Molecular cloning of complementary DNA for a novel human hepatoma-derived growth factor: its homology with high mobility group-1 protein. *J. Biol. Chem.* 269: 25143–25149, 1994.
- [39993] 13545. Wanschura, S.; Schoenmakers, E. F. P. M.; Huysmans, C.; Bartnitzke, S.; Van de Ven, W. J. M.; Bullerdiek, J.: Mapping of the gene encoding the human hepatoma-derived growth factor (HDGF) with homology to the high-mobility group (HMG)-1 protein to Xq25. *Genomics* 32: 298–300, 1996.
- [39994] 13546. Baens, M.; Marynen, P.: A human homologue

(BICD1) of the Drosophilabicaudal-D gene. Genomics 45: 601–606, 1997.

- [39995] 13547.Bullock, S. L.; Ish-Horowicz, D.: Conserved signals and machineryfor RNA transport in Drosophila oogenesis and embryogenesis. Nature 414:611–616, 2001.
- [39996] 13548.McMurtrie, E. B.; Barbosa, M. D. F. S.; Zerial, M.; Kingsmore,S. F.: Rab17 and Rab18, small GTPases with specificity for polarizedepithelial cells: genetic mapping in the mouse. Genomics 45: 623–625,1997.
- [39997] 13549.Schafer, U.; Seibold, S.; Schneider, A.; Neugebauer, E.: Isolationand characterisation of the human rab18 gene after stimulation ofendothelial cells with histamine. FEBS Lett. 466: 148–154, 2000.
- [39998] 13550.Doupnik, C. A.; Davidson, N.; Lester, H. A.: The inward rectifierpotassium channel family. Curr. Opin. Neurobiol. 5: 268–277, 1995.
- [39999] 13551.Tada, Y.; Horio, Y.; Takumi, T.; Terayama, M.; Tsuji, L.; Copeland,N. G.; Jenkins, N. A.; Kurachi, Y.: Assignment of the glial inwardlyrectifying potassium channel K(AB)–2/Kir4.1 (Kcnj10) gene to the distalregion of mouse chromosome 1. Genomics 45: 629–630, 1997.
- [40000] 13552.Takumi, T.; Ishii, T.; Horio, Y.; Morishige, K.–I.; Takahashi,N.; Yamada, M.; Yamashita, T.; Kiyama, H.;

Sohmiya, K.; Nakanishi, S.; Kurachi, Y.: A novel ATP-dependent inward rectifier potassium channel expressed predominantly in glial cells. *J. Biol. Chem.* 270:16339–16436, 1995.

[40001] 13553. Schenker, T.; Trueb, B.: Assignment of the gene for a developmentally regulated GTP-binding protein (DRG2) to human chromosome bands 17p13–p12 by in situ hybridization. *Cytogenet. Cell Genet.* 79: 274–275, 1997.

[40002] 13554. Heikkinen, J.; Risteli, M.; Wang, C.; Latvala, J.; Rossi, M.; Valtavaara, M.; Myllyla, R.: Lysyl hydroxylase 3 is a multifunctional protein possessing collagen glucosyl-transferase activity. *J. Biol. Chem.* 275:36158–36163, 2000.

[40003] 13555. Passoja, K.; Rautavuoma, K.; Ala-Kokko, L.; Kosonen, T.; Kivirikko, K. I.: Cloning and characterization of a third human lysyl hydroxylase isoform. *Proc. Nat. Acad. Sci.* 95: 10482–10486, 1998.

[40004] 13556. Valtavaara, M.; Szpirer, C.; Szpirer, J.; Myllyla, R.: Primary structure, tissue distribution, and chromosomal localization of a novel isoform of lysyl hydroxylase (lysyl hydroxylase 3). *J. Biol. Chem.* 273: 12881–12886, 1998.

[40005] 13557. Rohan, P. J.; Davis, P.; Moskaluk, C. A.; Kearns, M.; Kruttsch, H.; Siebenlist, U.; Kelly, K.: PAC-1: a mitogen-in-

duced nuclear protein tyrosine phosphatase. *Science* 259: 1763–1766, 1993.

- [40006] 13558. Ward, Y.; Gupta, S.; Jensen, P.; Wartmann, M.; Davis, R. J.; Kelly, K.: Control of MAP kinase activation by the mitogen-induced threonine/tyrosine phosphatase PAC1. *Nature* 367: 651–654, 1994.
- [40007] 13559. Yi, H.; Morton, C. C.; Weremowicz, S.; McBride, O. W.; Kelly, K.: Genomic organization and chromosomal localization of the DUSP2 gene, encoding a MAP kinase phosphatase, to human 2p11.2–q11. *Genomics* 28: 92–96, 1995.
- [40008] 13560. Ishibashi, T.; Bottaro, D. P.; Michieli, P.; Kelley, C. A.; Aaronson, S. A.: A novel dual specificity phosphatase induced by serum stimulation and heat shock. *J. Biol. Chem.* 269: 29897–29902, 1994.
- [40009] 13561. Kwak, S. P.; Dixon, J. E.: Multiple dual specificity protein tyrosine phosphatases are expressed and regulated differentially in liver cell lines. *J. Biol. Chem.* 270: 1156–1160, 1995.
- [40010] 13562. Kovalenko, O. V.; Golub, E. I.; Bray-Ward, P.; Ward, D. C.; Radding, C. M.: A novel nucleic acid-binding protein that interacts with human Rad51 recombinase. *Nucleic Acids Res.* 25: 4946–4953, 1997.

- [40011] 13563. Mizuta, R.; LaSalle, J. M.; Cheng, H.-L.; Shinohara, A.; Ogawa, H.; Copeland, N.; Jenkins, N. A.; Lalande, M.; Alt, F. W.: RAB22 and RAB163/mouse BRCA2: proteins that specifically interact with the RAD51 protein. *Proc. Nat. Acad. Sci.* 94: 6927–6932, 1997.
- [40012] 13564. Blasius, R.; Weber, R. G.; Lichter, P.; Ogilvie, A.: A novel orphan G protein-coupled receptor primarily expressed in the brain is localized on human chromosomal band 2q21. *J. Neurochem.* 70: 1357–1365, 1998.
- [40013] 13565. Raport, C. J.; Schweickart, V. L.; Chantry, D.; Eddy, R. L., Jr.; Shows, T. B.; Godiska, R.; Gray, P. W.: New members of the chemokine receptor gene family. *J. Leukoc. Biol.* 59: 18–23, 1996.
- [40014] 13566. Shindo, M.; Nakano, H.; Kuroyanagi, H.; Shirasawa, T.; Mihara, M.; Gilbert, D. J.; Jenkins, N. A.; Copeland, N. G.; Yagita, H.; Okumura, K.: cDNA cloning, expression, subcellular localization, and chromosomal assignment of mammalian aurora homologues, aurora-related kinase (ARK)1 and 2. *Biochem. Biophys. Res. Commun.* 244: 285–292, 1998.
- [40015] 13567. Adachi, H.; Tsujimoto, M.; Hattori, M.; Arai, H.; Inoue, K.: cDNA cloning of human cytosolic platelet-activating factor acetylhydrolase gamma-subunit and its

mRNA expression in human tissues. *Biochem. Biophys. Res. Commun.* 214: 180–187, 1995.

- [40016] 13568. Presky, D. H.; Yang, H.; Minetti, L. J.; Chua, A. O.; Nabavi, N.; Wu, C.-Y.; Gately, M. K.; Gubler, U.: A functional interleukin 12 receptor complex is composed of two beta-type cytokine receptor subunits. *Proc. Nat. Acad. Sci.* 93: 14002–14007, 1996.
- [40017] 13569. Yamamoto, K.; Kobayashi, H.; Miura, O.; Hirosawa, S.; Miyasaka, N.: Assignment of IL12RB1 and IL12RB2, interleukin-12 receptor beta-1 and beta-2 chains, to human chromosome 19 band p13.1 and chromosome 1 band p31.2, respectively, by in situ hybridization. *Cytogenet. Cell Genet.* 77: 257–258, 1997.
- [40018] 13570. Lafreniere, R. G.; Rochefort, D. L.; Kibar, Z.; Fon, E. A.; Han, F.-Y.; Cochius, J.; Kang, X.; Baird, S.; Korneluk, R. G.; Andermann, E.; Rommens, J. M.; Rouleau, G. A.: Isolation and characterization of GT335, a novel human gene conserved in *Escherichia coli* and mapping to 21q22.3. *Genomics* 38: 264–272, 1996.
- [40019] 13571. Nagamine, K.; Kudoh, J.; Minoshima, S.; Kawasaki, K.; Asakawa, S.; Ito, F.; Shimizu, N.: Isolation of cDNA for a novel human protein KNP-I that is homologous to the *E. coli* SCRP-27A protein from the autoimmune polyglandular

disease type I (APECED) region of chromosome 21q22.3.

Biochem. Biophys. Res. Commun. 225: 608–616, 1996.

[40020] 13572.Scott, H. S.; Chen, H.; Rossier, C.; Lalioti, M. D.; Antonarakis, S. E.: Isolation of a human gene (HES1) with homology to an Escherichia coli and a zebrafish protein that maps to chromosome 21q22.3. Hum.Genet. 99: 616–623, 1997.

[40021] 13573.Bernier–Villamor, V.; Sampson, D. A.; Matunis, M. J.; Lima, C.D.: Structural basis for E2–mediated SUMO conjugation revealed by a complex between ubiquitin–conjugating enzyme Ubc9 and RanGAP1. Cell 108:345–356, 2002.

[40022] 13574.Shi, Y.; Zou, M.; Farid, N. R.; Paterson, M. C.: Association of FHIT (fragile histidine triad), a candidate tumour suppressor gene, with the ubiquitin–conjugating enzyme hUBC9. Biochem. J. 352: 443–448, 2000.

[40023] 13575.Tachibana, M.; Iwata, N.; Watanabe, A.; Nobukuni, Y.; Ploplis, B.; Kajigaya, S.: Assignment of the gene for a ubiquitin–conjugating enzyme (UBE2I) to human chromosome band 16p13.3 by in situ hybridization. Cytogenet.Cell Genet. 75: 222–223, 1996.

[40024] 13576.Wang, Z.–Y.; Qiu, Q.–Q.; Seufert, W.; Taguchi, T.; Testa, J. R.; Whitmore, S. A.; Callen, D. F.; Welsh, D.; Shenk,

T.; Deuel, T. F.: Molecular cloning of the cDNA and chromosome localization of the gene for human ubiquitin-conjugating enzyme 9. *J. Biol. Chem.* 271:24811–24816, 1996.

[40025] 13577. Yasugi, T.; Howley, P. M.: Identification of the structural and functional human homolog of the yeast ubiquitin conjugating enzyme UBC9. *Nucleic Acids Res.* 24: 2005–2010, 1996.

[40026] 13578. Bord, S.; Horner, A.; Beavan, S.; Compston, J.: Estrogen receptors alpha and beta are differentially expressed in developing human bone. *J. Clin. Endocr. Metab.* 86: 2309–2314, 2001.

[40027] 13579. Watanabe, T. K.; Fujiwara, T.; Kawai, A.; Shimizu, F.; Takami, S.; Hirano, H.; Okuno, S.; Ozaki, K.; Takeda, S.; Shimada, Y.; Nagata, M.; Takaichi, A.; Takahashi, E.; Nakamura, Y.; Shin, S.: Cloning, expression, and mapping of UBE2I, a novel gene encoding a human homologue of yeast ubiquitin-conjugating enzymes which are critical for regulating the cell cycle. *Cytogenet. Cell Genet.* 72: 86–89, 1996.

[40028] 13580. Chaidarun, S. S.; Swearingen, B.; Alexander, J. M.: Differential expression of estrogen receptor-beta (ER-beta) in human pituitary tumors: functional interactions with ER-

alpha and a tumor-specific splice variant. J. Clin. Endocr. Metab. 83: 3308–3315, 1998.

[40029] 13581. Chu, S.; Mamers, P.; Burger, H. G.; Fuller, P. J.: Estrogen receptor isoform gene expression in ovarian stromal and epithelial tumors. J. Clin. Endocr. Metab. 85: 1200–1205, 2000.

[40030] 13582. Enmark, E.; Peltö-Huikko, M.; Grandien, K.; Lagercrantz, S.; Lagercrantz, J.; Fried, G.; Nordenskjöld, M.; Gustafsson, J.-A.: Human estrogen receptor beta-gene structure, chromosomal localization, and expression pattern. J. Clin. Endocr. Metab. 82: 4258–4265, 1997.

[40031] 13583. Esmaili, B.; Harvey, J. T.; Hewlett, B.: Immunohistochemical evidence for estrogen receptors in meibomian glands. Ophthalmology 107: 180–184, 2000.

[40032] 13584. Forsell, C.; Enmark, E.; Axelman, K.; Blomberg, M.; Wahlund, L.-O.; Gustafsson, J.-A.; Lannfelt, L.: Investigations of a CA repeat in the estrogen receptor beta gene in patients with Alzheimer's disease. Europ. J. Hum. Genet. 9: 802–804, 2001.

[40033] 13585. Korach, K. S.; Couse, J. F.; Curtis, S. W.; Washburn, T. F.; Lindzey, J.; Kimbro, K. S.; Eddy, E. M.; Migliaccio, S.; Snedeker, S. M.; Lubahn, D. B.; Schomberg, D. W.; Smith, E. P.: Estrogen receptor gene disruption: molecular charac-

terization and experimental and clinical phenotypes. Recent Prog. Horm. Res. 51: 159–188, 1996.

- [40034] 13586. Krege, J. H.; Hodgin, J. B.; Couse, J. F.; Enmark, E.; Warner, M.; Mahler, J. F.; Sar, M.; Korach, K. S.; Gustafsson, J.-A.; Smithies, O.: Generation and reproductive phenotypes of mice lacking estrogen receptor beta. Proc. Nat. Acad. Sci. 95: 15677–15682, 1998.
- [40035] 13587. Krezel, W.; Dupont, S.; Krust, A.; Chambon, P.; Chapman, P. F.: Increased anxiety and synaptic plasticity in estrogen receptor beta-deficient mice. Proc. Nat. Acad. Sci. 98: 12278–12282, 2001.
- [40036] 13588. Kuiper, G. G. J. M.; Enmark, E.; Peltö-Huikko, M.; Nilsson, S.; Gustafsson, J.-A.: Cloning of a novel estrogen receptor expressed in rat prostate and ovary. Proc. Nat. Acad. Sci. 93: 5925–5930, 1996.
- [40037] 13589. Mosialos, G.; Birkenbach, M.; Yalamanchili, R.; VanArsdale, T.; Ware, C.; Kieff, E.: The Epstein-Barr virus transforming protein LMP1 engages signaling proteins for the tumor necrosis factor receptor family. Cell 80: 389–399, 1995.
- [40038] 13590. Rothe, M.; Wong, S. C.; Henzel, W. J.; Goeddel, D. V.: A novel family of putative signal transducers associated with the cytoplasmic domain of the 75 kDa tumor necrosis

factor receptor. Cell 78: 681–692,1994.

[40039] 13591.Sieminski, K.; Peters, N.; Scheurich, P.; Wajant, H.: Organization of the human tumour necrosis factor receptor-associated factor 1 (TRAF1) gene and mapping to chromosome 9q33–34. Gene 195: 35–39, 1997.

[40040] 13592.Tsitsikov, E. N.; Laouini, D.; Dunn, I. F.; Sannikova, T. Y.; Davidson,L.; Alt, F. W.; Geha, R. S.: TRAF1 is a negative regulator of TNF signaling: enhanced TNF signaling in TRAF1-deficient mice. Immunity 15:647–657, 2001.

[40041] 13593.Jacquemin, P.; Hwang, J.-J.; Martial, J. A.; Dolle, P.; Davidson,I.: A novel family of developmentally regulated mammalian transcription factors containing the TEA/ATTS DNA binding domain. J. Biol. Chem. 271:21775–21785, 1996.

[40042] 13594.Bejjani, B. A.; Lewis, R. A.; Tomey, K. F.; Anderson, K. L.; Dueker,D. K.; Jabak, M.; Astle, W. F.; Otterud, B.; Leppert, M.; Lupski,J. R.: Mutations in CYP1B1, the gene for cytochrome P4501B1, are the predominant cause of primary congenital glaucoma in Saudi Arabia. Am.J. Hum. Genet. 62: 325–333, 1998.

[40043] 13595.Bejjani, B. A.; Stockton, D. W.; Lewis, R. A.; Tomey, K. F.; Dueker,D. K.; Jabak, M.; Astle, W. F.; Lupski, J. R.: Multiple CYP1B1 mutations and incomplete penetrance in

an inbred population segregating primary congenital glaucoma suggest frequent de novo events and a dominant-modifier locus. *Hum. Molec. Genet.* 9: 367–374, 2000.

[40044] 13596. Lin, J.; Arnold, H. B.; Della-Fera, M. A.; Azain, M. J.; Hartzell, D. L.; Baile, C. A.: Myostatin knockout in mice increases myogenesis and decreases adipogenesis. *Biochem. Biophys. Res. Commun.* 291:701–706, 2002.

[40045] 13597. McPherron, A. C.; Lawler, A. M.; Lee, S.-J.: Regulation of skeletal muscle mass in mice by a new TGF- β superfamily member. *Nature* 387:83–90, 1997.

[40046] 13598. Robins, P.; Lindahl, T.: DNA ligase IV from HeLa cell nuclei. *J. Biol. Chem.* 271: 24257–24261, 1996.

[40047] 13599. Roldan-Arjona, T.; Wei, Y.-F.; Carter, K. C.; Klungland, A.; Anselmino, C.; Wang, R.-P.; Augustus, M.; Lindahl, T.: Molecular cloning and functional expression of a human cDNA encoding the antimutator enzyme 8-hydroxyguanine-DNA glycosylase. *Proc. Nat. Acad. Sci.* 94: 8016–8020, 1997.

[40048] 13600. Guo, Z.; Turner, C.; Castle, D.: Relocation of the t-SNARE SNAP-23 from lamellipodia-like cell surface projections regulates compound exocytosis in mast cells. *Cell* 94: 537–548, 1998.

[40049] 13601. Lazo, P. A.; Nadal, M.; Ferrer, M.; Area, E.; Hernan-

dez-Torres,J.; Nabokina, S. M.; Mollinedo, F.; Estivill, X.: Genomic organization,chromosomal localization, alternative splicing, and isoforms of thehuman synaptosome-associated protein-23 gene implicated in vesicle-membranefusion processes. Hum. Genet. 108: 211-215, 2001.

[40050] 13602.Mollinedo, F.; Lazo, P. A.: Identification of two isoforms ofthe vesicle-membrane fusion protein SNAP-23 in human neutrophils andHL-60 cells. Biochem. Biophys. Res. Commun. 231: 808-812, 1997.

[40051] 13603.Ravichandran, V.; Chawla, A.; Roche, P. A.: Identification ofa novel syntaxin- and synaptobrevin/VAMP-binding protein, SNAP-23,expressed in non-neuronal tissues. J. Biol. Chem. 271: 13300-13303,1996.

[40052] 13604.Shukla, A.; Corydon, T. J.; Nielsen, S.; Hoffmann, H. J.; Dahl,R.: Identification of three new splice variants of the SNARE proteinSNAP-23. Biochem. Biophys. Res. Commun. 285: 320-327, 2001.

[40053] 13605.Mugita, N.; Kumura, Y.; Ogawa, M.; Saya, H.; Nakao, M.: Identificationof a novel, tissue-specific calpain htra-3; a human homologue of theCaenorhabditis elegans sex determination gene. Biochem. Biophys.Res. Commun. 239: 845-850, 1997.

- [40054] 13606.Sharpless, N. E.; Ferguson, D. O.; O'Hagan, R. C.; Castrillon,D. H.; Lee, C.; Farazi, P. A.; Alson, S.; Fleming, J.; Morton, C.C.; Frank, K.; Chin, L.; Alt, F. W.; DePinho, R. A.: Impaired nonhomologousend-joining provokes soft tissue sarcomas harboring chromosomal translocations,amplifications, and deletions. *Molec. Cell* 8: 1187-1196, 2001.
- [40055] 13607.Bohme, B.; Holtrich, U.; Wolf, G.; Luzius, H.; Grzeschik, K.-H.;Strebhardt, K.; Rubsamen-Waigmann, H.: PCR mediated detection ofa new human receptor-tyrosine-kinase, HEK 2. *Oncogene* 8: 2857-2862,1993.
- [40056] 13608.Ruiz, J. C.; Conlon, F. L.; Robertson, E. J.: Identification ofnovel protein kinases expressed in the myocardium of the developingmouse heart. *Mech. Dev.* 48: 153-164, 1994.
- [40057] 13609.Ashbourne, K. J.; Byth, B. C.; Meijers, J. C. M.; Cox, D. W.:Polymorphism of the protein C inhibitor (PCI) gene on chromosome 14. *Hum.Molec. Genet.* 2: 92 only, 1993.
- [40058] 13610.Sadler, J. E.: Combined factors V and VIII deficiency climbs ontothe map. (Editorial) *J. Clin. Invest.* 99: 555-556, 1997.
- [40059] 13611.Yasuda, T.; Nadano, D.; Iida, R.; Tanaka, Y.; Nakanaga, M.; Kishi,K.: Discovery of a genetic polymor-

phism of human plasma protein C inhibitor (PCI): genetic survey utilizing isoelectric focusing followed by immunoblotting, immunological and biochemical characterization. *Hum.Genet.* 89: 265–269, 1992.

- [40060] 13612. Eki, T.; Okumura, K.; Amin, A.; Ishiai, M.; Abe, M.; Nogami, M.; Taguchi, H.; Hurwitz, J.; Murakami, Y.; Hanaoka, F.: Mapping of the human homologue (ORC1L) of the yeast origin recognition complex subunit 1 gene to chromosome band 1p32. *Genomics* 36: 559–561, 1996.
- [40061] 13613. Mendez, J.; Zou-Yang, H.; Kim, S.-Y.; Hidaka, M.; Tansey, W. P.; Stillman, B.: Human origin recognition complex large subunit is degraded by ubiquitin-mediated proteolysis after initiation of DNA replication. *Molec.Cell* 9: 481–491, 2002.
- [40062] 13614. Bui, T. D.; Rankin, J.; Smith, K.; Huguet, E. L.; Ruben, S.; Strachan, T.; Harris, A. L.; Lindsay, S.: A novel human Wnt gene, WNT10B, maps to 12q13 and is expressed in human breast carcinomas. *Oncogene* 14: 1249–1253, 1997.
- [40063] 13615. Hardiman, G.; Kastelein, R. A.; Bazan, J. F.: Isolation, characterization and chromosomal localization of human WNT10B. *Cytogenet. Cell Genet.* 77: 278–282, 1997.
- [40064] 13616. Ross, S. E.; Hemati, N.; Longo, K. A.; Bennett, C. N.;

Lucas, P.C.; Erickson, R. L.; MacDougald, O. A.: Inhibition of adipogenesis by Wnt signaling. *Science* 289: 950–953, 2000.

[40065] 13617. Meyerhardt, J. A.; Look, A. T.; Bigner, S. H.; Fearon, E. R.: Identification and characterization of neogenin, a DCC-related gene. *Oncogene* 14:1129–1136, 1997.

[40066] 13618. Vielmetter, J.; Chen, X.-N.; Miskevich, F.; Lane, R. P.; Yamakawa, K.; Korenberg, J. R.; Dreyer, W. J.: Molecular characterization of human neogenin, a DCC-related protein, and the mapping of its gene (NEO1) to chromosomal position 15q22.3–q23. *Genomics* 41: 414–421, 1997.

[40067] 13619. Vielmetter, J.; Kayyem, J. F.; Roman, J. M.; Dreyer, W. J.: Neogenin, an avian cell surface protein expressed during terminal neuronal differentiation, is closely related to the human tumor suppressor molecule deleted in colorectal cancer. *J. Cell Biol.* 127: 2009–2020, 1994.

[40068] 13620. Rosenquist, T. A.; Zharkov, D. O.; Grollman, A. P.: Cloning and characterization of a mammalian 8-oxoguanine DNA glycosylase. *Proc. Nat. Acad. Sci.* 94: 7429–7434, 1997.

[40069] 13621. Tani, M.; Shinmura, K.; Kohno, T.; Shiroishi, T.; Wakama, S.; Kim, S.-R.; Nohmi, T.; Kasai, H.; Takenoshita, S.; Nagamachi, Y.; Yokota, J.: Genomic structure and chro-

mosomal localization of the mouse Ogg1 gene that is involved in the repair of 8-hydroxyguanine in DNA damage. Mammalian Genome 9: 32–37, 1998.

- [40070] 13622.Lim, H. N.; Hawkins, J. R.; Hughes, I. A.: Genetic evidence to exclude the androgen receptor co-factor, ARA70 (NCOA4) as a candidate gene for the causation of undermasculinised genitalia. (Letter) Clin.Genet. 59: 284–286, 2001.
- [40071] 13623.Santoro, M.; Dathan, N. A.; Berlingieri, M. T.; Bongarzone, I.; Paulin, C.; Grieco, M.; Pierotti, M. A.; Vecchio, G.; Fusco, A.: Molecular characterization of RET/PTC3: a novel rearranged version of the RET proto-oncogene in a human thyroid papillary carcinoma. Oncogene 9:509–516, 1994.
- [40072] 13624.Yeh, S.; Chang, C.: Cloning and characterization of a specific coactivator, ARA-70, for the androgen receptor in human prostate cells. Proc.Nat. Acad. Sci. 93: 5517–5521, 1996.
- [40073] 13625.Donghi, R.; Sozzi, G.; Pierotti, M. A.; Biunno, I.; Miozzo, M.; Fusco, A.; Grieco, M.; Santoro, M.; Vecchio, G.; Spurr, N. K.; Della Porta, G.: The oncogene associated with human papillary thyroid carcinoma (PTC) is assigned to chromosome 10 q11–q12 in the same region as multi-

pleendocrine neoplasia type 2A (MEN2A). *Oncogene* 4: 521–523, 1989.

[40074] 13626. Nikiforova, M. N.; Stringer, J. R.; Blough, R.; Medvedovic, M.; Fagin, J. A.; Nikiforov, Y. E.: Proximity of chromosomal loci that participate in radiation-induced rearrangements in human cells. *Science* 290:138–141, 2000.

[40075] 13627. Sozzi, G.; Pierotti, M. A.; Miozzo, M.; Donghi, R.; Radice, P.; De Benedetti, V.; Grieco, M.; Santoro, M.; Fusco, A.; Vecchio, G.; Mathew, C. G. P.; Ponder, B. A. J.; Spurr, N. K.: Refined localization to contiguous regions on chromosome 10q of the two genes (H4 and RET) that form the oncogenic sequence PTC. *Oncogene* 6: 339–342, 1991.

[40076] 13628. Tong, Q.; Li, Y.; Smanik, P. A.; Fithian, L. J.; Xing, S.; Mazzaferri, E. L.; Jhiang, S. M.: Characterization of the promoter region and oligomerization domain of H4 (D10S170), a gene frequently rearranged with the ret proto-oncogene. *Oncogene* 10: 1781–1787, 1995.

[40077] 13629. Araki, T.; Milbrandt, J.: Ninjurin, a novel adhesion molecule, is induced by nerve injury and promotes axonal growth. *Neuron* 17:353–361, 1996.

[40078] 13630. Araki, T.; Zimonjic, D. B.; Popescu, N. C.; Milbrandt, J.: Mechanism of homophilic binding mediated by ninjurin, a novel widely expressed adhesion molecule. *J. Biol. Chem.*

272: 21373–21380, 1997.

- [40079] 13631. Chadwick, B. P.; Heath, S. K.; Williamson, J.; Obermayr, F.; Patel, L.; Sheer, D.; Frischauf, A.-M.: The human homologue of the *ninjurin* gene maps to the candidate region of hereditary sensory neuropathy type I (HSN1). *Genomics* 47: 58–63, 1998.
- [40080] 13632. Mandich, P.; Bellone, E.; Di Maria, E.; Pigullo, S.; Pizzuti, A.; Schenone, A.; Soriani, S.; Varese, A.; Windbank, A. J.; Ajmar, F.: Exclusion of the *ninjurin* gene as a candidate for hereditary sensory neuropathies type I and type II. *Am. J. Med. Genet.* 83: 409–410, 1999.
- [40081] 13633. McAllister, G.; Whiting, P.; Hammond, E. A.; Knowles, M. R.; Atack, J. R.; Bailey, F. J.; Maigetter, R.; Ragan, C. I.: cDNA cloning of human and rat brain myo-inositol monophosphatase: expression and characterization of the human recombinant enzyme. *Biochem. J.* 284: 749–754, 1992.
- [40082] 13634. Sjoholt, G.; Molven, A.; Lovlie, R.; Wilcox, A.; Sikela, J. M.; Steen, V. M.: Genomic structure and chromosomal localization of a human myo-inositol monophosphatase gene (IMPA). *Genomics* 45: 113–122, 1997.
- [40083] 13635. Steen, V. M.; Gulbrandsen, A. K.; Eiken, H. G.; Berle, J. O.: Lack of genetic variation in the coding region of the

myo-inositolmonophosphatase gene in lithium-treated patients with manic depressive illness. *Pharmacogenetics* 6: 113–116, 1996.

[40084] 13636. McDonald, M. T.; Flejter, W.; Sheldon, S.; Putzi, M. J.; Gorski, J. L.: XY sex reversal and gonadal dysgenesis due to 9p24 monosomy. *Am. J. Med. Genet.* 73: 321–326, 1997.

[40085] 13637. Muroya, K.; Okuyama, T.; Goishi, K.; Ogiso, Y.; Fukuda, S.; Kameyama, J.; Sato, H.; Suzuki, Y.; Terasaki, H.; Gomyo, H.; Wakui, K.; Fukushima, Y.; Ogata, T.: Sex-determining gene(s) on distal 9p: clinical and molecular studies in six cases. *J. Clin. Endocr. Metab.* 85: 3094–3100, 2000.

[40086] 13638. Raymond, C. S.; Shamu, C. E.; Shen, M. M.; Seifert, K. J.; Hirsch, B.; Hodgkin, J.; Zarkower, D.: Evidence for evolutionary conservation of sex-determining genes. *Nature* 391: 691–695, 1998.

[40087] 13639. Shan, Z.; Zabel, B.; Trautmann, U.; Hillig, U.; Ottolenghi, C.; Wang, Y.; Haaf, T.: FISH mapping of the sex-reversal region on human chromosome 9p in two XY females and in primates. *Europ. J. Hum. Genet.* 8: 167–173, 2000.

[40088] 13640. Shen, M. M.; Hodgkin, J.: mab-3, a gene required

for sex-specific yolk protein expression and a male-specific lineage in *C. elegans*. *Cell* 54:1019–1031, 1988.

[40089] 13641. Smith, C. A.; McClive, P. J.; Western, P. S.; Reed, K. J.; Sinclair, A. H.: Conservation of a sex-determining gene. (Letter) *Nature* 402:601–602, 1999.

[40090] 13642. Veitia, R.; Nunes, M.; Brauner, R.; Doco-Fenzy, M.; Joanny-Flinois, O.; Jaubert, F.; Lortat-Jacob, S.; Fellous, M.; McElreavey, K.: Deletion of distal 9p associated with 46,XY male to female sex reversal: definition of the breakpoints at 9p23.3–p24.1. *Genomics* 41: 271–274, 1997.

[40091] 13643. Takekawa, M.; Posas, F.; Saito, H.: A human homolog of the yeast Ssk2/Ssk22 MAP kinase kinase kinases, MTK1, mediates stress-induced activation of the p38 and JNK pathways. *EMBO J.* 16: 4973–4982, 1997.

[40092] 13644. Chapman, D. L.; Agulnik, I.; Hancock, S.; Silver, L. M.; Papaioannou, V. E.: Tbx6, a mouse T-box gene implicated in paraxial mesoderm formation at gastrulation. *Dev. Biol.* 180: 534–542, 1996.

[40093] 13645. Chapman, D. L.; Papaioannou, V. E.: Three neural tubes in mouse embryos with mutations in the T-box gene Tbx6. *Nature* 391: 695–697, 1998.

[40094] 13646. Papapetrou, C.; Putt, W.; Fox, M.; Edwards, Y. H.: The human TBX6 gene: cloning and assignment to chro-

mosome 16p11.2. Genomics 55:238–241, 1999.

[40095] 13647.Spicer, A. P.; Olson, J. S.; McDonald, J. A.: Molecular cloning and characterization of a cDNA encoding the third putative mammalian hyaluronan synthase. J. Biol. Chem. 272: 8957–8961, 1997.

[40096] 13648.Bagri, A.; Marin, O.; Plump, A. S.; Mak, J.; Pleasure, S. J.; Rubenstein, J. L. R.; Tessier–Lavigne, M.: Slit proteins prevent midline crossing and determine the dorsoventral position of major axonal pathways in the mammalian fore–brain. Neuron 33: 233–248, 2002.

[40097] 13649.Dallol, A.; Forgacs, E.; Martinez, A.; Sekido, Y.; Walker, R.; Kishida, T.; Rabbitts, P.; Maher, E. R.; Minna, J. D.; Latif, F.: Tumour specific promoter region methylation of the human homologue of the Drosophila Roundabout gene DUTT1 (ROBO1) in human cancers. Oncogene 21:3020–3028, 2002.

[40098] 13650.Kidd, T.; Brose, K.; Mitchell, K. J.; Fetter, R. D.; Tessier–Lavigne, M.; Goodman, C. S.; Tear, G.: Roundabout controls axon crossing of the CNS midline and defines a novel subfamily of evolutionarily conserved guidance receptors. Cell 92: 205–215, 1998.

[40099] 13651.Sundaresan, V.; Chung, G.; Heppell–Parton, A.; Xiong, J.; Grundy, C.; Roberts, I.; James, L.; Cahn, A.;

Bench, A.; Douglas, J.; Minna, J.; Sekido, Y.; Lerman, M.; Latif, F.; Bergh, J.; Li, H.; Lowe, N.; Ogilvie, D.; Rabbitts, P.: Homozygous deletions at 3p12 in breast and lung cancer. *Oncogene* 17: 1723–1729, 1998.

[40100] 13652. Zallen, J. A.; Yi, B. A.; Bargmann, C. I.: The conserved immunoglobulin superfamily member SAX-3/Robo directs multiple aspects of axon guidance in *C. elegans*. *Cell* 92: 217–227, 1998.

[40101] 13653. Fischer, U.; Heckel, D.; Michel, A.; Janka, M.; Hulsebos, T.; Meese, E.: Cloning of a novel transcription factor-like gene amplified in human glioma including astrocytoma grade I. *Hum. Molec. Genet.* 6: 1817–1822, 1997.

[40102] 13654. Gracia, E.; Fischer, U.; Elkahoul, A.; Trent, J. M.; Meese, E.; Meltzer, P. S.: Isolation of genes amplified in human cancers by microdissection mediated cDNA capture. *Hum. Molec. Genet.* 5: 595–600, 1996.

[40103] 13655. Gerard, M.; Hernandez, L.; Wevrick, R.; Stewart, C. L.: Disruption of the mouse *necdin* gene results in early post-natal lethality. *Nature Genet.* 23: 199–202, 1999.

[40104] 13656. Hurst, L. D.; McVean, G.; Moore, T.: Imprinted genes have few and small introns. (Letter) *Nature Genet.* 12: 234–237, 1996.

[40105] 13657. Jay, P.; Rougeulle, C.; Massacrier, A.; Moncla, A.;

Mattei, M.-G.; Malzac, P.; Roeckel, N.; Taviaux, S.; Lefranc, J.-L. B.; Cau, P.; Berta, P.; Lalande, M.; Muscatelli, F.: The human necdin gene, NDN, is maternally imprinted and located in the Prader-Willi syndrome chromosomal region. *Nature Genet.* 17: 357-360, 1997.

[40106] 13658. MacDonald, H. R.; Wevrick, R.: The necdin gene is deleted in Prader-Willi syndrome and is imprinted in human and mouse. *Hum. Molec. Genet.* 6:1873-1878, 1997.

[40107] 13659. Maruyama, K.; Usami, M.; Aizawa, T.; Yoshikawa, K.: A novel brain-specific mRNA encoding nuclear protein (necdin) expressed in neurally differentiated embryonal carcinoma cells. *Biochem. Biophys. Res. Comm.* 178: 291-296, 1991.

[40108] 13660. Muscatelli, F.; Abrous, D. N.; Massacrier, A.; Boccaccio, I.; LeMoal, M.; Cau, P.; Cremer, H.: Disruption of the mouse Necdin gene results in hypothalamic and behavioral alterations reminiscent of the human Prader-Willi syndrome. *Hum. Molec. Genet.* 9: 3101-3110, 2000.

[40109] 13661. Nakada, Y.; Taniura, H.; Uetsuki, T.; Inazawa, J.; Yoshikawa, K.: The human chromosomal gene for necdin, a neuronal growth suppressor, in the Prader-Willi syndrome deletion region. *Gene* 213: 65-72, 1998.

[40110] 13662. Nicholls, R. D.: Incriminating gene suspects,

Prader–Willi style. *NatureGenet.* 23: 132–134, 1999.

[40111] 13663.Tsai, T.–F.; Armstrong, D.; Beaudet, A. L.: Necdin–deficient micedo not show lethality or the obesity and infertility of Prader–Willis syndrome. (Letter) *Nature Genet.* 22: 15–16, 1999.

[40112] 13664.Watrin, F.; Roeckel, N.; Lacroix, L.; Mignon, C.; Mattei, M.–G.; Distèche, C.; Muscatelli, F.: The mouse necdin gene is expressed from the paternal allele only and lies in the 7C region of the mouse chromosome 7, a region of conserved synteny to the human Prader–Willi syndrome region. *Europ. J. Hum. Genet.* 5: 324–332, 1997.

[40113] 13665.Delmas, V.; Stokes, D. G.; Perry, R. P.: A mammalian DNA–binding protein that contains a chromodomain and an SNF2/SWI2–like helicase domain. *Proc. Nat. Acad. Sci.* 90: 2414–2418, 1993.

[40114] 13666.Woodage, T.; Basrai, M. A.; Baxevanis, A. D.; Hieter, P.; Collins, F. S.: Characterization of the CHD family of proteins. *Proc. Nat. Acad. Sci.* 94: 11472–11477, 1997.

[40115] 13667.Ge, Q.; Nilasena, D. S.; O'Brien, C. A.; Frank, M. B.; Targoff, I. N.: Molecular analysis of a major antigenic region of the 240–kD protein of Mi–2 autoantigen. *J. Clin. Invest.* 96: 1730–1737, 1995.

[40116] 13668.Seelig, H. P.; Renz, M.; Targoff, I. N.; Ge, Q.; Frank,

M. B.:Two forms of the major antigenic protein of the dermatomyositis-specificMi-2 autoantigen. (Letter) Arthritis Rheum. 39: 1769–1771, 1996.

[40117] 13669.Hu, G.; Chung, Y.-L.; Glover, T.; Valentine, V.;

Look, A T.; Fearon,E. R.: Characterization of human homologs of the Drosophila sevenin absentia (sina) gene.

Genomics 46: 103–111, 1997.

[40118] 13670.Liu, J.; Stevens, J.; Rote, C. A.; Yost, H. J.; Hu, Y.;

Neufeld,K. L.; White, R. L.; Matsunami, N.: Siah-1 mediates a novel beta-catenin degradation pathway linking p53 to the adenomatous polyposis coliprotein. Molec. Cell 7:

927–936, 2001.

[40119] 13671.Nemani, M.; Linares-Cruz, G.; Bruzzoni-Giovanelli,

H.; Roperch,J.-P.; Tuynder, M.; Bougueleret, L.; Cherif, D.; Medhioub, M.; Pasturaud,P.; Alvaro, V.; der Sarkissan, H.;

Cazes, L.; Le Paslier, D.; Le Gall,I.; Israeli, D.; Dausset, J.;

Sigaux, F.; Chumakov, I.; Oren, M.;Calvo, F.; Amson, R. B.;

Cohen, D.; Telerman, A.: Activation of thehuman homologue of the Drosophila SINA gene in apoptosis and tumorsuppression. Proc. Nat. Acad. Sci. 93: 9039–9042,

1996.

[40120] 13672.Ozkaynak, E.; Schnegelsberg, P. N. J.; Jin, D. F.;

Clifford, G.M.; Warren, F. D.; Drier, E. A.; Oppermann, H.:

Osteogenic protein-2: a new member of the transforming growth factor-beta superfamily expressed in early embryogenesis. *J. Biol. Chem.* 267: 25220-25227, 1992.

[40121] 13673. Jang, W.; Weber, J. S.; Harkins, E. B.; Meisler, M. H.: Localization of the rhotekin gene RTKN on the physical maps of mouse chromosome 6 and human chromosome 2p13 and exclusion as a candidate for mnd2 and LGMD2B. *Genomics* 40: 506-507, 1997.

[40122] 13674. Reid, T.; Furuyashiki, T.; Ishizaki, T.; Watanabe, G.; Watanabe, N.; Fujisawa, K.; Morii, N.; Madaule, P.; Narumiya, S.: Rhotekin, a new putative target for Rho bearing homology to a serine/threonine kinase, PKN, and rhophilin in the Rho-binding domain. *J. Biol. Chem.* 271: 13556-13560, 1996.

[40123] 13675. Janssen, U.; Davis, E. M.; Le Beau, M. M.; Stoffel, W.: Human mitochondrial enoyl-CoA hydratase gene (ECHS1): structural organization and assignment to chromosome 10q26.2-q26.3. *Genomics* 40: 470-475, 1997.

[40124] 13676. Kanazawa, M.; Ohtake, A.; Abe, H.; Yamamoto, S.; Satoh, Y.; Takayanagi, M.; Niimi, H.; Mori, M.; Hashimoto, T.: Molecular cloning and sequence analysis of the cDNA for human mitochondrial short-chain enoyl-CoA hydratase. *Enzyme Protein* 47: 9-13, 1993.

- [40125] 13677.Bingle, C. D.; Gowan, S.: Molecular cloning of the forkhead transcriptionfactor HNF-3-alpha from a human pulmonary adenocarcinoma cell line. *Biochim.Biophys. Acta* 1307: 17-20, 1996.
- [40126] 13678.Hannigan, G. E.; Bayani, J.; Weksberg, R.; Beatty, B.; Pandita,A.; Dedhar, S.; Squire, J.: Mapping of the gene encoding the integrin-linkedkinase, ILK, to human chromosome 11p15.5-p15.4. *Genomics* 42: 177-179,1997.
- [40127] 13679.Hannigan, G. E.; Leung-Hagesteijn, C.; Fitz-Gibbon, L.; Coppolino,M. G.; Radeva, G.; Filmus, J.; Bell, J. C.; Dedhar, S.: Regulationof cell adhesion and anchorage-dependent growth by a new beta-1-integrin-linkedprotein kinase. *Nature* 379: 91-96, 1996.
- [40128] 13680.Omeis, I. A.; Hsu, Y.-C.; Perin, M. S.: Mouse and human neuronalpentraxin 1 (NPXT1): conservation, genomic structure, and chromosomallocalization. *Genomics* 36: 543-545, 1996.
- [40129] 13681.Babic, A. M.; Kireeva, M. L.; Kolesnikova, T. V.; Lau, L. F.:CYR61, a product of a growth factor-inducible immediate early gene,promotes angiogenesis and tumor growth. *Proc. Nat. Acad. Sci.* 95:6355-6360, 1998.
- [40130] 13682.Jay, P.; Berge-Lefranc, J. L.; Marsollier, C.; Mejean,

C.; Taviaux, S.; Berta, P.: The human growth factor-inducible immediate early gene, CYR61, maps to chromosome 1p. *Oncogene* 14: 1753–1757, 1997.

[40131] 13683. Kireeva, M. L.; Lam, S. C.-T.; Lau, L. F.: Adhesion of human umbilical vein endothelial cells to the immediate-early gene product Cyr61 is mediated through integrin α (V) β (3). *J. Biol. Chem.* 273: 3090–3096, 1998.

[40132] 13684. Martinerie, C.; Viegas-Pequignot, E.; Nguyen, V. C.; Perbal, B.: Chromosomal mapping and expression of the human cyr61 gene in tumour cells from the nervous system. *J. Clin. Path.* 50: 310–316, 1997.

[40133] 13685. Sampath, D.; Zhu, Y.; Winneker, R. C.; Zhang, Z.: Aberrant expression of Cyr61, a member of the CCN (CTGF/Cyr61/Cef10/NOVH) family, and dysregulation by 17- β -estradiol and basic fibroblast growth factor in human uterine leiomyomas. *J. Clin. Endocr. Metab.* 86: 1707–1715, 2001.

[40134] 13686. Fujino, T.; Kang, M.-J.; Suzuki, H.; Iijima, H.; Yamamoto, T.: Molecular characterization and expression of rat acyl-CoA synthetase 3. *J. Biol. Chem.* 271: 16748–16752, 1996.

[40135] 13687. Minekura, H.; Fujino, T.; Kang, M.-J.; Fujita, T.; Endo, Y.; Yamamoto, T. T.: Human acyl-coenzyme A syn-

thetase 3 cDNA and localization of its gene (ACS3) to chromosome band 2q34–q35. *Genomics* 42: 180–181, 1997.

[40136] 13688. Kubota, N.; Terauchi, Y.; Miki, H.; Tamemoto, H.; Yamauchi, T.; Komeda, K.; Satoh, S.; Nakano, R.; Ishii, C.; Sugiyama, T.; Eto, K.; Tsubamoto, Y.; and 17 others: PPAR- γ mediates high-fat diet-induced adipocyte hypertrophy and insulin resistance. *Molec. Cell* 4: 597–609, 1999.

[40137] 13689. Lapsys, N. M.; Kriketos, A. D.; Lim-Fraser, M.; Poynten, A. M.; Lowy, A.; Furler, S. M.; Chisholm, D. J.; Cooney, G. J.: Expression of genes involved in lipid metabolism correlate with peroxisome proliferator-activated receptor γ expression in human skeletal muscle. *J. Clin. Endocr. Metab.* 85: 4293–4297, 2000.

[40138] 13690. Lehmann, J. M.; Moore, L. B.; Smith-Oliver, T. A.; Wilkison, W. O.; Willson, T. M.; Kliewer, S. A.: An antidiabetic thiazolidinedione is a high affinity ligand for peroxisome proliferator-activated receptor γ (PPAR γ). *J. Biol. Chem.* 270: 12953–12956, 1995.

[40139] 13691. Lowell, B. B.: PPAR- γ : an essential regulator of adipogenesis and modulator of fat cell function. *Cell* 99: 239–242, 1999.

[40140] 13692. Martin, G.; Schoonjans, K.; Staels, B.; Auwerx, J.:

PPAR- γ activators improve glucose homeostasis by stimulating fatty acid uptake in the adipocytes.

Atherosclerosis 137: S75-S80, 1998.

[40141] 13693. Meirhaeghe, A.; Fajas, L.; Helbecque, N.; Cotel, D.; Lebel, P.; Dallongeville, J.; Deeb, S.; Auwerx, J.; Amouyel, P.: A genetic polymorphism of the peroxisome proliferator-activated receptor gamma gene influences plasma leptin levels in obese humans. Hum. Molec. Genet. 7: 435-440, 1998.

[40142] 13694. Miles, P. D. G.; Barak, Y.; He, W.; Evans, R. M.; Olefsky, J. M.: Improved insulin-sensitivity in mice heterozygous for PPAR- γ deficiency. J. Clin. Invest. 105: 287-292, 2000.

[40143] 13695. Mueller, E.; Sarraf, P.; Tontonoz, P.; Evans, R. M.; Martin, K. J.; Zhang, M.; Fletcher, C.; Singer, S.; Spiegelman, B. M.: Terminal differentiation of human breast cancer through PPAR- γ . Molec. Cell. 1: 465-470, 1998.

[40144] 13696. Mueller, E.; Smith, M.; Sarraf, P.; Kroll, T.; Aiyer, A.; Kaufman, D. S.; Oh, W.; Demetri, G.; Figg, W. D.; Zhou, X.-P.; Eng, C.; Spiegelman, B. M.; Kantoff, P. W.: Effects of ligand activation of peroxisome proliferator-activated receptor gamma in human prostate cancer. Proc. Nat. Acad. Sci. 97: 10990-10995, 2000.

- [40145] 13697. Nagy, L.; Tontonoz, P.; Alvarez, J. G. A.; Chen, H.; Evans, R.M.: Oxidized LDL regulates macrophage gene expression through ligand activation of PPAR- γ . *Cell* 93: 229–240, 1998.
- [40146] 13698. Oh, E. Y.; Min, K. M.; Chung, J. H.; Min, Y.-K.; Lee, M.-S.; Kim, K.-W.; Lee, M.-K.: Significance of pro12ala mutation in peroxisome proliferator-activated receptor- γ 2 in Korean diabetic and obese subjects. *J. Clin. Endocr. Metab.* 85: 1801–1804, 2000.
- [40147] 13699. Pritchard, J. K.; Rosenberg, N. A.: Use of unlinked genetic markers to detect population stratification in association studies. *Am. J. Hum. Genet.* 65: 220–228, 1999.
- [40148] 13700. Ristow, M.; Muller-Wieland, D.; Pfeiffer, A.; Krone, W.; Kahn, C. R.: Obesity associated with a mutation in a genetic regulator of adipocyte differentiation. *New Eng. J. Med.* 339: 953–959, 1998.
- [40149] 13701. Rocchi, S.; Picard, F.; Vamecq, J.; Gelman, L.; Potier, N.; Zeyer, D.; Dubuquoy, L.; Bac, P.; Champy, M.-F.; Plunket, K. D.; Leesnitzer, L. M.; Blanchard, S. G.; Desreumaux, P.; Moras, D.; Renaud, J.-P.; Auwerx, J.: A unique PPAR- γ ligand with potent insulin-sensitizing yet weak adipogenic activity. *Molec. Cell* 8: 737–747, 2001.
- [40150] 13702. Rosen, E. D.; Sarraf, P.; Troy, A. E.; Bradwin, G.;

Moore, K.; Milstone, D. S.; Spiegelman, B. M.; Mortensen, R. M.: PPAR- γ is required for the differentiation of adipose tissue in vivo and in vitro. *Molec. Cell* 4: 611–617, 1999.

[40151] 13703. Ricote, M.; Huang, J.; Fajas, L.; Li, A.; Welch, J.; Najib, J.; Witztum, J. L.; Auwerx, J.; Palinski, W.; Glass, C. K.: Expression of the peroxisome proliferator-activated receptor γ (PPAR- γ) in human atherosclerosis and regulation in macrophages by colony stimulating factors and oxidized low density lipoprotein. *Proc. Nat. Acad. Sci.* 95:7614–7619, 1998.

[40152] 13704. Savage, D. B.; Agostini, M.; Barroso, I.; Gurnell, M.; Luan, J.; Meirhaeghe, A.; Harding, A.-H.; Ihrke, G.; Rajanayagam, O.; Soos, M. A.; George, S.; Berger, D.; and 9 others: Digenic inheritance of severe insulin resistance in a human pedigree. *Nature Genet.* 31:379–384, 2002. Note: Erratum: *Nature Genet.* 32: 211 only, 2002.

[40153] 13705. Smith, W. M.; Zhou, X.-P.; Kurose, K.; Gao, X.; Latif, F.; Kroll, T.; Sugano, K.; Cannistra, S. A.; Clinton, S. K.; Maher, E. R.; Prior, T. W.; Eng, C.: Opposite association of two PPAR γ variants with cancer: overrepresentation of H449H in endometrial carcinoma cases and underrepresentation of P12A in renal cell carcinoma cases. *Hum. Genet.*

109: 146–151,2001.

- [40154] 13706.Tarrade, A.; Schoonjans, K.; Pavan, L.; Auwerx, J.; Rochette-Egly,C.; Evain-Brion, D.; Fournier, T.: PPAR-gamma/RXR-alpha heterodimerscontrol human trophoblast invasion. *J. Clin. Endocr. Metab.* 86:5017–5024, 2001.
- [40155] 13707.Tontonoz, P.; Hu, E.; Devine, J.; Beale, E. G.; Spiegelman, B.M.: PPAR gamma 2 regulates adipose expression of the phosphoenolpyruvatecarboxykinase gene. *Molec. Cell. Biol.* 15: 351–357, 1995.
- [40156] 13708.Tontonoz, P.; Hu, E.; Graves, R. A.; Budavari, A. I.; Spiegelman,B. M.: mPPAR gamma 2: tissue-specific regulator of an adipocyte enhancer. *GenesDev.* 8: 1224–1234, 1994.
- [40157] 13709.Tontonoz, P.; Nagy, L.; Alvarez, J. G. A.; Thomazy, V. A.; Evans,R. M.: PPAR-gamma promotes monocyte/macrophage differentiation anduptake of oxidized LDL. *Cell* 93: 241–252, 1998.
- [40158] 13710.Wang, M.: Isolation and characterization of a human gene encodinga single-strand-specific endoribonuclease. Ph.D. Thesis: StanfordUniv. , 1995.
- [40159] 13711.Wang, M.; Cohen, S. N.: ard-1: a human gene that reverses theeffects of temperature-sensitive and deletion

mutations in the Escherichiacoli rne gene and encodes an activity producing RNase E-like cleavages. Proc.Nat. Acad. Sci. 91: 10591–10595, 1994.

- [40160] 13712.Beck, S.; Hanson, I.; Kelly, A.; Pappin, D. J.; Trowsdale, J.:A homologue of the Drosophila female sterile homeotic (fsh) gene inthe class II region of the human MHC. DNA Sequence 2: 203–210, 1992.
- [40161] 13713.Denis, G. V.; Green, M. R.: A novel, mitogen-activated nuclearkinase is related to a Drosophila developmental regulator. GenesDev. 10: 261–271, 1996.
- [40162] 13714.Okamoto, N.; Ando, A.; Kawai, J.; Yoshiwara, T.; Tsuji, K.; Inoko,H.: Orientation of HLA–DNA gene and identification of a CpG island–associatedgene adjacent to DNA in human major histocompatibility complex classII region. Hum. Immun. 32: 221–228, 1991.
- [40163] 13715.Thorpe, K. L.; Abdulla, S.; Kaufman, J.; Trowsdale, J.; Beck, S.: Phylogeny and structure of the RING3 gene. Immunogenetics 44:391–396, 1996.
- [40164] 13716.Cushman, L. J.; Camper, S. A.: Molecular basis of pituitary dysfunctionin mouse and human. Mammalian Genome 12: 485–494, 2001.
- [40165] 13717.Kozak, M.: Interpreting cDNA sequences: some insights from studieson translation. Mammalian Genome 7:

563–574, 1996.

- [40166] 13718. Pellegrini–Bouiller, I.; Manrique, C.; Gunz, G.; Grino, M.; Zamora, A. J.; Figarella–Branger, D.; Grisoli, F.; Jaquet, P.; Enjalbert, A.: Expression of the members of the Ptx family of transcription factors in human pituitary adenomas. *J. Clin. Endocr. Metab.* 84:2212–2220, 1999.
- [40167] 13719. Brinke, A.; Green, P. M.; Giannelli, F.: Characterization of the gene (VBP1) and transcript for the von Hippel–Lindau binding protein and isolation of the highly conserved murine homologue. *Genomics* 45:105–112, 1997.
- [40168] 13720. Brinke, A.; Tagliavacca, L.; Naylor, J.; Green, P.; Giannelli, F.: Two chimaeric transcription units result from an inversion breaking intron 1 of the factor VIII gene and a region reportedly affected by reciprocal translocations in T–cell leukaemia. *Hum. Molec. Genet.* 5: 1945–1951, 1996.
- [40169] 13721. Clifford, S. C.; Walsh, S.; Hewson, K.; Green, E. K.; Brinke, A.; Green, P. M.; Giannelli, F.; Eng, C.; Maher, E. R.: Genomic organization and chromosomal localization of the human CUL2 gene and the role of von Hippel–Lindau tumor suppressor–binding protein (CUL2 and VBP1) mutation and loss in renal–cell carcinoma development. *Genes Chromosomes Cancer* 26: 20–28, 1999.

- [40170] 13722.Hemberger, M.; Himmelbauer, H.; Neumann, H. P. H.; Plate, K. H.;Schwarzkopf, G.; Fundele, R.: Expression of the von Hippel–Lindau–bindingprotein–1 (Vbp1) in fetal and adult mouse tissues. Hum. Molec. Genet. 8:229–236, 1999.
- [40171] 13723.Tsuchiya, H.; Iseda, T.; Hino, O.: Identification of a novel protein(VBP–1) binding to the von Hippel–Lindau (VHL) tumor suppressor geneproduct. Cancer Res. 56: 2881–2885, 1996.
- [40172] 13724.Corbaz, A.; ten Hove, T.; Herren, S.; Graber, P.; Schwartzburd,B.; Belzer, I.; Harrison, J.; Plitz, T.; Kosco–Vilbois, M. H.; Kim,S.–H.; Dinarello, C. A.; Novick, D.; van Deventer, S.; Chvatchko,Y.: IL–18–binding protein expression by endothelial cells and macrophagesis up–regulated during active Crohn's disease. J. Immun. 168: 3608–3616,2002.
- [40173] 13725.Turner, N.; Mason, P. J.; Brown, R.; Fox, M.; Povey, S.; Rees,A.; Pusey, C. D.: Molecular cloning of the human Goodpasture antigendemonstrates it to be the alpha–3 chain of type IV collagen. J. Clin.Invest. 89: 592–601, 1992.
- [40174] 13726.Hoffmann, M. M.; Jacob, S.; Luft, D.; Schmulling, R.–M.; Rett,K.; Marz, W.; Haring, H.–U.; Matthaei, S.: Type I

hyperlipoproteinemia due to a novel loss of function mutation of lipoprotein lipase, cys239→trp, associated with recurrent severe pancreatitis. *J. Clin. Endocr. Metab.* 85:4795–4798, 2000.

[40175] 13727. Holt, L. E., Jr.; Aylward, F. X.; Timbers, H. G.: Idiopathic familial lipemia. *Bull. Johns Hopkins Hosp.* 64: 279–314, 1939.

[40176] 13728. Ishimura-Oka, K.; Faustinella, F.; Kihara, S.; Smith, L. C.; Oka, K.; Chan, L.: A missense mutation (trp86→to→arg) in exon 3 of the lipoprotein lipase gene: a cause of familial chylomicronemia. *Am. J. Hum. Genet.* 50: 1275–1280, 1992.

[40177] 13729. Kastelein, J. J. P.; Groenemeyer, B. E.; Hallman, D. M.; Henderson, H.; Reymer, P. W. A.; Gagne, S. E.; Jansen, H.; Seidell, J. C.; Kromhout, D.; Jukema, J. W.; Bruschke, A. V. G.; Boerwinkle, E.; Hayden, M. R.; The Regress Study Group: The asn9 variant of lipoprotein lipase is associated with the –93G promoter mutation and an increased risk of coronary artery disease. *Clin. Genet.* 53: 27–33, 1998.

[40178] 13730. Kirchgessner, T. G.; Chuat, J.-C.; Heinzmann, C.; Etienne, J.; Guilhot, S.; Svenson, K.; Ameis, D.; Pilon, C.; d'Auriol, L.; Andalibi, A.; Schotz, M. C.; Galibert, F.; Lusi, A. J.: Organization of the human lipoprotein lipase gene

and evolution of the lipase gene family. *Proc.Nat. Acad. Sci.* 86: 9647–9651, 1989.

- [40179] 13731.Kirchgessner, T. G.; Svenson, K. L.; Lusis, A. J.; Schotz, M.C.: The sequence of cDNA encoding lipoprotein lipase: a member of a lipase gene family. *J. Biol. Chem.* 262: 8463–8466, 1987.
- [40180] 13732.Kobayashi, J.; Nishida, T.; Ameis, D.; Stahnke, G.; Schotz, M.C.; Hashimoto, H.; Fukamachi, I.; Shirai, K.; Saito, Y.; Yoshida, S.: A heterozygous mutation (the codon for ser477 to a stop codon) in lipoprotein lipase contributes to a defect in lipid interface recognition in a case with type I hyperlipidemia. *Biochem. Biophys. Res. Commun.* 182:70–77, 1992.
- [40181] 13733.Kobayashi, J.; Sasaki, N.; Tashiro, J.; Inadera, H.; Saito, Y.; Yoshida, S.: A missense mutation (ala334–to–thr) in exon 7 of the lipoprotein lipase gene in a case with type I hyperlipidemia. *Biochem. Biophys. Res. Commun.* 191: 1046–1054, 1993.
- [40182] 13734.Langlois, S.; Deeb, S.; Brunzell, J.; Kastelein, J. J.; Hayden, M. R.: A unique insertion accounts for a significant proportion of the mutations in the lipoprotein lipase (LPL) gene. (Abstract) *Am.J. Hum. Genet.* 43: A191, 1988.
- [40183] 13735.Langlois, S.; Deeb, S.; Brunzell, J. D.; Kastelein, J. J.;

Hayden, M. R.: A major insertion accounts for a significant proportion of mutations underlying human lipoprotein lipase deficiency. *Proc. Nat. Acad. Sci.* 86: 948–952, 1989.

- [40184] 13736. Levak–Frank, S.; Radner, H.; Walsh, A.; Stollberger, R.; Knipping, G.; Hoefler, G.; Sattler, W.; Weinstock, P. H.; Breslow, J. L.; Zechner, R.: Muscle–specific overexpression of lipoprotein lipase causes a severe myopathy characterized by proliferation of mitochondria and peroxisomes in transgenic mice. *J. Clin. Invest.* 96: 976–986, 1995.
- [40185] 13737. Li, S.; Oka, K.; Galton, D.; Stocks, J.: Pvu–II RFLP at the human lipoprotein lipase (LPL) gene locus. *Nucleic Acids Res.* 16: 2358, 1988.
- [40186] 13738. Lo, J. Y.; Smith, L. C.; Chan, L.: Lipoprotein lipase: role of intramolecular disulfide bonds in enzyme catalysis. *Biochem. Biophys. Res. Commun.* 206: 266–271, 1995.
- [40187] 13739. Ma, Y.; Henderson, H. E.; Julien, P.; Roederer, G.; Brunzell, J.; Hayden, M. R.: A missense mutation (pro–to–leu207) in the human lipoprotein lipase gene is the major cause of type I hyperlipoproteinemia in French Canadians. (Series) *Miami Short Reports. Advances in Gene Technology: The Molecular Biology of Human Genetic Disease.* New York: IRL Press (pub.) 1: 1991. Pp. 34 only.
- [40188] 13740. Ma, Y.; Henderson, H. E.; Ven Murthy, M. R.; Roed–

erer, G.; Monsalve, M. V.; Clarke, L. A.; Normand, T.; Julien, P.; Gagne, C.; Lambert, M.; Davignon, J.; Lupien, P. J.; Brunzell, J.; Hayden, M. R.: A mutation in the human lipoprotein lipase gene as the most common cause of familial chylomicronemia in French Canadians. *New Eng. J. Med.* 324: 1761–1766, 1991.

[40189] 13741. Ma, Y.; Liu, M.-S.; Chitayat, D.; Bruin, T.; Beisiegel, U.; Benlian, P.; Foubert, L.; De Gennes, J. L.; Funke, H.; Forsythe, I.; Blachman, S.; Papanikolaou, M.; Erkelens, D. W.; Kastelein, J.; Brunzell, J. D.; Hayden, M. R.: Recurrent missense mutations at the first and second base of codon arg243 in human lipoprotein lipase in patients of different ancestries. *Hum. Mutat.* 3: 52–58, 1994.

[40190] 13742. Ma, Y.; Liu, M.-S.; Ginzinger, D.; Frohlich, J.; Brunzell, J. D.; Hayden, M. R.: Gene–environment interaction in the conversion of a mild–to–severe phenotype in a patient homozygous for a ser172–to–cys mutation in the lipoprotein lipase gene. *J. Clin. Invest.* 91: 1953–1958, 1993.

[40191] 13743. Ma, Y.; Wilson, B. I.; Bijvoet, S.; Henderson, H. E.; Cramb, E.; Roederer, G.; Ven Murthy, M. R.; Julien, P.; Bakker, H. D.; Kastelein, J. J. P.; Brunzell, J. D.; Hayden, M. R.: A missense mutation (asp250–to–asn) in exon 6 of the human lipoprotein lipase gene causes chylomicronemia in

patients of different ancestries. *Genomics* 13: 649–653, 1992.

- [40192] 13744. Mattei, M. G.; Etienne, J.; Chuat, J. C.; Nguyen, V. C.; Brault, D.; Bernheim, A.; Galibert, F.: Assignment of the human lipoprotein lipase (LPL) gene to chromosome band 8p22. *Cytogenet. Cell Genet.* 63:45–46, 1993.
- [40193] 13745. Miesenbock, G.; Holzl, B.; Foger, B.; Brandstatter, E.; Paulweber, B.; Sandhofer, F.; Patsch, J. R.: Heterozygous lipoprotein lipase deficiency due to a missense mutation as the cause of impaired triglyceride tolerance with multiple lipoprotein abnormalities. *J. Clin. Invest.* 91:448–455, 1993.
- [40194] 13746. Nevin, D. N.; Brunzell, J. D.; Deeb, S. S.: The LPL gene in individuals with familial combined hyperlipidemia and decreased LPL activity. *Arteriosclerosis Thromb.* 14: 869–873, 1994.
- [40195] 13747. Nevin, N. C.; Slack, J.: Hyperlipidaemic xanthomatosis II: mode of inheritance in 55 families with essential hyperlipidaemia and xanthomatosis. *J. Med. Genet.* 5: 9–28, 1968.
- [40196] 13748. Nickerson, D. A.; Taylor, S. L.; Weiss, K. M.; Clark, A. G.; Hutchinson, R. G.; Stengard, J.; Salomaa, V.; Vartiainen, E.; Boerwinkle, E.; Sing, C. F.: DNA sequence diver-

sity in a 9.7-kb region of the human lipoprotein lipase gene. *Nature Genet.* 19: 233–240, 1998.

[40197] 13749. Lahn, B. T.; Page, D. C.: Retroposition of autosomal mRNA yielded testis-specific gene family on human Y chromosome. *Nature Genet.* 21:429–433, 1999.

[40198] 13750. Yen, P. H.: A long-range restriction map of deletion interval 6 of the human Y chromosome: a region frequently deleted in azoospermic males. *Genomics* 54: 5–12, 1998.

[40199] 13751. MacLean, H. E.; Chu, S.; Warne, G. L.; Zajac, J. D.: Related individuals with different androgen receptor gene deletions. *J. Clin. Invest.* 91: 1123–1128, 1993.

[40200] 13752. Madgar, I.; Green, L.; Kent-First, M.; Weissenberg, R.; Gershoni-Baruch, R.; Goldman, B.; Friedman, E.: Genotyping of Israeli infertile men with idiopathic oligozoospermia. *Clin. Genet.* 62: 203–207, 2002.

[40201] 13753. Marcelli, M.; Tilley, W. D.; Wilson, C. M.; Griffin, J. E.; Wilson, J. D.; McPhaul, M. J.: Definition of the human androgen receptor gene structure permits the identification of mutations that cause androgen resistance: premature termination of the receptor protein at amino acid residue 588 causes complete androgen resistance. *Molec. Endocr.* 4: 1105–1116, 1990.

[40202] 13754. Marcelli, M.; Tilley, W. D.; Wilson, C. M.; Wilson, J.

D.; Griffin, J. E.; McPhaul, M. J.: A single nucleotide substitution introduces a premature termination codon into the androgen receptor gene of a patient with receptor-negative androgen resistance. *J. Clin. Invest.* 85:1522–1528, 1990.

[40203] 13755. Marcelli, M.; Tilley, W. D.; Zoppi, S.; Griffin, J. E.; Wilson, J. D.; McPhaul, M. J.: Androgen resistance associated with a mutation of the androgen receptor at amino acid 772 (arg-to-cys) results from a combination of decreased messenger ribonucleic acid levels and impairment of receptor function. *J. Clin. Endocr.* 73: 318–325, 1991.

[40204] 13756. McCampbell, A.; Taylor, J. P.; Taye, A. A.; Robitschek, J.; Li, M.; Walcott, J.; Merry, D.; Chai, Y.; Paulson, H.; Sobue, G.; Fischbeck, K. H.: CREB-binding protein sequestration by expanded polyglutamine. *Hum. Molec. Genet.* 9: 2197–2202, 2000.

[40205] 13757. McPhaul, M. J.; Griffin, J. E.: Male pseudohermaphroditism caused by mutations of the human androgen receptor. *J. Clin. Endocr. Metab.* 84:3435–3441, 1999.

[40206] 13758. McPhaul, M. J.; Marcelli, M.; Tilley, W. D.; Griffin, J. E.; Isidro-Gutierrez, R. F.; Wilson, J. D.: Molecular basis of

androgen resistance in a family with a qualitative abnormality of the androgen receptor and responsive to high-dose androgen therapy. J. Clin. Invest. 87: 1413–1421, 1991.

[40207] 13759. McPhaul, M. J.; Marcelli, M.; Zoppi, S.; Griffin, J. E.; Wilson, J. D.: Genetic basis of endocrine disease 4: the spectrum of mutations in the androgen receptor gene that causes androgen resistance. J. Clin. Endocr. Metab. 76: 17–23, 1993.

[40208] 13760. McPhaul, M. J.; Marcelli, M.; Zoppi, S.; Wilson, C. M.; Griffin, J. E.; Wilson, J. D.: Mutations in the ligand-binding domain of the androgen receptor gene cluster in two regions of the gene. J. Clin. Invest. 90: 2097–2101, 1992.

[40209] 13761. McPhaul, M. J.; Schweikert, H.-U.; Allman, D. R.: Assessment of androgen receptor function in genital skin fibroblasts using a recombinant adenovirus to deliver an androgen-responsive reporter gene. J. Clin. Endocr. Metab. 82: 1944–1948, 1997.

[40210] 13762. Mifsud, A.; Ramirez, S.; Yong, E. L.: Androgen receptor gene CAG trinucleotide repeats in anovulatory infertility and polycystic ovaries. J. Clin. Endocr. Metab. 85: 3484–3488, 2000.

- [40211] 13763.Migeon, B. R.; Brown, T. R.; Axelman, J.; Migeon, C. J.: Studies of the locus for androgen receptor: localization on the human X and evidence for homology with the Tfm locus in the mouse. *Proc. Nat. Acad. Sci.* 78: 6339–6343, 1981.
- [40212] 13764.Mongan, N. P.; Jaaskelainen, J.; Green, K.; Schwabe, J. W.; Shimura, N.; Dattani, M.; Hughes, I. A.: Two de novo mutations in the AR gene cause the complete androgen insensitivity syndrome in a pair of monozygotic twins. *J. Clin. Endocr. Metab.* 87: 1057–1061, 2002.
- [40213] 13765.Mononen, N.; Syrjakoski, K.; Matikainen, M.; Tamela, T. L. J.; Schleutker, J.; Kallioniemi, O.-P.; Trapman, J.; Koivisto, P. A.: Two percent of Finnish prostate cancer patients have a germ-line mutation in the hormone-binding domain of the androgen receptor gene. *Cancer Res.* 60: 6479–6481, 2000.
- [40214] 13766.Murono, K.; Mendonca, B. B.; Arnhold, I. J. P.; Rigon, A. C. M.; Migeon, C. J.; Brown, T. R.: Human androgen insensitivity due to point mutations encoding amino acid substitutions in the androgen receptor steroid-binding domain. *Hum. Mutat.* 6: 152–162, 1995.
- [40215] 13767.Nakao, R.; Haji, M.; Yanase, T.; Ogo, A.; Takayanagi, R.; Katsube, T.; Fukumaki, Y.; Nawata, H.: A

single amino acid substitution (met786-to-val)in the steroid-binding domain of human androgen receptor leads to complete androgen insensitivity syndrome. J. Clin. Endocr. Metab. 74:1152–1157, 1992.

[40216] 13768. Newmark, J. R.; Hardy, D. O.; Tonb, D. C.; Carter, B. S.; Epstein, J. I.; Isaacs, W. B.; Brown, T. R.; Barrack, E. R.: Androgen receptor gene mutations in human prostate cancer. Proc. Nat. Acad. Sci. 89:6319–6323, 1992.

[40217] 13769. Nguyen, D.; Steinberg, S. V.; Rouault, E.; Chagnon, S.; Gottlieb, B.; Pinsky, L.; Trifiro, M.; Mader, S.: A G577R mutation in the human AR P box results in selective decreases in DNA binding and in partial androgen insensitivity syndrome. Molec. Endocr. 15: 1790–1802, 2001.

[40218] 13770. Ohno, S.: The Y-linked antigen locus and the X-linked Tfm locus as major regulatory genes of the mammalian sex determining mechanism. J. Steroid Biochem. 8: 585–592, 1977. 100. Ohno, S.: Simplicity of mammalian regulatory systems inferred by single gene determination of sex phenotypes. Nature 234: 134–137, 1971. 101. Ong, Y. C.; Wong, H. B.; Adaikan, G.; Yong, E. L.: Directed pharmacological therapy of ambiguous genitalia due to an androgen receptor gene mutation. (Letter) Lancet 354: 1444–1445, 1999. 102. Patterson, M. N.; Hughes, I. A.;

Gottlieb, B.; Pinsky, L.: The androgen receptor gene mutations database. *Nucleic Acids Res.* 22:3560–3562, 1994.103. Pinsky, L.; Kaufman, M.; Killinger, D. W.; Burko, B.; Shatz, D.; Volpe, R.: Human minimal androgen insensitivity with normal dihydrotestosterone-binding capacity in cultured genital skin fibroblasts: evidence for an androgen-selective qualitative abnormality of the receptor. *Am. J. Hum. Genet.* 36:965–978, 1984.104. Pinsky, L.; Kaufman, M.; Levitsky, L. L.: Partial androgen resistance due to a distinctive qualitative defect of the androgen receptor. *Am. J. Med. Genet.* 27: 459–466, 1987.105. Pinsky, L.; Kaufman, M.; Summitt, R. L.: Congenital androgen insensitivity due to a qualitatively abnormal androgen receptor. *Am. J. Med. Genet.* 10: 91–99, 1981.106. Pinsky, L.; Trifiro, M.; Sebbaghian, N.; Kaufman, M.; Chang, C.; Trapman, J.; Brinkmann, A. O.; Kuiper, G. G. J. M.; Ris, C. J.; Brown, C. J.; Willard, H. F.; Sergovich, F.: A deletional alteration of the androgen receptor (AR) gene in a sporadic patient with complete androgen insensitivity (CAI) who is mentally retarded. (Abstract) *Am. J. Hum. Genet.* 45 (suppl.): A212, 1989.107. Prior, L.; Bordet, S.; Trifiro, M. A.; Mhatre, A.; Kaufman, M.; Pinsky, L.; Wrogeman, K.; Belsham, D. D.; Pereira, F.; Greenberg, C.; Trapman, J.; Brinkman, A. O.;

Chang, C.; Liao, S.: Replacement of arginine 773 by cysteine or histidine in the human androgen receptor causes complete androgen insensitivity with different receptor phenotypes. *Am. J. Hum. Genet.* 51: 143–155, 1992.108.

Quigley, C. A.; Friedman, K. J.; Johnson, A.; Lafreniere, R. G.; Silverman, L. M.; Lubahn, D. B.; Brown, T. R.; Wilson, E. M.; Willard, H. F.; French, F. S.: Complete deletion of the androgen receptor gene: definition of the null phenotype of the androgen insensitivity syndrome and determination of carrier status. *J. Clin. Endocr. Metab.* 74:927–933, 1992.109.

Ris-Stalpers, C.; Kuiper, G. G. J. M.; Faber, P. W.; Schweikert, H. U.; van Rooij, H. C. J.; Zegers, N. D.; Hodgins, M. B.; Degenhart, H. J.; Trapman, J.; Brinkmann, A. O.: Aberrant splicing of androgen receptor mRNA results in synthesis of a nonfunctional receptor protein in a patient with androgen insensitivity. *Proc. Nat. Acad. Sci.* 87:7866–7870, 1990.110.

Rodien, P.; Mebarki, F.; Mowszowicz, I.; Chaussain, J.-L.; Young, J.; Morel, Y.; Schaison, G.: Different phenotypes in a family with androgen insensitivity caused by the same M780I point mutation in the androgen receptor gene. *J. Clin. Endocr. Metab.* 81: 2994–2998, 1996.111.

Sai, T.; Seino, S.; Chang, C.; Trifiro, M.; Pinsky, L.; Mhatre, A.; Kaufman, M.; Lambert, B.; Trap-

man, J.; Brinkmann, A. O.; Rosenfield, R. L.; Liao, S.: An exonic point mutation of the androgen receptor gene in a family with complete androgen insensitivity. *Am. J. Hum. Genet.* 46: 1095–1100, 1990.112. Sammarco, I.; Grimaldi, P.; Rossi, P.; Cappa, M.; Moretti, C.; Frajese, G.; Geremia, R.: Novel point mutation in the splice donor site of exon–intron junction 6 of the androgen receptor gene in a patient with partial androgen insensitivity syndrome. *J. Clin. Endocr. Metab.* 85: 3256–3261, 2000.113. Schoenberg, M. P.; Hakimi, J. M.; Wang, S.; Bova, G. S.; Epstein, J. I.; Fischback, K. H.; Isaacs, W. B.; Walsh, P. C.; Barrack, E. R.: Microsatellite mutation (CAG (24–to–18)) in the androgen receptor gene in human prostate cancer *Biochem. Biophys. Res. Commun.* 198:74–80, 1994.114. Shang, Y.; Myers, M.; Brown, M.: Formation of the androgen receptor transcription complex. *Molec. Cell* 9: 601–610, 2002.115. Simeoni, S.; Mancini, M. A.; Stenoien, D. L.; Marcelli, M.; Weigel, N. L.; Zanisi, M.; Martini, L.; Poletti, A.: Motoneuronal cell death is not correlated with aggregate formation of androgen receptors containing an elongated polyglutamine tract. *Hum. Molec. Genet.* 9: 133–144, 2000.116. Sullivan, D. A.; Sullivan, B. D.; Ullman, M. D.; Rocha, E. M.; Krenzer, K. L.; Cermak, J. M.; Toda, I.;

Doane, M. G.; Evans, J. E.; Wickham, L. A.: Androgen influence on the meibomian gland. *Invest. Ophthalm. Vis. Sci.* 41: 3732–3742, 2000.117. Sultan, C.; Lumbroso, S.; Poujol, N.; Belon, C.; Boudon, C.; Lobaccaro, J.–M.: Mutations of androgen receptor gene in androgen insensitivity syndromes. *J. Steroid Biochem. Molec. Biol.* 46: 519–530, 1993.118. Sutherland, R. W.; Wiener, J. S.; Hicks, J. P.; Marcelli, M.; Gonzales, E. T.; Roth, D. R.; Lamb, D. J.: Androgen receptor gene mutations are rarely associated with isolated penile hypospadias. *J. Urol.* 156: 828–831, 1996.119. Taplin, M.–E.; Bubley, G. J.; Shuster, T. D.; Frantz, M. E.; Spooner, A. E.; Ogata, G. K.; Keer, H. N.; Balk, S. P.: Mutation of the androgen–receptor gene in metastatic androgen–independent prostate cancer. *New Eng. J. Med.* 332: 1393–1398, 1995.120. Tilley, W. D.; Marcelli, M.; Wilson, J. D.; McPhaul, M. J.: Characterization and expression of a cDNA encoding the human androgen–receptor. *Proc. Nat. Acad. Sci.* 86: 327–331, 1989.121. Trifiro, M.; Prior, L.; Pinsky, L.; Kaufman, M.; Chang, C.; Trapman, J.; Brinkmann, A. O.; Kuiper, G. G. J. M.; Ris, C.: A single transition at an exonic CpG site apparently abolishes androgen receptor (AR)–binding activity in a family with complete androgen insensitivity (CAI). (Abstract)

Am.J. Hum. Genet. 45 (suppl.): A225, 1989.122. Trifiro, M.; Prior, R. L.; Sabbaghian, N.; Pinsky, L.; Kaufman, M.; Nylén, E. G.; Belsham, D. D.; Greenberg, C. R.; Wrogermann, K.: Amber mutation creates a diagnostic Mael site in the androgen receptorgene of a family with complete androgen insensitivity. Am. J. Med.Genet. 40: 493–499, 1991.123. Tut, T. G.; Ghadessy, F. J.; Trifiro, M. A.; Pinsky, L.; Yong, E. L.: Long polyglutamine tracts in the androgen receptor are associatedwith reduced trans-activation, impaired sperm production, and maleinfertility. J. Clin. Endocr. Metab. 82: 3777–3782, 1997.124. Visakorpi, T.; Hyytinen, E.; Koivisto, P.; Tanner, M.; Keinänen, R.; Palmberg, C.; Palotie, A.; Tammela, T.; Isola, J.; Kallioniemi, O.-P.: In vivo amplification of the androgen receptor gene and progressionof human prostate cancer. Nature Genet. 9: 401–406, 1995.125. Von Eckardstein, S.; Syska, A.; Gromoll, J.; Kamischke, A.; Simoni, M.; Nieschlag, E.: Inverse correlation between sperm concentrationand number of androgen receptor CAG repeats in normal men. J. Clin.Endocr. Metab. 86: 2585–2590, 2001.126. Wang, Q.; Ghadessy, F. J.; Trounson, A.; de Kretser, D.; McLachlan, R.; Ng, S. C.; Yong, E. L.: Azoospermia associated with a mutationin the ligand-binding domain of an androgen

receptor displaying normal ligand binding, but defective trans-activation. *J. Clin. Endocr. Metab.* 83: 4303–4309, 1998.127. Weidemann, W.; Peters, B.; Romalo, G.; Spindler, K.-D.; Schweikert, H.-U.: Response to androgen treatment in a patient with partial androgen insensitivity and a mutation in the deoxyribonucleic acid-binding domain of the androgen receptor. *J. Clin. Endocr. Metab.* 83: 1173–1176, 1998.128. Welch, W. J.; Diamond, M. I.: Glucocorticoid modulation of androgen receptor nuclear aggregation and cellular toxicity is associated with distinct forms of soluble expanded polyglutamine protein. *Hum. Molec. Genet.* 10: 3063–3074, 2001.129. Westberg, L.; Baghaei, F.; Rosmond, R.; Hellstrand, M.; Landen, M.; Jansson, M.; Holm, G.; Bjorntorp, P.; Eriksson, E.: Polymorphisms of the androgen receptor gene and the estrogen receptor beta gene are associated with androgen levels in women. *J. Clin. Endocr. Metab.* 86: 2562–2568, 2001.130. Wieacker, P.; Breckwoldt, M.; Gal, A.: Testicular feminization: diagnosis and search for closely linked restriction fragment length polymorphism. *Dis. Markers* 3: 213–218, 1985.131. Wieacker, P.; Griffin, J. E.; Wienker, T.; Lopez, J. M.; Wilson, J. D.; Breckwoldt, M.: Linkage analysis with RFLPs in families with androgen resistance syndromes: evi-

dence for close linkage between the androgen receptor locus and the DXS1 segment. *Hum. Genet.* 76:248–252, 1987.132. Wilson, C. M.; McPhaul, M. J.: A and B forms of the androgen receptor are present in human genital skin fibroblasts. *Proc. Nat. Acad. Sci.* 91: 1234–1238, 1994.133. Wilson, J. D.: The promiscuous receptor: prostate cancer comes of age. (Editorial) *New Eng. J. Med.* 332: 1440–1441, 1995.134. Wilson, J. D.; Carlson, B. R.; Weaver, D. D.; Kovacs, W. J.; Griffin, J. E.: Endocrine and genetic characterization of cousins with male pseudohermaphroditism: evidence that the Lubs phenotype can result from a mutation that alters the structure of the androgen receptor. *Clin. Genet.* 26: 363–370, 1984.135. Wooster, R.; Mangion, J.; Eeles, R.; Smith, S.; Dowsett, M.; Averill, D.; Barrett-Lee, P.; Easton, D. F.; Ponder, B. A. J.; Stratton, M. R.: A germline mutation in the androgen receptor gene in two brothers with breast cancer and Reifenshtein syndrome. *Nature Genet.* 2: 132–134, 1992.136. Zhang, L.; Leeflang, E. P.; Yu, J.; Arnheim, N.: Studying human mutations by sperm typing: instability of CAG trinucleotide repeats in the human androgen receptor gene. *Nature Genet.* 7: 531–535, 1994.137. Zhu, Y.-S.; Cai, L.-Q.; Cordero, J. J.; Canovatchel, W. J.; Katz, M. D.; Imper-

ato-McGinley, J.: A novel mutation in the CAG triplet region of exon 1 of androgen receptor gene causes complete androgeninsensitivity syndrome in a large kindred. *J. Clin. Endocr. Metab.* 84:1590–1594, 1999.138. Zoppi, S.; Wilson, C. M.; Harbison, M. D.; Griffin, J. E.; Wilson, J. D.; McPhaul, M. J.; Marcelli, M.: Complete testicular feminization caused by an amino-terminal truncation of the androgen receptor with downstream initiation. *J. Clin. Invest.* 91: 1105–1112, 1993.

[40219] 13771. Konig, A.; Happle, R.; Fink-Puches, R.; Soyer, H. P.; Bornholdt, D.; Engel, H.; Grzeschik, K.-H.: A novel missense mutation of NSDHL in an unusual case of CHILD syndrome showing bilateral, almost symmetric involvement. *J. Am. Acad. Derm.* 46: 594–596, 2002.

[40220] 13772. Labit-Le Bouteiller, C.; Jamme, M. F.; David, M.; Silve, S.; Lanau, C.; Dhers, C.; Picard, C.; Rahier, A.; Taton, M.; Loison, G.; Caput, D.; Ferrara, P.; Lupker, J.: Antiproliferative effects of SR31747A in animal cell lines are mediated by inhibition of cholesterol biosynthesis at the sterol isomerase step. *Europ. J. Biochem.* 256: 342–349, 1998.

[40221] 13773. Liu, X. Y.; Dangel, A. W.; Kelley, R. I.; Zhao, W.; Denny, P.; Botcherby, M.; Cattanach, B.; Peters, J.; Hunsicker, P. R.; Mallon, A.-M.; Strivens, M. A.; Bate, R.; Miller,

W.; Rhodes, M.; Brown, S.D. M.; Herman, G. E.: The gene mutated in bare patches and striated mice encodes a novel 3-beta-hydroxysteroid dehydrogenase. *Nature Genet.* 22: 182-187, 1999.

[40222] 13774. Schindelhauer, D.; Hellebrand, H.; Grimm, L.; Bader, I.; Meitinger, T.; Wehnert, M.; Ross, M.; Meindl, A.: Long-range map of a 3.5-Mb region in Xp11.23-22 with a sequence-ready map from a 1.1-Mb gene-rich interval. *Genome Res.* 6: 1056-1069, 1996.

[40223] 13775. Silve, S.; Dupuy, P. H.; Labit-Lebouteiller, C.; Kaghad, M.; Chalon, P.; Rahier, A.; Taton, M.; Lupker, J.; Shire, D.; Loison, G.: Emopamil-binding protein, a mammalian protein that binds a series of structurally diverse neuroprotective agents, exhibits delta(8)-delta(7) sterol isomerase activity in yeast. *J. Biol. Chem.* 271: 22434-22440, 1996.

[40224] 13776. Traupe, H.; Muller, D.; Atherton, D.; Kalter, D. C.; Cremers, F. P. M.; van Oost, B. A.; Ropers, H.-H.: Exclusion mapping of the X-linked dominant chondrodysplasia punctata/ichthyosis/cataract/short stature (Happle) syndrome: possible involvement of an unstable pre-mutation. *Hum. Genet.* 89: 659-665, 1992.

[40225] 13777. Jin, H.; Gardner, R. J.; Viswesvariah, R.; Muntoni,

F.; Roberts,R. G.: Two novel members of the interleukin-1 receptor gene family,one deleted in Xp22.1-Xp21.3 mental retardation. *Europ. J. Hum. Genet.* 8:87-94, 2000.

[40226] 13778.Biamonti, G.; Ruggiu, M.; Saccone, S.; Della Valle, G.; Riva, S.: Two homologous genes, originated by duplication, encode the humanhnRNP proteins A2 and A1. *Nucleic Acids Res.* 22: 1996-2002, 1994.

[40227] 13779.Stoddart, K. L.; Jermak, C.; Nagaraja, R.; Schlessinger, D.; Bech-Hansen,N. T.: Physical map covering a 2 Mb region in human Xp11.3 distalto DX6849. *Gene* 227: 111-116, 1999.

[40228] 13780.Swanson, D. A.; Freund, C. L.; Ploder, L.; McInnes, R. R.; Valle,D.: A ubiquitin C-terminal hydrolase gene on the proximal short armof the X chromosome: implications for X-linked retinal disorders. *Hum.Molec. Genet.* 5: 533-538, 1996.

[40229] 13781.Narayanan, V.; Olinsky, S.; Dahle, E.; Naidu, S.; Zoghbi, H. Y.: Mutation analysis of the M6b gene in patients with Rett syndrome. *Am.J. Med. Genet.* 78: 165-168, 1998.

[40230] 13782.Olinsky, S.; Loop, B. T.; DeKosky, A.; Ripepi, B.; Weng, W.; Cummins,J.; Wenger, S. L.; Yan, Y.; Lagenaur, C.; Narayanan, V.: Chromosomalmapping of the human

M6 genes. Genomics 33: 532–536, 1996.

[40231] 13783.Yan, Y.; Lagenaur, C.; Narayanan, V.: Molecular cloning of M6:identification of a PLP/DM20 gene family. Neuron 11: 423–431, 1993.

[40232] 13784.Ciccodicola, A.; D'Esposito, M.; Esposito, T.; Gianfrancesco, F.;Migliaccio, C.; Miano, M. G.; Matarazzo, M. R.; Vacca, M.; Franze,A.; Cuccurese, M.; Cocchia, M.; Curci, A.: Differentially regulatedand evolved genes in the fully sequenced Xq/Yq pseudoautosomal region. Hum.Molec. Genet. 9: 395–401, 2000.

[40233] 13785.D'Esposito, M.; Ciccodicola, A.; Gianfrancesco, F.; Esposito, T.;Flagiello, L.; Mazzarella, R.; Schlessinger, D.; D'Urso, M.: A synaptobrevin–likegene in the Xq28 pseudoautosomal region undergoes X inactivation. Nature–Genet. 13: 227–229, 1996.

[40234] 13786.Muller, D. J.; Schulze, T. G.; Jahnes, E.; Cichon, S.; Krauss,H.; Kesper, K.; Held, T.; Maier, W.; Propping, P.; Nothen, M. M.;Rietschel, M.: Association between a polymorphism in the pseudoautosomalX–linked gene SYBL1 and bipolar affective disorder. Am. J. Med. Genet.(Neuropsychiat. Genet.) 114: 74–78, 2002.

[40235] 13787.Saito, T.; Parsia, S.; Papolos, D. F.; Lachman, H. M.: Analysisof the pseudoautosomal X–linked gene SYBL1 in

bipolar affective disorder:description of a new candidate allele for psychiatric disorders. Am.J. Med. Genet. (Neuropsychiat. Genet.) 96: 317–323, 2000.

- [40236] 13788.Schaefer, L.; Ballabio, A.; Zoghbi, H. Y.: Cloning and characterization of a putative human holocytochrome c-type synthetase gene (HCCS) isolated from the critical region for microphthalmia with linear skin defects (MLS). Genomics 34: 166–172, 1996.
- [40237] 13789.Van den Veyver, I. B.; Subramanian, S.; Zoghbi, H. Y.: Genomic structure of a human holocytochrome c-type synthetase gene in Xp22.3 and mutation analysis in patients with Rett syndrome. Am. J. Med. Genet. 78: 179–181, 1998.
- [40238] 13790.Andersson, M.; Page, D. C.; Pettay, D.; Subrt, I.; Turleau, C.; de Grouchy, J.; de la Chapelle, A.: Y;autosome translocations and mosaicism in the aetiology of 45,X maleness: assignment of fertility factor to distal Yq11. Hum. Genet. 79: 2–7, 1988.
- [40239] 13791.Chandley, A. C.; Gosden, J. R.; Hargreave, T. B.; Spowart, G.; Speed, R. M.; McBeath, S.: Deleted Yq in the sterile son of a man with a satellited Y chromosome (Yqs). J. Med. Genet. 26: 145–153, 1989.
- [40240] 13792.Franco, B.; Meroni, G.; Parenti, G.; Levilliers, J.;

Bernard, L.;Gebbia, M.; Cox, L.; Maroteaux, P.; Sheffield, L.; Rappold, G. A.;Andria, G.; Petit, C.; Ballabio, A.: A cluster of sulfatase genes on Xp22.3: mutations in chondrodysplasia punctata (CDPX) and implications for warfarin embryopathy. *Cell* 81: 1–20, 1995.

[40241] 13793.Meroni, G.; Franco, B.; Archidiacono, N.; Messali, S.; Andolfi, G.; Rocchi, M.; Ballabio, A.: Characterization of a cluster of sulfatase genes on Xp22.3 suggests gene duplications in an ancestral pseudoautosomal region. *Hum. Molec. Genet.* 5: 423–431, 1996.

[40242] 13794.Lee, C. C.; Yazdani, A.; Wehnert, M.; Zhao, Z. Y.; Lindsay, E.A.; Bailey, J.; Coolbaugh, M. I.; Couch, L.; Xiong, M.; Chinault, A. C.; Baldini, A.; Caskey, C. T.: Isolation of chromosome-specific genes by reciprocal probing of arrayed cDNA and cosmid libraries. *Hum. Molec. Genet.* 4: 1373–1380, 1995.

[40243] 13795.Patzak, D.; Zhuchenko, O.; Lee, C.–C.; Wehnert, M.: Identification, mapping, and genomic structure of a novel X-chromosomal human gene (SMPX) encoding a small muscular protein. *Hum. Genet.* 105: 506–512, 1999.

[40244] 13796.Lahn, B. T.; Page, D. C.: A human sex-chromosomal gene family expressed in male germ cells and encoding variably charged proteins. *Hum. Molec.*

Genet. 9: 311–319, 2000.

- [40245] 13797. Nagase, T.; Seki, N.; Ishikawa, K.; Ohira, M.; Kawarabayashi, Y.; Ohara, O.; Tanaka, A.; Kotani, H.; Miyajima, N.; Nomura, N.: Prediction of the coding sequences of unidentified human genes. VI. The coding sequences of 80 new genes (KIAA0201–KIAA0280) deduced by analysis of cDNA clones from cell line KG–1 and brain. DNA Res. 3: 321–329, 1996. Note: Supplement: DNA Res. 3: 341–354, 1996.
- [40246] 13798. Numata, M.; Petrecca, K.; Lake, N.; Orlowski, J.: Identification of a mitochondrial Na⁺/H⁺ exchanger. J. Biol. Chem. 273: 6951–6959, 1998.
- [40247] 13799. Buchner, G.; Orfanelli, U.; Quaderi, N.; Bassi, M. T.; Andolfi, G.; Ballabio, A.; Franco, B.: Identification of a new EGF-repeat-containing gene from human Xp22: a candidate for developmental disorders. Genomics 65:16–23, 2000.
- [40248] 13800. Yeung, G.; Mulero, J. J.; Berntsen, R. P.; Loeb, D. B.; Drmanac, R.; Ford, J. E.: Cloning of a novel epidermal growth factor repeat-containing gene EGFL6: expressed in tumor and fetal tissues. Genomics 62:304–307, 1999.
- [40249] 13801. Asao, H.; Okuyama, C.; Kumaki, S.; Ishii, N.; Tsuchiya, S.; Foster, D.; Sugamura, K.: Cutting edge: the

common gamma-chain is an indispensable subunit of the IL-21 receptor complex. *J. Immun.* 167: 1-5, 2001.

- [40250] 13802. Lankes, W.; Griesmacher, A.; Grunwald, J.; Schwartz-Albiez, R.; Keller, R.: A heparin-binding protein involved in inhibition of smooth-muscle cell proliferation. *Biochem. J.* 251: 831-842, 1988.
- [40251] 13803. Lankes, W. T.; Furthmayr, H.: Moesin: a member of the protein 4.1-talin-ezrin family of proteins. *Proc. Nat. Acad. Sci.* 88: 8297-8301, 1991.
- [40252] 13804. Shcherbina, A.; Bretscher, A.; Rosen, F. S.; Kenney, D. M.; Remold-O'Donnell, E.: The cytoskeletal linker protein moesin: decreased levels in Wiskott-Aldrich syndrome platelets and identification of a cleavage pathway in normal platelets. *Brit. J. Haemat.* 106: 216-223, 1999.
- [40253] 13805. Wilgenbus, K. K.; Hsieh, C.-L.; Lankes, W. T.; Milatovich, A.; Francke, U.; Furthmayr, H.: Structure and localization on the X chromosome of the gene coding for the human filopodial protein moesin (MSN). *Genomics* 19: 326-333, 1994.
- [40254] 13806. Deng, L.; Wang, C.; Spencer, E.; Yang, L.; Braun, A.; You, J.; Slaughter, C.; Pickart, C.; Chen, Z. J.: Activation of the I-kappa-B complex by TRAF6 requires a dimeric ubiquitin-conjugating enzyme complex and a unique polyubiq-

uitin chain. Cell 103: 351–361, 2000.

- [40255] 13807.Wang, C.; Deng, L.; Hong, M.; Akkaraju, G. R.; Inoue, J.; Chen,Z. J.: TAK1 is a ubiquitin–dependent kinase of MKK and IKK. Nature 412:346–351, 2001.
- [40256] 13808.Wong, B. R.; Besser, D.; Kim, N.; Arron, J. R.; Vologodskaya, M.; Hanafusa, H.; Choi, Y.: TRANCE, a TNF family member, activates Akt/PKB through a signaling complex involving TRAF6 and c-Src. Molec. Cell 4:1041–1049, 1999.
- [40257] 13809.Nakano, H.; Oshima, H.; Chung, W.; Williams–Abbott, L.; Ware, C.F.; Yagita, H.; Okumura, K.: TRAF5, an activator of NF–kappaB and putative signal transducer for the lymphotoxin–beta receptor. J.Biol. Chem. 271: 14661–14664, 1996.
- [40258] 13810.Nakano, H.; Sakon, S.; Koseki, H.; Takemori, T.; Tada, K.; Matsumoto, M.; Munechika, E.; Sakai, T.; Shirasawa, T.; Akiba, H.; Kobata, T.; Santee, S. M.; Ware, C. F.; Renner, P. D.; Taniguchi, M.; Yagita, H.; Okumura, K.: Targeted disruption of Traf5 gene causes defects in CD40– and CD27–mediated lymphocyte activation. Proc. Nat. Acad.Sci. 96: 9803–9808, 1999.
- [40259] 13811.Nakano, H.; Shindo, M.; Yamada, K.; Yoshida, M. C.; Santee, S.M.; Ware, C. F.; Jenkins, N. A.; Gilbert, D. J.;

Yagita, H.; Copeland, N. G.; Okumura, K.: Human TNF receptor-associated factor 5 (TRAF5): cDNA cloning, expression and assignment of the TRAF5 gene to chromosome 1q32. *Genomics* 42: 26–32, 1997.

[40260] 13812. Yost, C.; Farr, G. H., III; Pierce, S. B.; Ferkey, D. M.; Chen, M. M.; Kimelman, D.: GBP, an inhibitor of GSK-3, is implicated in *Xenopus* development and oncogenesis. *Cell* 93: 1031–1041, 1998.

[40261] 13813. De Baere, E.; Speleman, F.; Van Roy, N.; De Paepe, A.; Messiaen, L.: Assignment of SHOX2 (alias OG12X and SHOT) to human chromosome bands 3q25–q26.1 by in situ hybridization. *Cytogenet. Cell Genet.* 82: 228–229, 1998.

[40262] 13814. Fernandez-Valle, C.; Tang, Y.; Ricard, J.; Rodenas-Ruano, A.; Taylor, A.; Hackler, E.; Biggerstaff, J.; Iacovelli, J.: Paxillin binds schwannomin and regulates its density-dependent localization and effect on cell morphology. *Nature Genet.* 31: 354–362, 2002.

[40263] 13815. Glenney, J. R., Jr.; Zokas, L.: Novel tyrosine kinase substrates from Rous sarcoma virus-transformed cells are present in the membrane skeleton. *J. Cell Biol.* 108: 2401–2408, 1989.

[40264] 13816. Mazaki, Y.; Hashimoto, S.; Sabe, H.: Monocyte cells

and cancer cells express novel paxillin isoforms with different binding properties to focal adhesion proteins. *J. Biol. Chem.* 272: 7437–7444, 1997.

[40265] 13817. Salgia, R.; Li, J.-L.; Lo, S. H.; Brunkhorst, B.; Kansas, G. S.; Sobhany, E. S.; Sun, Y.; Pisick, E.; Hallek, M.; Ernst, T.; Tantravahi, R.; Chen, L. B.; Griffin, J. D.: Molecular cloning of human paxillin, a focal adhesion protein phosphorylated by P210(BCR/ABL). *J. Biol. Chem.* 270: 5039–5047, 1995.

[40266] 13818. Turner, C. E.; Glenney, J. R., Jr.; Burridge, K.: Paxillin: a new vinculin-binding protein present in focal adhesions. *J. Cell Biol.* 111: 1059–1068, 1990.

[40267] 13819. Nguyen, V. T.; Kiss, T.; Michels, A. A.; Bensaude, O.: 7SK small nuclear RNA binds to and inhibits the activity of CDK9/cyclin T complexes. *Nature* 414:322–325, 2001.

[40268] 13820. Peng, J.; Zhu, Y.; Milton, J. T.; Price, D. H.: Identification of multiple cyclin subunits of human P-TEFb. *Genes Dev.* 12: 755–762, 1998.

[40269] 13821. Yang, Z.; Zhu, Q.; Luo, K.; Zhou, Q.: The 7SK small nuclear RNA inhibits the CDK9/cyclin T1 kinase to control transcription. *Nature* 414:317–322, 2001.

[40270] 13822. Adachi, H.; Tsujimoto, M.; Hattori, M.; Arai, H.; Inoue, K.: Differential tissue distribution of the beta- and

gamma-subunits of human cytosolic platelet-activating factor acetylhydrolase (isoform 1). *Biochem. Biophys. Res. Commun.* 233: 10–13, 1997.

[40271] 13823. Moro, F.; Arrigo, G.; Fogli, A.; Bernard, L.; Carrozzo, R.: The beta and gamma subunits of the human platelet-activating factor acetylhydrolase isoform Ib (PAFAH1B2 and PAFAH1B3) map to chromosome 11q23 and 19q13.1, respectively. *Genomics* 51: 157–159, 1998.

[40272] 13824. Daigo, Y.; Isomura, M.; Nishiwaki, T.; Tamari, M.; Ishikawa, S.; Kai, M.; Murata, Y.; Takeuchi, K.; Yamane, Y.; Hayashi, R.; Minami, M.; Fujino, M. A.; Hojo, Y.; Uchiyama, I.; Takagi, T.; Nakamura, Y.: Characterization of a 1200-kb genomic segment of chromosome 3p22–p21.3. *DNAREs.* 6: 37–44, 1999.

[40273] 13825. Erlich, R.; Gleeson, P. A.; Campbell, P.; Dietzsch, E.; Toh, B.-H.: Molecular characterization of trans-Golgi p230: a human peripheral membrane protein encoded by a gene on chromosome 6p12–22 contains extensive coiled-coil alpha-helical domains and a granin motif. *J. Biol. Chem.* 271: 8328–8337, 1996.

[40274] 13826. Barr, F. A.; Nakamura, N.; Warren, G.: Mapping the interaction between GRASP65 and GM130, components of

a protein complex involved in the stacking of Golgi cisternae. EMBO J. 17: 3258–3268, 1998.

[40275] 13827. Boguski, M. S.; Lowe, T. M. J.; Tolstocher, C. M.: dbEST: database for 'expressed sequence tags.' (Letter) Nature Genet. 4: 332–333, 1993.

[40276] 13828. Izon, D. J.; Aster, J. C.; He, Y.; Weng, A.; Karnell, F. G.; Patriub, V.; Xu, L.; Bakkour, S.; Rodriguez, C.; Allman, D.; Pear, W. S.: Deltex1 redirects lymphoid progenitors to the B cell lineage by antagonizing Notch1. Immunity 16: 231–243, 2002.

[40277] 13829. Lee, L.; Dowhanick–Morrisette, J.; Katz, A.; Jukofsky, L.; Krantz, I. D.: Chromosomal localization, genomic characterization, and mapping to the Noonan syndrome critical region of the human Deltex (DTX1) gene. Hum. Genet. 107: 577–581, 2000.

[40278] 13830. Matsuno, K.; Eastman, D.; Mitsiades, T.; Quinn, A. M.; Carcanciu, M. L.; Ordentlich, P.; Kadesch, T.; Artavanis–Tsakonas, S.: Human Deltex is a conserved regulator of Notch signalling. Nature Genet. 19: 74–78, 1998.

[40279] 13831. Chen, J. J.; Reid, C. E.; Band, V.; Androphy, E. J.: Interaction of papillomavirus E6 oncoproteins with a putative calcium-binding protein. Science 269: 529–531, 1995.

[40280] 13832. Imai, T.; Matsuda, K.; Shimojima, T.; Hashimoto, T.;

Masuhira,Y.; Kitamoto, T.; Sugita, A.; Suzuki, K.; Matsumoto, H.; Masushige,S.; Nogi, Y.; Muramatsu, M.; Handa, H.; Kato, S.: ERC-55, a bindingprotein for the papilloma virus E6 oncoprotein, specifically interactswith vitamin D receptor among nuclear receptors. *Biochem. Biophys.Res. Commun.* 233: 765-769, 1997.

[40281] 13833.Wang, J. Y.; Zhen, D. K.; Bianchi, D. W.; Androphy, E. J.; Chen,J. J.: Assignment of the gene for ERC-55 (RCN2) to human chromosomeband 15q22.33-q24.1 by in situ hybridization. *Cytogenet. Cell Genet.* 79:60-61, 1997.

[40282] 13834.Weis, K.; Griffiths, G.; Lamond, A. I.: The endoplasmic reticulumcalcium-binding protein of 55 kDa is a novel EF-hand protein retainedin the endoplasmic reticulum by a carboxyl-terminal His-Asp-Glu-Leumotif. *J. Biol. Chem.* 269: 19142-19150, 1994.

[40283] 13835.Dempsey, C. E.; Sakurai, H.; Sugita, T.; Guesdon, F.: Alternativesplicing and gene structure of the transforming growth factor beta-activatedkinase 1. *Biochim. Biophys. Acta* 1517: 46-52, 2000.

[40284] 13836.Kondo, M.; Osada, H.; Uchida, K.; Yanagisawa, K.; Masuda, A.; Takagi,K.; Takahashi, T.; Takahashi, T.: Molecular cloning of human TAK1and its mutational analysis in human lung cancer. *Int. J. Cancer* 75:559-563,

1998.

- [40285] 13837.Sakurai, H.; Shigemori, N.; Hasegawa, K.; Sugita, T.: TGF-beta-activatedkinase 1 stimulates NF-kappa-B activation by an NF-kappa-B-inducingkinase-independent mechanism. *Biochem. Biophys. Res. Commun.* 243:545-549, 1998.
- [40286] 13838.Yamaguchi, K.; Shirakabe, K.; Shibuya, H.; Irie, K.; Oishi, I.;Ueno, N.; Taniguchi, T.; Nishida, E.; Matsumoto, K.: Identificationof a member of the MAPKKK family as a potential mediator of TGF-betasignal transduction. *Science* 270: 2008-2011, 1995.
- [40287] 13839.Chan, A. M.-L.; Chedid, M.; McGovern, E. S.; Popescu, N. C.; Miki,T.; Aaronson, S. A.: Expression cDNA cloning of a serine kinase transforminggene. *Oncogene* 8: 1329-1333, 1993.
- [40288] 13840.Frank, K. M.; Sekiguchi, J. M.; Seidl, K. J.; Swat, W.; Rathbun,G. A.; Cheng, H.-L.; Davidson, L.; Kangaloo, L.; Alt, F. W.: Lateembryonic lethality and impaired V(D)J recombination in mice lackingDNA ligase IV. *Nature* 396: 173-177, 1998.
- [40289] 13841.Frank, K. M.; Sharpless, N. E.; Gao, Y.; Sekiguchi, J. M.; Ferguson,D. O.; Zhu, C.; Manis, J. P.; Horner, J.; De-Pinho, R. A.; Alt, F.W.: DNA ligase IV deficiency in mice

leads to defective neurogenesis and embryonic lethality via the p53 pathway. *Molec. Cell* 5: 993–1002, 2000.

[40290] 13842. Grawunder, U.; Zimmer, D.; Fugmann, S.; Schwarz, K.; Lieber, M.R.: DNA ligase IV is essential for V(D)J recombination and DNA double-strandbreak repair in human precursor lymphocytes. *Molec. Cell* 2: 477–484, 1998.

[40291] 13843. O'Driscoll, M.; Cerosaletti, K. M.; Girard, P.-M.; Dai, Y.; Stumm, M.; Kysela, B.; Hirsch, B.; Gennery, A.; Palmer, S. E.; Seidel, J.; Gatti, R. A.; Varon, R.; Oettinger, M. A.; Neitzel, H.; Jeggo, P.A.; Concannon, P.: DNA ligase IV mutations identified in patients exhibiting developmental delay and immunodeficiency. *Molec. Cell* 8: 1175–1185, 2001.

[40292] 13844. Riballo, E.; Critchlow, S. E.; Teo, S.-H.; Doherty, A. J.; Priestley, A.; Broughton, B.; Kysela, B.; Beamish, H.; Plowman, N.; Arlett, C.F.; Lehmann, A. R.; Jackson, S. P.; Jeggo, P. A.: Identification of a defect in DNA ligase IV in a radiosensitive leukaemia patient. *Curr. Biol.* 9: 699–702, 1999.

[40293] 13845. Riballo, E.; Doherty, A. J.; Dai, Y.; Stiff, T.; Oettinger, M.A.; Jeggo, P. A.; Kysela, B.: Cellular and biochemical impact of a mutation in DNA ligase IV conferring clinical radiosensitivity. *J. Biol. Chem.* 276: 31124–31132,

2001.

- [40294] 13846. Giachino, C.; Lantelme, E.; Lanzetti, L.; Saccone, S.; Della Valle, G.; Migone, N.: A novel SH3-containing human gene family preferentially expressed in the central nervous system. *Genomics* 41: 427–434, 1997.
- [40295] 13847. Furlan, M.; Robles, R.; Galbusera, M.; Remuzzi, G.; Kyrle, P. A.; Brenner, B.; Krause, M.; Scharer, I.; Aumann, V.; Mittler, U.; Solenthaler, M.; Lammle, B.: Von Willebrand factor-cleaving protease in thrombotic thrombocytopenic purpura and the hemolytic-uremic syndrome. *New Eng. J. Med.* 339: 1578–1584, 1998.
- [40296] 13848. Furlan, M.; Robles, R.; Lammle, B.: Partial purification and characterization of a protease from human plasma cleaving von Willebrand factor to fragments produced by in vivo proteolysis. *Blood* 87: 4223–4234, 1996.
- [40297] 13849. Nichols, A. F.; Ong, P.; Linn, S.: Mutations specific to the xeroderma pigmentosum group E Ddb- phenotype. *J. Biol. Chem.* 271: 24317–24320, 1996.
- [40298] 13850. Shisheva, A.; Sudhof, T. C.; Czech, M. P.: Cloning, characterization, and expression of a novel GDP dissociation inhibitor isoform from skeletal muscle. *Molec. Cell. Biol.* 14: 3459–3468, 1994.
- [40299] 13851. Grieff, M.; Whyte, M. P.; Thakker, R. V.; Mazzarella,

R.: Sequence analysis of 139 kb in Xp22.1 containing spermine synthase and the 5'-prime region of PEX. *Genomics* 44: 227-231, 1997.

[40300] 13852. Korhonen, V. P.; Halmekyto, M.; Kauppinen, L.; Myohanen, S.; Wahlfors, J.; Keinänen, T.; Hyvonen, T.; Alhonen, L.; Eloranta, T.; Janne, J.: Molecular cloning of a cDNA encoding human spermine synthase. *DNACell Biol.* 14: 841-847, 1995.

[40301] 13853. Banfi, S.; Borsani, G.; Bulfone, A.; Ballabio, A.: *Drosophila*-related expressed sequences. *Hum. Molec. Genet.* 6: 1745-1753, 1997.

[40302] 13854. Castrillon, D. H.; Wasserman, S. A.: Diaphanous is required for cytokinesis in *Drosophila* and shares domains of similarity with the products of the limb deformity gene. *Development* 120: 3367-3377, 1994.

[40303] 13855. Philippe, C.; Cremers, F. P. M.; Chery, M.; Bach, I.; Abbadi, N.; Ropers, H. H.; Gilgenkrantz, S.: Physical mapping of DNA markers in the q13-q22 region of the human X chromosome. *Genomics* 17: 147-152, 1993.

[40304] 13856. Bione, S.; Sala, C.; Manzini, C.; Arrigo, G.; Zuffardi, O.; Banfi, S.; Borsani, G.; Jonveaux, P.; Philippe, C.; Zucotti, M.; Ballabio, A.; Toniolo, D.: A human homologue of the *Drosophila melanogaster* diaphanous gene is disrupted

in a patient with premature ovarian failure:evidence for conserved function in oogenesis and implications for human sterility. *Am. J. Hum. Genet.* 62: 533–541, 1998.

- [40305] 13857.Sala, C.; Arrigo, G.; Torri, G.; Martinazzi, F.; Riva, P.; Larizza,L.; Philippe, C.; Jonveaux, P.; Sloan, F.; Labella, T.; Toniolo, D.: Eleven X chromosome breakpoints associated with premature ovarianfailure (POF) map to a 15-Mb YAC contig spanning Xq21. *Genomics* 40:123–131, 1997.
- [40306] 13858.Hanaoka, F.; Tandai, M.; Miyazawa, H.; Murakami, Y.; Hori, T.;Yamada, M.: Assignment of human DNA polymerase alpha gene (POLA)to the X chromosome. (Abstract) *Cytogenet. Cell Genet.* 40: 647 only,1985.
- [40307] 13859.Miyazawa, H.; Tandai, M.; Hanaoka, F.; Yamada, M.; Hori, T.; Shimizu,K.; Sekiguchi, M.: Identification of a DNA segment containing thehuman DNA polymerase alpha gene. *Biochem. Biophys. Res. Commun.* 139:637–643, 1986.
- [40308] 13860.Wang, T. S.–F.; Pearson, B. E.; Suomalainen, H. A.; Mohandas, T.;Shapiro, L. J.; Schroder, J.; Korn, D.: Assignment of the gene forhuman DNA polymerase alpha to the X chromosome. *Proc. Nat. Acad.Sci.* 82: 5270–5274, 1985.
- [40309] 13861.Watson, J. M.; Spencer, J. A.; Riggs, A. D.; Graves, J.

A. M.:Sex chromosome evolution: platypus gene mapping suggests that part of the human X chromosome was originally autosomal. *Proc. Nat. Acad.Sci.* 88: 11256–11260, 1991.

- [40310] 13862.Lachner, M.; O'Carroll, D.; Rea, S.; Mechtler, K.; Jenuwein, T.: Methylation of histone H3 lysine 9 creates a binding site for HP1proteins. *Nature* 410: 116–120, 2001.
- [40311] 13863.Melcher, M.; Schmid, M.; Aagaard, L.; Selenko, P.; Laible, G.;Jenuwein, T.: Structure–function analysis of SUV39H1 reveals a dominantrole in heterochromatin organization, chromosome segregation, andmitotic progression. *Molec. Cell Biol.* 20: 3728–3841, 2000.
- [40312] 13864.Nielsen, S. J.; Schneider, R.; Bauer, U.–M.; Bannister, A. J.;Morrison, A.; O'Carroll, D.; Firestein, R.; Cleary, M.; Jenuwein,T.; Herrera, R. E.; Kouzarides, T.: Rb targets histone H3 methylationand HP1 to promoters. *Nature* 412: 561–565, 2001.
- [40313] 13865.Peters, A. H. F. M.; O'Carroll, D.; Scherthan, H.; Mechtler, K.;Sauer, S.; Schofer, C.; Weipoltshammer, K.; Pagani, M.; Lachner, M.;Kohlmaier, A.; Opravil, S.; Doyle, M.; Sibilia, M.; Jenuwein, T.:Loss of the Suv39h histone methyltransferases impairs mammalian heterochromatinand genome stability. *Cell* 107: 323–337, 2001.

- [40314] 13866.Rea, S.; Eisenhaber, F.; O'Carroll, D.; Strahl, B. D.; Sun, Z.-W.;Schmid, M.; Opravil, S.; Mechtler, K.; Ponting, C. P.; Allis, C. D.;Jenuwein, T.: Regulation of chromatin structure by site-specific histone H3 methyltransferases. *Nature* 406: 593–599, 2000.
- [40315] 13867.Scott, A. F.: Personal Communication. Baltimore, Md. 8/7/2000.
- [40316] 13868.Haltiwanger, R. S.; Blomberg, M. A.; Hart, G. W.: Glycosylation of nuclear and cytoplasmic proteins: purification and characterization of a uridine diphosphate-N-acetylglucosamine:polypeptide beta-N-acetylglucosaminyltransferase. *J.Biol. Chem.* 267: 9005–9013, 1992.
- [40317] 13869.Kreppel, L. K.; Blomberg, M. A.; Hart, G. W.: Dynamic glycosylation of nuclear and cytosolic proteins: cloning and characterization of a unique O-GlcNAc transferase with multiple tetratricopeptide repeats. *J.Biol. Chem.* 272: 9308–9315, 1997.
- [40318] 13870.Lubas, W. A.; Frank, D. W.; Krause, M.; Hanover, J. A.: O-linked GlcNAc transferase is a conserved nucleocytoplasmic protein containing tetratricopeptide repeats. *J. Biol. Chem.* 272: 9316–9324, 1997.
- [40319] 13871.Shafi, R.; Iyer, S. P. N.; Ellies, L. G.; O'Donnell, N.;

Marek, K. W.; Chui, D.; Hart, G. W.; Marth, J. D.: The O-GlcNAc transferase gene resides on the X chromosome and is essential for embryonic stem cell viability and mouse ontogeny. *Proc. Nat. Acad. Sci.* 97: 5735–5739, 2000.

[40320] 13872. Yang, X.; Zhang, F.; Kudlow, J. E.: Recruitment of O-GlcNAc transferase to promoters by corepressor mSin3A: coupling protein O-GlcNAcylation to transcriptional repression. *Cell* 110: 69–80, 2002.

[40321] 13873. Furuta, S.; Kobayashi, A.; Miyazawa, S.; Hashimoto, T.: Cloning and expression of cDNA for a newly identified isozyme of bovine liver 3-hydroxyacyl-CoA dehydrogenase and its import into mitochondria. *Biochim. Biophys. Acta* 1350: 317–324, 1997.

[40322] 13874. Hansis, C.; Jahner, D.; Spiess, A.-N.; Boettcher, K.; Ivell, R.: The gene for the Alzheimer-associated beta-amyloid-binding protein (ERAB) is differentially expressed in the testicular Leydig cells of the azoospermic by w/w(v) mouse. *Europ. J. Biochem.* 258: 53–60, 1998.

[40323] 13875. He, X.-Y.; Schulz, H.; Yang, S.-Y.: A human brain L-3-hydroxyacyl-coenzyme A dehydrogenase is identical to an amyloid beta-peptide-binding protein involved in Alzheimer's disease. *J. Biol. Chem.* 273: 10741–10746, 1998.

- [40324] 13876. Yan, S. D.; Fu, J.; Soto, C.; Chen, X.; Zhu, H.; Al-Mohanna, F.; Collison, K.; Zhu, A.; Stern, E.; Saido, T.; Tohyama, M.; Ogawa, S.; Roher, A.; Stern, D.: An intracellular protein that binds amyloid- β peptide and mediates neurotoxicity in Alzheimer's disease. *Nature* 389:689-695, 1997.
- [40325] 13877. Chamberlin, M. E.; Ubagai, T.; Mudd, S. H.; Wilson, W. G.; Leonard, J. V.; Chou, J. Y.: Demyelination of the brain is associated with methionine adenosyltransferase I/III deficiency. *J. Clin. Invest.* 98:1021-1027, 1996.
- [40326] 13878. Nishiyama, K.; Funai, T.; Katafuchi, R.; Hattori, F.; Onoyama, K.; Ichiyama, A.: Primary hyperoxaluria type I due to a point mutation of T to C in the coding region of the serine:pyruvate aminotransferase gene. *Biochem. Biophys. Res. Commun.* 176: 1093-1099, 1991.
- [40327] 13879. Pirulli, D.; Puzzer, D.; Ferri, L.; Crovella, S.; Amoroso, A.; Ferretti, C.; Marangella, M.; Mazzola, G.; Florian, F.: Molecular analysis of hyperoxaluria type 1 in Italian patients reveals eight new mutations in the alanine:glyoxylate aminotransferase gene. *Hum. Genet.* 104: 523-525, 1999.
- [40328] 13880. Watts, R. W. E.; Calne, R. Y.; Rolles, K.; Danpure, C. J.; Morgan, S. H.; Mansell, M. A.; Williams, R.; Purkiss, P.:

Successful treatment of primary hyperoxaluria type I by combined hepatic and renal transplantation. *Lancet* II:474–475, 1987.

- [40329] 13881. Cramer, S. D.; Ferree, P. M.; Lin, K.; Milliner, D. S.; Holmes, R. P.: The gene encoding hydroxypyruvate reductase (GRHPR) is mutated in patients with primary hyperoxaluria type II. *Hum. Molec. Genet.* 8:2063–2069, 1999.
- [40330] 13882. Natt, E.; Kida, K.; Odievre, M.; Di Rocco, M.; Scherer, G.: Point mutations in the tyrosine aminotransferase gene in tyrosinemia type II. *Proc. Nat. Acad. Sci.* 89: 9297–9301, 1992.
- [40331] 13883. Johnson, W. G.: Metabolic interference and the +/- heterozygote: a hypothetical form of simple inheritance which is neither dominant nor recessive. *Am. J. Hum. Genet.* 32: 374–386, 1980.
- [40332] 13884. Brenner, V.; Nyakatura, G.; Rosenthal, A.; Platzer, M.: Genomic organization of two novel genes on human Xq28: compact head to head arrangement of IDH-gamma and TRAP-delta is conserved in rat and mouse. *Genomics* 44:8–14, 1997.
- [40333] 13885. Achen, M. G.; Jeltsch, M.; Kukk, E.; Makinen, T.; Vitali, A.; Wilks, A. F.; Alitalo, K.; Stacker, S. A.: Vascular endothelial growth factor D (VEGF-D) is a ligand for the tyro-

sine kinases VEGF receptor 2 (Flk1) and VEGF receptor 3 (Flt4). *Proc. Nat. Acad. Sci.* 95: 548–553, 1998.

[40334] 13886. Stacker, S. A.; Caesar, C.; Baldwin, M. E.; Thornton, G. E.; Williams, R. A.; Prevo, R.; Jackson, D. G.; Nishikawa, S.; Kubo, H.; Achen, M. G.: VEGF-D promotes the metastatic spread of tumor cells via the lymphatics. *Nature Med.* 7: 186–191, 2001.

[40335] 13887. Yamada, Y.; Nezu, J.; Shimane, M.; Hirata, Y.: Molecular cloning of a novel vascular endothelial growth factor, VEGF-D. *Genomics* 42:483–488, 1997.

[40336] 13888. Davies, P. A.; Hanna, M. C.; Hales, T. G.; Kirkness, E. F.: Insensitivity to anaesthetic agents conferred by a class of GABA(A) receptor subunit. *Nature* 385:820–823, 1997.

[40337] 13889. Sinkkonen, S. T.; Hanna, M. C.; Kirkness, E. F.; Korppe, E. R.: GABA-A receptor epsilon and theta subunits display unusual structural variation between species and are enriched in the rat locus ceruleus. *J. Neurosci.* 20: 3588–3595, 2000.

[40338] 13890. Wilke, K.; Gaul, R.; Klauck, S. M.; Poustka, A.: A gene in human chromosome band Xq28 (GABRE) defines a putative new subunit class of the GABA(A) neurotransmitter receptor. *Genomics* 45: 1–10, 1997.

- [40339] 13891.Gecz, J.; Baker, E.; Donnelly, A.; Ming, J. E.; McDonald-McGinn, D. M.; Spinner, N. B.; Zackai, E. H.; Sutherland, G. R.; Mulley, J.C.: Fibroblast growth factor homologous factor 2 (FHF2): gene structure, expression and mapping to the Borjeson-Forssman-Lehmann syndrome region in Xq26 delineated by a duplication breakpoint in a BFLS-like patient. *Hum.Genet.* 104: 56-63, 1999.
- [40340] 13892.Lovec, H.; Hartung, H.; Verdier, A.-S.; Mattei, M.-G.; Birnbaum, D.; Goldfarb, M.; Coulier, F.: Assignment of FGF13 to human chromosome band Xq21 by in situ hybridization. *Cytogenet. Cell Genet.* 76: 183-184, 1997.
- [40341] 13893.Smallwood, P. M.; Munoz-Sanjuan, I.; Tong, P.; Macke, J. P.; Hendry, S. H. C.; Gilbert, D. J.; Copeland, N. G.; Jenkins, N. A.; Nathans, J.: Fibroblast growth factor (FGF) homologous factors: new members of the FGF family implicated in nervous system development. *Proc.Nat. Acad. Sci.* 93: 9850-9857, 1996.
- [40342] 13894.Jones, M. H.; Furlong, R. A.; Burkin, H.; Chalmers, I. J.; Brown, G. M.; Khwaja, O.; Affara, N. A.: The Drosophila developmental gene fat facets has a human homologue in Xp11.4 which escapes X-inactivation and has related sequences on Yq11.2. *Hum. Molec. Genet.* 5: 1695-1701, 1996.

- [40343] 13895.Stoffers, D. A.; Zinkin, N. T.; Stanojevic, V.; Clarke, W. L.;Habener, J. F.: Pancreatic agenesis attributable to a single nucleotidedeletion in the human IPF1 gene coding sequence. *Nature Genet.* 15:106–110, 1997.
- [40344] 13896.Theophilos, M. B.; Cox, D. W.; Mercer, J. F. B.: The toxic milkmouse is a murine model of Wilson disease. *Hum. Molec. Genet.* 5:1619–1624, 1996.
- [40345] 13897.Natt, E.; Westphal, E.–M.; Toth–Fejel, S. E.; Magenis, R. E.;Buist, N. R. M.; Rettenmeier, R.; Scherer, G.: Inherited and de novodeletion of the tyrosine aminotransferase gene locus at 16q22.1–q22.3in a patient with tyrosinemia type II. *Hum. Genet.* 77: 352–358,1987.
- [40346] 13898.Pelet, B.; Antener, I.; Faggioni, R.; Spahr, A.; Gautier, E.:Tyrosinemia without liver or renal damage with plantar and palmarkeratosis and keratitis (hypertyrosinemia type II). *Helv. Paediat.Acta* 34: 177–183, 1979.
- [40347] 13899.Rehak, A.; Selim, M. M.; Yadav, G.: Richner–Hanhart syndrome(tyrosinaemia–II) (report of four cases without ocular involvement). *Brit.J. Derm.* 104: 469–475, 1981.
- [40348] 13900.Rettenmeier, R.; Natt, E.; Zentgraf, H.; Scherer, G.: Isolationand characterization of the human tyrosine aminotransferase gene. *NucleicAcids Res.* 18: 3853–3861,

1990.

- [40349] 13901. Richner, H.: Hornhautaffektion bei Keratoma palmare et plantare hereditarium. *Klin. Mbl. Augenheilk.* 100: 580–588, 1938.
- [40350] 13902. Schmid, W.; Muller, G.; Schutz, G.; Gluecksohn-Waelsch, S.: Deletions near the albino locus on chromosome 7 of the mouse affect the level of tyrosine aminotransferase mRNA. *Proc. Nat. Acad. Sci.* 82: 2866–2869, 1985.
- [40351] 13903. Tallab, T.M.: Richner–Hanhart syndrome: importance of early diagnosis and early intervention. *J. Am. Acad. Dermatol.* 35: 857–859, 1996.
- [40352] 13904. Ventura, G.; Biasini, G.; Petrozzi, M.: Cheratomia palmoplantare dissipatum associato a lesioni corneali in due fratelli. *Boll. Oculist.* 44: 497–510, 1965.
- [40353] 13905. Waardenburg, P. J.; Franceschetti, A.; Klein, D.: *Genetics and Ophthalmology*. Springfield, Ill.: Charles C Thomas (pub.) 1: 1961. Pp. 515–517.
- [40354] 13906. Westphal, E.-M.; Natt, E.; Grimm, T.; Odievre, M.; Scherer, G.: The human tyrosine aminotransferase gene: characterization of restriction fragment length polymorphisms and haplotype analysis in a family with tyrosinemia type II. *Hum. Genet.* 79: 260–264, 1988.

- [40355] 13907.Perheentupa, J.: Autoimmune polyendocrinopathy–candidosis–ectodermaldystrophy (APECED).In: Eriksson, A. W.; Forsius, H. R.; Nevanlinna,H. R.; Workman, P. L.; Norio, R. K.: Population Structure and Genetic Disorders. New York: Academic Press (pub.) 1980. Pp. 583–587.
- [40356] 13908.Prader, A.: Personal Communication. Zurich, Switzerland 1972.
- [40357] 13909.Ramsay, C.; Winqvist, O.; Puhakka, L.; Halonen, M.; Moro, A.;Kampe, O.; Eskelin, P.; Pelto–Huikko, M.; Peltonen, L.: Aire deficient mice develop multiple features of APECED phenotype and show altered immune response. Hum. Molec. Genet. 11: 397–409, 2002.
- [40358] 13910.Rinderle, C.; Christensen, H.–M.; Schweiger, S.; Lehrach, H.;Yaspo, M.–L.: AIRE encodes a nuclear protein co–localizing with cytoskeletal filaments: altered sub–cellular distribution of mutants lacking PHD zinc fingers. Hum. Molec. Genet. 8: 277–290, 1999.
- [40359] 13911.Rosatelli, M. C.; Meloni, A.; Meloni, A.; Devoto, M.; Cao, A.;Scott, H. S.; Peterson, P.; Heino, M.; Krohn, K. J.; Nagamine, K.;Kudoh, J.; Shimizu, N.; Antonarakis, S. E.: A common mutation in Sardinian autoimmune polyendocrinopathy–candidiasis–ectodermal dystrophy patients. Hum. Genet. 103: 428–434, 1998.

- [40360] 13912.Saenger, P.: Primary hypoaldosteronism due to zona glomerulosa defect. (Letter) New Eng. J. Med. 310: 1394 only, 1984.
- [40361] 13913.Saenger, P.; Levine, L. S.; Irvine, W. J.; Gottesdiener, K.; Rauh, W.; Sonino, N.; Chow, D.; New, M. I.: Progressive adrenal failure in polyglandular autoimmune disease. J. Clin. Endocr. Metab. 54:863–868, 1982.
- [40362] 13914.Scott, H. S.; Heino, M.; Peterson, P.; Mittaz, L.; Lalioti, M.D.; Betterle, C.; Cohen, A.; Seri, M.; Lerone, M.; Romeo, G.; Collin, P.; Salo, M.; and 9 others: Common mutations in autoimmune polyendocrinopathy–candidiasis–ectodermal dystrophy patients of different origins. Molec. Endocr. 12: 1112–1119, 1998.
- [40363] 13915.Shapiro, M. S.; Zamir, R.; Weiss, E.; Radnay, J.; Shenkman, L.: The polyglandular deficiency syndrome: a new variant in Persian Jews. J. Endocr. Invest. 10: 1–7, 1987.
- [40364] 13916.Spinner, M. W.; Blizzard, R. M.; Childs, B.: Clinical and genetic heterogeneity in idiopathic Addison's disease and hypoparathyroidism. J. Clin. Endocr. 28: 795–804, 1968.
- [40365] 13917.Sweetnam, W. P.: Juvenile familial endocrinopathy. Lancet I:463–465, 1966.

- [40366] 13918.Wang, C.-Y.; Davoodi-Semiromi, A.; Huang, W.; Connor, E.; Shi,J.-D.; She, J.-X.: Characterization of mutations in patients with autoimmune polyglandular syndrome type 1 (APS1). *Hum. Genet.* 103:681-685, 1998.
- [40367] 13919.Whitaker, J. A.; Landing, B. H.; Esselborn, V. M.; Williams, R.R.: Syndrome of familial juvenile hypoadrenocorticism, hypoparathyroidism and superficial moniliasis. *J. Clin. Endocr.* 16: 1374-1387, 1956.
- [40368] 13920.Wirfalt, A.: Genetic heterogeneity in autoimmune polyglandular failure. *Acta Med. Scand.* 210: 7-13, 1981.
- [40369] 13921.Zlotogora, J.; Shapiro, M. S.: Polyglandular autoimmune syndrometype I among Iranian Jews. *J. Med. Genet.* 29: 824-826, 1992.
- [40370] 13922.Hediger, M. A.: New view at C. *Nature Med.* 8: 445-446, 2002.
- [40371] 13923.Sotiriou, S.; Gispert, S.; Cheng, J.; Wang, Y.; Chen, A.; Hoogstraten-Miller,S.; Miller, G. F.; Kwon, O.; Levine, M.; Guttentag, S. H.; Nussbaum,R. L.: Ascorbic-acid transporter Slc23a1 is essential for vitaminC transport into the brain and for perinatal survival. *Nature Med.* 8:514-517, 2002.
- [40372] 13924.Nauseef, W. M.; Brigham, S.; Cogley, M.: Hereditary myeloperoxidase deficiency due to a missense mutation of

arginine 569 to tryptophan. J.Biol. Chem. 269:
1212–1216, 1994.

- [40373] 13925. Busard, B. L. S. M.; Renier, W. O.; Gabreels, F. J. M.; Jaspar, H. H. J.; van Haelst, U. J. G.; Slooff, J. L.: Lafora's disease: comparison of inclusion bodies in skin and in brain. Arch. Neurol. 43:296–299, 1986.
- [40374] 13926. Busard, H. L. S. M.; Gabreels–Festen, A. A. W. M.; Renier, W. O.; Gabreels, F. J. M.; Stadhouders, A. M.: Axilla skin biopsy: a reliable test for the diagnosis of Lafora's disease. Ann. Neurol. 21: 599–601, 1987.
- [40375] 13927. Fluharty, A. L.; Porter, M. T.; Hirsh, G. A.; Pevida, E.; Kihara, H.: Metachromasia in fibroblasts from a patient with Lafora's disease. (Letter) Lancet II: 109–110, 1970.
- [40376] 13928. Ganesh, S.; Agarwala, K. L.; Ueda, K.; Akagi, T.; Shoda, K.; Usui, T.; Hashikawa, T.; Osada, H.; Delgado–Escueta, A. V.; Yamakawa, K.: Laforin, defective in the progressive myoclonus epilepsy of Lafora type, is a dual–specificity phosphatase associated with polyribosomes. Hum. Molec. Genet. 9: 2251–2261, 2000.
- [40377] 13929. Gomez–Garre, P.; Sanz, Y.; Rodriguez de Cordoba, S.; Serratosa, J. M.: Mutational spectrum of the EPM2A gene in progressive myoclonus epilepsy of Lafora: high degree of allelic heterogeneity and prevalence of deletions. Europ.

J. Hum. Genet. 8: 946–954, 2000.

- [40378] 13930.Harriman, D. G. F.; Millar, J. H. D.: Progressive familial myoclonicepilepsy in 3 families: its clinical features and pathological basis. Brain 78:325–349, 1955.
- [40379] 13931.Janeway, R.; Ravens, J. R.; Pearce, L. A.; Odor, D. L.; Suzuki,K.: Progressive myoclonus epilepsy with Lafora inclusion bodies.I. Clinical, genetic, histopathologic and biochemical aspects. Arch.Neurol. 16: 565–582, 1967.
- [40380] 13932.Lehesjoki, A.–E.; Koskiniemi, M.; Pandolfo, M.; Antonelli, A.;Kyllerman, M.; Wahlstrom, J.; Nergardh, A.; Burmeister, M.; Sistonen,P.; Norio, R.; de la Chapelle, A.: Linkage studies in progressivemyoclonus epilepsy: Unverricht–Lundborg and Lafora's diseases. Neurology 42:1545–1550, 1992.
- [40381] 13933.Maddox, L. O.; Descartes, M.; Collins, J.; Keating, J.; Rosenfeld,S.; Palmer, C.; Carroll, A. J.; Kuzniecky, R.: Identification ofa recombination event narrowing the Lafora disease gene region. J.Med. Genet. 34: 590–591, 1997.
- [40382] 13934.Amadou, C.; Ribouchon, M. T.; Mattei, M. G.; Jenkins, N. A.; Gilbert,D. J.; Copeland, N. G.; Avoustin, P.; Pontarotti, P.: Localizationof new genes and markers to the distal part of the human major histocompatibilitycomplex (MHC) region and comparison with the mouse: new in–

sights into the evolution of mammalian genomes. *Genomics* 26: 9–20, 1995.

[40383] 13935. Solinas-Toldo, S.; Lengauer, C.; Fries, R.: Comparative genome map of human and cattle. *Genomics* 27: 489–496, 1995.

[40384] 13936. Olives, B.; Martial, S.; Mattei, M.-G.; Matassi, G.; Rousselet, G.; Ripoche, P.; Cartron, J.-P.; Bailly, P.: Molecular characterization of a new urea transporter in the human kidney. *FEBS Lett.* 386: 156–160, 1996.

[40385] 13937. Ranade, K.; Wu, K.-W.; Hwu, C.-M.; Ting, C.-T.; Pei, D.; Pesich, R.; Hebert, J.; Chen, Y.-D. I.; Pratt, R.; Olshen, R.; Masaki, K.; Risch, N.; Cox, D. R.; Botstein, D.: Genetic variation in the human urea transporter-2 is associated with variation in blood pressure. *Hum. Molec. Genet.* 10: 2157–2164, 2001.

[40386] 13938. Ansel, K. M.; Ngo, V. N.; Hyman, P. L.; Luther, S. A.; Forster, R.; Sedgwick, J. D.; Browning, J. L.; Lipp, M.; Cyster, J. G.: A chemokine-driven positive feedback loop organizes lymphoid follicles. *Nature* 406: 309–314, 2000.

[40387] 13939. Galko, M. J.; Tessier-Lavigne, M.: Function of an axonal chemoattractant modulated by metalloprotease activity. *Science* 289: 1365–1367, 2000.

[40388] 13940. Stein, E.; Tessier-Lavigne, M.: Hierarchical organi-

zation of guidance receptors: silencing of netrin attraction by Slit through a Robo/DCC receptor complex. *Science* 291: 1928–1938, 2001.

- [40389] 13941. Stein, E.; Zou, Y.; Poo, M.; Tessier-Lavigne, M.: Binding of DCC by netrin-1 to mediate axon guidance independent of adenosine A_{2B} receptor activation. *Science* 291: 1976–1982, 2001.
- [40390] 13942. Lubahn, D. B.; Moyer, J. S.; Golding, T. S.; Couse, J. F.; Korach, K. S.; Smithies, O.: Alteration of reproductive function but not prenatal sexual development after insertional disruption of the mouse estrogen receptor gene. *Proc. Nat. Acad. Sci.* 90: 11162–11166, 1993.
- [40391] 13943. Hanna, I. H.; Dawling, S.; Roodi, N.; Guengerich, F. P.; Parl, F. F.: Cytochrome P450 1B1 (CYP1B1) pharmacogenetics: association of polymorphisms with functional differences in estrogen hydroxylation activity. *Cancer Res.* 60: 3440–3444, 2000.
- [40392] 13944. Moore, J. T.; McKee, D. D.; Slentz-Kesler, K.; Moore, L. B.; Jones, S. A.; Horne, E. L.; Su, J.-L.; Kliwer, S. A.; Lehmann, J. M.; Willson, T. M.: Cloning and characterization of human estrogen receptor beta isoforms. *Biochem. Biophys. Res. Commun.* 247: 75–78, 1998.
- [40393] 13945. Mosselman, S.; Polman, J.; Dijkema, R.: ER-beta:

identification and characterization of a novel human estrogen receptor. FEBS Lett. 392:49–53, 1996.

[40394] 13946.Ogawa, S.; Emi, M.; Shiraki, M.; Hosoi, T.; Ouchi, Y.; Inoue, S.: Association of estrogen receptor beta (ESR2) gene polymorphism with blood pressure. J. Hum. Genet. 45: 327–330, 2000.

[40395] 13947.Ogawa, S.; Inoue, S.; Watanabe, T.; Orimo, A.; Hosoi, T.; Ouchi, Y.; Muramatsu, M.: Molecular cloning and characterization of human estrogen receptor beta-cx: a potential inhibitor of estrogen action in human. Nucleic Acids Res. 26: 3505–3512, 1998.

[40396] 13948.Osterlund, M. K.; Gustafsson, J. A.; Keller, E.; Hurd, Y. L.: Estrogen receptor beta (ER-beta) messenger ribonucleic acid (mRNA) expression within the human forebrain: distinct distribution pattern to ER-alpha mRNA. J. Clin. Endocr. Metab. 85: 3840–3846, 2000.

[40397] 13949.Pasquali, D.; Rossi, V.; Esposito, D.; Abbondanza, C.; Puca, G.A.; Bellastella, A.; Sinisi, A. A.: Loss of estrogen receptor beta expression in malignant human prostate cells in primary cultures and in prostate cancer tissues. J. Clin. Endocr. Metab. 86: 2051–2055, 2001.

[40398] 13950.Petersen, D. N.; Tkalcovic, G. T.; Koza-Taylor, P. H.; Turi, T.G.; Brown, T. A.: Identification of estrogen receptor

beta-2, a functional variant of estrogen receptor beta expressed in normal rat tissues. *Endocrinology* 139:1082–1092, 1998.

- [40399] 13951. Rissman, E. F.; Heck, A. L.; Leonard, J. E.; Shupnik, M. A.; Gustafsson, J.-A.: Disruption of estrogen receptor beta gene impairs spatial learning in female mice. *Proc. Nat. Acad. Sci.* 99: 3996–4001, 2002.
- [40400] 13952. Rosenkranz, K.; Hinney, A.; Ziegler, A.; Hermann, H.; Fichter, M.; Mayer, H.; Siegfried, W.; Young, J. K.; Remschmidt, H.; Hebebrand, J.: Systematic mutation screening of the estrogen receptor beta gene in probands of different weight extremes: identification of several genetic variants. *J. Clin. Endocr. Metab.* 83: 4524–4527, 1998.
- [40401] 13953. Speirs, V.; Adams, I. P.; Walton, D. S.; Atkin, S. L.: Identification of wild-type and exon 5 deletion variants of estrogen receptor beta in normal human mammary gland. *J. Clin. Endocr. Metab.* 85: 1601–1605, 2000.
- [40402] 13954. Tsukamoto, K.; Inoue, S.; Hosoi, T.; Orimo, H.; Emi, M.: Isolation and radiation hybrid mapping of dinucleotide repeat polymorphism at the human estrogen receptor beta locus. *J. Hum. Genet.* 43: 73–74, 1998.
- [40403] 13955. Weihua, Z.; Saji, S.; Makinen, S.; Cheng, G.; Jensen, E. V.; Warner, M.; Gustafsson, J.-A.: Estrogen receptor

(ER)-beta, a modulator of ER-alpha in the uterus. Proc. Nat. Acad. Sci. 97: 5936-5941, 2000.

[40404] 13956. Westberg, L.; Baghaei, F.; Rosmond, R.; Hellstrand, M.; Landen, M.; Jansson, M.; Holm, G.; Bjorntorp, P.; Eriksson, E.: Polymorphisms of the androgen receptor gene and the estrogen receptor gene are associated with androgen levels in women. J. Clin. Endocr. Metab. 86: 2562-2568, 2001.

[40405] 13957. Zhu, Y.; Bian, Z.; Lu, P.; Karas, R. H.; Bao, L.; Cox, D.; Hodgins, J.; Shaul, P. W.; Thoren, P.; Smithies, O.; Gustafsson, J.-A.; Mendelsohn, M. E.: Abnormal vascular function and hypertension in mice deficient in estrogen receptor beta. Science 295: 505-508, 2002.

[40406] 13958. DesGroseillers, L.; Lemieux, N.: Localization of a human double-stranded RNA-binding protein gene (STAU) to band 20q13.1 by fluorescence in situ hybridization. Genomics 36: 527-529, 1996.

[40407] 13959. Marion, R. M.; Fortes, P.; Beloso, A.; Dotti, C.; Ortin, J.: A human sequence homologue of stau60 is an RNA-binding protein that is associated with polysomes and localizes to the rough endoplasmic reticulum. Molec. Cell. Biol. 19: 2212-2219, 1999.

[40408] 13960. Wickham, L.; Duchaine, T.; Luo, M.; Nabi, I. R.;

DesGroseillers,L.: Mammalian staufen is a double-stranded-RNA- and tubulin-bindingprotein which localizes to the rough endoplasmic reticulum. Molec.Cell. Biol. 19: 2220-2230, 1999.

[40409] 13961.Lee, J. E.; Hollenberg, S. M.; Snider, L.; Turner, D. L.; Lipnick,N.; Weintraub, H.: Conversion of Xenopus ectoderm into neurons byNeuroD, a basic helix-loop-helix protein. Science 268: 836-844,1995.

[40410] 13962.Liu, M.; Pleasure, S. J.; Collins, A. E.; Noebels, J. L.; Naya,F. J.; Tsai, M.-J.; Lowenstein, D. H.: Loss of BETA2/NeuroD leadsto malformation of the dentate gyrus and epilepsy. Proc. Nat. Acad.Sci. 97: 865-870, 2000.

[40411] 13963.Malecki, M. T.; Jhala, U. S.; Antonellis, A.; Fields, L.; Doria,A.; Orban, T.; Saad, M.; Warram, J. H.; Montminy, M.; Krolewski, A.S.: Mutations in NEUROD1 are associated with the development of type2 diabetes mellitus. Nature Genet. 23: 323-328, 1999.

[40412] 13964.Naya, F. J.; Huang, H.-P.; Qiu, Y.; Mutoh, H.; De-Mayo, F. J.; Leiter,A. B.; Tsai, M.-J.: Diabetes, defective pancreatic morphogenesis,and abnormal enteroendocrine differentiation in BETA2/neurod-deficientmice. Genes Dev. 11: 2323-2334, 1997.

[40413] 13965.Naya, F. J.; Stellrecht, C. M.; Tsai, M. J.: Tissue-

specific regulation of the insulin gene by a novel basic helix-loop-helix transcription factor. *Genes Dev.* 9: 1009–1019, 1995.

- [40414] 13966. Plasilova, M.; Stoilov, I.; Sarfarazi, M.; Kadasi, L.; Ferakova, E.; Ferak, V.: Identification of a single ancestral CYP1B1 mutation in Slovak Gypsies (Roms) affected with primary congenital glaucoma. *J. Med. Genet.* 36: 290–294, 1999.
- [40415] 13967. Schwartzman, M. L.; Balazy, M.; Masferrer, J.; Abraham, N. G.; McGiff, J. C.; Murphy, R. C.: 12(R)-hydroxyicosatetraenoic acid: a cytochrome P450-dependent arachidonate metabolite that inhibits Na⁺, K⁺-ATPase in the cornea. *Proc. Nat. Acad. Sci.* 84: 8125–8129, 1987.
- [40416] 13968. Stoilov, I.; Akarsu, A. N.; Alozie, I.; Child, A.; Barsoum-Homsy, M.; Turacli, M. E.; Or, M.; Lewis, R. A.; Ozdemir, N.; Brice, G.; Aktan, S. G.; Chevrette, L.; Coca-Prados, M.; Sarfarazi, M.: Sequence analysis and homology modeling suggest that primary congenital glaucoma on 2p21 results from mutations disrupting either the hinge region or the conserved core structures of cytochrome P4501B1. *Am. J. Hum. Genet.* 62: 573–584, 1998.
- [40417] 13969. Stoilov, I.; Akarsu, A. N.; Sarfarazi, M.: Identifica-

tion of three different truncating mutations in cytochrome P4501B1 (CYP1B1) as the principal cause of primary congenital glaucoma (buphthalmos) in families linked to the GLC3A locus on chromosome 2p21. *Hum. Molec. Genet.* 6:641–647, 1997.

[40418] 13970. Sutter, T. R.; Tang, Y. M.; Hayes, C. L.; Wo, Y.-Y. P.; Jabs, E. W.; Li, X.; Yin, H.; Cody, C. W.; Greenlee, W. F.: Complete cDNA sequence of a human dioxin-inducible mRNA identifies a new gene subfamily of cytochrome P450 that maps to chromosome 2. *J. Biol. Chem.* 269:13092–13099, 1994.

[40419] 13971. Tang, Y. M.; Wo, Y.-Y. P.; Stewart, J.; Hawkins, A. L.; Griffin, C. A.; Sutter, T. R.; Greenlee, W. F.: Isolation and characterization of the human cytochrome P450 CYP1B1 gene. *J. Biol. Chem.* 271: 28324–28330, 1996.

[40420] 13972. Vincent, A.; Billingsley, G.; Priston, M.; Williams-Lyn, D.; Sutherland, J.; Glaser, T.; Oliver, E.; Walter, M. A.; Heathcote, G.; Levin, A.; Heon, E.: Phenotypic heterogeneity of CYP1B1: mutations in a patient with Peters' anomaly. *J. Med. Genet.* 38: 324–326, 2001.

[40421] 13973. Chen, H. T.; Bhandoola, A.; DiFilippantonio, M. J.; Zhu, J.; Brown, M. J.; Tai, X.; Rogakou, E. P.; Brotz, T. M.; Bonner, W. M.; Ried, T.; Nussenzweig, A.: Response to

RAG-mediated V(D)J cleavage by NBS1 and gamma-H2AX.
Science 290: 1962–1964, 2000.

[40422] 13974. Petersen, S.; Casellas, R.; Reina-San-Martin, B.; Chen, H. T.; Difilippantonio, M. J.; Wilson, P. C.; Hanitsch, L.; Celeste, A.; Muramatsu, M.; Pilch, D. R.; Redon, C.; Ried, T.; Bonner, W. M.; Honjo, T.; Nussenzweig, M. C.; Nussenzweig, A.: AID is required to initiate Nbs1/gamma-H2AX focus formation and mutations at sites of class switching. Nature 414:660–665, 2001.

[40423] 13975. Morasso, M. I.; Yonescu, R.; Griffin, C. A.; Sargent, T. D.: Localization of human DLX8 to chromosome 17q21.3–q22 by fluorescence in situ hybridization. Mammalian Genome 8: 302–303, 1997.

[40424] 13976. Quinn, L. M.; Johnson, B. V.; Nicholl, J.; Sutherland, G. R.; Kalionis, B.: Isolation and identification of homeobox genes from the human placenta including a novel member of the Distal-less family, DLX4. Gene 187:55–61, 1997.

[40425] 13977. Boddy, M. N.; Howe, K.; Etkin, L. D.; Solomon, E.; Freemont, P. S.: PIC 1, a novel ubiquitin-like protein which interacts with the PML component of a multiprotein complex that is disrupted in acute promyelocytic leukaemia. Oncogene 13: 971–982, 1996.

[40426] 13978. Desterro, J. M. P.; Rodriguez, M. S.; Hay, R. T.:

SUMO-1 modification of I-kappa-B-alpha inhibits NF-kappa-B activation. *Molec. Cell* 2:233-239, 1998.

[40427] 13979. Lapenta, V.; Chiurazzi, P.; van der Spek, P.; Pizzuti, A.; Hanaoka, F.; Brahe, C.: SMT3A, a human homologue of the *S. cerevisiae* SMT3 gene, maps to chromosome 21qter and defines a novel gene family. *Genomics* 40:362-366, 1997.

[40428] 13980. Mao, Y.; Sun, M.; Desai, S. D.; Liu, L. F.: SUMO-1 conjugation to topoisomerase I: a possible repair response to topoisomerase-mediated DNA damage. *Proc. Nat. Acad. Sci.* 97: 4046-4051, 2000.

[40429] 13981. Okura, T.; Gong, L.; Kamitani, T.; Wada, T.; Okura, I.; Wei, C.-F.; Chang, H.-M.; Yeh, E. T. H.: Protection against Fas/APO-1- and tumor necrosis factor-mediated cell death by a novel protein, sentrin. *J. Immun.* 157: 4277-4281, 1996.

[40430] 13982. Shen, Z.; Pardington-Purtymun, P. E.; Comeaux, J. C.; Moyzis, R. K.; Chen, D. J.: UBL1, a human ubiquitin-like protein associating with human RAD51/RAD52 proteins. *Genomics* 36: 271-279, 1996.

[40431] 13983. Bengtsson, E.; Neame, P. J.; Heinegard, D.; Sommarin, Y.: The primary structure of a basic leucine-rich repeat protein, PRELP, found in connective tissues. *J. Biol.*

Chem. 270: 25639–25644, 1995.

- [40432] 13984. Grover, J.; Chen, X.-N.; Korenberg, J. R.; Recklies, A. D.; Roughley, P. J.: The gene organization, chromosome location, and expression of a 55-kDa matrix protein (PRELP) of human articular cartilage. *Genomics* 38:109–117, 1996.
- [40433] 13985. Bigg, H. F.; Shi, Y. E.; Liu, Y. E.; Steffensen, B.; Overall, C. M.: Specific, high affinity binding of tissue inhibitor of metalloproteinases-4 (TIMP-4) to the COOH-terminal hemopexin-like domain of human gelatinase A. *J. Biol. Chem.* 272: 15496–15500, 1997.
- [40434] 13986. Greene, J.; Wang, M.; Liu, Y. E.; Raymond, L. A.; Rosen, C.; Shi, Y. E.: Molecular cloning and characterization of human tissue inhibitor of metalloproteinase 4. *J. Biol. Chem.* 271: 30375–30380, 1996.
- [40435] 13987. Leco, K. J.; Apte, S. S.; Taniguchi, G. T.; Hawkes, S. P.; Khokha, R.; Schultz, G. A.; Edwards, D. R.: Murine tissue inhibitor of metalloproteinases-4 (Timp-4): cDNA isolation and expression in adult mouse tissues. *FEBS Lett.* 401: 213–217, 1997.
- [40436] 13988. Olson, T. M.; Hirohata, S.; Ye, J.; Leco, K.; Seldin, M. F.; Apte, S. S.: Cloning of the human tissue inhibitor of metalloproteinase-4 gene (TIMP4) and localization of the

TIMP4 and Timp4 gene to human chromosome 3p25 and mouse chromosome 6, respectively. Genomics 51:148–151, 1998.

[40437] 13989.Wang, M.; Liu, Y. E.; Greene, J.; Sheng, S.; Fuchs, A.; Rosen, E. M.; Shi, Y. E.: Inhibition of tumor growth and metastasis of human breast cancer cells transfected with tissue inhibitor of metalloproteinase4. Oncogene 14: 2767–2774, 1997.

[40438] 13990.Hsu, L. C.; Chang, W.-C.: Sequencing and expression of the human ALDH8 encoding a new member of the aldehyde dehydrogenase family. Gene 174:319–322, 1996.

[40439] 13991.Hsu, L. C.; Chang, W.-C.; Lin, S. W.; Yoshida, A.: Cloning and characterization of genes encoding four additional human aldehyde dehydrogenase isozymes. Adv. Exp. Med. Biol. 372: 159–168, 1995.

[40440] 13992.Rawson, R. B.; Zelenski, N. G.; Nijhawan, D.; Ye, J.; Sakai, J.; Hasan, M. T.; Chang, T. Y.; Brown, M. S.; Goldstein, J. L.: Complementation cloning of S2P, a gene encoding a putative metalloprotease required for intramembrane cleavage of SREBPs. Molec. Cell 1: 47–57, 1997.

[40441] 13993.Ye, J.; Rawson, R. B.; Komuro, R.; Chen, X.; Dave, U. P.; Prywes, R.; Brown, M. S.; Goldstein, J. L.: ER stress in–

duces cleavage of membrane-bound ATF6 by the same proteases that process SREBPs. *Molec.Cell* 6: 1355–1364, 2000.

[40442] 13994. Zelenski, N. G.; Rawson, R. B.; Brown, M. S.; Goldstein, J. L.: Membrane topology of S2P, a protein required for intramembraneous cleavage of sterol regulatory element-binding proteins. *J. Biol.Chem.* 274: 21973–21980, 1999.

[40443] 13995. Kim, V. N.; Kataoka, N.; Dreyfuss, G.: Role of the nonsense-mediated decay factor hUpf3 in the splicing-dependent exon-exon junction complex. *Science* 293:1832–1836, 2001.

[40444] 13996. Lykke-Andersen, J.; Shu, M.-D.; Steitz, J. A.: Human Upf protein target an mRNA for nonsense-mediated decay when bound downstream of a termination codon. *Cell* 103: 1121–1131, 2000.

[40445] 13997. Scott, A. F.: Personal Communication. Baltimore, Md. 2/9/2001.

[40446] 13998. Serin, G.; Gersappe, A.; Black, J. D.; Aronoff, R.; Maquat, L.E.: Identification and characterization of human orthologues to *Saccharomyces cerevisiae* Upf2 protein and Upf3 protein (*Caenorhabditis elegans* SMG-4). *Molec.Cell. Biol.* 21: 209–223, 2001.

- [40447] 13999.de Leeuw, B.; Balemans, M.; Geurts van Kessel, A.: A novel Kruppel-associated box containing the SSX gene (SSX3) on the human X chromosome is not implicated in t(X;18)-positive synovial sarcomas. *Cytogenet. Cell Genet.* 73: 179–183, 1996.
- [40448] 14000.Gure, A. O.; Tureci, O.; Sahin, U.; Tsang, S.; Scanlan, M. J.; Jager, E.; Knuth, A.; Pfreundschuh, M.; Old, L. J.; Chen, Y.-T.: SSX: a multigene family with several members transcribed in normal testis and human cancer. *Int. J. Cancer* 72: 965–971, 1997.
- [40449] 14001.Kawai, A.; Woodruff, J.; Healey, J. H.; Brennan, M. F.; Antonescu, C. R.; Ladanyi, M.: SYT-SSX gene fusion as a determinant of morphology and prognosis in synovial sarcoma. *New Eng. J. Med.* 338: 153–160, 1998.
- [40450] 14002.Skytting, B.; Nilsson, G.; Brodin, B.; Xie, Y.; Lundeberg, J.; Uhlen, M.; Larsson, O.: A novel fusion gene, SYT-SSX4, in synovial sarcoma. (Letter) *J. Nat. Cancer Inst.* 91: 974–975, 1999.
- [40451] 14003.Yang, K.; Lui, W.-O.; Xie, Y.; Zhang, A.; Skytting, B.; Mandahl, N.; Larsson, C.; Larsson, O.: Co-existence of SYT-SSX1 and SYT-SSX2 fusions in synovial sarcomas. *Oncogene* 21: 4181–4190, 2002.
- [40452] 14004.Piccini, M.; Vitelli, F.; Seri, M.; Galletta, L. J. V.;

Moran, O.; Bulfone, A.; Banfi, S.; Pober, B.; Renieri, A.: KCNE1-like gene is deleted in AMME contiguous gene syndrome: identification and characterization of the human and mouse homologs. *Genomics* 60: 251–257, 1999.

[40453] 14005. Koda, T.; Kakinuma, M.: Molecular cloning of a cDNA encoding a novel small GTP-binding protein. *FEBS Lett.* 328: 21–24, 1993.

[40454] 14006. Scott, A. F.: Personal Communication. Baltimore, Md. 5/14/2001.

[40455] 14007. Zheng, J. Y.; Koda, T.; Arimura, Y.; Kishi, M.; Kakinuma, M.: Structure and expression of the mouse S10 gene. *Biochim. Biophys. Acta* 1351: 47–50, 1997.

[40456] 14008. Tipnis, S. R.; Hooper, N. M.; Hyde, R.; Karran, E.; Christie, G.; Turner, A. J.: A human homolog of an angiotensin-converting enzyme: cloning and functional expression as a captopril-insensitive carboxypeptidase. *J. Biol. Chem.* 275: 33238–33243, 2000.

[40457] 14009. Berta, P.; Hawkins, J. R.; Sinclair, A. H.; Taylor, A.; Griffiths, B. L.; Goodfellow, P. N.; Fellous, M.: Genetic evidence equating SRY and the testis-determining factor. *Nature* 348: 448–450, 1990.

[40458] 14010. Disteche, C. M.; Casanova, M.; Saal, H.; Friedman, C.; Sybert, V.; Graham, J.; Thuline, H.; Page, D.; Fellous, M.:

Small deletions of the short arm of the Y chromosome in 46,XY females. *Proc. Nat. Acad. Sci.* 83: 7841–7844, 1986.

[40459] 14011. De Plaen, E.; De Backer, O.; Arnaud, D.; Bonjean, B.; Chomez, P.; Martelange, V.; Avner, P.; Baldacci, P.; Babinet, C.; Hwang, S.-Y.; Knowles, B.; Boon, T.: A new family of mouse genes homologous to the human MAGE genes. *Genomics* 55: 176–184, 1999.

[40460] 14012. Andersson, M.; Page, D. C.; de la Chapelle, A.: Chromosome Y-specific DNA is transferred to the short arm of X chromosome in human XX males. *Science* 233: 786–788, 1986.

[40461] 14013. Burgoyne, P. S.; Levy, E. R.; McLaren, A.: Spermatogenic failure in male mice lacking H-Y antigen. *Nature* 320: 170–172, 1986.

[40462] 14014. Ferguson-Smith, M. A.: X-Y chromosomal interchange in the aetiology of true hermaphroditism and of XX Klinefelter's syndrome. *Lancet* II: 475–476, 1966.

[40463] 14015. Simpson, E.; Chandler, P.; Goulmy, E.; Distèche, C. M.; Ferguson-Smith, M. A.; Page, D. C.: Separation of the genetic loci for the H-Y antigen and for testis determination on human Y chromosome. *Nature* 326: 876–878, 1987.

[40464] 14016. Tiepolo, L.; Zuffardi, O.: Localization of factors

controllingspermatogenesis in the nonfluorescent portion of the human Y chromosomelong arm. Hum. Genet. 34: 119–124, 1976.

- [40465] 14017.Aalfs, C. M.; van den Berg, H.; Barth, P. G.; Hennekam, R. C. M.: The Hoyeraal–Hreidarsson syndrome: the fourth case of a separate entity with prenatal growth retardation, progressive pancytopeniaand cerebellar hypoplasia. Europ. J. Pediat. 154: 304–308, 1995.
- [40466] 14018.Devriendt, K.; Matthijs, G.; Legius, E.; Schollen, E.; Blockmans,D.; van Geet, C.; Degreef, H.; Cassiman, J.–J.; Fryns, J.–P.: SkewedX–chromosome inactivation in female carriers of dyskeratosis congenita. Am.J. Hum. Genet. 60: 581–587, 1997.
- [40467] 14019.Hassock, S.; Vetrie, D.; Giannelli, F.: Mapping and characterizationof the X–linked dyskeratosis congenita (DKC) gene. Genomics 55:21–27, 1999.
- [40468] 14020.Heiss, N. S.; Knight, S. W.; Vulliamy, T. J.; Klauck, S. M.; Wiemann,S.; Mason, P. J.; Poustka, A.; Dokal, I.: X–linked dyskeratosis congenita is caused by mutations in a highly conserved gene with putative nucleolarfunctions. Nature Genet. 19: 32–38, 1998.
- [40469] 14021.Heiss, N. S.; Megarbane, A.; Klauck, S. M.; Kreuz, F. R.; Makhoul,E.; Majewski, F.; Poustka, A.: One novel and

two recurrent missenseDKC1 mutations in patients with dyskeratosis congenita (DKC). Genet.Counsel. 12: 129–136, 2001.

[40470] 14022.Abedinia, M.; Layfield, R.; Jones, S. M.; Nixon, P. F.; Mattick,J. S.: Nucleotide and predicted amino acid sequence of a cDNA clone encoding part of human transketolase. Biochem. Biophys. Res. Commun. 183:1159–1166, 1992.

[40471] 14023.Coy, J. F.; Dubel, S.; Kioschis, P.; Thomas, K.; Micklem, G.; Delius,H.; Poustka, A.: Molecular cloning of tissue-specific transcripts of a transketolase-related gene: implications for the evolution of new vertebrate genes. Genomics 32: 309–316, 1996.

[40472] 14024.McCool, B. A.; Plonk, S. G.; Martin, P. R.; Singleton, C. K.:Cloning of human transketolase cDNAs and comparison of the nucleotide sequence of the coding region in Wernicke–Korsakoff and non–Wernicke–Korsakoff individuals. J. Biol. Chem. 268: 1397–1404, 1993.

[40473] 14025.van der Maarel, S. M.; Scholten, I. H. J. M.; Huber, I.; Philippe,C.; Suijkerbuijk, R. F.; Gilgenkrantz, S.; Kere, J.; Cremers, F. P.M.; Ropers, H.–H.: Cloning and characterization of DXS6673E, a candidate gene for X-linked mental

retardation in Xq13.1. Hum. Molec. Genet. 5:887–897, 1996.

- [40474] 14026. Jacobi, F. K.; Broghammer, M.; Pesch, K.; Zrenner, E.; Berger, W.; Meindl, A.; Pusch, C. M.: Physical mapping and exclusion of GPR34 as the causative gene for congenital stationary night blindness type 1. Hum. Genet. 107: 89–91, 2000.
- [40475] 14027. Marchese, A.; Sawzdargo, M.; Nguyen, T.; Cheng, R.; Heng, H. H. Q.; Nowak, T.; Im, D.-S.; Lynch, K. R.; George, S. R.; O'Dowd, B. F.: Discovery of three novel orphan G-protein-coupled receptors. Genomics 56:12–21, 1999.
- [40476] 14028. Schöneberg, T.; Schulz, A.; Grosse, R.; Schade, R.; Henklein, P.; Schultz, G.; Gudermann, T.: A novel subgroup of class I G-protein-coupled receptors. Biochim. Biophys. Acta 1446: 57–70, 1999.
- [40477] 14029. Sanchis, D.; Fleury, C.; Chomiki, N.; Goubern, M.; Huang, Q.; Neverova, M.; Gregoire, F.; Easlick, J.; Raimbault, S.; Levi-Meyrueis, C.; Miroux, B.; Collins, S.; Seldin, M.; Richard, D.; Warden, C.; Bouillaud, F.; Ricquier, D.: BMCP1, a novel mitochondrial carrier with high expression in the central nervous system of humans and rodents, and respiration uncoupling activity in recombinant yeast. J.

Biol. Chem. 273: 34611–34615,1998.

- [40478] 14030. Blanco, P.; Sargent, C. A.; Boucher, C. A.; Mitchell, M.; Affara, N. A.: Conservation of PCDHX in mammals; expression of X/Y genes predominantly in brain. *Mammalian Genome* 11: 906–914, 2000.
- [40479] 14031. Ciccodicola, A.; D'Esposito, M.; Esposito, T.; Gianfrancesco, F.; Migliaccio, C.; Miano, M. G.; Matarazzo, M. R.; Vacca, M.; Franze, A.; Cuccurese, M.; Cocchia, M.; Curci, A.; and 9 others: Differentially regulated and evolved genes in the fully sequenced Xq/Yq pseudoautosomal region. *Hum. Molec. Genet.* 9: 395–401, 2000.
- [40480] 14032. Mumm, S.; Molini, B.; Terrell, J.; Srivastava, A.; Schlessinger, D.: Evolutionary features of the 4-Mb Xq21.3 XY homology region revealed by a map at 60-kb resolution. *Genome Res.* 7: 307–314, 1997.
- [40481] 14033. Schwartz, A.; Chan, D. C.; Brown, L. G.; Alagappan, R.; Pettay, D.; Distech, C.; McGillivray, B.; de la Chapelle, A.; Page, D. C.: Reconstructing hominid Y evolution: X-homologous block, created by X–Y transposition, was disrupted by Yp inversion through LINE–LINE recombination. *Hum. Molec. Genet.* 7: 1–11, 1998.
- [40482] 14034. Tilford, C. A.; Kuroda-Kawaguchi, T.; Skaletsky, H.; Rozen, S.; Brown, L. G.; Rosenberg, M.; McPherson, J. D.;

Wylie, K.; Sekhon, M.; Kucaba, T. A.; Waterston, R. H.; Page, D. C.: A physical map of the human Y chromosome. *Nature* 409: 943–945, 2001.

[40483] 14035. Roberts, R. G.; Kendall, E.; Vetrie, D.; Bobrow, M.: Sequence and chromosomal location of a human homologue of LRPR1, an FSH primary response gene. *Genomics* 37: 122–124, 1996.

[40484] 14036. Slegtenhorst–Eegdeeman, K. E.; Post, M.; Baarends, W. M.; Themmen, A. P. N.; Grootegoed, J. A.: Regulation of gene expression in Sertoli cells by follicle-stimulating hormone (FSH): cloning and characterization of LRPR1, a primary response gene encoding a leucine-rich protein. *Molec. Cell. Endocr.* 108: 115–124, 1995.

[40485] 14037. Batch, J. A.; Evans, B. A. J.; Hughes, I. A.; Patterson, M. N.: Mutations of the androgen receptor gene identified in perineal hypospadias. *J. Med. Genet.* 30: 198–201, 1993.

[40486] 14038. Borkhardt, A.; Repp, R.; Haas, O. A.; Leis, T.; Harbott, J.; Kreuder, J.; Hammermann, J.; Henn, T.; Lampert, F.: Cloning and characterization of AFX, the gene that fuses to MLL in acute leukemias with a t(X;11)(q13;q23). *Oncogene* 14:195–202, 1997.

[40487] 14039. Chatterjee, A.; Faust, C. J.; Herman, G. E.: Genetic

and physical mapping of the biglycan gene on the mouse X chromosome. *Mammalian Genome* 4: 33–36, 1993.

[40488] 14040. Das, S.; Metzenberg, A.; Pai, G. S.; Gitschier, J.: Mutational analysis of the biglycan gene excludes it as a candidate for X-linked dominant chondrodysplasia punctata, dyskeratosis congenita, and incontinentia pigmenti. (Letter) *Am. J. Hum. Genet.* 54: 922–925, 1994.

[40489] 14041. Fisher, L. W.; Heegaard, A.-M.; Vetter, U.; Vogel, W.; Just, W.; Termine, J. D.; Young, M. F.: Human biglycan gene: putative promoter, intron–exon junctions, and chromosomal localization. *J. Biol. Chem.* 266:14371–14377, 1991.

[40490] 14042. Fisher, L. W.; Termine, J. D.; Young, M. F.: Deduced–protein sequence of bone small proteoglycan I (biglycan) shows homology with proteoglycan II (decorin) and several nonconnective tissue proteins in a variety of species. *J. Biol. Chem.* 264: 4571–4576, 1989.

[40491] 14043. Geerkens, C.; Vetter, U.; Just, W.; Fedarko, N. S.; Fisher, L. W.; Young, M. F.; Termine, J. D.; Gehron Robey, P.; Woehle, D.; Vogel, W.: The X–chromosomal human biglycan gene BGN is subject to X inactivation but is transcribed like an X–Y homologous gene. *Hum. Genet.* 96:44–52, 1995.

- [40492] 14044.McBride, O. W.; Fisher, L. W.; Young, M. F.: Localization of PGI(biglycan, BGN) and PGII (decorin, DCN, PG-40) genes on human chromosomesXq13-qter and 12q, respectively. *Genomics* 6: 219-225, 1990.
- [40493] 14045.Traupe, H.; van den Ouweland, A. M. W.; van Oost, B. A.; Vogel,W.; Vetter, U.; Warren, S. T.; Rocchi, M.; Darli-son, M. G.; Ropers,H.-H.: Fine mapping of the human biglycan (BGN) gene within the Xq28region employing a hybrid cell panel. *Genomics* 13: 481-483, 1992.
- [40494] 14046.Wegrowski, Y.; Pillarisetti, J.; Danielson, K. G.; Suzuki, S.;Iozzo, R. V.: The murine biglycan: complete cDNA cloning, genomicorganization, promoter function, and expression. *Genomics* 30: 8-17,1995.
- [40495] 14047.Xu, T.; Bianco, P.; Fisher, L. W.; Longenecker, G.; Smith, E.;Goldstein, S.; Bonadio, J.; Boskey, A.; Heegaard, A.-M.; Sommer, B.;Satomura, K.; Dominguez, P.; Zhao, C.; Kulkarni, A. B.; Robey, P.G.; Young, M. F.: Targeted disruption of the biglycan gene leadsto an osteoporosis-like phenotype in mice. *Nature Genet.* 20: 78-82,1998.
- [40496] 14048.Yoshida, K.; Sugano, S.: Identification of a novel protocadheringene (PCDH11) on the human XY homology region in Xq21.3. *Genomics* 62:540-543, 1999.
- [40497] 14049.de la Chapelle, A.; Sankila, E.-M.; Lindlof, M.; Aula,

P.; Norio,R.: Norrie disease caused by a gene deletion allowing carrier detectionand prenatal diagnosis. Clin. Genet. 28: 317–320, 1985.

- [40498] 14050.Sims, K. B.; de la Chapelle, A.; Norio, R.; Sankila, E.–M.; Hsu,Y.–P. P.; Rinehart, W. B.; Corey, T. J.; Ozelius, L.; Powell, J. F.;Bruns, G.; Gusella, J. F.; Murphy, D. L.; Breakefield, X. O.: Monoamineoxidase deficiency in males with an X chromosome deletion. Neuron 2:1069–1076, 1989.
- [40499] 14051.Grino, P. B.; Griffin, J. E.; Cushard, W. G., Jr.; Wilson, J.D.: A mutation of the androgen receptor associated with partial androgenresistance, familial gynecomastia, and fertility. J. Clin. Endocr.Metab. 66: 754–761, 1988.
- [40500] 14052.Holterhus, P.–M.; Sinnecker, G. H. G.; Hiort, O.: Phenotypicdiversity and testosterone–induced normalization of mutant L712F androgenreceptor function in a kindred with androgen insensitivity. J. Clin.Endocr. Metab. 85: 3245–3250, 2000.
- [40501] 14053.Hughes, I. A.; Evans, B. A. J.: Complete androgen insensitivitysyndrome characterized by increased concentration of a normal androgenreceptor in genital skin fibroblasts. J. Clin. Endocr. Metab. 63:309–315, 1986.
- [40502] 14054.Imperato–McGinley, J.; Ip, N. Y.; Gautier, T.;

Neuweiler, J.; Gruenspan, H.; Liao, S.; Chang, C.; Balazs, I.: DNA linkage analysis and studies of the androgen receptor gene in a large kindred with complete androgen insensitivity. *Am. J. Med. Genet.* 36: 104–108, 1990.

[40503] 14055. Jukier, L.; Kaufman, M.; Pinsky, L.; Peterson, R. E.: Partial androgen resistance associated with secondary 5- α -reductase deficiency: identification of a novel qualitative androgen receptor defect and clinical implications. *J. Clin. Endocr. Metab.* 59: 679–688, 1984.

[40504] 14056. Kaufman, M.; Pinsky, L.; Bowin, A.; Au, M. W. S.: Familial external genital ambiguity due to a transformation defect of androgen-receptor complexes that is expressed with 5- α -dihydrotestosterone and the synthetic androgen methyltrienolone. *Am. J. Med. Genet.* 18: 493–507, 1984.

[40505] 14057. Kaufman, M.; Straisfeld, C.; Pinsky, L.: Male pseudohermaphroditism presumably due to target organ unresponsiveness to androgens: deficient 5- α -dihydrotestosterone binding in cultured skin fibroblasts. *J. Clin. Invest.* 58: 345–350, 1976.

[40506] 14058. Keenan, B. S.; Meyer, W. J., III; Hadjian, A. J.; Jones, H. W.; Migeon, C. J.: Syndrome of androgen insensitivity in man: absence of 5- α -dihydrotestosterone binding

protein in skin fibroblasts. J.Clin. Endocr. 38: 1143–1146, 1974.

[40507] 14059.Liao, S.; Witte, D.: Autoimmune anti-androgen-receptor antibodies in human serum. Proc. Nat. Acad. Sci. 82: 8345–8348, 1985.

[40508] 14060.Lin, S.-Y.; Ohno, S.: The binding of androgen receptor to DNA and RNA. Biochim. Biophys. Acta 654: 181–186, 1981.

[40509] 14061.Aradhya, S.; Bardaro, T.; Galgoczy, P.; Yamagata, T.; Esposito, T.; Patlan, H.; Ciccodicola, A.; Munnich, A.; Kenwrick, S.; Platzer, M.; D'Urso, M.; Nelson, D. L.: Multiple pathogenic and benign genomic rearrangements occur at a 35 kb duplication involving the NEMO and LAGE2 genes. Hum. Molec. Genet. 10: 2557–2567, 2001.

[40510] 14062.Aradhya, S.; Courtois, G.; Rajkovic, A.; Lewis, R. A.; Levy, M.; Israel, A.; Nelson, D. L.: Atypical forms of incontinentia pigmenti in male individuals result from mutations of a cytosine tract in exon 10 of NEMO (IKK- γ). Am. J. Hum. Genet. 68: 765–771, 2001.

[40511] 14063.Aradhya, S.; Woffendin, H.; Jakins, T.; Bardaro, T.; Esposito, T.; Smahi, A.; Shaw, C.; Levy, M.; Munnich, A.; D'Urso, M.; Lewis, R. A.; Kenwrick, S.; Nelson, D. L.: A recurrent deletion in the ubiquitously expressed NEMO

(IKK- γ) gene accounts for the vast majority of *continentia pigmenti* mutations. *Hum. Molec. Genet.* 10: 2171–2179, 2001.

- [40512] 14064. Doffinger, R.; Smahi, A.; Bessia, C.; Geissmann, F.; Feinberg, J.; Durandy, A.; Bodemer, C.; Kenwrick, S.; Dupuis-Girod, S.; Blanche, S.; Wood, P.; Rabia, S. H.; and 16 others: X-linked anhidrotic ectodermal dysplasia with immunodeficiency is caused by impaired NF- κ B signaling. *Nature Genet.* 27: 277–285, 2001.
- [40513] 14065. Jain, A.; Ma, C. A.; Liu, S.; Brown, M.; Cohen, J.; Strober, W.: Specific missense mutations in NEMO result in hyper-IgM syndrome with hypohidrotic (sic) ectodermal dysplasia. *Nature Immun.* 2: 223–228, 2001.
- [40514] 14066. Jin, D. Y.; Jeang, K. T.: Isolation of full-length cDNA and chromosomal localization of human NF- κ B modulator NEMO to Xq28. *J. Biomed. Sci.* 6: 115–120, 1999.
- [40515] 14067. Kosaki, K.; Shimasaki, N.; Fukushima, H.; Hara, M.; Ogata, T.; Matsuo, N.: Female patient showing hypohidrotic ectodermal dysplasia and immunodeficiency (HED-ID). (Letter) *Am. J. Hum. Genet.* 69: 664–665, 2001.
- [40516] 14068. Li, Q.; Van Antwerp, D.; Mercurio, F.; Lee, K.-F.; Verma, I. M.: Severe liver degeneration in mice lacking the I- κ B kinase 2 gene. *Science* 284: 321–325, 1999.

- [40517] 14069.Li, Y.; Kang, J.; Friedman, J.; Tarassishin, L.; Ye, J.; Kovalenko,A.; Wallach, D.; Horwitz, M. S.: Identification of a cell protein(FIP-3) as a modulator of NF-kappa-B activity and as a target of anadenovirus inhibitor of tumor necrosis factor alpha-induced apoptosis. Proc.Nat. Acad. Sci. 96: 1042-1047, 1999.
- [40518] 14070.Makris, C.; Godfrey, V. L.; Krahn-Senftleben, G.; Takahashi, T.;Roberts, J. L.; Schwarz, T.; Feng, L.; Johnson, R. S.; Karin, M.:Female mice heterozygous for IKK-gamma/NEMO deficiencies develop a dermatopathy similar to the human X-linked disorder incontinentia pigmenti. Molec. Cell 5: 969-979, 2000.
- [40519] 14071.May, M. J.; D'Acquisto, F.; Madge, L. A.; Glockner, J.; Pober,J. S.; Ghosh, S.: Selective inhibition of NF-kappa-B activation by a peptide that blocks the interaction of NEMO with the I-kappa-B kinase complex. Science 289: 1550-1554, 2000.
- [40520] 14072.Roberts, J. L.; Morrow, B.; Vega-Rich, C.; Salafia, C. M.; Nitowsky,H. M.: Incontinentia pigmenti in a newborn male infant with DNA confirmation. Am.J. Med. Genet. 75: 159-163, 1998.
- [40521] 14073.Rothwarf, D. M.; Zandi, E.; Natoli, G.; Karin, M.: IKK-gamma is an essential regulatory subunit of the I-

kappa-B kinase complex. *Nature* 395:297–300, 1998.

[40522] 14074. Rudolph, D.; Yeh, W.-C.; Wakeham, A.; Rudolph, B.; Nallainathan, D.; Potter, J.; Elia, A. J.; Mak, T. W.: Severe liver degeneration and lack of NF-kappa-B activation in NEMO/IKK-gamma-deficient mice. *Genes Dev.* 14: 854–862, 2000.

[40523] 14075. Schmidt-Suppran, M.; Bloch, W.; Courtois, G.; Ad-dicks, K.; Israel, A.; Rajewsky, K.; Pasparakis, M.: NEMO/IKK-gamma-deficient mice model incontinentia pigmenti. *Molec. Cell* 5: 981–992, 2000.

[40524] 14076. The International Incontinentia Pigmenti Consortium: Genomic rearrangement in NEMO impairs NF-kappa-B activation and is a cause of incontinentia pigmenti. *Nature* 405: 466–472, 2000.

[40525] 14077. The International IP Consortium: Survival of male patients with incontinentia pigmenti carrying a lethal mutation can be explained by somatic mosaicism or Klinefelter syndrome. *Am. J. Hum. Genet.* 69:1210–1217, 2001.

[40526] 14078. Yamaoka, S.; Courtois, G.; Bessia, C.; Whiteside, S. T.; Weil, R.; Agou, F.; Kirk, H. E.; Kay, R. J.; Israel, A.: Complementation cloning of NEMO, a component of the I-kappa-B kinase complex essential for NF-kappa-B activation. *Cell* 93: 1231–1240, 1998.

- [40527] 14079.Zonana, J.; Elder, M. E.; Schneider, L. C.; Orlow, S. J.; Moss,C.; Golabi, M.; Shapira, S. K.; Farndon, P. A.; Wara, D. W.; Emmal,S. A.; Ferguson, B. M.: A novel X-linked disorder of immune deficiencyand hypohidrotic ectodermal dysplasia is allelic to incontinentiapigmenti and due to mutations in IKK-gamma (NEMO). *Am. J. Hum. Genet.* 67:1555–1562, 2000.
- [40528] 14080.Bauer, M. F.; Gempel, K.; Reichert, A. S.; Rappold, G. A.; Lichtner,P.; Gerbitz, K.–D.; Neupert, W.; Brunner, M.; Hofmann, S.: Geneticand structural characterization of the human mitochondrial inner membranetranslocase. *J. Molec. Biol.* 289: 69–82, 1999.
- [40529] 14081.Moller, D. E.; Xia, C. H.; Tang, W.; Zhu, A. X.; Jakubowski, M.: Human rsk isoforms: cloning and characterization of tissue-specificexpression. *Am. J. Physiol.* 266: C351–C359, 1994.
- [40530] 14082.Zhao, Y.; Bjorbaek, C.; Weremowicz, S.; Morton, C. C.; Moller,D. E.: RSK3 encodes a novel pp90rsk isoform with a unique N-terminalsequence: growth factor-stimulated kinase function and nuclear translocation. *Molec.Cell. Biol.* 15: 4353–4363, 1995.
- [40531] 14083.Au, H. C.; Seo, B. B.; Matsuno-Yagi, A.; Yagi, T.; Scheffler, I.E.: The NDUFA1 gene product (MWFE protein)

is essential for activity of complex I in mammalian mitochondria. Proc. Nat. Acad. Sci. 96:4354–4359, 1999.

- [40532] 14084. Zhuchenko, O.; Wehnert, M.; Bailey, J.; Sun, Z. S.; Lee, C. C.: Isolation, mapping, and genomic structure of an X-linked gene for a subunit of human mitochondrial complex I. Genomics 37: 281–288, 1996.
- [40533] 14085. Liu, Z.; Sun, C.; Olejniczak, E. T.; Meadows, R. P.; Betz, S. F.; Oost, T.; Herrmann, J.; Wu, J. C.; Fesik, S. W.: Structural basis for binding of Smac/DIABLO to the XIAP BIR3 domain. Nature 408:1004–1008, 2000.
- [40534] 14086. Srinivasula, S. M.; Hegde, R.; Saleh, A.; Datta, P.; Shiozaki, E.; Chai, J.; Lee, R.-A.; Robbins, P. D.; Fernandes-Alnemri, T.; Shi, Y.; Alnemri, E. S.: A conserved XIAP-interaction motif in caspase-9 and Smac/DIABLO regulates caspase activity and apoptosis. Nature 410:112–116, 2001.
- [40535] 14087. Bomont, P.; Cavalier, L.; Blondeau, F.; Ben Hamida, C.; Belal, S.; Tazir, M.; Demir, E.; Topaloglu, H.; Korinthenberg, R.; Tuysuz, B.; Landrieu, P.; Hentati, F.; Koenig, M.: The gene encoding gigaxonin, a new member of the cytoskeletal BTB/kelch repeat family, is mutated in giant axonal neuropathy. Nature Genet. 26: 370–374, 2000.
- [40536] 14088. Treiber-Held, S.; Budjarjo-Welim, H.; Riemann, D.;

Richter, J.;Kretzschmar, H. A.; Hanefeld, F.: Giant axonal neuropathy: a generalized disorder of intermediate filaments with longitudinal grooves in the hair. *Neuropediatrics* 25: 89–93, 1994.

[40537] 14089.Lieuallen, K.; Pennacchio, L. A.; Park, M.; Myers, R. M.; Lennon, G. G.: Cystatin B-deficient mice have increased expression of apoptosis and glial activation genes. *Hum. Molec. Genet.* 10: 1867–1871, 2001.

[40538] 14090.Mazarib, A.; Xiong, L.; Neufeld, M. Y.; Birnbaum, M.; Korczyn, A. D.; Pandolfo, M.; Berkovic, S. F.: Unverricht-Lundborg disease in a five-generation Arab family: instability of dodecamer repeats. *Neurology* 57:1050–1054, 2001.

[40539] 14091.Pennacchio, L. A.; Bouley, D. M.; Higgins, K. M.; Scott, M. P.; Noebels, J. L.; Myers, R. M.: Progressive ataxia, myoclonic epilepsy and cerebellar apoptosis in cystatin B-deficient mice. *Nature Genet.* 20:251–258, 1998.

[40540] 14092.Virtaneva, K.; D'Amato, E.; Miao, J.; Koskiniemi, M.; Norio, R.; Avanzini, G.; Franceschetti, S.; Michelucci, R.; Tassinari, C. A.; Omer, S.; Pennacchio, L. A.; Myers, R. M.; Dieguez-Lucena, J. L.; Krahe, R.; de la Chapelle, A.; Lehesjoki, A.-E.: Unstable minisatellite expansion causing recessively inherited myoclonus epilepsy, EPM1. *Nature*–

Genet. 15: 393–396, 1997.

- [40541] 14093.Cannizzaro, L. A.; Chen, Y. Q.; Rafi, M. A.; Wenger, D. A.: Regional mapping of the human galactocerebrosidase gene (GALC) to 14q31 by in situ hybridization. Cytogenet. Cell Genet. 66: 244–245, 1994.
- [40542] 14094.De Gasperi, R.; Sosa, M. A. G.; Sartorato, E. L.; Battistini, S.; MacFarlane, H.; Gusella, J. F.; Krivit, W.; Kolodny, E. H.: Molecular heterogeneity of late-onset forms of globoid-cell leukodystrophy. Am.J. Hum. Genet. 59: 1233–1242, 1996.
- [40543] 14095.Duchen, L. W.; Eicher, E. M.; Jacobs, J. M.; Scaravilli, F.; Teixeira, F.: Hereditary leucodystrophy in the mouse: the new mutant twitcher. Brain 103:695–710, 1980.
- [40544] 14096.Minassian, B. A.; Lee, J. R.; Herbrick, J.-A.; Huizenga, J.; Soder, S.; Mungall, A. J.; Dunham, I.; Gardner, R.; Fong, C. G.; Carpenter, S.; Jardim, L.; Satishchandra, P.; Andermann, E.; Snead, O. C., III; Lopes-Cendes, I.; Tsui, L.-C.; Delgado-Escueta, A. V.; Rouleau, G.A.; Scherer, S. W.: Mutations in a gene encoding a novel protein tyrosine phosphatase cause progressive myoclonus epilepsy. Nature Genet. 20: 171–174, 1998.
- [40545] 14097.Norio, R.; Koskiniemi, M.: Progressive myoclonus epilepsy: genetic and nosological aspects with special ref-

erence to 107 Finnish patients. Clin.Genet. 15: 382–398, 1979.

[40546] 14098.Ortiz–Hidalgo, C.: The man behind Lafora's bodies. Am. J. Surg.Path. 10: 358–361, 1986.

[40547] 14099.Sainz, J.; Minassian, B. A.; Serratosa, J. M.; Gee, M. N.; Sakamoto,L. M.; Iranmanesh, R.; Bohlega, S.; Baumann, R. J.; Ryan, S.; Sparkes,R. S.; Delgado–Escueta, A. V.: Lafora progressive myoclonus epilepsy:narrowing the chromosome 6q24 locus by recombinations and homozygosity.(Letter) Am. J. Hum. Genet. 61: 1205–1209, 1997.

[40548] 14100.Sarlin, M. B.; Kloepper, H. W.; Mickle, W. A.; Heath, R. G.:The detection of carriers in hereditary myoclonic epilepsy. ActaGenet. Med. Gemellol. 9: 466–471, 1960.

[40549] 14101.Schwarz, G. A.; Yanoff, M.: Lafora's disease, distinct clinico–pathologicform of Unverricht's syndrome. Arch. Neurol. 12: 172–188, 1965.

[40550] 14102.Serratosa, J. M.; Delgado–Escueta, A. V.; Posada, I.; Shih, S.;Drury, I.; Berciano, J.; Zabala, J. A.; Antunez, M. C.; Sparkes, R.S.: The gene for progressive myoclonus epilepsy of the Lafora typemaps to chromosome 6q. Hum. Molec. Genet. 4: 1657–1663, 1995.

[40551] 14103.Serratosa, J. M.; Gomez–Garre, P.; Gallardo, M. E.; Anta, B.;Beltran–Valero de Bernabe, D.; Lindhout, D.; Au–

gustijn, P. B.; Tassinari, C. A.; Michelucci, R.; Malafosse, A.; Topcu, M.; Grid, D.; Dravet, C.; Berkovic, S. F.; Rodriguez de Cordoba, S.: A novel protein tyrosinephosphatase gene is mutated in progressive myoclonus epilepsy of the Lafora type (EPM2). *Hum. Molec. Genet.* 8: 345–352, 1999.

[40552] 14104. Yanoff, M.; Schwarz, G. A.: Lafora's disease: a distinct genetically determined form of Unverricht's syndrome. *Genet. Hum.* 14: 235–244, 1965.

[40553] 14105. Yokoi, S.; Austin, J.; Witmer, F.; Sakai, M.: Studies in myoclonus epilepsy (Lafora body forms). I. Isolation and preliminary characterization of Lafora bodies in two cases. *Arch. Neurol.* 19: 15–33, 1968.

[40554] 14106. Antonarakis, S.: Personal Communication. Geneva, Switzerland 4/8/1997.

[40555] 14107. Lafreniere, R. G.; Rochefort, D. L.; Chretien, N.; Rommens, J. M.; Cochius, J. I.; Kalviainen, R.; Nousiainen, U.; Patry, G.; Farrell, K.; Soderfeldt, B.; Federico, A.; Hale, B. R.; Cossio, O. H.; Sorensen, T.; Pouliot, M. A.; Kmiec, T.; Uldall, P.; Janszky, J.; Pranzatelli, M. R.; Andermann, F.; Andermann, E.; Rouleau, G. A.: Unstable insertion of the 5-prime flanking region of the cystatin B gene is the most common mutation in progressive myoclonus epilepsy type 1, EPM1. *Nature Genet.* 15: 298–302, 1997.

- [40556] 14108.Lalioti, M. D.; Mirotsoy, M.; Buresi, C.; Peitsch, M. C.; Rossier,C.; Ouazzani, R.; Baldy-Moulinier, M.; Bottani, A.; Malafosse, A.;Antonarakis, S. E.: Identification of mutations in cystatin B, the gene responsible for the Unverricht-Lundborg type of progressive myoclonusepilepsy (EPM1). *Am. J. Hum. Genet.* 60: 342-351, 1997.
- [40557] 14109.Lalioti, M. D.; Scott, H. S.; Buresi, C.; Rossier, C.; Bottani,A.; Morris, M. A.; Malafosse, A.; Antonarakis, S. E.: Dodecamer repeatexpansion in cystatin B gene in progressive myoclonus epilepsy. *Nature* 386:847-851, 1997.
- [40558] 14110.Carmel, R.: Gastric juice in congenital pernicious anemia containsno immunoreactive intrinsic factor molecule: study of three kindredswith variable ages at presentation, including a patient first diagnosedin adulthood. *Am. J. Hum. Genet.* 35: 67-77, 1983.
- [40559] 14111.Lyerla, T. A.; Konola, J. T.; Skiba, M. C.; Raghavan, S.: Galactocerebrosidaseactivity in somatic cell hybrids derived from twitcher mouse/controlhuman fibroblasts is associated with human chromosome 17. *Am. J.Hum. Genet.* 44: 198-207, 1989.
- [40560] 14112.Sakai, N.; Inui, K.; Fujii, N.; Fukushima, H.; Nishimoto, J.;Yanagihara, I.; Isegawa, Y.; Iwamatsu, A.; Okada, S.: Krabbe disease:isolation and characterization of a full-

length cDNA for human galactocerebrosidase.

Biochem.Biophys. Res. Commun. 198: 485–491, 1994.

[40561] 14113.Sweet, H.: Twitcher (twi) is on chromosome 12.
Mouse Newsletter 75:30, 1986.

[40562] 14114.Roume, J.; Genin, E.; Cormier–Daire, V.; Ma, H. W.;
Mehaye, B.;Attie, T.; Razavi–Encha, F.; Fallet–Bianco, C.;
Buenerd, A.; Clerget–Darpoux,F.; Munnich, A.; Le Merrer,
M.: A gene for Meckel syndrome maps tochromosome
11q13. Am. J. Hum. Genet. 63: 1095–1101, 1998.

[40563] 14115.Fernandes, M.; Poirier, C.; Lespinasse, F.; Carle, G.
F.: Themouse homologs of human GIF, DDB1, and CFL1
genes are located on chromosome19. Mammalian Genome
9: 339–342, 1998.

[40564] 14116.Hewitt, J. E.; Gordon, M. M.; Taggart, R. T.; Mohan–
das, T. K.;Alpers, D. H.: Human gastric intrinsic factor:
characterization ofcDNA and genomic clones and localiza–
tion to human chromosome 11. Genomics 10:432–440,
1991.

[40565] 14117.Katz, M.; Lee, S. K.; Cooper, B. A.: Vitamin B(12)
malabsorptiondue to biologically inert intrinsic factor.
New Eng. J. Med. 287:425–429, 1972.

[40566] 14118.Katz, M.; Mehlman, C. S.; Allen, R. H.: Isolation and
characterizationof an abnormal human intrinsic factor. J.

Clin. Invest. 53: 1274–1283,1974.

- [40567] 14119. Levine, J. S.; Yang, Y.; Ducos, R.; Rosenberg, A. J.; Catron, P.G.; Podell, E. R.; Allen, R. H.: Juvenile pernicious anemia (JPA) due to an abnormal intrinsic factor (IF) that is markedly susceptible to acid and proteolysis. (Abstract) Gastroenterology 80: 1210 only, 1981.
- [40568] 14120. McNicholl, B.; Egan, B.: Congenital pernicious anemia: effect on growth, brain, and absorption of B12. Pediatrics 42: 149–156, 1968.
- [40569] 14121. Remacha, A. F.; Sambeat, M. A.; Barcelo, M. J.; Mones, J.; Garcia-Die, J.; Gimferrer, E.: Congenital intrinsic factor deficiency in a Spanish patient. Ann. Hemat. 64: 202–204, 1992.
- [40570] 14122. Yang, Y. M.; Ducos, R.; Rosenberg, A. J.; Catrou, P. G.; Levine, J. S.; Podell, E. R.; Allen, R. H.: Cobalamin malabsorption in three siblings due to an abnormal intrinsic factor that is markedly susceptible to acid and proteolysis. J. Clin. Invest. 76: 2057–2065, 1985.
- [40571] 14123. Carney, J. P.; Maser, R. S.; Olivares, H.; Davis, E. M.; Le Beau, M.; Yates, J. R., III; Hays, L.; Morgan, W. F.; Petrini, J. H. J.: The hMre11/hRad50 protein complex and Nijmegen breakage syndrome: linkage of double-strand break repair to the cellular DNA damage response. Cell

93:477–486, 1998.

- [40572] 14124. Maily, F.; Palmen, J.; Muller, D. P. R.; Gibbs, T.; Lloyd, J.; Brunzell, J.; Durrington, P.; Mitropoulos, K.; Betteridge, J.; Watts, G.; Lithell, H.; Angelico, F.; Humphries, S. E.; Talmud, P. J.: Familial lipoprotein lipase (LPL) deficiency: a catalogue of LPL gene mutations identified in 20 patients from the UK, Sweden, and Italy. *Hum. Mutat.* 10:465–473, 1997.
- [40573] 14125. Normand, T.; Bergeron, J.; Fernandez–Margallo, T.; Bharucha, A.; Ven Murthy, M. R.; Julien, P.; Gagne, C.; Dionne, C.; De Braekeleer, M.; Ma, R.; Hayden, M. R.; Lupien, P. J.: Geographic distribution and genealogy of mutation 207 of the lipoprotein lipase gene in the French Canadian population of Quebec. *Hum. Genet.* 89: 671–675, 1992.
- [40574] 14126. Monsalve, M. V.; Henderson, H.; Roederer, G.; Julien, P.; Deeb, S.; Kastelein, J. J. P.; Peritz, L.; Devlin, R.; Bruin, T.; Murthy, M. R. V.; Gagne, C.; Davignon, J.; Lupien, P. J.; Brunzell, J. D.; Hayden, M. R.: A missense mutation at codon 188 of the human lipoprotein lipase gene is a frequent cause of lipoprotein lipase deficiency in persons of different ancestries. *J. Clin. Invest.* 86: 728–734, 1990.
- [40575] 14127. Cobben, J. M.; van der Steege, G.; Grootscholten,

P.; de Visser, M.; Scheffer, H.; Buys, C. H. C. M.: Deletions of the survival motor neuron gene in unaffected siblings of patients with spinal muscular atrophy. *Am. J. Hum. Genet.* 57: 805–808, 1995.

[40576] 14128. Hahnen, E.; Schonling, J.; Rudnik-Schoneborn, S.; Zerres, K.; Wirth, B.: Hybrid survival motor neuron genes in patients with autosomal recessive spinal muscular atrophy: new insights into molecular mechanisms responsible for the disease. *Am. J. Hum. Genet.* 59: 1057–1065, 1996.

[40577] 14129. Lefebvre, S.; Burglen, L.; Reboullet, S.; Clermont, O.; Burlet, P.; Viollet, L.; Benichou, B.; Cruaud, C.; Millasseau, P.; Zeviani, M.; Le Paslier, D.; Frezal, J.; Cohen, D.; Weissenbach, J.; Munnich, A.; Melki, J.: Identification and characterization of a spinal muscular atrophy-determining gene. *Cell* 80: 155–165, 1995.

[40578] 14130. Matthijs, G.; Schollen, E.; Legius, E.; Devriendt, K.; Goemans, N.; Kayserili, H.; Apak, M. Y.; Cassiman, J.-J.: Unusual molecular findings in autosomal recessive spinal muscular atrophy. *J. Med. Genet.* 33: 469–474, 1996.

[40579] 14131. Handschug, K.; Sperling, S.; Yoon, S.-J. K.; Hennig, S.; Clark, A. J. L.; Huebner, A.: Triple A syndrome is caused by mutations in AAAS, a new WD-repeat protein gene. *Hum. Molec. Genet.* 10: 283–290, 2001.

- [40580] 14132.Kaplan, J.; Gerber, S.; Larget-Piet, D.; Rozet, J.-M.; Dollfus, H.; Dufier, J.-L.; Odent, S.; Postel-Vinay, A.; Janin, N.; Briard, M.-L.; Frezal, J.; Munnich, A.: A gene for Star-gardt's disease (fundusflavimaculatus) maps to the short arm of chromosome 1. *Nature Genet.* 5:308-311, 1993.
- [40581] 14133.Coppa, G. V.; Giorgi, P. L.; Felici, L.; Gabrielli, O.; Donti, E.; Bernasconi, S.; Kresse, H.; Paschke, E.; Mastropaolo, C.: Clinical heterogeneity in Sanfilippo disease (mucopolysaccharidosis III) type D: presentation of two new cases. *Europ. J. Pediat.* 140: 130-133, 1983.
- [40582] 14134.Dhoot, G. K.; Gustafsson, M. K.; Ai, X.; Sun, W.; Standiford, D.M.; Emerson, C. P., Jr.: Regulation of Wnt signaling and embryo patterning by an extracellular sulfatase. *Science* 293: 1663-1666, 2001.
- [40583] 14135.Freeman, C.; Clements, P. R.; Hopwood, J. J.: Human liver N-acetylglucosamine-6-sulphate sulphatase: purification and characterization. *Biochem. J.* 246:347-354, 1987.
- [40584] 14136.Freeman, C.; Hopwood, J. J.: Human liver N-acetylglucosamine-6-sulphate sulphatase: catalytic properties. *Biochem. J.* 246: 355-365, 1987.
- [40585] 14137.Gatti, R.; Borrone, C.; Durand, P.; De Virgiliis, S.; Sanna, G.; Cao, A.; von Figura, K.; Kresse, H.; Paschke, E.:

Sanfilippo type D disease: clinical findings in two patients with a new variant of mucopolysaccharidosis III. *Europ. J. Pediat.* 138: 168–171, 1982.

[40586] 14138. Kaplan, P.; Wolfe, L. S.: Sanfilippo syndrome type D. *J. Pediat.* 110:267–271, 1987.

[40587] 14139. Kresse, H.; Paschke, E.; von Figura, K.; Gilberg, W.; Fuchs, W.: Sanfilippo disease type D: deficiency of N-acetylglucosamine-6-sulfate sulfatase required for heparan sulfate degradation. *Proc. Nat. Acad. Sci.* 77: 6822–6826, 1980.

[40588] 14140. Robertson, D. A.; Callen, D. F.; Baker, E. G.; Morris, C. P.; Hopwood, J. J.: Chromosomal localization of the gene for human glucosamine-6-sulphatase to 12q14. *Hum. Genet.* 79: 175–178, 1988.

[40589] 14141. Robertson, D. A.; Freeman, C.; Nelson, P. V.; Morris, C. P.; Hopwood, J. J.: Human glucosamine-6-sulfatase cDNA reveals homology with steroid sulfatase. *Biochem. Biophys. Res. Commun.* 157: 218–224, 1988.

[40590] 14142. Siciliano, L.; Fiumara, A.; Pavone, L.; Freeman, C.; Robertson, D.; Morris, C. P.; Hopwood, J. J.; Di Natale, P.; Musumeci, S.; Horwitz, A. L.: Sanfilippo syndrome type D in two adolescent sisters. *J. Med. Genet.* 28: 402–405, 1991.

[40591] 14143. Thompson, J. N.; Jones, M. Z.; Dawson, G.; Huff-

man, P. S.: N-acetylglucosamine 6-sulphatase deficiency in a Nubian goat: a model of Sanfilippo syndrome type D (mucopolysaccharidosis IIID). *J. Inherit. Metab. Dis.* 15:760–768, 1992.

[40592] 14144. Faiella, A.; Brunelli, S.; Granata, T.; D'Incerti, L.; Cardini, R.; Lenti, C.; Battaglia, G.; Boncinelli, E.: A number of schizencephaly patients including 2 brothers are heterozygous for germline mutations in the homeobox gene EMX2. *Europ. J. Hum. Genet.* 5: 186–190, 1997.

[40593] 14145. Bespalova, I. N.; Adkins, S.; Pranzatelli, M.; Burmeister, M.: Novel cystatin B mutation and diagnostic PCR assay in an Unverricht-Lundborg progressive myoclonus epilepsy patient. *Am. J. Med. Genet.* 74: 467–471, 1997.

[40594] 14146. Jarvinen, M.; Rinne, A.: Human spleen cysteine proteinase inhibitor: purification, fractionation into isoelectric variants and some properties of the variants. *Biochim. Biophys. Acta* 708: 210–217, 1982.

[40595] 14147. Jerala, R.; Trstenjak, M.; Lenarcic, B.; Turk, V.: Cloning a synthetic gene for human stefin B and its expression in *E. coli*. *FEBS Lett.* 239:41–44, 1988.

[40596] 14148. Lalioti, M. D.; Scott, H. S.; Antonarakis, S. E.: What is expanded in progressive myoclonus epilepsy? (Letter)

Nature Genet. 17: 17only, 1997.

- [40597] 14149.Lalioi, M. D.; Scott, H. S.; Genton, P.; Grid, D.; Ouazzani, R.;M'Rabet, A.; Ibrahim, S.; Gouider, R.; Dravet, C.; Chkili, T.; Bottani,A.; Buresi, C.; Malafosse, A.; Antonarakis, S. E.: A PCR amplificationmethod reveals instability of the dodecamer repeat in progressivemyoclonus epilepsy (EPM1) and no correlation between the size of therepeat and age at onset. Am. J. Hum. Genet. 62: 842–847, 1998.
- [40598] 14150.Pennacchio, L. A.; Lehesjoki, A.–E.; Stone, N. E.; Willour, V.L.; Virtaneva, K.; Miao, J.; D'Amato, E.; Ramirez, L.; Faham, M.;Koskiniemi, M.; Warrington, J. A.; Norio, R.; de la Chapelle, A.;Cox, D. R.; Myers, R. M.: Mutations in the gene encoding cystatinB in progressive myoclonus epilepsy (EPM1). Science 271: 1731–1733,1996.
- [40599] 14151.Pennacchio, L. A.; Myers, R. M.: Isolation and characterizationof the mouse cystatin B gene. Genome Res. 6: 1103–1109, 1996.
- [40600] 14152.Pranzatelli, M. R.; Tate, E.; Huang, Y.; Haas, R. H.; Bodensteiner,J.; Ashwal, S.; Franz, D.: Neuropharmacology of progressive myoclonusepilepsy: response to 5–hydroxy–L–tryptophan. Epilepsia 36: 783–791,1995.
- [40601] 14153.Turk, V.; Bode, W.: The cystatins: protein inhibitors

of cysteineproteinases. FEBS Lett. 285: 213–219, 1991.

- [40602] 14154. Chang, S. C.; Hoang, B.; Thomas, J. T.; Vukicevic, S.; Luyten, F. P.; Ryba, N. J. P.; Kozak, C. A.; Reddi, A. H.; Moos, M., Jr.: Cartilage-derived morphogenetic proteins: new members of the transforming growth factor-beta superfamily predominantly expressed in long bones during human embryonic development. J. Biol. Chem. 269: 28227–28234, 1994.
- [40603] 14155. Faiyaz-Ul-Haque, M.; Ahmad, W.; Wahab, A.; Haque, S.; Azim, A. C.; Zaidi, S. H. E.; Teebi, A. S.; Ahmad, M.; Cohn, D. H.; Siddique, T.; Tsui, L.-C.: Frameshift mutation in the cartilage-derived morphogenetic protein 1 (CDMP1) gene and severe acromesomelic chondrodysplasia resembling Grebe-type chondrodysplasia. Am. J. Med. Genet. 111: 31–37, 2002.
- [40604] 14156. Rousseau-Merck, M.-F.; Duro, D.; Berger, R.; Thiesen, H. J.: Chromosomal localization of two KIX zinc finger genes on chromosome bands 7q21–q22. Ann. Genet. 38: 81–84, 1995.
- [40605] 14157. Tommerup, N.; Vissing, H.: Isolation and fine mapping of 16 novel human zinc finger-encoding cDNAs identify putative candidate genes for developmental and malignant disorders. Genomics 27: 259–264, 1995.

- [40606] 14158.Canman, C. E.; Radany, E. H.; Parsels, L. A.; Davis, M. A.; Lawrence,T. S.; Maybaum, J.: Induction of resistance to fluorodeoxyuridinecytotoxicity and DNA damage in human tumor cells by expression ofEscherichia coli deoxyuridinetriphosphatase. Cancer Res. 54: 2296–2298,1994.
- [40607] 14159.Canman, C. E.; Tang, H. Y.; Normolle, D. P.; Lawrence, T. S.; Maybaum,J.: Variations in patterns of DNA damage induced in human colorectal tumor cells by 5–fluorodeoxyuridine: implications for mechanisms of resistance and cytotoxicity. Proc. Nat. Acad. Sci. 89: 10474–10478,1992.
- [40608] 14160.Chu, R.; Lin, Y.; Rao, M. S.; Reddy, J. K.: Cloning and identificationof rat deoxyuridine triphosphatase as an inhibitor of peroxisome proliferator–activatedreceptor alpha. J. Biol. Chem. 271: 27670–27676, 1996.
- [40609] 14161.Cohen, D.; Heng, H. H. Q.; Shi, X.–M.; McIntosh, E. M.; Tsui, L.–C.;Pearlman, R. E.: Assignment of the human dUTPase gene (DUT) to chromosome15q15–q21.1 by fluorescence in situ hybridization. Genomics 40:213–215, 1997.
- [40610] 14162.el–Hajj, H. H.; Zhang, H.; Weiss, B.: Lethality of a dut (deoxyuridinetriphosphatase) mutation in Escherichia

coli. J. Bacteriol. 170:1069–1075, 1988.

[40611] 14163.Ladner, R. D.; Caradonna, S. J.: The human dUT-Pase gene encodes both nuclear and mitochondrial isoforms: differential expression of the isoforms and characterization of a cDNA encoding the mitochondrial species. J. Biol. Chem. 272: 19072–19080, 1997.

[40612] 14164.Ladner, R. D.; McNulty, D. E.; Carr, S. A.; Roberts, G. D.; Caradonna, S. J.: Characterization of distinct nuclear and mitochondrial forms of human deoxyuridine triphosphate nucleotidohydrolase. J. Biol. Chem. 271: 7745–7751, 1996.

[40613] 14165.Lindahl, T.: DNA repair enzymes. Annu. Rev. Biochem. 51: 61–87, 1982.

[40614] 14166.McIntosh, E. M.; Ager, D. D.; Gadsden, M. H.; Haynes, R. H.: Human dUTP pyrophosphatase: cDNA sequence and potential biological importance of the enzyme. Proc. Nat. Acad. Sci. 89: 8020–8024, 1992. Note: Erratum: Proc. Nat. Acad. Sci.: 90: 4328 only, 1993.

[40615] 14167.Boring, L.; Gosling, J.; Cleary, M.; Charo, I. F.: Decreased lesion formation in CCR2 $-/-$ mice reveals a role for chemokines in the initiation of atherosclerosis. Nature 394: 894–897, 1998.

[40616] 14168.Charo, I. F.; Myers, S. J.; Herman, A.; Franci, C.;

Connolly, A.J.; Coughlin, S. R.: Molecular cloning and functional expression of two monocyte chemoattractant protein 1 receptors reveals alternative splicing of the carboxyl-terminal tails. *Proc. Nat. Acad. Sci.* 91:2752–2756, 1994.

- [40617] 14169. Barrett, T. E.; Savva, R.; Panayotou, G.; Barlow, T.; Brown, T.; Jiricny, J.; Pearl, L. H.: Crystal structure of a G:T/U mismatch-specific DNA glycosylase: mismatch recognition by complementary-strand interactions. *Cell* 92:117–129, 1998.
- [40618] 14170. De Gregorio, L.; Gallinari, P.; Gariboldi, M.; Marenti, G.; Pierotti, M. A.; Jiricny, J.; Dragani, T. A.: Genetic mapping of thymine DNA glycosylase (Tdg) gene and of one pseudogene in the mouse. *Mammalian Genome* 7: 909–910, 1996.
- [40619] 14171. Lindahl, T.: DNA repair enzymes. *Ann. Rev. Biochem.* 51: 61–87, 1982.
- [40620] 14172. Neddermann, P.; Gallinari, P.; Lettieri, T.; Schmid, D.; Truong, O.; Hsuan, J. J.; Wiebauer, K.; Jiricny, J.: Cloning and expression of human G/T mismatch-specific thymine-DNA glycosylase. *J. Biol. Chem.* 271: 12767–12774, 1996.
- [40621] 14173. Sard, L.; Tornielli, S.; Gallinari, P.; Minoletti, F.; Jiricny, J.; Lettieri, T.; Pierotti, M. A.; Sozzi, G.; Radice, P.:

Chromosomal localizations and molecular analysis of TDG gene-related sequences. *Genomics* 44:222–226, 1997.

- [40622] 14174. DiMarco, S. P.; Glover, T. W.; Miller, D. E.; Reines, D.; Warren, S. T.: Transcription elongation factor SII (TCEA) maps to human chromosome 3p22–p21.3. *Genomics* 36: 185–188, 1996.
- [40623] 14175. Park, H.; Baek, K.; Jeon, C.; Agarwal, K.; Yoo, O.: Characterization of the gene encoding the human transcriptional elongation factor TFIIS. *Gene* 139:263–267, 1994.
- [40624] 14176. Reines, D.: Nascent RNA cleavage by transcription elongation complexes. In: Conaway, R. C.; Conaway, J. W. (eds.): *Transcription: Mechanisms and Regulation*. New York: Raven Press 1994. Pp. 263–278.
- [40625] 14177. Thomas, M. J.; Platas, A. A.; Hawley, D. K.: Transcriptional fidelity and proofreading by RNA polymerase II. *Cell* 93: 627–637, 1998.
- [40626] 14178. Chang, C.; da Silva, S. L.; Ideta, R.; Lee, Y.; Yeh, S.; Burbach, J. P. H.: Human and rat TR4 orphan receptors specify a subclass of the steroid receptor superfamily. *Proc. Nat. Acad. Sci.* 91: 6040–6044, 1994.
- [40627] 14179. Chang, C.; Kokontis, J.; Acakpo-Satchivi, L.; Liao, S.; Takeda, H.; Chang, Y.: Molecular cloning of new human

TR2 receptors: a class of steroid receptor with multiple ligand-binding domains. *Biochem. Biophys. Res. Commun.* 165: 735–741, 1989.

- [40628] 14180. Hirose, T.; Fujimoto, W.; Yamaai, T.; Kim, K. H.; Matsuura, H.; Jetten, A. M.: TAK1: Molecular cloning and characterization of a new member of the nuclear receptor superfamily. *Molec. Endocr.* 1667–1680, 1994.
- [40629] 14181. Yoshikawa, T.; DuPont, B. R.; Leach, R. J.; Detera-Wadleigh, S. D.: New variants of the human and rat nuclear hormone receptor, TR4: expression and chromosomal localization of the human gene. *Genomics* 35: 361–366, 1996.
- [40630] 14182. Applequist, S. E.; Selg, M.; Raman, C.; Jack, H.-M.: Cloning and characterization of HUPF1, a human homolog of the *Saccharomyces cerevisiae* nonsense mRNA-reducing UPF1 protein. *Nucleic Acids Res.* 25: 814–821, 1997.
- [40631] 14183. Medghalchi, S. M.; Frischmeyer, P. A.; Mendell, J. T.; Kelly, A. G.; Lawler, A. M.; Dietz, H. C.: Rent1, a trans-effector of nonsense-mediated mRNA decay, is essential for mammalian embryonic viability. *Hum. Molec. Genet.* 10: 99–105, 2001.
- [40632] 14184. Mendell, J. T.; ap Rhys, C. M. J.; Dietz, H. C.: Separable roles for rent1/hUpf1 in altered splicing and decay of

nonsense transcripts. *Science* 298:419–371, 2002.

[40633] 14185. Perlick, H. A.; Medghalchi, S. M.; Spencer, F. A.; Dietz, H. C.: Cloning and characterization of a human regulator of nonsense transcript stability. (Abstract) *Am. J. Hum. Genet.* 59 (suppl.): A32 only, 1996.

[40634] 14186. Perlick, H. A.; Medghalchi, S. M.; Spencer, F. A.; Kendzior, R. J., Jr.; Dietz, H. C.: Mammalian orthologues of a yeast regulator of nonsense transcript stability. *Proc. Nat. Acad. Sci.* 93: 10928–10932, 1996.

[40635] 14187. Sun, X.; Perlick, H. A.; Dietz, H. C.; Maquat, L. E.: A mutated human homologue to yeast Upf1 protein has a dominant-negative effect on the decay of nonsense-containing mRNAs in mammalian cells. *Proc. Nat. Acad. Sci.* 95: 10009–10014, 1998.

[40636] 14188. Eng, C.; Myers, S. M.; Kogon, M. D.; Sanicola, M.; Hession, C.; Cate, R. L.; Mulligan, L. M.: Genomic structure and chromosomal localization of the human GDNFR- α gene. *Oncogene* 16: 597–601, 1998.

[40637] 14189. GFR- α Nomenclature Committee: Nomenclature of GPI-linked receptors for the GDNF ligand family. *Neuron* 19: 485 only, 1997.

[40638] 14190. Gorodinsky, A.; Zimonjic, D. B.; Popescu, N. C.; Milbrandt, J.: Assignment of the GDNF family receptor al-

pha-1 (GFRA1) to human chromosome band 10q26 by in situ hybridization. *Cytogenet. Cell Genet.* 78:289–290, 1997.

- [40639] 14191. Jing, S.; Wen, D.; Yu, Y.; Holst, P. L.; Luo, Y.; Fang, M.; Tamir, R.; Antonio, L.; Hu, Z.; Cupples, R.; Louis, J.-C.; Hu, S.; Altrock, B. W.; Fox, G. M.: GDNF-induced activation of the Ret protein tyrosine kinase is mediated by GDNFR- α , a novel receptor for GDNF. *Cell* 85:1113–1124, 1996.
- [40640] 14192. Paratcha, G.; Ledda, F.; Baars, L.; Couplier, M.; Besset, V.; Anders, J.; Scott, R.; Ibanez, C. F.: Released GFR- α -1 potentiates downstream signaling, neuronal survival, and differentiation via a novel mechanism of recruitment of c-Ret to lipid rafts. *Neuron* 29: 171–184, 2001.
- [40641] 14193. Puliti, A.; Cinti, R.; Seri, M.; Ceccherini, I.; Romeo, G.: Assignment of mouse Gfra1, the homologue of a new human HSCR candidate gene, to the telomeric region of mouse chromosome 19. *Cytogenet. Cell Genet.* 78:291–294, 1997.
- [40642] 14194. Shefelbine, S. E.; Khorana, S.; Schultz, P. N.; Huang, E.; Thobe, N.; Hu, Z. J.; Fox, G. M.; Jing, S.; Cote, G. J.; Gagel, R. F.: Mutational analysis of the GDNF/RET-GDNFR- α signaling complex in a kindred with vesicoureteral reflux. *Hum. Genet.* 102: 474–478, 1998.

- [40643] 14195.Chen, H.; Chrast, R.; Rossier, C.; Gos, A.; Antonarakis, S. E.;Kudoh, J.; Yamaki, A.; Shindoh, N.; Maeda, H.; Minoshima, S.; Shimizu,N.: Single-minded and Down syndrome? (Letter) *Nature Genet.* 10:9–10, 1995.
- [40644] 14196.Chrast, R.; Scott, H. S.; Madani, R.; Huber, L.; Wolfer, D. P.;Prinz, M.; Aguzzi, A.; Lipp, H.–P.; Antonarakis, S. E.: Mice trisomicfor a bacterial artificial chromosome with the single-minded 2 gene(Sim2) show phenotypes similar to some of those present in the partialtrisomy 16 mouse models of Down syndrome. *Hum. Molec. Genet.* 9:1853–1864, 2000.
- [40645] 14197.Dahmane, N.; Charron, G.; Lopes, C.; Yaspo, M.–L.; Maunoury, C.;Decorte, L.; Sinet, P.–M.; Bloch, B.; Delabar, J.–M.: Down syndrome–criticalregion contains a gene homologous to *Drosophila sim* expressed duringrat and human central nervous system development. *Proc. Nat. Acad.Sci.* 92: 9191–9195, 1995.
- [40646] 14198.Ema, M.; Ikegami, S.; Hosoya, T.; Mimura, J.; Ohtani, H.; Nakao,K.; Inokuchi, K.; Katsuki, M.; Fujii–Kuriyama, Y.: Mild impairmentof learning and memory in mice overexpressing the mSim2 gene locatedon chromosome 16: an animal model of Down's syndrome. *Hum. Molec.Genet.* 8: 1409–1415, 1999.

- [40647] 14199.Moffett, P.; Dayo, M.; Reece, M.; McCormick, M. K.; Pelletier,J.: Characterization of msim, a murine homologue of the Drosophilasim transcription factor. *Genomics* 35: 144–155, 1996.
- [40648] 14200.Muenke, M.; Bone, L. J.; Mitchell, H. F.; Hart, I.; Walton, K.;Hall–Johnson, K.; Ippel, E. F.; Dietz–Band, J.; Kvaloy, K.; Fan, C.–M.;Tessier–Lavigne, M.; Patterson, D.: Physical mapping of the holoprosencephalycritical region in 21q22.3, exclusion of SIM2 as a candidate gene for holoprosencephaly, and mapping of SIM2 to a region of chromosome21 important for Down syndrome. *Am. J. Hum. Genet.* 57: 1074–1079,1995.
- [40649] 14201.Yamaki, A.; Noda, S.; Kudoh, J.; Shindoh, N.; Maeda, H.; Minoshima,S.; Kawasaki, K.; Shimizu, Y.; Shimizu, N.: The mammalian single–minded(SIM) gene: mouse cDNA structure and diencephalic expression indicate a candidate gene for Down syndrome. *Genomics* 35: 136–143, 1996.
- [40650] 14202.de Winter, J. P.; Leveille, F.; van Berkel, C. G. M.; Rooimans,M. A.; van der Weel, L.; Steltenpool, J.; Demuth, I.; Morgan, N. V.;Alon, N.; Bosnoyan–Collins, L.; Lightfoot, J.; Leegwater, P. A.; Waisfisz,Q.; Komatsu, K.; Arwert, F.; Pronk, J. C.; Mathew, C. G.; Digweed,M.; Buchwald, M.;

Joenje, H.: Isolation of a cDNA representing the Fanconi anemia complementation group E gene. *Am. J. Hum. Genet.* 67:1306–1308, 2000. Note: Erratum: *Am. J. Hum. Genet.* 67: 1365 only, 2000.

[40651] 14203. Sosa–Pineda, B.; Wigle, J. T.; Oliver, G.: Hepatocyte migration during liver development requires Prox1. *Nature Genet.* 25: 254–255, 2000.

[40652] 14204. Wigle, J. T.; Chowdhury, K.; Gruss, P.; Oliver, G.: Prox1 function is crucial for mouse lens–fibre elongation. *Nature Genet.* 21: 318–322, 1999.

[40653] 14205. Wigle, J. T.; Oliver, G.: Prox1 function is required for the development of the murine lymphatic system. *Cell* 98: 769–778, 1999.

[40654] 14206. Zinovieva, R. D.; Duncan, M. K.; Johnson, T. R.; Torres, R.; Polymeropoulos, M. H.; Tomarev, S. I.: Structure and chromosomal localization of the human homeobox gene Prox 1. *Genomics* 35: 517–522, 1996.

[40655] 14207. Ladenburger, E. M.; Fackelmayer, F. O.; Hameister, H.; Knippers, R.: MCM4 and PRKDC, human genes encoding proteins MCM4 and DNA–PKcs, are close neighbours located on chromosome 8q12–q13. *Cytogenet. Cell Genet.* 77: 268–270, 1997.

[40656] 14208. Ma, Y.; Pannicke, U.; Schwarz, K.; Lieber, M. R.:

Hairpin opening and overhang processing by an Artemis/DNA-dependent protein kinase complex in nonhomologous end joining and V(D)J recombination. *Cell* 108:781–794, 2002.

[40657] 14209. Satoh, T.; Tsuruga, H.; Yabuta, N.; Ishidate, M., Jr.; Nojima, H.: Assignment of the human CDC21 (MCM4) gene to chromosome 8q11.2. *Genomics* 46:525–526, 1997.

[40658] 14210. Joenje, H.; Lo Ten Foe, J. R.; Oostra, A. B.; van Berkel, C. G. M.; Rooimans, M. A.; Schroeder-Kurth, T.; Wegner, R.-D.; Gille, J. J. P.; Buchwald, M.; Arwert, F.: Classification of Fanconi anemia patients by complementation analysis: evidence for a fifth genetic subtype. *Blood* 86: 2156–2160, 1995.

[40659] 14211. Byk, T.; Dobransky, T.; Cifuentes-Diaz, C.; Sobel, A.: Identification and molecular characterization of Unc-33-like phosphoprotein (Ulip), a putative mammalian homolog of the axonal guidance-associated unc-33 gene product. *J. Neurosci.* 16: 688–701, 1996.

[40660] 14212. Choe, H.; Farzan, M.; Sun, Y.; Sullivan, N.; Rollins, B.; Ponath, P. D.; Wu, L.; Mackay, C. R.; LaRosa, G.; Newman, W.; Gerard, N.; Gerard, C.; Sodroski, J.: The beta-chemokine receptors CCR3 and CCR5 facilitate infection by primary HIV-1 isolates. *Cell* 85: 1135–1148, 1996.

- [40661] 14213.Diriong, S.; Lory, P.; Williams, M. E.; Ellis, S. B.; Harpold,M. M.; Taviaux, S.: Chromosomal localization of the human genes for α -1A, α -1B, and α -1E voltage-dependent Ca^{2+} channel subunits. *Genomics* 30: 605–609, 1995.
- [40662] 14214.Ben-Porath, I.; Benvenisty, N.: Characterization of a tumor-associated gene, a member of a novel family of genes encoding membrane glycoproteins. *Gene* 183:69–75, 1996.
- [40663] 14215.Hong, Y.; Ohishi, K.; Inoue, N.; Endo, Y.; Fujita, T.; Takeda,J.; Kinoshita, T.: Structures and chromosomal localizations of the glycosylphosphatidylinositol synthesis gene PIGC and its pseudogene PIGCP1. *Genomics* 44: 347–349, 1997.
- [40664] 14216.Gaetano, C; Matsuo, T.; Thiele, C. J.: Identification and characterization of a retinoic acid-regulated human homologue of the unc-33-like phosphoprotein gene (hUlip) from neuroblastoma cells. *J. Biol. Chem.* 272: 12195–12201, 1997.
- [40665] 14217.Matsuo, T.; Stauffer, J. K.; Walker, R. L.; Meltzer, P.; Thiele,C. J.: Structure and promoter analysis of the human unc-33-like phosphoprotein gene: E-box required for maximal expression in neuroblastoma and myoblasts.

J.Biol. Chem. 275: 16560–16568, 2000.

- [40666] 14218.Behm, F. G.; Smith, F. O.; Raimondi, S. C.; Pui, C.–H.; Bernstein, I. D.: Human homologue of the rat chondroitin sulfate proteoglycan, NG2, detected by monoclonal antibody 7.1, identifies childhood acutelymphoblastic leukemias with t(4;11)(q21;q23) or t(11;19)(q23;p13) and MLL gene rearrangements. Blood 87: 1134–1139, 1996.
- [40667] 14219.Pluschke, G.; Vanek, M.; Evans, A.; Dittmar, T.; Schmid, P.; Itin, P.; Filardo, E. J.; Reisfeld, R. A.: Molecular cloning of a human melanoma–associated chondroitin sulfate proteoglycan. Proc. Nat.Acad. Sci. 93: 9710–9715, 1996.
- [40668] 14220.Rettig, W. J.; Dracopoli, N. C.; Goetzger, T. A.; Spengler, B.A.; Biedler, J. L.; Oettgen, H. F.; Old, L. J.: Somatic cell genetic analysis of human cell surface antigens: chromosomal assignments and regulation of expression in rodent–human hybrid cells. Proc. Nat.Acad. Sci. 81: 6437–6441, 1984.
- [40669] 14221.Rettig, W. J.; Real, F. X.; Spengler, B. A.; Biedler, J. L.; Old, L. J.: Human melanoma proteoglycan: expression in hybrids controlled by intrinsic and extrinsic signals. Science 231: 1281–1284, 1986.
- [40670] 14222.Smith, F. O.; Rauch, C.; Williams, D. E.; March, C. J.;

Arthur,D.; Hilden, J.; Lampkin, B. C.; Buckley, J. D.; Buckley, C. V.; Woods,W. G.; Dinndorf, P. A.; Sorensen, P.; Kersey, J.; Hammond, D.; Bernstein,I. D.: The human homologue of rat NG2, a chondroitin sulfate proteoglycan, is not expressed on the cell surface of normal hematopoietic cells but is expressed by acute myeloid leukemia blasts from poor-prognosis patients with abnormalities of chromosome band 11q23. *Blood* 87:1123–1133, 1996.

[40671] 14223. Gipp, J. J.; Bailey, H. H.; Mulcahy, R. T.: Cloning and sequencing of the cDNA for the light subunit of human liver gamma-glutamylcysteine synthetase and relative mRNA levels for heavy and light subunits in human normal tissues. *Biochem. Biophys. Res. Commun.* 206: 584–589, 1995.

[40672] 14224. Huang; C.-S.; Anderson; M. E.; Meister, A.: Amino acid sequence and function of the light subunit of rat kidney gamma-glutamylcysteine synthetase. *J. Biol. Chem.* 268: 20578–20583, 1993.

[40673] 14225. Sierra-Rivera, E.; Dasouki, M.; Summar, M. L.; Krishnamani, M.R. S.; Meredith, M.; Rao, P. N.; Phillips, J. A., III; Freeman, M.L.: Assignment of the human gene (GLCLR) that encodes the regulatory subunit of gamma-glutamylcysteine synthetase to chromosome 1p21. *Cyto-*

genet.Cell Genet. 72: 252–254, 1996.

- [40674] 14226.Tsuchiya, K.; Mulcahy, R. T.; Reid, L. L.; Disteché, C. M.; Kavanagh,T. J.: Mapping of the glutamate–cysteine ligase catalytic subunitgene (GLCLC) to human chromosome 6p12 and mouse chromosome 9D–E andof the regulatory subunit gene (GLCLR) to human chromosome 1p21–p22and mouse chromosome 3H1–3. Genomics 30: 630–632, 1995.
- [40675] 14227.Rozet, J.–M.; Gerber, S.; Perrault, I.; Calvas, P.; Souied, E.;Chatelin, S.; Viegas–Pequignot, E.; Molina–Gomez, D.; Munnich, A.;Kaplan, J.: Structure and refinement of the physical mapping of thegamma–glutamylcysteine ligase regulatory subunit (GLCLR) gene to chromosome1pp22.1 within the critically deleted region of human malignant mesothelioma. Cytogenet.Cell Genet. 82: 91–94, 1998.
- [40676] 14228.Ellis, J. A.; Luzio, J. P.: Identification and characterizationof a novel protein (p137) which transcytoses bidirectionally in Caco–2cells. J. Biol. Chem. 270: 20717–20723, 1995.
- [40677] 14229.Gessler, M.; Klamt, B.; Tsaoussidou, S.; Ellis, J. A.; Luzio, J.P.: The gene encoding the GPI–anchored membrane protein p137(GPI)(M11S1) maps to human chromo–

some 11p13 and is highly conserved in the mouse. *Genomics* 32: 169–170, 1996.

[40678] 14230. Inoue, N.; Watanabe, R.; Takeda, J.; Kinoshita, T.: PIG-C, one of the three human genes involved in the first step of glycosylphosphatidylinositol biosynthesis is a homologue of *Saccharomyces cerevisiae* GPI2.

Biochem. Biophys. Res. Commun. 226: 193–199, 1996.

[40679] 14231. Ring, H. Z.; Vameghi-Meyers, V.; Wang, W.; Crabtree, G. R.; Francke, U.: Five SWI/SNF-related, matrix-associated, actin-dependent regulators of chromatin (SMARC) genes are dispersed in the human genome. *Genomics* 51: 140–143, 1998.

[40680] 14232. Wang, W.; Xue, Y.; Zhou, S.; Kuo, A.; Cairns, B. R.; Crabtree, G. R.: Diversity and specialization of mammalian SWI/SNF complexes. *Genes Dev.* 10: 2117–2130, 1996.

[40681] 14233. Kim, B.-T.; Kitagawa, H.; Tamura, J.; Saito, T.; Kusche-Gullberg, M.; Lindahl, U.; Sugahara, K.: Human tumor suppressor EXT gene family members EXTL1 and EXTL3 encode alpha-1,4-N-acetylglucosaminyl transferases that likely are involved in heparan sulfate/heparin biosynthesis. *Proc. Nat. Acad. Sci.* 98: 7176–7181, 2001.

[40682] 14234. Wise, C. A.; Clines, G. A.; Massa, H.; Trask, B. J.;

Lovett, M.: Identification and localization of the gene for EXTL, a third member of the multiple exostoses gene family. *Genome Res.* 7: 10–16, 1997.

[40683] 14235. Moskow, J. J.; Bullrich, F.; Huebner, K.; Daar, I. O.; Buchberg, A. M.: Meis1, a PBX1-related homeobox gene involved in myeloid leukemia in BXH-2 mice. *Molec. Cell. Biol.* 15: 5434–5443, 1995.

[40684] 14236. Steelman, S.; Moskow, J. J.; Muzynski, K.; North, C.; Druck, T.; Montgomery, J. C.; Huebner, K.; Daar, I. O.; Buchberg, A. M.: Identification of a conserved family of Meis1-related homeobox genes. *Genome Res.* 7: 142–156, 1997.

[40685] 14237. Thorsteinsdottir, U.; Kroon, E.; Jerome, L.; Blasi, F.; Sauvageau, G.: Defining roles for HOX and MEIS1 genes in induction of acute myeloid leukemia. *Molec. Cell. Biol.* 21: 224–234, 2001.

[40686] 14238. Capdevila, J.; Tsukui, T.; Esteban, C. R.; Zappavigna, V.; Belmonte, J. C. I.: Control of vertebrate limb outgrowth by the proximal factor Meis2 and distal antagonism of BMPs by Gremlin. *Molec. Cell* 4: 839–849, 1999.

[40687] 14239. Nakamura, T.; Jenkins, N. A.; Copeland, N. G.: Identification of a new family of Pbx-related homeobox genes. *Oncogene* 13: 2235–2242, 1996.

- [40688] 14240.Smith, J. E.; Afonja, O.; Yee, H. T.; Inghirami, G.; Takeshita,K.: Chromosomal mapping to 15q14 and expression analysis of the humanMEIS2 homeobox gene. *Mammalian Genome* 8: 951–952, 1997.
- [40689] 14241.Kobayashi, S.; Uemura, H.; Kohda, T.; Nagai, T.; Chinen, Y.; Naritomi,K.; Kinoshita, E.; Ohashi, H.; Imaizumi, K.; Tsukahara, M.; Sugio,Y.; Tonoki, H.; Kishino, T.; Tanaka, T.; Yamada, M.; Tsutsumi, O.;Niikawa, N.; Kaneko–Ishino, T.; Ishino, F.: No evidence of PEG1/MESTgene mutations in Silver–Russell syndrome patients. *Am. J. Med. Genet.* 104:225–231, 2001.
- [40690] 14242.Mizuno, K.; Hasegawa, K.; Katagiri, T.; Ogimoto, M.; Ichikawa,T.; Yakura, H.: MPTP–delta, a putative murine homolog of HPTP–delta,is expressed in specialized regions of the brain and in the B–celllineage. *Molec. Cell. Biol.* 13: 5513–5523, 1993.
- [40691] 14243.Uetani, N.; Kato, K.; Ogura, H.; Mizuno, K.; Kawano, K.; Mikoshiba,K.; Yakura, H.; Asano, M.; Iwakura, Y.: Impaired learning with enhancedhippocampal long–term potentiation in PTP–delta–deficient mice. *EMBOJ.* 19: 2775–2785, 2000.
- [40692] 14244.Heighway, J.; Betticher, D. C.; Hoban, P. R.; Altermatt, H. J.;Cowen, R.: Coamplification in tumors of KRAS2,

type 2 inositol 1,4,5triphosphate receptor gene, and a novel human gene, KRAG. Genomics 35:207–214, 1996.

- [40693] 14245.Scott, A. F.; Elizaga, A.; Morrell, J.; Bergen, A.; Penno, M. B.: Characterization of a gene coamplified with Ki-ras in Y1 murineadrenal carcinoma cells that codes for a putative membrane protein. Genomics 20:227–230, 1994.
- [40694] 14246.Monte, D.; Baert, J. L.; Defossez, P. A.; de Launoit, Y.; Stehelin,D.: Molecular cloning and characterization of human ERM, a new memberof the Ets family closely related to mouse PEA3 and ER81 transcriptionfactors. Oncogene 9: 1397–1406, 1994.
- [40695] 14247.Monte, D.; Coutte, L.; Dewitte, F.; Defossez, P.–A.; Le Coniat,M.; Stehelin, D.; Berger, R.; de Launoit, Y.: Genomic organizationof the human ERM (ETV5) gene, a PEA3 group member of ETS transcriptionfactors. Genomics 35: 236–240, 1996.
- [40696] 14248.Protopopova, M. V.; Vorobieva, N. V.; Protopopov, A. I.; Gizatullin,R. Z.; Kashuba, V. I.; Klein, G.; Zabarovsky, E. R.; Graphodatsky,A. S.: Assignment of the ERM gene (ETV5) coding for the ets-relatedprotein to human chromosome band 3q28 by in situ hybridization. Cyto-genet.Cell Genet. 74: 220 only, 1996.

- [40697] 14249.Bosher, J. M.; Williams, T.; Hurst, H. C.: The developmentally regulated transcription factor AP-2 is involved in c-erbB-2 overexpression in human mammary carcinoma. *Proc. Nat. Acad. Sci.* 92: 744-747, 1995.
- [40698] 14250.Combadiere, C.; Ahuja, S. K.; Murphy, P. M.: Cloning, chromosomal localization, and RNA expression of a human beta chemokine receptor-like gene. *DNA Cell Biol.* 14: 673-680, 1995.
- [40699] 14251.Combadiere, C.; Ahuja, S. K.; Van Damme, J.; Tiffany, H. L.; Gao, J.-L.; Murphy, P. M.: Monocyte chemoattractant protein-3 is a functional ligand for CC chemokine receptors 1 and 2B. *J. Biol. Chem.* 270:29671-29675, 1995.
- [40700] 14252.Doranz, B. J.; Rucker, J.; Yi, Y.; Smyth, R. J.; Samson, M.; Peiper, S. C.; Parmentier, M.; Collman, R. G.; Doms, R. W.: A dual-tropic primary HIV-1 isolate that uses fusin and the beta-chemokine receptors CKR-5, CKR-3, and CKR-2b as fusion cofactors. *Cell* 85: 1149-1158, 1996.
- [40701] 14253.Mummidi, S.; Ahuja, S. S.; Gonzalez, E.; Anderson, S. A.; Santiago, E. N.; Stephan, K. T.; Craig, F. E.; O'Connell, P.; Tryon, V.; Clark, R. A.; Dolan, M. J.; Ahuja, S. K.: Genealogy of the CCR5 locus and chemokine system gene

variants associated with altered rates of HIV-1 disease progression. *Nature Med.* 4: 786–793, 1998.

[40702] 14254. Peters, W.; Dupuis, M.; Charo, I. F.: A mechanism for the impaired IFN- γ production in C-C chemokine receptor 2 (CCR2) knockout mice: role of CCR2 in linking the innate and adaptive immune responses. *J. Immun.* 165: 7072–7077, 2000.

[40703] 14255. Peters, W.; Scott, H. M.; Chambers, H. F.; Flynn J. L.; Charo, I. F.; Ernst, J. D.: Chemokine receptor 2 serves an early and essential role in resistance to *Mycobacterium tuberculosis*. *Proc. Nat. Acad. Sci.* 98: 7958–7963, 2001.

[40704] 14256. Samson, M.; Labbe, O.; Mollereau, C.; Vassart, G.; Parmentier, M.: Molecular cloning and functional expression of a new human CC-chemokine receptor gene. *Biochemistry* 35: 3362–3367, 1996.

[40705] 14257. Samson, M.; Soularue, P.; Vassart, G.; Parmentier, M.: The genes encoding the human CC-chemokine receptors CC-CKR1 to CC-CKR5 (CMKBR1–CMKBR5) are clustered in the p21.3–p24 region of chromosome 3. *Genomics* 36: 522–526, 1996.

[40706] 14258. Sanders, S. K.; Crean, S. M.; Boxer, P. A.; Kellner, D.; LaRosa, G. J.; Hunt, S. W., III.: Functional differences between monocyte chemotactic protein-1 receptor A and

monocyte chemotactic protein-1 receptor B expressed in a Jurkat T cell. *J. Immun.* 165: 4877–4883, 2000.

[40707] 14259. Smith, M. W.; Dean, M.; Carrington, M.; Winkler, C.; Huttley, G. A.; Lomb, D. A.; Goedert, J. J.; O'Brien, T. R.; Jacobson, L. P.; Kaslow, R.; Buchbinder, S.; Vittinghoff, E.; Vlahov, D.; Hoots, K.; Hilgartner, M. W.; Hemophilia Growth and Development Study (HGDS); Multicenter AIDS Cohort Study (MACS); Multicenter Hemophilia Cohort Study (MHCS); San Francisco City Cohort (SFCC); ALIVE Study; O'Brien, S. J.: Contrasting genetic influence of CCR2 and CCR5 variants on HIV-1 infection and disease progression. *Science* 277: 959–965, 1997.

[40708] 14260. Wong, L.-M.; Myers, S. J.; Tsou, C.-L.; Gosling, J.; Arai, H.; Charo, I. F.: Organization and differential expression of the human monocyte chemoattractant protein 1 receptor gene: evidence for the role of the carboxyl-terminal tail in receptor trafficking. *J. Biol. Chem.* 272: 1038–1045, 1997.

[40709] 14261. Donald, L. J.; Wang, H. S.; Hamerton, J. L.: Assignment of the gene for cystathionase (CYS) to human chromosome 16. (Abstract) *Cytogenet. Cell Genet.* 32: 268 only, 1982.

[40710] 14262. Frimpter, G. W.: Cystathioninuria: nature of the de-

fect. Science 149:1095–1096, 1965.

- [40711] 14263.Frimpter, G. W.: Cystathioninuria, sulfite oxidase deficiency, and 'beta-mercaptolactate-cysteine disulfiduria. In: Stanbury, J. B.; Wyngaarden, J. B.; Fredrickson, D. S.: The Metabolic Basis of Inherited Disease. New York: McGraw-Hill (pub.) (3rd ed.): 1972. Pp. 413–425.
- [40712] 14264.Frimpter, G. W.; Haymovitz, A.; Horwith, M.: Cystathioninuria. New Eng. J. Med. 268: 333–339, 1963.
- [40713] 14265.Harris, H.; Penrose, L. S.; Thomas, D. H. H.: Cystathioninuria. Ann.Hum. Genet. 23: 442–453, 1959.
- [40714] 14266.Lu, Y.; O'Dowd, B. F.; Orrego, H.; Israel, Y.: Cloning and nucleotide sequence of human liver cDNA encoding for cystathionine gamma-lyase. Biochem.Biophys. Res. Commun. 189: 749–758, 1992.
- [40715] 14267.Lyon, I. C. T.; Procopis, P. G.; Turner, B.: Cystathioninuria in a well baby population. Acta Paediat. Scand. 60: 324–328, 1971.
- [40716] 14268.Mongeau, J.-G.; Hilgartner, M.; Worthen, H. G.; Frimpter, G. W.: Cystathioninuria: study of an infant with normal mentality, thrombocytopenia, and renal calculi. J. Pediat. 69: 1113–1120, 1967.
- [40717] 14269.Pascal, T. A.; Gaull, G. E.; Beratis, N. G.; Gillam, B. M.; Tallan, H. H.: Cystathionase deficiency: evidence for

genetic heterogeneity in primary cystathioninuria. *Pediat. Res.* 12: 125–133, 1978.

[40718] 14270. Perry, T. L.; Hardwick, D. F.; Hansen, S.; Love, D. L.; Israels, S.: Cystathioninuria in two healthy siblings. *New Eng. J. Med.* 278:590–592, 1968.

[40719] 14271. Schneiderman, L. J.: Latent cystathioninuria. *J. Med. Genet.* 4:260–263, 1967.

[40720] 14272. Scott, C. R.; Dassell, S. W.; Clark, S. H.; Chiang-Teng, C.; Swedberg, K. R.: Cystathioninemia: a benign genetic condition. *J. Pediat.* 76:571–577, 1970.

[40721] 14273. Shaw, K. N. F.; Lieberman, E.; Koch, R.; Donnell, G. N.: Cystathioninuria. *Am. J. Dis. Child.* 113: 119–128, 1967.

[40722] 14274. Tada, K.; Yoshida, T.; Yokoyama, Y.; Sato, T.; Nakagawa, H.; Arakawa, T.: Cystathioninuria not associated with vitamin B6 dependency: a probably new type of cystathioninuria. *Tohoku J. Exp. Med.* 95: 235–242, 1968.

[40723] 14275. Whelan, D. T.; Scriver, C. R.: Cystathioninuria and renal iminoglycinuria in a pedigree: a perspective on counseling. *New Eng. J. Med.* 278:924–927, 1968.

[40724] 14276. Joenje, H.: Fanconi anaemia complementation groups in Germany and The Netherlands. *Hum. Genet.* 97: 280–282, 1996.

- [40725] 14277.Joenje, H.; Oostra, A. B.; Wijker, M.; di Summa, F. M.; van Berkel,C. G. M.; Rooimans, M. A.; Ebell, W.; van Weel, M.; Pronk, J. C.;Buchwald, M.; Arwert, F.: Evidence for at least eight Fanconi anemiagenes. *Am. J. Hum. Genet.* 61: 940–944, 1997.
- [40726] 14278.Bongiovanni, A. M.: Disorders of adrenocortical steroid biogenesis.In:Stanbury, J. B.; Wyngaarden, J. B.; Fredrickson, D. S.: *The MetabolicBasis of Inherited Dis–ease*. New York: McGraw–Hill (pub.) (3rded.): 1972. Pp. 857–885.
- [40727] 14279.Bongiovanni, A. M.; Root, A. W.: The adrenogenital syndrome. *NewEng. J. Med.* 268: 1283–1289; 1342–1351; 1391–1399, 1963.
- [40728] 14280.Boudreaux, D.; Waisman, J.; Skinner, D. G.; Low, R.: Giant adrenalmyelolipoma and testicular interstitial cell tumor in a man with congenital21–hydroxylase deficiency. *Am. J. Surg. Path.* 3: 109–123, 1979.
- [40729] 14281.Carroll, M. C.; Campbell, R. D.; Porter, R. R.: Map–ping of steroid21–hydroxylase genes adjacent to comple–ment component C4 genes inHLA, the major histocompat–ibility complex in man. *Proc. Nat. Acad.Sci.* 82: 521–525, 1985.
- [40730] 14282.Chaplin, D. D.; Galbraith, L. J.; Seidman, J. G.;

White, P. C.;Parker, K. L.: Nucleotide sequence analysis of murine 21-hydroxylasegene: mutations affecting gene expression. Proc. Nat. Acad. Sci. 83:9601-9605, 1986.

- [40731] 14283.Charmandari, E.; Hindmarsh, P. C.; Johnston, A.; Brook, C. G.D.: Congenital adrenal hyperplasia due to 21-hydroxylase deficiency:alterations in cortisol pharmacokinetics at puberty. J. Clin. Endocr.Metab. 86: 2701-2708, 2001.
- [40732] 14284.Childs, B.; Grumbach, M. M.; Van Wyk, J. J.: Virilizing adrenalhyperplasia: genetic and hormonal studies. J. Clin. Invest. 35:213-222, 1956.
- [40733] 14285.Chiou, S.-H.; Hu, M.-C.; Chung, B.: A missense mutation at ile172-to-asnor arg356-to-trp causes steroid 21-hydroxylase deficiency. J. Biol.Chem. 265: 3549-3552, 1990.
- [40734] 14286.Chrousos, G. P.; Loriaux, D. L.; Mann, D. L.; Cutler, G. B., Jr.: Late-onset 21-hydroxylase deficiency mimicking idiopathic hirsutismor polycystic ovarian disease: an allelic variant of congenital virilizingadrenal hyperplasia with a milder enzymatic defect. Ann. Intern.Med. 96: 143-148, 1982.
- [40735] 14287.Collier, S.; Tassabehji, M.; Strachan, T.: A de novo pathologicalpoint mutation at the 21-hydroxylase locus:

implications for geneconversion in the human genome.
Nature Genet. 3: 260–265, 1993.

[40736] 14288.Cooper, D. Y.; Levin, S.; Narasimhulu, S.; Rosen-
thal, O.; Estabrook,R. W.: Photochemical action spectrum
of the terminal oxidase of mixedfunction oxidase sys-
tems. Science 147: 400–402, 1965.

[40737] 14289.Cornean, R. E.; Hindmarsh, P. C.; Brook, C. G. D.:
Obesity in21–hydroxylase deficient patients. Arch. Dis.
Child. 78: 261–263,1998.

[40738] 14290.Couillin, P.; Kottler–Missonnier, M. L.; Grisard, M.
C.; Hors,J.; Feingold, J.; Boue, J.; Boue, A.: HLA–A, B, C, DR
alleles incongenital adrenal hyperplasia. Hum. Genet. 53:
389–392, 1980.

[40739] 14291.Cutfield, R. G.; Bateman, J. M.; Odell, W. D.: Infer-
tility causedby bilateral testicular masses secondary to
congenital adrenal hyperplasia(21–hydroxylase defi-
ciency). Fertil. Steril. 40: 809–814, 1983.

[40740] 14292.Cutler, G. B., Jr.; Laue, L.: Congenital adrenal hy-
perplasiadue to 21–hydroxylase deficiency. New Eng. J.
Med. 323: 1806–1813,1990.

[40741] 14293.Dacou–Voutetakis, C.; Dracopoulou, M.: High inci-
dence of moleculardefects of the CYP21 gene in patients
with premature adrenarche. J.Clin. Endocr. Metab. 84:

1570–1574, 1999.

- [40742] 14294. Day, D. J.; Speiser, P. W.; Schulze, E.; Bettendorf, M.; Fitness, J.; Barany, F.; White, P. C.: Identification of non-amplifying CYP21 genes when using PCR-based diagnosis of 21-hydroxylase deficiency in congenital adrenal hyperplasia (CAH) affected pedigrees. *Hum. Molec. Genet.* 5: 2039–2048, 1996.
- [40743] 14295. Deneuve, C.; Tardy, V.; Dib, A.; Mornet, E.; Billaud, L.; Charron, D.; Morel, Y.; Kuttann, F.: Phenotype-genotype correlation in 56 women with nonclassical congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *J. Clin. Endocr. Metab.* 86: 207–213, 2001.
- [40744] 14296. Donohoue, P. A.; Jospe, N.; Migeon, C. J.; Van Dop, C.: Two distinct areas of unequal crossing over within the steroid 21-hydroxylase genes produce absence of CYP21B. *Genomics* 5: 397–406, 1989.
- [40745] 14297. Dracopoulou-Vabouli, M.; Maniati-Christidi, M.; Dacou-Voutetakis, C.: The spectrum of molecular defects of the CYP21 gene in the Hellenic population: variable concordance between genotype and phenotype in the different forms of congenital adrenal hyperplasia. *J. Clin. Endocr. Metab.* 86: 2845–2848, 2001.
- [40746] 14298. Duck, S. C.: Malignancy associated with congenital

adrenal hyperplasia. *J.Pediat.* 99: 423–424, 1981.

[40747] 14299.Dumic, M.; Brkljacic, L.; Mardesic, D.; Plavsic, V.; Lukenda,M.; Kastelan, A.: 'Cryptic' form of congenital adrenal hyperplasiadue to 21–hydroxylase deficiency in the Yugoslav population. *ActaEndocr.* 109: 386–392, 1985.

[40748] 14300.Dupont, B.; Smithwick, E. M.; Oberfield, S. E.; Lee, T. D.; Levine,L. S.: Close genetic linkage between HLA and congenital adrenal hyperplasia(21–hydroxylase deficiency). *Lancet II*: 1309–1312, 1977.

[40749] 14301.Ezquieta, B.; Oliver, A.; Gracia, R.; Gancedo, P. G.: Analysisof steroid 21–hydroxylase gene mutations in the Spanish population. *Hum.Genet.* 96: 198–204, 1995.

[40750] 14302.Ferenczi, A.; Garami, M.; Kiss, E.; Pek, M.; Sasvari–Szekely,M.; Barta, C.; Staub, M.; Solyom, J.; Fekete, G.: Screening for mutationsof 21–hydroxylase gene in Hungarian patients with congenital adrenalhyperplasia. *J. Clin. Endocr. Metab.* 84: 2369–2372, 1999.

[40751] 14303.Fitness, J.; Dixit, N.; Webster, D.; Torresani, T.; Pergolizzi,R.; Speiser, P. W.; Day, D. J.: Genotyping of CYP21, linked chromosome6p markers, and a sex–specific gene in neonatal screening for congenitaladrenal hyperplasia. *J. Clin. Endocr. Metab.* 84: 960–966, 1999.

- [40752] 14304.Fleischnick, E.; Awdeh, Z. L.; Raum, D.; Granados, J.; Alosco, S. M.; Crigler, J. F., Jr.; Gerald, P. S.; Giles, C. M.; Yunis, E.J.; Alper, C. A.: Extended MHC haplotypes in 21-hydroxylase-deficiency congenital adrenal hyperplasia: shared genotypes in unrelated patients. *Lancet* I:152-156, 1983.
- [40753] 14305.Flori, J.; Tongio, M. M.; Kurtz, F.; Mayer, S.; Juif, J. G.: Liaison entre le gene responsable de l'hyperplasie sur-renalienne par deficit en 21 hydroxylase et les marqueurs du bras court du chromosome 6. *J. Genet. Hum.* 28: 185-193, 1980.
- [40754] 14306.Forest, M. G.; Betuel, H.; David, M.: Prenatal treatment in congenital adrenal hyperplasia due to 21-hydroxylase deficiency: up-date 88 of the French multicentric study. *Endocr. Res.* 15: 277-301, 1989.
- [40755] 14307.Galal, O. M.; Rudd, B. T.; Drayer, N. M.: Evaluation of deficiency of 21-hydroxylation in patients with congenital adrenal hyperplasia. *Arch. Dis. Child.* 43: 410-414, 1968.
- [40756] 14308.Ghanem, N.; Lobaccaro, J. M.; Buresi, C.; Abbal, M.; Halaby, G.; Sultan, C.; Lefranc, G.: Defective, deleted or converted CYP21B gene and negative association with a rare restriction fragment length polymorphism allele of the

factor B gene in congenital adrenal hyperplasia.

Hum.Genet. 86: 117–125, 1990.

[40757] 14309.Rassool, F. V.; Le Beau, M. M.; Shen, M.–L.; Neilly, M. E.; Espinosa,R., III; Ong, S. T.; Boldog, F.; Drabkin, H.; McCarroll, R.; McKeithan,T. W.: Direct cloning of DNA sequences from the common fragile siteregion at chromosome band 3p14.2. Genomics 35: 109–117, 1996.

[40758] 14310.Rudduck, C.; Franzen, G.: A new heritable fragile site on human chromosome 3. Hereditas 98: 297–299, 1983.

[40759] 14311.Shi, Y.; Zou, M.; Farid, N. R.; Paterson, M. C.: Association of FHIT (fragile histidine triad), a candidate tumour suppressor gene, with the ubiquitin–conjugating enzyme hUBC9. Biochem. J. 352: 443–448, 2000.

[40760] 14312.Shiraishi, T.; Druck, T.; Mimori, K.; Flomenberg, J.; Berk, L.; Alder, H.; Miller, W.; Huebner, K.; Croce, C. M.: Sequence conservation at human and mouse orthologous common fragile regions, FRA3B/FHIT and Fra14A2/Fhit. Proc. Nat. Acad. Sci. 98: 5722–5727, 2001.

[40761] 14313.Sozzi, G.; Veronese, M. L.; Negrini, M.; Baffa, R.; Cotticelli, M. G.; Inoue, H.; Tornielli, S.; Pilotti, S.; De Gregorio, L.; Pastorino, U.; Pierotti, M. A.; Ohta, M.; Huebner, K.; Croce, C. M.: The FHIT gene at 3p14.2 is abnormal in

lung cancer. Cell 85: 17–26, 1996.

[40762] 14314.Stein, C. K.; Glover, T. W.; Palmer, J. L.; Glisson, B. S.: Directcorrelation between FRA3B expression and cigarette smoking. GenesChromosomes Cancer 34: 333–340, 2002.

[40763] 14315.Virgilio, L.; Shuster, M.; Gollin, S. M.; Veronese, M. L.; Ohta,M.; Huebner, K.; Croce, C. M.: FHIT gene alterations in head andneck squamous cell carcinomas. Proc. Nat. Acad. Sci. 93: 9770–9775,1996.

[40764] 14316.Wegner, R.–D.: A new inducible fragile site on chromosome 3 (p14.2)in human lymphocytes. Hum. Genet. 63: 297–298, 1983.

[40765] 14317.Wegner, R.–D.: Reply to the letter of A. Markkanen, S. Knuutila,and A. de la Chapelle. (Letter) Hum. Genet. 65: 218 only, 1983.

[40766] 14318.Zanesi, N.; Fidanza, V.; Fong, L. Y.; Mancini, R.; Druck, T.;Valtieri, M.; Rudiger, T.; McCue, P. A.; Croce, C. M.; Huebner, K.: The tumor spectrum in FHIT–deficient mice. Proc. Nat. Acad. Sci. 98:10250–10255, 2001.

[40767] 14319.Nagase, T.; Seki, N.; Ishikawa, K.; Ohira, M.; Kawarabayasi, Y.;Ohara, O.; Tanaka, A.; Kotani, H.; Miyajima, N.; Nomura, N.: Predictionof the coding sequences of unidentified human genes. VI. The codingsequences of

80 new genes (KIAA0201–KIAA0280) deduced by analysis of cDNA clones from cell line KG–1 and brain. DNA Res. 3: 321–329, 1996.

- [40768] 14320. Palfree, R. G. E.; Sadro, L. C.; Solomon, S.: The gene encoding the human corticostatin HP–4 precursor contains a recent 86–base duplication and is located on chromosome 8. Molec. Endocr. 7: 199–205, 1993.
- [40769] 14321. Singh, A.; Bateman, A.; Zhu, Q. Z.; Shimasaki, S.; Esch, F.; Solomon, S.: Structure of the novel human granulocyte peptide with anti–ACTH activity. Biochem. Biophys. Res. Commun. 155: 524–529, 1988.
- [40770] 14322. Lee, E.; Marcucci, M.; Daniell, L.; Pypaert, M.; Weisz, O. A.; Ochoa, G.–C.; Farsad, K.; Wenk, M. R.; De Camilli, P.: Amphiphysin2 (Bin1) and T–tubule biogenesis in muscle. Science 297: 1193–1196, 2002.
- [40771] 14323. Negorev, D.; Riethman, H.; Wechsler–Reya, R.; Sakamuro, D.; Prendergast, G. C.; Simon, D.: The Bin1 gene localizes to human chromosome 2q14 by PCR analysis of somatic cell hybrids and fluorescence in situ hybridization. Genomics 33: 329–331, 1996.
- [40772] 14324. Sakamuro, D.; Elliott, K. J.; Wechsler–Reya, R.; Prendergast, G. C.: BIN1 is a novel MYC–interacting protein with features of a tumour suppressor. Nature Genet. 14:

69–77, 1996.

- [40773] 14325. Ishizuka, T.; Ahmad, I.; Kita, K.; Sonoda, T.; Ishijima, S.; Sawa, K.; Suzuki, N.; Tatibana, M.: The human phosphoribosylpyrophosphatesynthetase-associated protein 39 gene (PRPSAP1) is located in the chromosome region 17q24–q25. *Genomics* 33: 332–334, 1996.
- [40774] 14326. Ishizuka, T.; Kita, K.; Sonoda, T.; Ishijima, S.; Sawa, K.; Suzuki, N.; Tatibana, M.: Cloning and sequencing of human complementary DNA for the phosphoribosylpyrophosphate synthetase-associated protein 39. *Biochim. Biophys. Acta* 1306: 27–30, 1996.
- [40775] 14327. Kita, K.; Ishizuka, T.; Ishijima, S.; Sonoda, T.; Tatibana, M.: A novel 39-kDa phosphoribosylpyrophosphate synthetase-associated protein of rat liver: cloning, high sequence similarity to the catalytic subunits, and a negative regulatory role. *J. Biol. Chem.* 269: 8334–8340, 1994.
- [40776] 14328. Endo, Y.; Sato, Y.; Matsushita, M.; Fujita, T.: Cloning and characterization of the human lectin P35 gene and its related gene. *Genomics* 36: 515–521, 1996.
- [40777] 14329. Wayne, S.; Robertson, N. G.; DeClau, F.; Chen, N.; Verhoeven, K.; Prasad, S.; Tranebjarg, L.; Morton, C. C.; Ryan, A. F.; Van Camp, G.; Smith, R. J. H.: Mutations in the transcriptional activator EYA4 cause late-onset deafness at

the DFNA10 locus. *Hum. Molec. Genet.* 10:195–200, 2001.

- [40778] 14330. Kanai, Y.; Miura, K.; Uehara, T.; Amagai, M.; Takeda, O.; Tanuma, S.; Kurosawa, Y.: Natural occurrence of Nuc in the sera of autoimmune-prone MRL/lpr mice. *Biochem. Biophys. Res. Commun.* 196: 729–736, 1993.
- [40779] 14331. Miura, K.; Hirai, M.; Kanai, Y.; Kurosawa, Y.: Organization of the human gene for nucleobindin (NUC) and its chromosomal assignment to 19q13.2–q13.4. *Genomics* 34: 181–186, 1996.
- [40780] 14332. Miura, K.; Titani, K.; Kurosawa, Y.; Kanai, Y.: Molecular cloning of nucleobindin, a novel DNA-binding protein that contains both a signal peptide and a leucine zipper structure. *Biochem. Biophys. Res. Commun.* 187: 375–380, 1992.
- [40781] 14333. Connelly, M. A.; Grady, R. C.; Mushinski, J. F.; Marcu, K. B.: PANG, a gene encoding a neuronal glycoprotein, is ectopically activated by intracisternal A-type particle long terminal repeats in murine plasmacytomas. *Proc. Nat. Acad. Sci.* 91: 1337–1341, 1994.
- [40782] 14334. Mock, B. A.; Connelly, M. A.; McBride, O. W.; Kozak, C. A.; Marcu, K. B.: Plasmacytoma-associated neuronal glycoprotein, Pang, maps to mouse chromosome 6 and

human chromosome 3. Genomics 34: 226–228,1996.

- [40783] 14335.Noben–Trauth, K.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N.A.; Sonoda, G.; Testa, J. R.; Klemptner, K.–H.: Mybl2 (Bmyb) maps to mouse chromosome 2 and human chromosome 20q13.1. Genomics 35:610–612, 1996.
- [40784] 14336.Ando, A.; Kikuti, Y. Y.; Shigenari, A.; Kawata, H.; Okamoto, N.; Shiina, T.; Chen, L.; Ikemura, T.; Abe, K.; Kimura, M.; Inoko, H.: cDNA cloning of the human homologues of the mouse Ke4 and Ke6 genes at the centromeric end of the human MHC region. Genomics 35: 600–602,1996.
- [40785] 14337.Aziz, N.; Maxwell, M. M.; Brenner, B. M.: Coordinate regulation of 11–beta–HSD and Ke6 gene in cpk mouse: implications for steroid metabolic defect in PKD. Am. J. Physiol. 267: F791–F797, 1994.
- [40786] 14338.Aziz, N.; Maxwell, M. M.; St.–Jacques, B.; Brenner, B. M.: Downregulation of Ke 6, a novel gene encoded within the major histocompatibility complex, in murine polycystic kidney disease. Molec. Cell. Biol. 13:1847–1853, 1993.

- [40787] 14339.Kikuti, Y. Y.; Tamiya, G.; Ando, A.; Chen, L.; Kimura, M.; Ferreira,E.; Tsuji, K.; Trowsdale, J.; Inoko, H.: Physical mapping 220 kbcentromeric of the human MHC and DNA sequence analysis of the 43-kbsegment including the RING1, HKE6, and HKE4 genes. *Genomics* 42:422–435, 1997.
- [40788] 14340.Aziz, N.; Anderson, E.; Lee, G. Y.; Woo, D. D. L.: Arrested testisdevelopment in the cpk mouse may be the result of abnormal steroidmetabolism. *Molec. Cell. Endocr.* 171: 83–88, 2001.
- [40789] 14341.Fomitcheva, J.; Baker, M. E.; Anderson, E.; Lee, G. Y.; Aziz, N.: Characterization of Ke 6, a new 17-beta-hydroxysteroid dehydrogenase,and its expression in gonadal tissues. *J. Biol. Chem.* 273: 22664–22671,1998.
- [40790] 14342.Cheung, A. H.; Stewart, R. J.; Marsden, P. A.: Endothelial Tie2/Tekligands angiopoietin–1 (ANGPT1) and angiopoietin–2 (ANGPT2): regionallocalization of the human genes to 8q22.3–q23 and 8p23. *Genomics* 48:389–391, 1998.
- [40791] 14343.Davis, S.; Aldrich, T. H.; Jones, P. F.; Acheson, A.; Compton,D. L.; Jain, V.; Ryan, T. E.; Bruno, J.; Radziejewski, C.; Maisonpierre,P. C.; Yancopoulos, G. D.: Isolation of

angiopoietin-1, a ligand for the TIE2 receptor, by secretion-trap expression cloning. *Cell* 87:1161-1169, 1996.

[40792] 14344. Folkman, J.; D'Amore, P. A.: Blood vessel formation: what is its molecular basis? *Cell* 87: 1153-1155, 1996.

[40793] 14345. Marziliano, N.; Crovella, S.; Audero, E.; Pecile, V.; Bussolino, F.; Amoroso, A.; Garagna, S.: Genetic mapping of the mouse homologue of the human angiopoietin-1 gene (Agpt) to mouse chromosome 9E2 by in situ hybridization. *Cytogenet. Cell Genet.* 87: 199-200, 1999.

[40794] 14346. Sato, T. N.; Tozawa, Y.; Deutsch, U.; Wolburg-Buchholz, K.; Fujiwara, Y.; Gendron-Maguire, M.; Gridley, T.; Wolburg, H.; Risau, W.; Qin, Y.: Distinct roles of the receptor tyrosine kinases Tie-1 and Tie-2 in blood vessel formation. *Nature* 376: 70-73, 1995.

[40795] 14347. Suri, C.; Jones, P. F.; Patan, S.; Bartunkova, S.; Maisonpierre, P. C.; Davis, S.; Sato, T. N.; Yancopoulos, G. D.: Requisite role of angiopoietin-1, a ligand for the TIE2 receptor, during embryonic angiogenesis. *Cell* 87: 1171-1180, 1996.

[40796] 14348. Suri, C.; McClain, J.; Thurston, G.; McDonald, D. M.; Zhou, H.; Oldmixon, E. H.; Sato, T. N.; Yancopoulos, G. D.: Increased vascularization in mice overexpressing angiopoietin-1. *Science* 282: 468-471, 1998.

- [40797] 14349.Valenzuela, D. M.; Griffiths, J. A.; Rojas, J.; Aldrich, T. H.; Jones, P. F.; Zhou, H.; McClain, J.; Copeland, N. G.; Gilbert, D.J.; Jenkins, N. A.; Huang, T.; Papadopoulos, N.; Maisonpierre, P.C.; Davis, S.; Yancopoulos, G. D.: An-
giopoietins 3 and 4: diverging gene counterparts in mice
and humans. *Proc. Nat. Acad. Sci.* 96:1904–1909, 1999.
- [40798] 14350.Chestkov, A. V.; Baka, I. D.; Kost, M. V.; Georgiev,
G. P.; Buchman, V. L.: The d4 gene family in the human
genome. *Genomics* 36: 174–177, 1996.
- [40799] 14351.Gabig, T. G.; Crean, C. D.; Klenk, A.; Long, H.;
Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Quincey, D.;
Parente, F.; Lespinasse, F.; Carle, G. F.; Gaudray, P.; and 13
others: Expression and chromosomal localization of the
Requiem gene. *Mammalian Genome* 9: 660–665, 1998.
- [40800] 14352.Gabig, T. G.; Mantel, P. L.; Rosli, R.; Crean, C. D.:
Requiem: a novel zinc finger gene essential for apoptosis
in myeloid cells. *J. Biol. Chem.* 269: 29515–29519, 1994.
- [40801] 14353.Smith, J. E., Jr.; Bollekens, J. A.; Inghirami, G.;
Takeshita, K.: Cloning and mapping of the MEIS1 gene, the
human homolog of murine leukemogenic gene. *Ge-
nomics* 43: 99–103, 1997.
- [40802] 14354.Duperat, V. G.; Jacquelin, B.; Boisseau, P.; Arveiler,
B.; Nurden, A. T.: Protease-activated receptor genes are

clustered on 5q13. Blood 92:25–31, 1998.

- [40803] 14355. Ishihara, H.; Connolly, A. J.; Zeng, D.; Kahn, M. L.; Zheng, Y.W.; Timmons, C.; Tram, T.; Coughlin, S. R.: Protease-activated receptor3 is a second thrombin receptor in humans. Nature 386: 502–506, 1997.
- [40804] 14356. Kahn, M. L.; Zheng, Y. W.; Huang, W.; Bigornia, V.; Zeng, D.; Moff, S.; Farese, R. V., Jr.; Tam, C.; Coughlin, S. R.: A dual thrombin receptor system for platelet activation. Nature 394: 690–694, 1998.
- [40805] 14357. Nakanishi–Matsui, M.; Zheng, Y.–W.; Sulciner, D. J.; Weiss, E.J.; Ludeman, M. J.; Coughlin, S. R.: PAR3 is a co-factor for PAR4 activation by thrombin. Nature 404: 609–613, 2000.
- [40806] 14358. Sambrano, G. R.; Weiss, E. J.; Zheng, Y.–W.; Huang, W.; Coughlin, S. R.: Role of thrombin signalling in platelets in haemostasis and thrombosis. Nature 413: 74–78, 2001.
- [40807] 14359. Schmidt, V.; Wainer, J.; Bahou, W.: Identification and characterization of the human proteolytically activated receptor–3 (PAR–3) gene within the PAR gene cluster by positional mapping. (Abstract) Blood (suppl.1) 90: 284a only, 1997.
- [40808] 14360. Paulweber, B.; Wiebusch, H.; Miesenboeck, G.; Funke, H.; Assmann, G.; Hoelzl, B.; Sippl, M. J.; Friedl, W.;

Patsch, J. R.; Sandhofer, F.: Molecular basis of lipoprotein lipase deficiency in two Austrian families with type I hyperlipoproteinemia. *Atherosclerosis* 86: 239–250, 1991.

[40809] 14361. Abe, M.; Suzuki, T.; Deguchi, T.: An improved method for genotyping of N-acetyltransferase polymorphism by polymerase chain reaction. *Jpn. J. Hum. Genet.* 38: 163–168, 1993.

[40810] 14362. Azad Khan, A. K.; Nurazzaman, M.; Truelove, S. C.: The effect of the acetylator phenotype on the metabolism of sulphasalazine in man. *J. Med. Genet.* 20: 30–36, 1983.

[40811] 14363. Bell, D. A.; Taylor, J. A.; Butler, M. A.; Stephens, E. A.; Wiest, J.; Brubaker, L. H.; Kadlubar, F. F.; Lucier, G. W.: Genotype/phenotype discordance for human arylamine N-acetyltransferase (NAT2) reveals a new slow-acetylator allele common in African-Americans. *Carcinogenesis* 14: 1689–1692, 1993.

[40812] 14364. Blum, M.; Grant, D.; Demierre, A.; Meyer, U. A.: N-acetylation pharmacogenetics: a gene deletion causes absence of arylamine N-acetyltransferase in liver of slow acetylator rabbits. *Proc. Nat. Acad. Sci.* 86: 9554–9557, 1989.

[40813] 14365. Burrows, A. W.; Hockaday, T. D. R.; Mann, J. I.; Taylor, J. G.: Diabetic dimorphism according to acetylator sta-

tus. Brit. Med.J. 1: 208–210, 1978.

- [40814] 14366.Cascorbi, I.; Brockmoller, J.; Bauer, S.; Reum, T.; Roots, I.:NAT2*12A (803A–G) codes for rapid arylamine N–acetylation in humans. Pharmacogenetics 6:257–259, 1996.
- [40815] 14367.Cascorbi, I.; Drakoulis, N.; Brockmoller, J.; Maurer, A.; Sperling,K.; Roots, I.: Arylamine N–acetyltransferase (NAT2) mutations andtheir allelic linkage in unrelated Caucasian individuals: correlationwith phenotypic activity. Am. J. Hum. Genet. 57: 581–592, 1995.
- [40816] 14368.Delomenie, C.; Sica, L.; Grant, D. M.; Krishnamoorthy, R.; Dupret,J.–M.: Genotyping of the polymorphic N–acetyltransferase (NAT2*)gene locus in two native African populations. Pharmacogenetics 6:177–185, 1996.
- [40817] 14369.Evans, D. A. P.: N–acetyltransferase. Pharm. Therap. 42: 157–234,1989.
- [40818] 14370.Evans, D. A. P.; Manley, K. A.; McKusick, V. A.: Genetic controlof isoniazid metabolism in man. Brit. Med. J. 2: 485–491, 1960.
- [40819] 14371.Evans, D. A. P.; White, T. A.: Human acetylation polymorphism. J.Lab. Clin. Med. 63: 394–403, 1964.
- [40820] 14372.Frymoyer, J. W.; Jacox, R. F.: Studies of genetically controlledsulfadiazine acetylation in rabbit livers: possible

identification of the heterozygous trait. J. Lab. Clin. Med. 62: 905–909, 1963.

- [40821] 14373. Glowinski, I. B.; Weber, W. W.: Biochemical characterization of genetically variant aromatic amine N-acetyltransferases in A/J and C57BL/6J mice. J. Biol. Chem. 257: 1431–1437, 1982.
- [40822] 14374. Glowinski, I. B.; Weber, W. W.: Genetic regulation of aromatic amine N-acetylation in inbred mice. J. Biol. Chem. 257: 1424–1430, 1982.
- [40823] 14375. Grant, D. M.; Morike, K.; Eichelbaum, M.; Meyer, U. A.: Acetylation pharmacogenetics: the slow acetylator phenotype is caused by decreased or absent arylamine N-acetyltransferase in human liver. J. Clin. Invest. 85: 968–972, 1990.
- [40824] 14376. Grant, D. M.; Tang, B. K.; Kalow, W.: A simple test for acetylator phenotype using caffeine. Brit. J. Clin. Pharm. 17: 459–464, 1984.
- [40825] 14377. Hoo, J. J.; Hussein, L.; Goedde, H. W.: A simplified micromethod for the determination of the acetylator phenotype. J. Clin. Chem. Clin. Biochem. 15: 329–331, 1977.
- [40826] 14378. Ilett, K. F.; David, B. M.; Detchon, P.; Castleden, W. M.; Kwa, R.: Acetylation phenotype in colorectal carcinoma. Cancer Res. 47: 1466–1469, 1987.

- [40827] 14379.Iselius, L.; Evans, D. A. P.: Formal genetics of isoniazid metabolism in man. Clin. Pharmacokinetics 8: 541–544, 1983.
- [40828] 14380.Kukongviriyapan, V.; Lulitanond, V.; Areejitranusorn, C.; Kongyingyose, B.; Laupattarakasem, P.: N-acetyltransferase polymorphism in Thailand. Hum.Hered. 34: 246–249, 1984.
- [40829] 14381.Kutt, H.; Brennan, R.; Dehejia, H.; Verebely, K.: Diphenylhydantoin intoxication: a complication of isoniazid therapy. Am. Rev. Resp.Dis. 101: 377–384, 1970.
- [40830] 14382.Lang, N. P.; Chu, D. Z. J.; Hunter C. F.; Kendall, D. C.; Flammang, T. J.; Kadlubar, F. F.: Role of aromatic amine acetyltransferase in human colorectal cancer. Arch. Surg. 121: 1259–1261, 1986.
- [40831] 14383.Lee, E. J. D.; Zhao, B.; Seow-Choen, F.: Relationship between polymorphism of N-acetyltransferase gene and susceptibility to colorectal carcinoma in a Chinese population. Pharmacogenetics 8: 513–517, 1998.
- [40832] 14384.Lin, H. J.; Han, C.-Y.; Lin, B. K.; Hardy, S.: Ethnic distribution of slow acetylator mutations in the polymorphic N-acetyltransferase (NAT2) gene. Pharmacogenetics 4: 125–134, 1994.
- [40833] 14385.Lin, H. J.; Han, C.-Y.; Lin, B. K.; Hardy, S.: Slow

acetylator mutations in the human polymorphic N-acetyltransferase gene in 786 Asians, blacks, Hispanics, and whites: application to metabolic epidemiology. *Am. J. Hum. Genet.* 52: 827–834, 1993.

- [40834] 14386. Mashimo, M.; Suzuki, T.; Abe, M.; Deguchi, T.: Molecular genotyping of N-acetylation polymorphism to predict phenotype. *Hum. Genet.* 90:139–143, 1992.
- [40835] 14387. Mattano, S. S.; Weber, W. W.: Kinetics of arylamine N-acetyltransferase in tissues from rapid and slow acetylator mice. *Carcinogenesis* 8:133–137, 1987.
- [40836] 14388. McLaren, E. H.; Burden, A. C.; Moorhead, P. J.: Acetylator phenotype in diabetic neuropathy. *Brit. Med. J.* 2: 291–293, 1977.
- [40837] 14389. McQueen, C. A.; Maslansky, C. J.; Glowinski, I. B.; Crescenzi, S. B.; Weber, W. W.; Williams, G. M.: Relationship between the genetically determined acetylator phenotype and DNA damage induced by hydralazine and 2-aminofluorene in cultured rabbit hepatocytes. *Proc. Nat. Acad. Sci.* 79: 1269–1272, 1982.
- [40838] 14390. Giralt, M.; Park, E. A.; Gurney, A. L.; Liu, J.; Hakimi, P.; Hanson, R. W.: Identification of a thyroid hormone response element in the phosphoenolpyruvate carboxykinase (GTP) gene: evidence for synergistic interaction between

thyroid hormone and cAMP cis-regulatory elements. J.Biol. Chem. 266: 21991–21996, 1991.

- [40839] 14391. Pilz, A. J.; Willer, E.; Povey, S.; Abbott, C. M.: The genes coding for phosphoenolpyruvate carboxykinase-1 (PCK1) and neuronal nicotinic acetylcholine receptor alpha-4 subunit (CHRNA4) map to human chromosome 20, extending the known region of homology with mouse chromosome 2. Ann.Hum. Genet. 56: 289–293, 1992.
- [40840] 14392. Olswang, Y.; Cohen, H.; Papo, O.; Cassuto, H.; Croniger, C. M.; Hakimi, P.; Tilghman, S. M.; Hanson, R. W.; Reshef, L.: A mutation in the peroxisome proliferator-activated receptor gamma-binding site in the gene for the cytosolic form of phosphoenolpyruvate carboxykinase reduces adipose tissue size and fat content in mice. Proc. Nat. Acad.Sci. 99: 625–630, 2002.
- [40841] 14393. Stoffel, M.; Xiang, K.; Espinosa, R., III; Cox, N. J.; Le Beau, M. M.; Bell, G. I.: cDNA sequence and localization of polymorphic human cytosolic phosphoenolpyruvate carboxykinase gene (PCK1) to chromosome 20, band q13.31: PCK1 is not tightly linked to maturity-onset diabetes of the young. Hum. Molec. Genet. 2: 1–4, 1993.
- [40842] 14394. Ting, C.-N.; Burgess, D. L.; Chamberlain, J. S.; Keith, T. P.; Falls, K.; Meisler, M. H.: Phosphoenolpyruvate

carboxykinase (GTP):characterization of the human PCK1 gene and localization distal toMODY on chromosome 20. Genomics 16: 698–706, 1993.

- [40843] 14395.Vidnes, J.; Sovik, O.: Gluconeogenesis in infancy and childhood.III. Deficiency of the extramitochondrial form of hepatic phosphoenolpyruvatecarboxykinase in a case of persistent neonatal hypoglycaemia. ActaPaediat. Scand. 65: 301–312, 1976.
- [40844] 14396.Yu, H.; Thun, R.; Chandrasekharappa, S.; Trent, J. M.; Zhang, J.;Meisler, M. H.: Human PCK1 encoding phosphoenolpyruvate carboxykinaseis located on chromosome 20q13.2. Genomics 15: 219–221, 1993.
- [40845] 14397.Pepe, G.; Chimienti, G.; Resta, F.; Di Perna, V.; Taricone, C.;Lovecchio, M.; Colacicco, A. M.; Capurso, A.: A new Italian caseof lipoprotein lipase deficiency: a leu365-to-val change resultingin loss of enzyme activity. Biochem. Biophys. Res. Commun. 199:570–576, 1994.
- [40846] 14398.Reymer, P. W. A.; Groenemeyer, B. E.; Gagne, E.; Miao, L.; Appelman,E. E. G.; Seidel, J. C.; Kromhout, D.; Bijvoet, S. M.; van de Oever,K.; Bruin, T.; Hayden, M. R.; Kastelein, J. J. P.: A frequently occurringmutation in the lipoprotein lipase gene (Asn291Ser) contributes tothe expression of familial combined hyperlipidemia. Hum.

Molec. Genet. 4:1543–1549, 1995.

- [40847] 14399.Samuels, M. E.; Forbey, K. C.; Reid, J. E.; Abkevich, V.; Bulka,K.; Wardell, B. R.; Bowen, B. R.; Hopkins, P. N.; Hunt, S. C.; Ballinger,D. G.; Skolnick, M. H.; Wagner, S.: Identification of a common variant in the lipoprotein lipase gene in a large Utah kindred ascertained for coronary heart disease: the –93G/D9N variant predisposes to lowHDL–C/high triglycerides. Clin. Genet. 59: 88–98, 2001.
- [40848] 14400.Schreibman, P. H.; Arons, D. L.; Saudek, C. D.; Arky, R. A.:Abnormal lipoprotein lipase in familial exogenous hypertriglyceridemia. J.Clin. Invest. 52: 2074–2082, 1973.
- [40849] 14401.Sprecher, D. L.; Kobayashi, J.; Rymaszewski, M.; Goldberg, I.J.; Harris, B. V.; Bellet, P. S.; Ameis, D.; Yunker, R. L.; Black,D. M.; Stein, E. A.; Schotz, M. C.; Wiginton, D. A.: Trp64–to–nonsense mutation in the lipoprotein lipase gene. J. Lipid Res. 33: 859–866,1992.
- [40850] 14402.Sternowsky, H. J.; Gaertner, U.; Stahnkel, N.; Kaukel, E.: Juvenilefamilial hypertriglyceridemia and growth retardation: clinical andbiochemical observations in three siblings. Europ. J. Pediat. 125:59–70, 1977.
- [40851] 14403.Takagi, A.; Ikeda, Y.; Tsutsumi, Z.; Shoji, T.; Yamamoto, A.:Molecular studies on primary lipoprotein li-

pase (LPL) deficiency:one base deletion (G-916) in exon 5 of LPL gene causes no detectableLPL protein due to the absence of LPL mRNA transcript. J. Clin. Invest. 89:581-591, 1992.

- [40852] 14404.Weinstock, P. H.; Bisgaier, C. L.; Aalto-Setala, K.; Radner, H.;Ramakrishnan, R.; Levak-Frank, S.; Essenburg, A. D.; Zechner, R.;Breslow, J. L.: Severe hypertriglyceridemia, reduced high densitylipoprotein, and neonatal death in lipoprotein lipase knockout mice:mild hypertriglyceridemia with impaired very low density lipoprotein clearance in heterozygotes. J. Clin. Invest. 96: 2555-2568, 1995.
- [40853] 14405.Wessler, S.; Avioli, L. A.: Familial hyperlipoproteinemia. J.A.M.A. 207:929-937, 1969.
- [40854] 14406.Wilson, D. E.; Edwards, C. Q.; Chan, I.-F.: Phenotypic heterogeneityin the extended pedigree of a proband with lipoprotein lipase deficiency. Metabolism 32:1107-1114, 1983.
- [40855] 14407.Wilson, D. E.; Emi, M.; Iverius, P.-H.; Hata, A.; Wu, L. L.; Hillas,E.; Williams, R. R.; Lalouel, J.-M.: Phenotypic expression of heterozygouslipoprotein lipase deficiency in the extended pedigree of a probandhomozygous for a missense mutation. J. Clin. Invest. 86: 735-750,1990.

- [40856] 14408. Wilson, D. E.; Hata, A.; Kwong, L. K.; Lingam, A.; Shuhua, J.; Ridinger, D. N.; Yeager, C.; Kaltenborn, K. C.; Iverius, P.-H.; Lalouel, J.-M.: Mutations in exon 3 of the lipoprotein lipase gene segregating in a family with hypertriglyceridemia, pancreatitis, and non-insulin-dependent diabetes. *J. Clin. Invest.* 92: 203–211, 1993.
- [40857] 14409. Wion, K. L.; Kirchgessner, T. G.; Lusis, A. J.; Schotz, M. C.; Lawn, R. M.: Human lipoprotein lipase complementary DNA sequence. *Science* 235:1638–1641, 1987.
- [40858] 14410. Wittrup, H. H.; Tybjaerg-Hansen, A.; Abildgaard, S.; Steffensen, R.; Schnohr, P.; Nordestgaard, B. G.: A common substitution (asn291ser) in lipoprotein lipase is associated with increased risk of ischemic heart disease. *J. Clin. Invest.* 99: 1606–1613, 1997.
- [40859] 14411. Wittrup, H. H.; Tybjaerg-Hansen, A.; Steffensen, R.; Deeb, S.S.; Brunzell, J. D.; Jensen, G.; Nordestgaard, B. G.: Mutations in the lipoprotein lipase gene associated with ischemic heart disease in men: the Copenhagen City Heart Study. *Arterioscler. Thromb. Vasc. Biol.* 19: 1535–1540, 1999.
- [40860] 14412. Wood, S.; Schertzer, M.; Hayden, M.; Ma, Y.: Support for founder effect for two lipoprotein lipase (LPL) gene

mutations in French Canadians by analysis of GT microsatellites flanking the LPL gene. Hum. Genet. 91:312–316, 1993.

- [40861] 14413. McQueen, C. A.; Weber, W. W.: Characterization of human lymphocyte N-acetyltransferase and its relationship to the isoniazid acetylator polymorphism. Biochem. Genet. 18: 889–904, 1980.
- [40862] 14414. Nhachi, C. F. B.: Polymorphic acetylation of sulphamethazine in a Zimbabwe population. J. Med. Genet. 25: 29–31, 1988.
- [40863] 14415. Ohsako, S.; Deguchi, T.: Cloning and expression of cDNAs for polymorphic and monomorphic arylamine N-acetyltransferases from human liver. J. Biol. Chem. 265: 4630–4634, 1990.
- [40864] 14416. Parker, J. M.: Human variability in the metabolism of sulfamethazine. Hum. Hered. 19: 402–409, 1969.
- [40865] 14417. Penketh, R. J. A.; Gibney, S. F. A.; Nurse, G. T.; Hopkinson, D. A.: Acetylator phenotypes in Papua New Guinea. J. Med. Genet. 20: 37–40, 1983.
- [40866] 14418. Roberts–Thomson, I. C.; Ryan, P.; Khoo, K. K.; Hart, W. J.; McMichael, A. J.; Butler, R. N.: Diet, acetylator phenotype, and risk of colorectal neoplasia. Lancet 347: 1372–1374, 1996.

- [40867] 14419. Roots, I.; Drakoulis, N.; Brockmoller, J.; Janicke, I.; Cuprunov, M.; Ritter, J.: Hydroxylation and acetylation phenotypes as genetic risk factors in certain malignancies. In: Kato, W.; Estabrook, R. W.; Cayen, M. N. (eds.): Xenobiotic Metabolism and Disposition. London: Taylor and Francis 1989. Pp. 499–506.
- [40868] 14420. Schloot, W.; Goedde, H. W.: Studies on the polymorphism of isoniazid (INH) acetylation in rhesus monkeys (*Macaca mulatta*). *Acta Genet. Statist. Med.* 18: 394–398, 1968.
- [40869] 14421. Schnakenberg, E.; Ehlers, C.; Feyerabend, W.; Werdin, R.; Hubotter, R.; Dreikorn, K.; Schloot, W.: Genotyping of the polymorphic N-acetyltransferase (NAT2) and loss of heterozygosity in bladder cancer patients. *Clin. Genet.* 53: 396–402, 1998.
- [40870] 14422. Sonnhag, C.; Karlsson, E.; Hed, J.: Procainamide-induced lupus erythematosus-like syndrome in relation to acetylator phenotype and plasma levels of procainamide. *Acta Med. Scand.* 206: 245–251, 1979.
- [40871] 14423. Sunahara, S.; Urano, M.; Ogawa, M.: Genetical and geographic studies on isoniazid inactivation. *Science* 134: 1530–1531, 1961.
- [40872] 14424. Tannen, R. H.; Weber, W. W.: Inheritance of acety-

lator phenotype in mice. J. Pharm. Exp. Therap. 213: 480–484, 1980.

- [40873] 14425. Timbrell, J. A.; Wright, J. M.; Baillie, T. A.: Monoacetylhydrazine as a metabolite of isoniazid in man. Clin. Pharm. Therap. 22: 602–609, 1977.
- [40874] 14426. Vansant, J.; Woosley, R. L.; John, J. T.; Sergent, J. S.: Normal distribution of acetylation phenotypes in systemic lupus erythematosus. Arthritis Rheum. 21: 192–195, 1978.
- [40875] 14427. Weber, W. W.: The Acetylator Genes and Drug Response. New York: Oxford Univ. Press (pub.) 1987.
- [40876] 14428. Woosley, R. L.; Drayer, D. E.; Reidenberg, M. M.; Nies, A. S.; Carr, K.; Oates, J. A.: Effect of acetylator phenotype on the rate at which procainamide induces antinuclear antibodies and the lupus syndrome. New Eng. J. Med. 298: 1157–1159, 1978.
- [40877] 14429. Pace, P.; Johnson, M.; Tan, W. M.; Mosedale, G.; Sng, C.; Hoatlin, M.; de Winter, J.; Joenje, H.; Gergely, F.; Patel, K. J.: FANCE: the link between Fanconi anaemia complex assembly and activity. EMBO J. 21: 3414–3423, 2002.
- [40878] 14430. Hodge, S. E.; Anderson, C. E.; Neiswanger, K.; Field, L. L.; Spence, M. A.; Sparkes, R. S.; Sparkes, M. C.; Crist, M.; Terasaki, P. I.; Rimoim, D. L.; Rotter, J. I.: Close genetic linkage between diabetes mellitus and Kidd blood group.

Lancet II: 893–895, 1981.

- [40879] 14431. Faiyaz–Ul–Haque, M.; Ahmad, W.; Zaidi, S. H. E.; Haque, S.; Teebi, A. S.; Ahmad, M.; Cohn, D. H.; Tsui, L. C.: Mutation in the cartilage–derived morphogenetic protein–1 (CDMP1) gene in a kindred affected with fibular hypoplasia and complex brachydactyly (DuPan syndrome). Clin. Genet. 61:454–458, 2002.
- [40880] 14432. Touchman, J. W.; Anikster, Y.; Dietrich, N. L.; Braden Maduro, V. V.; McDowell, G.; Shotelersuk, V.; Bouffard, G. G.; Beckstrom–Sternberg, S. M.; Gahl, W. A.; Green, E. D.: The genomic region encompassing the nephropathic cystinosis gene (CTNS): complete sequencing of a 200–kb segment and discovery of a novel gene within the common cystinosis–causing deletion. Genome Res. 10: 165–173, 2000.
- [40881] 14433. Shapiro, M. B.; Senapathy, P.: RNA splice junctions of different classes of eukaryotes: sequence statistics and functional implications in gene expression. Nucleic Acids Res. 15: 7155–7174, 1987.
- [40882] 14434. Jobard, F.; Lefevre, C.; Karaduman, A.; Blanchet–Bardon, C.; Emre, S.; Weissenbach, J.; Ozguc, M.; Lathrop, M.; Prud'homme, J.–F.; Fischer, J.: Lipoxxygenase–3 (ALOXE3) and 12(R)–lipoxxygenase (ALOX12B) are mutated in non–

bullous congenital ichthyosiform erythroderma

(NCIE)linked to chromosome 17p13.1. Hum. Molec. Genet. 11: 107–113, 2002.

[40883] 14435.Varon, R.; Vissinga, C.; Platzer, M.; Cerosaletti, K. M.; Chrzanowska,K. H.; Saar, K.; Beckmann, G.; See-manova, E.; Cooper, P. R.; Nowak,N. J.; Stumm, M.; Weemaes, C. M. R.; Gatti, R. A.; Wilson, R. K.;Digweed, M.; Rosenthal, A.; Sperling, K.; Concannon, P.; Reis, A.: Nibrin, a novel DNA double-strand break repair protein, is mutated in Nijmegen breakage syndrome. Cell 93: 467–476, 1998.

[40884] 14436.ten Hove, T.; Corbaz, A.; Amitai, H.; Aloni, S.; Belzer, I.; Graber,P.; Drillenburger, P.; van Deventer, S. J.; Chvatchko, Y.; Te Velde,A. A.: Blockade of endogenous IL-18 ameliorates TNBS-induced colitisby decreasing local TNF-alpha production in mice. Gastroenterology 121:1372–1379, 2001.

[40885] 14437.Ishikawa, K.; Nagase, T.; Suyama, M.; Miyajima, N.; Tanaka, A.;Kotani, H.; Nomura, N.; Ohara, O.: Prediction of the coding sequencesof unidentified human genes. X. The complete sequences of 100 newcDNA clones from brain which can code for large proteins in vitro. DNARes. 5: 169–176, 1998.

- [40886] 14438.Kipreos, E. T.; Lander, L. E.; Wing, J. P.; He, W. W.; Hedgecock, E. M.: cul-1 is required for cell cycle exit in *C. elegans* and identifies a novel gene family. *Cell* 85: 829–839, 1996.
- [40887] 14439.Rasooly, R. S.: Personal Communication. Baltimore, Md. 9/29/1998.
- [40888] 14440.Wang, P. J.; McCarrey, J. R.; Yang, F.; Page, D. C.: An abundance of X-linked genes expressed in spermatogonia. *Nature Genet.* 27:422–426, 2001.
- [40889] 14441.Herold, A.; Suyama, M.; Rodrigues, J. P.; Braun, I. C.; Kutay, U.; Carmo-Fonseca, M.; Bork, P.; Izaurralde, E.: TAP (NXF1) belongs to a multigene family of putative RNA export factors with a conserved modular architecture. *Molec. Cell. Biol.* 20: 8996–9008, 2000.
- [40890] 14442.Yang, J.; Bogerd, H. P.; Wang, P. J.; Page, D. C.; Cullen, B. R.: Two closely related human nuclear export factors utilize entirely distinct export pathways. *Molec. Cell* 8: 397–406, 2001.
- [40891] 14443.Ikeda, M.; Ishida, O.; Hinoi, T.; Kishida, S.; Kikuchi, A.: Identification and characterization of a novel protein interacting with Ral-binding protein 1, a putative effector protein of Ral. *J. Biol. Chem.* 273:814–821, 1998.
- [40892] 14444.Koshiba, S.; Kigawa, T.; Iwahara, J.; Kikuchi, A.;

Yokoyama, S.: Solution structure of the Eps15 homology domain of a human POB1(partner of RalBP1). FEBS Lett. 442: 138–142, 1999.

[40893] 14445.Crackower, M. A.; Sarao, R.; Oudit, G. Y.; Yagil, C.; Kozieradzki, I.; Scanga, S. E.; Oliveira–dos–Santos, A. J.; da Costa, J.; Zhang, L.; Pei, Y.; Scholey, J.; Ferrario, C. M.; Manoukian, A. S.; Chappell, M. C.; Backx, P. H.; Yagil, Y.; Penninger, J. M.: Angiotensin–converting enzyme 2 is an essential regulator of heart function. Nature 417:822–828, 2002.

[40894] 14446.Scanlan, M. J.; Gordan, J. D.; Williamson, B.; Stockert, E.; Bander, N. H.; Jongeneel, V.; Gure, A. O.; Jager, D.; Jager, E.; Knuth, A.; Chen, Y.–T.; Old, L. J.: Antigens recognized by autologous antibody in patients with renal–cell carcinoma. Int. J. Cancer 83: 456–464, 1999.

[40895] 14447.Yan, Z.; Fedorov, S. A.; Mumby, M. C.; Williams, R. S.: PR48, a novel regulatory subunit of protein phosphatase 2A, interacts with Cdc6 and modulates DNA replication in human cells. Molec. Cell. Biol. 20:1021–1029, 2000.

[40896] 14448.Gitzelmann, R.: Hereditary galactokinase deficiency, a newly recognized cause of juvenile cataracts. Pediat. Res. 1: 14–23, 1967.

- [40897] 14449.Kalaydjieva, L.; Perez-Lezaun, A.; Angelicheva, D.; Onengut, S.;Dye, D.; Bosshard, N. U.; Jordanova, A.; Savov, A.; Yanakiev, P.;Kremensky, I.; Radeva, B.; Hallmayer, J.; Markov, A.; Nedkova, V.;Tournev, I.; Aneva, L.; Gitzelmann, R.: A founder mutation in theGK1 gene is responsible for galactokinase deficiency in Roma (Gypsies). *Am.J. Hum. Genet.* 65: 1299–1307, 1999.
- [40898] 14450.Stratakis, C. A.; Lin, J.–P.; Pras, E.; Rennert, O. M.; Bourdony,C. J.; Chan, W.–Y.: Segregation of Allgrove (triple–A) syndrome inPuerto Rican kindreds with chromosome 12 (12q13) polymorphic markers. *Proc.Assoc. Am. Phys.* 109: 478–482, 1997.
- [40899] 14451.Tullio–Pelet, A.; Salomon, R.; Hadj–Rabia, S.; Mugnier, C.; deLaet, M.–H.; Chaouachi, B.; Bakiri, F.; Brottier, P.; Cattolico, L.;Penet, C.; Begeot, M.; Naville, D.; Nicolino, M.; Chaussain, J.–L.;Weissenbach, J.; Munnich, A.; Lyonnet, S.: Mutant WD–repeat proteinin triple–A syndrome. *Nature Genet.* 26: 332–335, 2000.
- [40900] 14452.Camacho, J. A.; Obie, C.; Biery, B.; Goodman, B. K.; Hu, C.–A.;Almashanu, S.; Steel, G.; Casey, R.; Lambert, M.; Mitchell, G. A.;Valle, D.: Hyperornithinaemia–hyperammonaemia–homocitrullinuria syndromeis caused by mutations in a gene encoding a mitochondrial ornithinetrans–

porter. *Nature Genet.* 22: 151–158, 1999.

[40901] 14453.Darnell, J. E., Jr.: Reflections on STAT3, STAT5, and STAT6 as fat STATs. *Proc. Nat. Acad. Sci.* 93: 6221–6224, 1996.

[40902] 14454.Ghilardi, N.; Ziegler, S.; Wiestner, A.; Stoffel, R.; Heim, M.H.; Skoda, R. C.: Defective STAT signaling by the leptin receptor in diabetic mice. *Proc. Nat. Acad. Sci.* 93: 6231–6235, 1996.

[40903] 14455.Mohandas, T. K.; Chen, X.-N.; Rowe, L. B.; Birkenmeier, E. H.; Fanning, A. S.; Anderson, J. M.; Korenberg, J. R.: Localization of the tight junction protein gene TJP1 to human chromosome 15q13, distal to the Prader-Willi/Angelman region, and to mouse chromosome 7. *Genomics* 30:594–597, 1995.

[40904] 14456.Willott, E.; Balda, M. S.; Fanning, A. S.; Jameson, B.; Van Itallie, C.; Anderson, J. M.: The tight junction protein ZO-1 is homologous to the *Drosophila* discs-large tumor suppressor protein of septate junctions. *Proc. Nat. Acad. Sci.* 90: 7834–7838, 1993.

[40905] 14457.Blanar, M. A.; Crossley, P. H.; Peters, K. G.; Steingrimsson, E.; Copeland, N. G.; Jenkins, N. A.; Martin, G. R.; Rutter, W. J.: Meso1, a basic helix-loop-helix protein involved in mammalian presomitic mesoderm development.

Proc. Nat. Acad. Sci. 92: 5870–5874, 1995.

[40906] 14458. Burgess, R.; Rawls, A.; Brown, D.; Bradley, A.; Olson, E. N.: Requirement of the paraxis gene for somite formation and musculoskeletal patterning. *Nature* 384: 570–573, 1996.

[40907] 14459. Hidai, H.; Quertermous, E. E.; Espinosa, R., III; Le Beau, M. M.; Quertermous, T.: Genomic organization and chromosomal localization of the gene TCF15 encoding the early mesodermal basic helix–loop–helix factor bHLH-EC2. *Genomics* 30: 598–601, 1995.

[40908] 14460. Quertermous, E. E.; Hidai, H.; Blonar, M. A.; Quertermous, T.: Cloning and characterization of a basic helix–loop–helix protein expressed in early mesoderm and the developing somites. *Proc. Nat. Acad. Sci.* 91: 7066–7070, 1994.

[40909] 14461. Quertermous, T.: Personal Communication. Nashville, Tenn. 1/22/1996.

[40910] 14462. Clevidence, D. E.; Overdier, D. G.; Peterson, R. S.; Porcella, A.; Ye, H.; Paulson, K. E.; Costa, R. H.: Members of the HNF-3/forkhead family of transcription factors exhibit distinct cellular expression patterns in lung and regulate the surfactant protein B promoter. *Dev. Biol.* 166: 195–209, 1994.

- [40911] 14463.Hellqvist, M.; Mahlapuu, M.; Blixt, A.; Enerback, S.; Carlsson,P.: The human forkhead protein FREAC-2 contains two functionally redundant activation domains and interacts with TBP and TFIIB. J.Biol. Chem. 273: 23335-23343, 1998.
- [40912] 14464.Hellqvist, M.; Mahlapuu, M.; Samuelsson, L.; Enerback, S.; Carlsson,P.: Differential activation of lung-specific genes by two forkhead proteins, FREAC-1 and FREAC-2. J. Biol. Chem. 271: 4482-4490, 1996.
- [40913] 14465.Larsson, C.; Hellqvist, M.; Pierrou, S.; White, I.; Enerback, S.; Carlsson, P.: Chromosomal localization of six human forkhead genes, freac-1 (FKHL5), -3 (FKHL7), -4 (FKHL8), -5 (FKHL9), -6 (FKHL10), and -8 (FKHL12). Genomics 30: 464-469, 1995.
- [40914] 14466.Pierrou, S.; Hellqvist, M.; Samuelsson, L.; Enerback, S.; Carlsson,P.: Cloning and characterization of seven human forkhead proteins: binding site specificity and DNA bending. EMBO J. 13: 5002-5012, 1994.
- [40915] 14467.Ernstsson, S.; Pierrou, S.; Hulander, M.; Cederberg, A.; Hellqvist,M.; Carlsson, P.; Enerback, S.: Characterization of the human forkhead gene FREAC-4. J. Biol. Chem. 271: 21094-21099, 1996.
- [40916] 14468.Hulander, M.; Wurst, W.; Carlsson, P.; Enerback, S.:

The wingedhelix transcription factor Fkh10 is required for normal development of the inner ear. *Nature Genet.* 20: 374–376, 1998.

- [40917] 14469. De Smaele, E.; Zazzeroni, F.; Papa, S.; Nguyen, D. U.; Jin, R.; Jones, J.; Cong, R.; Franzoso, G.: Induction of gadd45–beta by NF–kappa–B downregulates pro–apoptotic JNK signaling. *Nature* 414: 308–313, 2001.
- [40918] 14470. Dong, C.; Yang, D. D.; Tournier, C.; Whitmarsh, A. J.; Xu, J.; Davis, R. J.; Flavell, R. A.: JNK is required for effector T–cell function but not for T–cell activation. *Nature* 405: 91–94, 2000.
- [40919] 14471. Gupta, S.; Barrett, T.; Whitmarsh, A. J.; Cavanagh, J.; Sluss, H. K.; Derijard, B.; Davis, R. J.: Selective interaction of JNK protein kinase isoforms with transcription factors. *EMBO J.* 15: 2760–2770, 1996.
- [40920] 14472. Tournier, C.; Hess, P.; Yang, D. D.; Xu, J.; Turner, T. K.; Nimnual, A.; Bar–Sagi, D.; Jones, S. N.; Flavell, R. A.; Davis, R. J.: Requirement of JNK for stress–induced activation of the cytochrome c–mediated death pathway. *Science* 288: 870–874, 2000.
- [40921] 14473. Daugherty, B. L.; Springer, M. S.: The beta–chemokine receptor genes CCR1 (CMKBR1), CCR2 (CMKBR2), and CCR3 (CMKBR3) cluster within 285 kb on

human chromosome 3p21. *Genomics* 41: 294–295, 1997.

[40922] 14474. Ayyanathan, K.; Naylor, S. L.; Kunapuli, S. P.: Structural characterization and fine chromosomal mapping of the human P2Y(1) purinergic receptor gene (P2RY1). *Somat. Cell Molec. Genet.* 22: 419–424, 1996.

[40923] 14475. Ayyanathan, K.; Webbs, T. E.; Sandhu, A. K.; Athwal, R. S.; Barnard, E. A.; Kunapuli, S. P.: Cloning and chromosomal localization of the human P2Y1 purinoceptor. *Biochem. Biophys. Res. Commun.* 218: 783–788, 1996.

[40924] 14476. Leon, C.; Hechler, B.; Freund, M.; Eckly, A.; Vial, C.; Ohlmann, P.; Dierich, A.; LeMeur, M.; Cazenave, J.-P.; Gachet, C.: Defective platelet aggregation and increased resistance to thrombosis in purinergic P2Y1 receptor-null mice. *J. Clin. Invest.* 104: 1731–1737, 1999.

[40925] 14477. Leon, C.; Hechler, B.; Vial, C.; Leray, C.; Cazenave, J.-P.; Gachet, C.: The P2Y(1) receptor is an ADP receptor antagonized by ATP and expressed in platelets and megakaryoblastic cells. *FEBS Letts.* 403:26–30, 1997.

[40926] 14478. Leon, C.; Vial, C.; Cazenave, J.-P.; Gachet, C.: Cloning and sequencing of a human cDNA encoding endothelial P2Y1 purinoceptor. *Gene* 171:295–297, 1996.

[40927] 14479. Matsuda, C.; Hayashi, Y. K.; Ogawa, M.; Aoki, M.; Murayama, K.; Nishino, I.; Nonaka, I.; Arahata, K.; Brown,

R. H., Jr.: The sarcolemmal proteins dysferlin and caveolin-3 interact in skeletal muscle. *Hum.Molec. Genet.* 10: 1761–1766, 2001.

[40928] 14480. Furlong, R. A.; Zhou, C. Y.; Ferguson-Smith, M. A.; Affara, N.A.: Characterization of a kinesin-related gene ATSV, within the tuberous sclerosis locus (TSC1) candidate region on chromosome 9q34. *Genomics* 33:421–429, 1996.

[40929] 14481. Keller, M. P.; Seifried, B. A.; Rabin, B. A.; Chance, P. F.: Mapping of the kinesin-related gene ATSV to chromosome 2q37. *Hum. Genet.* 104:254–256, 1999.

[40930] 14482. Kikkawa, M.; Okada, Y.; Hirokawa, N.: 15-angstrom resolution model of the monomeric kinesin motor, KIF1A. *Cell* 100: 241–252, 2000.

[40931] 14483. Kikkawa, M.; Sablin, E. P.; Okada, Y.; Yajima, H.; Fletterick, R. J.; Hirokawa, N.: Switch-based mechanism of kinesin motors. *Nature* 411:439–445, 2001.

[40932] 14484. Klopfenstein, D. R.; Tomishige, M.; Stuurman, N.; Vale, R. D.: Role of phosphatidylinositol(4,5)bisphosphate organization in membrane transport by the Unc104 kinesin motor. *Cell* 109: 347–358, 2002.

[40933] 14485. Okada, Y.; Yamazaki, H.; Sekine-Aizawa, Y.; Hirokawa, N.: The neuron-specific kinesin superfamily pro-

tein KIF1A is a unique monomeric motor for anterograde axonal transport of synaptic vesicle precursors. *Cell* 81:769–780, 1995.

[40934] 14486. Yonekawa, Y.; Harada, A.; Okada, Y.; Funakoshi, T.; Kanai, Y.; Takei, Y.; Terada, S.; Noda, T.; Hirokawa, N.: Defect in synaptic vesicle precursor transport and neuronal cell death in KIF1A motor protein-deficient mice. *J. Cell Biol.* 141: 431–441, 1998.

[40935] 14487. Kikuta, Y.; Sogawa, K.; Haniu, M.; Kinoshita, M.; Kusunose, E.; Nojima, Y.; Yamamoto, S.; Ichihara, K.; Kusunose, M.; Fujii-Kuriyama, Y.: A novel species of cytochrome P-450 (P-450ib) specific for the small intestine of rabbits: cDNA cloning and its expression in COS cells. *J. Biol. Chem.* 266: 17821–17825, 1991.

[40936] 14488. Ma, J.; Ramachandran, S.; Fiedorek, F. T., Jr.; Zeldin, D. C.: Mapping of the CYP2J cytochrome P450 genes to human chromosome 1 and mouse chromosome 4. *Genomics* 49: 152–155, 1998.

[40937] 14489. Wu, S.; Moomaw, C. R.; Tomer, K. B.; Falck, J. R.; Zeldin, D. C.: Molecular cloning and expression of CYP2J2, a human cytochrome P450 arachidonic acid epoxygenase highly expressed in heart. *J. Biol. Chem.* 271: 3460–3468, 1996.

- [40938] 14490. Chiang, P.-W.; Wang, S.; Smithivas, P.; Song, W.-J.; Ramamoorthy, S.; Hillman, J.; Puett, S.; Van Keuren, M. L.; Crombez, E.; Kumar, A.; Glover, T. W.; Miller, D. E.; Tsai, C.-H.; Blackburn, C. C.; Chen, X.-N.; Sun, Z.; Cheng, J.-F.; Korenberg, J. R.; Kurnit, D. M.: Identification and analysis of the human and murine putative chromatin structure regulator SUPT6H and Supt6h. *Genomics* 34: 328–333, 1996.
- [40939] 14491. Segre, J. A.; Nemhauser, J. L.; Taylor, B. A.; Nadeau, J. H.; Lander, E. S.: Positional cloning of the nude locus: genetic, physical and transcription maps of the region and mutations in the mouse and rat. *Genomics* 28: 549–559, 1995.
- [40940] 14492. Han, J.; Knops, J. F.; Longshore, J. W.; King, P. H.: Localization of human elav-like neuronal protein 1 (Hel-N1) on chromosome 9p21 by chromosome microdissection polymerase chain reaction and fluorescence in situ hybridization. *Genomics* 36: 189–191, 1996.
- [40941] 14493. King, P. H.: Hel-N2: a novel isoform of Hel-N1 which is conserved in rat neural tissue and produced in early embryogenesis. *Gene* 151: 261–265, 1994.
- [40942] 14494. King, P. H.; Levine, T. D.; Fremeau, R. T., Jr.; Keene, J. D.: Mammalian homologs of *Drosophila* ELAV localized

to a neuronal subset can bind in vitro to the 3' UTR of mRNA encoding the Id transcriptional repressor. *J. Neurosci.* 14: 1943–52, 1994.

[40943] 14495. Abel, K. J.; Brody, L. C.; Valdes, J. M.; Erdos, M. R.; McKinley, D. R.; Castilla, L. H.; Merajver, S. D.; Couch, F. J.; Friedman, L. S.; Ostermeyer, E. A.; Lynch, E. D.; King, M.-C.; Welcsh, P. L.; Osborne-Lawrence, S.; Spillman, M.; Bowcock, A. M.; Collins, F. S.; Weber, B. L.: Characterization of EZH1, a human homolog of *Drosophila* enhancer of zeste near BRCA1. *Genomics* 37:161–171, 1996.

[40944] 14496. Akazawa, K.; Yamane, S.; Shiota, H.; Naito, E.: A case of retinoblastoma associated with Rieger's anomaly and 13q deletion. *Jpn. J. Ophthalmol.* 25:321–325, 1981.

[40945] 14497. Stathacopoulos, R. A.; Bateman, J. B.; Sparkes, R. S.; Hepler, R. S.: The Rieger syndrome and a chromosome 13 deletion. *J. Pediatr. Ophthalmol. Strabismus* 24: 198–203, 1987.

[40946] 14498. Ferbus, D.; Le Chalony, C.; Prosperi, M.-T.; Muleris, M.; Vincent-Salomon, A.; Goubin, G.: Identification, nuclear localization, and binding activities of OZF, a human protein solely composed of zinc finger motifs. *Europ. J. Biochem.* 236: 991–995, 1996.

[40947] 14499. Le Chalony, C.; Apiou, F.; Pibouin, L.; Dutrillaux, B.;

Goubin,G.: Constitutive amplification of a zinc finger protein gene in cattle. *DNACell Biol.* 15: 83–88, 1996.

[40948] 14500.Le Chalony, C.; Prosperi, M.–T.; Haluza, R.; Apiou, F.; Dutrillaux,B.; Goubin, G.: The OZF gene encodes a protein consisting essentially of zinc–finger motifs. *J. Molec. Biol.* 236: 399–404, 1994.

[40949] 14501.Dell'Angelica, E. C.; Ohno, H.; Ooi, C. E.; Rabinovich, E.; Roche,K. W.; Bonifacino, J. S.: AP–3: an adaptor–like protein complex with ubiquitous expression. *EMBO J.* 16: 917–928, 1997.

[40950] 14502.Watanabe, T. K.; Shimizu, F.; Nagata, M.; Takaichi, A.; Fujiwara,T.; Nakamura, Y.; Takahashi, E.; Hirai, Y.: Cloning, expression pattern and mapping to 12p13.2–p13.1 of CLAPS3, a gene encoding a novel clathrin–adaptor small chain. *Cytogenet. Cell Genet.* 73: 214–217, 1996.

[40951] 14503.Angrist, M.; Wells, D. E.; Chakravarti, A.; Pandey, A.: Chromosomal localization of the mouse Src–like adapter protein (Slap) gene and its putative human homolog SLA. *Genomics* 30: 623–625, 1995.

[40952] 14504.Holland, S. J.; Liao, X. C.; Mendenhall, M. K.; Zhou, X.; Pardo,J.; Chu, P.; Spencer, C.; Fu, A.; Sheng, N.; Yu, P.; Pali, E.; Nagin,A.; and 14 others: Functional cloning of

Src-like adapter protein-2(SLAP-2), a novel inhibitor of antigen receptor signaling. J. Exp.Med. 194: 1263–1276, 2001.

[40953] 14505.Kratchmarova, I.; Sosinowski, T.; Weiss, A.; Witter, K.; Vincenz,C.; Pandey, A.: Characterization of promoter region and genomic structureof the murine and human genes encoding Src like adapter protein. Gene 262:267–273, 2001.

[40954] 14506.Meijerink, P. H. S.; Yanakiev, P.; Zorn, I.; Grierson, A. J.; Bikker,H.; Dye, D.; Kalaydjieva, L.; Baas, F.: The gene for the human Src-likeadaptor protein (hSLAP) is located within the 64–kb intron of the thyroglobulin gene. Europ. J. Biochem. 254: 297–303, 1998.

[40955] 14507.Sosinowski, T.; Pandey, A.; Dixit, V. M.; Weiss, A.: Src-likeadaptor protein (SLAP) is a negative regulator of T cell receptorsignaling. J. Exp. Med. 191: 463–474, 2000.

[40956] 14508.Bailey, S. M.; Cornforth, M. N.; Kurimasa, A.; Chen, D. J.; Goodwin,E. H.: Strand-specific postreplicative processing of mammalian telomeres. Science 293:2462–2465, 2001.

[40957] 14509.Maniatis, T.: A ubiquitin ligase complex essential for the NF-kappa-B,Wnt/Wingless, and Hedgehog signaling pathways. Genes Dev. 13: 505–510,1999.

- [40958] 14510. Winston, J. T.; Strack, P.; Beer-Romero, P.; Chu, C. Y.; Elledge, S. J.; Harper, J. W.: The SCF(beta-TRCP)-ubiquitin ligase complex associates specifically with phosphorylated destruction motifs in I-kappa-B-alpha and beta-catenin and stimulates I-kappa-B-alpha ubiquitination in vitro. *Genes Dev.* 13: 270-283, 1999.
- [40959] 14511. Ahouse, J. J.; Hagerman, C. L.; Mittal, P.; Gilbert, D. J.; Copeland, N. G.; Jenkins, N. A.; Simister, N. E.: Mouse MHC class I-like Fc receptor encoded outside the MHC. *J. Immun.* 151: 6076-6088, 1993.
- [40960] 14512. Junghans, R. P.; Anderson, C. L.: The protection receptor for IgG catabolism is the beta-2-microglobulin-containing neonatal intestinal transport receptor. *Proc. Nat. Acad. Sci.* 93: 5512-5516, 1996.
- [40961] 14513. Junghans, R. P.; Ebralidze, A.; Tiwari, B.: Does (CUG)_n repeat in DMPK mRNA 'paint' chromosome 19 to suppress distant genes to create the diverse phenotype of myotonic dystrophy? A new hypothesis of long-range cis autosomal inactivation. *Neurogenetics* 3: 59-67, 2001.
- [40962] 14514. Kandil, E.; Egashira, M.; Miyosi, O.; Niikawa, N.; Ishibashi, T.; Kasahara, M.: The human gene encoding the heavy chain of the major histocompatibility complex class

I-like Fc receptor (FCGRT) maps to 19q13.3. Cytogenet. Cell Genet. 73: 97–98, 1996.

- [40963] 14515. Story, C. M.; Mikulska, J. E.; Simister, N. E.: A major histocompatibility complex class I-like Fc receptor cloned from human placenta: possible role in transfer of immunoglobulin G from mother to fetus. J. Exp. Med. 180: 2377–2381, 1994.
- [40964] 14516. Waldmann, T. A.; Strober, W.: Metabolism of immunoglobulins. Prog. Allergy 13: 1–110, 1969.
- [40965] 14517. Hart, T. C.; Price, J. A.; Bobby, P. L.; Pettenati, M. J.; Shashi, V.; Von Kap Herr, C.; Van Dyke, T. E.: Cytogenetic assignment and physical mapping of the human DGKE gene to chromosome 17q22. Genomics 56: 233–235, 1999.
- [40966] 14518. Tang, W.; Bunting, M.; Zimmerman, G. A.; McIntyre, T. M.; Prescott, S. M.: Molecular cloning of a novel human diacylglycerol kinase highly selective for arachidonate-containing substrates. J. Biol. Chem. 271: 10237–10241, 1996.
- [40967] 14519. Beddow, A. L.; Richards, S. A.; Orem, N. R.; Macara, I. G.: The Ran/TC4 GTPase-binding domain: identification by expression cloning and characterization of a conserved sequence motif. Proc. Nat. Acad. Sci. 92: 3328–3332,

1995.

- [40968] 14520.Fauser, S.; Aslanukov, A.; Roepman, R.; Ferreira, P. A.: Genomic organization, expression, and localization of murine Ran-binding protein2 (RanBP2) gene. *Mammalian Genome* 12: 406–415, 2001.
- [40969] 14521.Krebber, H.; Bastians, H.; Hoheisel, J.; Lichter, P.; Ponstingl, H.; Joos, S.: Localization of the gene encoding the Ran-binding protein RanBP2 to human chromosome 2q11–q13 by fluorescence in situ hybridization. *Genomics* 43:247–248, 1997.
- [40970] 14522.Pichler, A.; Gast, A.; Seeler, J. S.; Dejean, A.; Melchior, F.: The nucleoporin RanBP2 has SUMO1 E3 ligase activity. *Cell* 108:109–120, 2002.
- [40971] 14523.Wu, J.; Matunis, M. J.; Kraemer, D.; Blobel, G.; Coutavas, E.: Nup358, a cytoplasmically exposed nucleoporin with peptide repeats, Ran-GTP binding sites, zinc fingers, a cyclophilin A homologous domain, and a leucine-rich region. *J. Biol. Chem.* 270: 14209–14213, 1995.
- [40972] 14524.Yokoyama, N.; Hayashi, N.; Seki, T.; Pante, N.; Ohba, T.; Nishii, K.; Kuma, K.; Hayashida, T.; Miyata, T.; Abei, U.; Fukui, M.; Nishimoto, T.: A giant nucleopore protein that binds Ran/TC4. *Nature* 376:184–188, 1995.

- [40973] 14525.Dhar, S. K.; Yoshida, K.; Machida, Y.; Khaira, P.; Chaudhuri, B.; Wohlschlegel, J. A.; Leffak, M.; Yates, J.; Dutta, A.: Replication from oriP of Epstein–Barr virus requires human ORC and is inhibited by geminin. *Cell* 106: 287–296, 2001.
- [40974] 14526.Gavin, K. A.; Hidaka, M.; Stillman, B.: Conserved initiator proteins in eukaryotes. *Science* 270: 1667–1671, 1995.
- [40975] 14527.Ohtani, K.; DeGregori, J.; Leone, G.; Herendeen, D. R.; Kelly, T. J.; Nevins, J. R.: Expression of the HsOrc1 gene, a human ORC1 homolog, is regulated by cell proliferation via the E2F transcription factor. *Molec. Cell. Biol.* 16: 6977–6984, 1996.
- [40976] 14528.Takahara, K.; Bong, M.; Brevard, R.; Eddy, R. L.; Haley, L. L.; Sait, S. J.; Shows, T. B.; Hoffman, G. G.; Greenspan, D. S.: Mouse and human homologues of the yeast origin of replication recognition complex subunit ORC2 and chromosomal localization of the cognate human gene ORC2L. *Genomics* 31: 119–122, 1996.
- [40977] 14529.Chang, A. C.–M.; Janosi, J.; Hulsbeek, M.; de Jong, D.; Jeffrey, K. J.; Noble, J. R.; Reddel, R. R.: A novel human cDNA highly homologous to the fish hormone stanniocalcin. *Molec. Cell. Endocr.* 112: 241–247, 1995.

- [40978] 14530.Chang, A. C.-M.; Jeffrey, K. J.; Tokutake, Y.; Shimamoto, A.; Neumann,A. A.; Dunham, M. A.; Cha, J.; Sugawara, M.; Furuichi, Y.; Reddel,R. R.: Human stanniocalcin (STC): genomic structure, chromosomallocalization, and the presence of CAG trinucleotide repeats. *Genomics* 47:393–398, 1998.
- [40979] 14531.Jellinek, D. A.; Chang, A. C.; Larsen, M. R.; Wang, X.; Robinson,P. J.; Reddel, R. R.: Stanniocalcin 1 and 2 are secreted as phosphoproteinsfrom human fibrosarcoma cells. *Biochem. J.* 350: 453–461, 2000.
- [40980] 14532.Olsen, H. S.; Cepeda, M. A.; Zhang, Q.-Q.; Rosen, C. A.; Vozzolo,B. L.; Wagner, G. F.: Human stanniocalcin: a possible hormonal regulatorof mineral metabolism. *Proc. Nat. Acad. Sci.* 93: 1792–1796, 1996.
- [40981] 14533.Varghese, R.; Wong, C. K. C.; Deol, H.; Wagner, G. F.; DiMattia,G. E.: Comparative analysis of mammalian stanniocalcin genes. *Endocrinology* 139:4714–4725, 1998.
- [40982] 14534.Wagner, G. F.; Guiraudon, C. C.; Milliken, C.; Copp, D. H.: Immunologicaland biological evidence for a stanniocalcin–like hormone in humankidney. *Proc. Nat. Acad. Sci.* 92: 1871–1875, 1995.
- [40983] 14535.Bartsch, O.; Wagner, A.; Hinkel, G. K.; Lichtner, P.;

Murken, J.;Schuffenhauer, S.: No evidence for chromosomal microdeletions at the second DiGeorge syndrome locus on 10p near D10S585. (Letter) *Am.J. Med. Genet.* 83: 425–426, 1999.

[40984] 14536.Daw, S. C. M.; Taylor, C.; Kraman, M.; Call, K.; Mao, J.; Schuffenhauer,S.; Meitinger, T.; Lipson, T.; Goodship, J.; Scambler, P.: A common region of 10p deleted in DiGeorge and velocardiofacial syndromes. *NatureGenet.* 13: 458–461, 1996.

[40985] 14537.Lichtner, P.; Konig, R.; Hasegawa, T.; Van Esch, H.; Meitinger,T.; Schuffenhauer, S.: An HDR (hypoparathyroidism, deafness, renal dysplasia) syndrome locus maps distal to the DiGeorge syndrome region on 10p13/14. *J. Med. Genet.* 37: 33–37, 2000.

[40986] 14538.Sorensen, O. E.; Follin, P.; Johnsen, A. H.; Calafat, J.; Tjabringa,G. S.; Hiemstra, P. S.; Borregaard, N.: Human cathelicidin, hCAP–18, is processed to the antimicrobial peptide LL–37 by extracellular cleavage with proteinase 3. *Blood* 97: 3951–3959, 2001.

[40987] 14539.Zanetti, M.; Del Sal, G.; Storici, P.; Schneider, C.; Romeo, D.: The cDNA of the neutrophil antibiotic Bac5 predicts a pro–sequence homologous to a cysteine proteinase inhibitor that is common to other neutrophil an–

tibiotics. J. Biol. Chem. 268: 522–526, 1993.

- [40988] 14540.Chen, C.-Y.; Gherzi, R.; Ong, S.-E.; Chan, E. L.; Raijmakers, R.;Pruijn, G. J. M.; Stoecklin, G.; Moroni, C.; Mann, M.; Karin, M.:AU binding proteins recruit the exosome to degrade ARE-containingmRNAs. Cell 107: 451–464, 2001.
- [40989] 14541.Dangel, A. W.; Shen, L.; Mendoza, A. R.; Wu, L.-c.; Yu, C. Y.:Human helicase gene SKI2W in the HLA class III region exhibits strikingstructural similarities to the yeast antiviral gene SKI2 and to thehuman gene KIAA0052: emergence of a new gene family. Nucleic AcidsRes. 23: 2120–2126, 1995.
- [40990] 14542.Lee, S.-G.; Lee, I.; Park, S. H.; Kang, C.; Song, K.: Identificationand characterization of a human cDNA homologous to yeast SKI2. Genomics 25:660–666, 1995.
- [40991] 14543.Shen, L.; Wu, L. C.; Sanlioglu, S.; Chen, R.; Mendoza, A. R.; Dangel,A. W.; Carroll, M. C.; Zipf, W. B.; Yu, C. Y.: Structure and geneticsof the partially duplicated gene RP located immediately upstream ofthe complement C4A and the C4B genes in the HLA class III region:molecular cloning, exon–intron structure, composite retroposon, andbreakpoint of gene duplication. J. Biol. Chem. 269: 8466–8476, 1994.

- [40992] 14544. Yang, Z.; Shen, L.; Dangel, A. W.; Wu, L.-C.; Yu, C. Y.: Four ubiquitously expressed genes, RD (D6S45)–SKI2W (SKIV2L)–DOM3Z–RP1(D6S60E), are present between complement component genes factor Band C4 in the class III region of the HLA. *Genomics* 53: 338–347, 1998.
- [40993] 14545. Doyle, K.; Zhang, Y.; Baer, R.; Bina, M.: Distinguishable patterns of protein–DNA interactions involving complexes of basic helix–loop–helix proteins. *J. Biol. Chem.* 269: 12099–12105, 1994.
- [40994] 14546. Hu, J.-S.; Olson, E. N.; Kingston, R. E.: HEB, a helix–loop–helix protein related to E2A and ITF2 that can modulate the DNA–binding ability of myogenic regulatory factors. *Molec. Cell. Biol.* 12: 1031–1042, 1992.
- [40995] 14547. Sawada, S.; Littman, D. R.: A heterodimer of HEB and an E12–related protein interacts with the CD4 enhancer and regulates its activity in T–cell lines. *Molec. Cell. Biol.* 13: 5620–5628, 1993.
- [40996] 14548. Zhang, Y.; Babin, J.; Feldhaus, A. L.; Singh, H.; Sharp, P. A.; Bina, M.: HTF4: a new human helix–loop–helix protein. *Nucleic Acids Res.* 19: 4555 only, 1991.
- [40997] 14549. Zhang, Y.; Bina, M.: The nucleotide sequence of the human transcription factor HTF4a cDNA. *DNA Sequence* 2: 397–403, 1992.

- [40998] 14550.Zhang, Y.; Flejter, W. L.; Barcroft, C. L.; Riviere, M.; Szpirer,J.; Szpirer, C.; Bina, M.: Localization of the human HTF4 transcriptionfactors 4 gene (TCF12) to chromosome 15q21. Cytogenet. Cell Genet. 68:235–238, 1995.
- [40999] 14551.Dubrulle, J.; McGrew, M. J.; Pourquie, O.: FGF signaling controlssomite boundary position and regulates segmentation clock controlof spatiotemporal Hox gene activation. Cell 106: 219–232, 2001.
- [41000] 14552.Fukuchi–Shimogori, T.; Grove, E. A.: Neocortex patterning by thesecreted signaling molecule FGF8. Science 294: 1071–1074, 2001.
- [41001] 14553.Gemel, J.; Gorry, M.; Ehrlich, G. D.; MacArthur, C. A.: Structureand sequence of human FGF8. Genomics 35: 253–257, 1996.
- [41002] 14554.Rowen, L.; Young, J.; Birditt, B.; Kaur, A.; Madan, A.; Philipps,D. L.; Qin, S.; Minx, P.; Wilson, R. K.; Hood, L.; Graveley, B. R.: Analysis of the human neurexin genes: alternative splicing and thegeneration of protein diversity. Genomics 79: 587–597, 2002.
- [41003] 14555.Tabuchi, K.; Sudhof, T. C.: Structure and evolution of neurexingenes: insight into the mechanism of alternative splicing. Genomics 79:849–859, 2002.
- [41004] 14556.Ullrich, B.; Ushkaryov, Y. A.; Sudhof, T. C.: Cartog–

raphy of neurexins: more than 1000 isoforms generated by alternative splicing and expressed in distinct subsets of neurons. *Neuron* 14: 497–507, 1995.

- [41005] 14557. Ushkaryov, Y. A.; Petrenko, A. G.; Geppert, M.; Sudhof, T. C.: Neurexins: synaptic cell surface proteins related to the α -latrotoxin receptor and laminin. *Science* 257: 50–56, 1992.
- [41006] 14558. Bergman, L.; Silins, G.; Grimmond, S.; Hummerich, H.; Stewart, C.; Little, P.; Hayward, N.: A 500-kb sequence-ready cosmid contig and transcript map of the MEN1 region on 11q13. *Genomics* 55: 49–56, 1999.
- [41007] 14559. Ushkaryov, Y. A.; Petrenko, A. G.; Geppert, M.; Sudhof, T. C.: Neurexins: synaptic cell surface proteins related to the α -latrotoxin receptor and laminin. *Science* 257: 50–56, 1992.
- [41008] 14560. Scheiffele, P.; Fan, J.; Choih, J.; Fetter, R.; Serafini, T.: Neuroligin expressed in nonneuronal cells triggers presynaptic development in contacting axons. *Cell* 101: 657–669, 2000.
- [41009] 14561. Abderrahmani, A.; Steinmann, M.; Plaisance, V.; Niederhauser, G.; Haefliger, J.-A.; Mooser, V.; Bonny, C.; Nicod, P.; Waeber, G.: The transcriptional repressor REST determines the cell-specific expression of the human

MAPK8IP1 gene encoding IB1 (JIP-1). *Molec. Cell. Biol.* 21:7256–7627, 2001.

- [41010] 14562.Chen, Z.-F.; Paquette, A. J.; Anderson, D. J.: NRSF/REST is required *in vivo* for repression of multiple neuronal target genes during embryogenesis. *Nature Genet.* 20: 136–142, 1998.
- [41011] 14563.Palm, K.; Metsis, M.; Timmusk, T.: Neuron-specific splicing of zinc finger transcription factor REST/NRSF/XBR is frequent in neuroblastomas and conserved in human, mouse and rat. *Molec. Brain Res.* 72: 30–39, 1999.
- [41012] 14564.Schoenherr, C. J.; Anderson, D. J.: The neuron-restrictive silencer factor (NRSF): a coordinate repressor of multiple neuron-specific genes. *Science* 267: 1360–1363, 1995.
- [41013] 14565.Thiel, G.; Lietz, M.; Cramer, M.: Biological activity and modulator structure of RE-1-silencing transcription factor (REST), a repressor of neuronal genes. *J. Biol. Chem.* 273: 26891–26899, 1998.
- [41014] 14566.Cavaloc, Y.; Popielarz, M.; Fuchs, J.-P.; Gattoni, R.; Stevenin, J.: Characterization and cloning of the human splicing factor 9G8: a novel 35 kDa factor of the serine/arginine protein family. *EMBO J.* 13: 2639–2649, 1994.
- [41015] 14567.Popielarz, M.; Cavaloc, Y.; Mattei, M.-G.; Gattoni,

R.; Stevenin,J.: The gene encoding human splicing factor 9G8: structure, chromosomal localization, and expression of alternatively processed transcripts. J.Biol. Chem. 270: 17830–17835, 1995.

[41016] 14568.Chiang, C.–M.; Roeder, R. G.: Cloning of an intrinsic human TFIID subunit that interacts with multiple transcriptional activators. Science 267:531–536, 1995.

[41017] 14569.Wu, Q.; Zhang, T.; Cheng, J.–F.; Kim, Y.; Grimwood, J.; Schmutz,J.; Dickson, M.; Noonan, J. P.; Zhang, M. Q.; Myers, R. M.; Maniatis,T.: Comparative DNA sequence analysis of mouse and human protocadherin gene clusters. Genome Res. 11: 389–404, 2001.

[41018] 14570.Aoki, K.; Inazawa, J.; Takahashi, T.; Nakahara, K.; Kasai, M.: Genomic structure and chromosomal localization of the gene encoding translin, a recombination hotspot binding protein. Genomics 43:237–241, 1997.

[41019] 14571.Aoki, K.; Nakahara, K.; Ikegawa, C.; Seto, M.; Takahashi, T.; Minowada,J.; Strominger, J. L.; Maziarz, R. T.; Kasai, M.: Nuclear proteins binding to a novel target sequence within the recombination hotspot regions of Bcl-2 and the immunoglobulin D(H) gene family. Oncogene 9:1109–1115, 1994.

[41020] 14572.Blair, I. P.; Gibson, R. R.; Bennett, C. L.; Chance, P.

F.: Searchfor genes involved in Joubert syndrome: evidence that one or moremajor loci are yet to be identified and exclusion of candidate genesEN1, EN2, FGF8, and BARHL1. Am. J. Med. Genet. 107: 190–196, 2002.

[41021] 14573.Berge, K. E.; Tian, H.; Graf, G. A.; Yu, L.; Grishin, N. V.; Schultz,J.; Kwiterovich, P.; Shan, B.; Barnes, R.; Hobbs, H. H.: Accumulationof dietary cholesterol in sitosterolemia caused by mutations in adjacentABC transporters. Science 290: 1771–1775, 2000.

[41022] 14574.Bodzioch, M.; Orso, E.; Klucken, J.; Langmann, T.; Bottcher, A.;Diederich, W.; Drobnik, W.; Barlage, S.; Buchler, C.; Porsch–Ozcurumez,M.; Kaminski, W. E.; Hahmann, H. W.; Oette, K.; Rothe, G.; Aslanidis,C.; Lackner, K. J.; Schmitz, G.: The gene encoding ATP–binding cassette–transporter 1 is mutated in Tangier disease. Nature Genet. 22: 347–351,1999.

[41023] 14575.Brooks–Wilson, A.; Marcil, M.; Clee, S. M.; Zhang, L.–H.; Roomp,K.; van Dam, M.; Yu, L.; Brewer, C.; Collins, J. A.; Molhuizen, H.O. F.; Loubser, O.; Ouelette, B. F. F.; and 14 others: Mutationsin ABC1 in Tangier disease and familial high–density lipoprotein deficiency. NatureGenet. 22: 336–345, 1999.

[41024] 14576.Asamoah, A.; Wilson, A. F.; Elston, R. C.; Dalferes,

E., Jr.; Berenson, G. S.: Segregation and linkage analyses of dopamine-beta-hydroxylase activity in a six-generation pedigree. *Am. J. Med. Genet.* 27: 613-621, 1987.

[41025] 14577. Biaggioni, I.; Goldstein, D. S.; Atkinson, T.; Robertson, D.: Dopamine-beta-hydroxylase deficiency in humans. *Neurology* 40: 370-373, 1990.

[41026] 14578. Biaggioni, I.; Robertson, D.: Endogenous restoration of noradrenaline by precursor therapy in dopamine-beta-hydroxylase deficiency. *Lancet* II: 1170-1172, 1987.

[41027] 14579. Craig, S. P.; Buckle, V. J.; Lamouroux, A.; Mallet, J.; Craig, I. W.: Localization of the human dopamine beta hydroxylase (DBH) gene to chromosome 9q34. *Cytogenet. Cell Genet.* 48: 48-50, 1988.

[41028] 14580. Dunnette, J.; Weinshilboum, R.: Human serum dopamine beta-hydroxylase: correlation of enzymatic activity with immunoreactive protein in genetically defined samples. *Am. J. Hum. Genet.* 28: 155-166, 1976.

[41029] 14581. Dunnette, J.; Weinshilboum, R.: Inheritance of low immunoreactive human plasma dopamine-beta-hydroxylase: radioimmunoassay studies. *J. Clin. Invest.* 60: 1080-1087, 1977.

[41030] 14582. Elston, R. C.; Namboodiri, K. K.; Hames, C. G.: Segregation and linkage analysis of dopamine-

beta-hydroxylase activity. Hum. Hered. 29:284-292, 1979.

[41031] 14583. Gershon, E. S.; Goldin, L. R.: Segregation and linkage studies of plasma dopamine-beta-hydroxylase (DBH), erythrocyte catechol-O-methyltransferase (COMT) and platelet monoamine oxidase (MAO): possible linkage between the ABO locus and a gene controlling DBH activity. (Abstract) Am. J. Hum. Genet. 33: 136A only, 1981.

[41032] 14584. Goldin, L. R.; Gershon, E. S.; Lake, C. R.; Murphy, D. L.; McGinniss, M.; Sparkes, R. S.: Segregation and linkage studies of plasma dopamine-beta-hydroxylase (DBH), erythrocyte catechol-O-methyltransferase (COMT), and platelet monoamine oxidase (MAO): possible linkage between the ABO locus and a gene controlling DBH activity. Am. J. Hum. Genet. 34: 250-262, 1982.

[41033] 14585. Joh, T. H.; Baetge, E. E.; Reis, D. J.: Evidence for the existence of a single gene or linked genes coding for catecholamine biosynthetic enzymes. Trans. Assoc. Am. Phys. 96: 38-43, 1983.

[41034] 14586. Joh, T. H.; Baetge, E. E.; Ross, M. E.; Albert, V. R.; Moon, H. M.; Reis, D. J.: Existence of catecholamine biosynthetic enzyme gene family. (Abstract) Clin. Res. 31: 528 only, 1983.

- [41035] 14587.Joh, T. H.; Baetge, E. E.; Ross, M. E.; Reis, D. J.: Biochemistry and molecular biology of catecholamine neurons: a single gene or gene family hypothesis. Clin. Exp. Hypertension 6A: 11-21, 1984.
- [41036] 14588.Kobayashi, K.; Kurosawa, Y.; Fujita, K.; Nagatsu, T.: Human dopamine beta-hydroxylase gene: two mRNA types having different 3-prime-terminal regions are produced through alternative polyadenylation. Nucleic Acids Res. 17: 1089-1102, 1989.
- [41037] 14589.Lamouroux, A.; Vigny, A.; Faucon Biguet, N.; Darnon, M. C.; Franck, R.; Henry, J.-P.; Mallet, J.: The primary structure of human dopamine-beta-hydroxylase: insights into the relationship between the soluble and the membrane-bound forms of the enzyme. EMBO J. 6: 3931-3937, 1987.
- [41038] 14590.Lea, R. A.; Dohy, A.; Jordan, K.; Quinlan, S.; Brimage, P. J.; Griffiths, L. R.: Evidence for allelic association of the dopamine beta-hydroxylase gene (DBH) with susceptibility to typical migraine. Neurogenetics 3:35-40, 2000.
- [41039] 14591.Mathias, C. J.; Bannister, R. B.; Cortelli, P.; Heslop, K.; Polak, J. M.; Raimbach, S.; Springall, D. R.; Watson, L.: Clinical, autonomic and therapeutic observations in two siblings with postural hypotension and sympathetic failure

due to an inability to synthesize noradrenaline from dopamine because of a deficiency of dopamine beta hydroxylase. Quart.J. Med. 75: 617–633, 1990.

- [41040] 14592. McKinney, E. F.; Walton, R. T.; Yudkin, P.; Fuller, A.; Haldar, N. A.; Mant, D.; Murphy, M.; Welsh, K. I.; Marshall, S. E.: Association between polymorphisms in dopamine metabolic enzymes and tobacco consumption in smokers. Pharmacogenetics 10: 483–491, 2000.
- [41041] 14593. O'Malley, K. L.; Maunon, A.; Raese, J.; Barchas, J. D.; Kedes, L.: Genes for catecholamine biosynthesis: cloning by expression and identification of the cDNA for rat dopamine beta-hydroxylase. Proc. Nat. Acad. Sci. 80: 2161–2165, 1983.
- [41042] 14594. Ogiwara, T.; Nugent, C. A., Jr.; Shen, S.-W.; Goldfein, S.: Serum dopamine-beta-hydroxylase activity in parents and children. J. Lab. Clin. Med. 85: 566–573, 1975.
- [41043] 14595. Perry, S. E.; Summar, M. L.; Phillips, J. A., III; Robertson, D.: Linkage analysis of the human dopamine beta-hydroxylase gene. Genomics 10: 493–495, 1991.
- [41044] 14596. Robertson, D.; Haile, V.; Perry, S. E.; Robertson, R. M.; Phillips, J. A., III; Biaggioni, I.: Dopamine beta-hydroxylase deficiency: a genetic disorder of cardiovascular regulation. Hypertension 18: 1–8, 1991.

- [41045] 14597.Ross, S. B.; Wetterberg, L.; Myrhed, M.: Genetic control of plasmadopamine-beta-hydroxylase. Life Sci. 12: 529-532, 1973.
- [41046] 14598.Schanberg, S. M.; Stone, R. A.; Kirshner, N.; Gunnells, J. C.;Robinson, R. R.: Plasma dopamine beta-hydroxylase: a possible aidin the study and evaluation of hypertension. Science 183: 523-525,1974.
- [41047] 14599.Marlar, R. A.; Griffin, J. H.: Deficiency of protein C inhibitorin combined factor V/VIII deficiency disease. J. Clin. Invest. 66:1186-1189, 1980.
- [41048] 14600.Meijers, J. C.; Chung, D. W.: Organization of the gene codingfor human protein C inhibitor (plasminogen activator inhibitor-3):assignment of the gene to chromosome 14. J. Biol. Chem. 266: 15028-15034,1991.
- [41049] 14601.Nichols, W. C.; Seligsohn, U.; Zivelin, A.; Terry, V. H.; Arnold,N. D.; Siemieniak, D. R.; Kaufman, R. J.; Ginsburg, D.: Linkage ofcombined factors V and VIII deficiency to chromosome 18q by homozygositymapping. J. Clin. Invest. 99: 596-601, 1997.
- [41050] 14602.Suzuki, K.; Deyashiki, Y.; Nishioka, J.; Kurachi, K.; Akira, M.;Yamamoto, S.; Hashimoto, S.: Characterization of a cDNA for humanprotein C inhibitor: a new member of the plasma serine protease inhibitorsuperfamily. J. Biol.

Chem. 262: 611–616, 1987.

- [41051] 14603. Cathelineau, L.; Dinh, D. P.; Briand, P.; Kamoun, P.: Studies on complementation in argininosuccinate synthetase and argininosuccinatelyase deficiencies in human fibroblasts. Hum. Genet. 57: 282–284, 1981.
- [41052] 14604. Kromberg, J. G. R.; Castle, D.; Zwane, E. M.; Jenkins, T.: Albinism and skin cancer in southern Africa. Clin. Genet. 36: 43–52, 1989.
- [41053] 14605. El Kahloun, A.; Chauvel, B.; Mauvieux, V.; Dorval, I.; Jouanolle, A.-M.; Gicquel, I.; Le Gall, J.-Y.; David, V.: Localization of seven new genes around the HLA-A locus. Hum. Molec. Genet. 2: 55–60, 1993.
- [41054] 14606. Beattie, E. C.; Stellwagen, D.; Morishita, W.; Bresnahan, J. C.; Ha, B. K.; Von Zastrow, M.; Beattie, M. S.; Malenka, R. C.: Control of synaptic strength by glial TNF- α . Science 295: 2282–2285, 2002.
- [41055] 14607. Beutler, B.; Krochin, N.; Milsark, I. W.; Luedke, C.; Cerami, A.: Control of cachectin (tumor necrosis factor) synthesis: mechanisms of endotoxin resistance. Science 232: 977–980, 1986.
- [41056] 14608. Brenner, D. A.; O'Hara, M.; Angel, P.; Chojkier, M.; Karin, M.: Prolonged activation of JUN and collagenase genes by tumour necrosis factor- α . Nature 337:

661–663, 1989.

- [41057] 14609. Broudy, V. C.; Kaushansky, K.; Segal, G. M.; Harlan, J. M.; Adamson, J. W.: Tumor necrosis factor type alpha stimulates human endothelial cells to produce granulocyte/macrophage colony-stimulating factor. *Proc. Nat. Acad. Sci.* 83: 7467–7471, 1986.
- [41058] 14610. Bruce, A. J.; Boling, W.; Kindy, M. S.; Peschon, J.; Kraemer, P. J.; Carpenter, M. K.; Holtsberg, F. W.; Mattson, M. P.: Altered neuronal and microglial responses to excitotoxic and ischemic brain injury in mice lacking TNF receptors. *Nature Med.* 2: 788–794, 1996.
- [41059] 14611. Cabrera, M.; Shaw, M. A.; Sharples, C.; Williams, H.; Castes, M.; Convit, J.; Blackwell, J. M.: Polymorphism in tumor necrosis factor genes associated with mucocutaneous leishmaniasis. *J. Exp. Med.* 182: 1259–1264, 1995.
- [41060] 14612. Conway, D. J.; Holland, M. J.; Bailey, R. L.; Campbell, A. E.; Mahdi, O. S.; Jennings, R.; Mbena, E.; Mabey, D. C.: Scarring trachoma is associated with polymorphism in the tumor necrosis factor alpha (TNF-alpha) gene promoter and with elevated TNF-alpha levels in tear fluid. *Infect. Immun.* 65: 1003–1006, 1997.
- [41061] 14613. Davis, J. M.; Narachi, M. A.; Alton, N. K.; Arakawa, T.: Structure of human tumor necrosis factor alpha derived

from recombinant DNA. *Biochemistry* 26:1322–1326, 1987.

- [41062] 14614. Escobar–Morreale, H. F.; Calvo, R. M.; Sancho, J.; San Millan, J. L.: TNF–alpha and hyperandrogenism: a clinical, biochemical, and molecular genetic study. *J. Clin. Endocr. Metab.* 86: 3761–3767, 2001.
- [41063] 14615. Gorman, J. D.; Sack, K. E.; Davis, J. C., Jr.: Treatment of ankylosing spondylitis by inhibition of tumor necrosis factor–alpha. *New Eng. J. Med.* 346: 1349–1356, 2002.
- [41064] 14616. Herrmann, S.–M.; Ricard, S.; Nicaud, V.; Mallet, C.; Arveiler, D.; Evans, A.; Ruidavets, J.–B.; Luc, G.; Bara, L.; Parra, H.–J.; Poirier, O.; Cambien, F.: Polymorphisms of the tumour necrosis factor–alpha gene, coronary heart disease and obesity. *Europ. J. Clin. Invest.* 28:59–66, 1998.
- [41065] 14617. Inoko, H.; Trowsdale, J.: Linkage of TNF genes to the HLA–B locus. *Nucleic Acids Res.* 15: 8957–8962, 1987.
- [41066] 14618. Knight, J. C.; Udalova, I.; Hill, A. V. S.; Greenwood, B. M.; Peshu, N.; Marsh, K.; Kwiatkowski, D.: A polymorphism that affects OCT–1 binding to the TNF promoter region is associated with severe malaria. *Nature Genet.* 22: 145–150, 1999.
- [41067] 14619. Koss, K.; Satsangi, J.; Fanning, G. C.; Welsh, K. I.;

Jewell,D. P.: Cytokine (TNF- α , LT- α , and IL-10) polymorphisms in inflammatory bowel diseases and normal controls: differential effects on production and allele frequencies. *Genes Immun.* 1: 185-190, 2000.

- [41068] 14620. Marino, M. W.; Dunn, A.; Grail, D.; Inglese, M.; Noguchi, Y.; Richards, E.; Jungbluth, A.; Wada, H.; Moore, M.; Williamson, B.; Basu, S.; Old, L. J.: Characterization of tumor necrosis factor-deficient mice. *Proc. Nat. Acad. Sci.* 94: 8093-8098, 1997.
- [41069] 14621. McCusker, S. M.; Curran, M. D.; Dynan, K. B.; McCullagh, C. D.; Urquhart, D. D.; Middleton, D.; Patterson, C. C.; McIlroy, S. P.; Passmore, A. P.: Association between polymorphism in regulatory region of gene encoding tumour necrosis factor- α and risk of Alzheimer's disease and vascular dementia: a case-control study. *Lancet* 357:436-439, 2001.
- [41070] 14622. Remaley, A. T.; Rust, S.; Rosier, M.; Knapper, C.; Naudin, L.; Broccardo, C.; Peterson, K. M.; Koch, C.; Arnould, I.; Prades, C.; Duverger, N.; Funke, H.; Assman, G.; Dinger, M.; Dean, M.; Chimini, G.; Santamarina-Fojo, S.; Fredrickson, D. S.; Deneffe, P. Brewer, H. B., Jr.: Human ATP-binding cassette transporter 1 (ABC1): genomic organization and identification of the genetic defect in the

originalTangier disease kindred. Proc. Nat. Acad. Sci. 96: 12685–12690,1999.

- [41071] 14623.Rust, S.; Rosier, M.; Funke, H.; Real, J.; Amoura, Z.; Piette,J.–C.; Deleuze, J.–F.; Brewer, H. B.; Duverger, N.; Deneffe, P.; Assmann,G.: Tangier disease is caused by mutations in the gene encoding ATP–bindingcassette transporter 1. Nature Genet. 22: 352–355, 1999.
- [41072] 14624.Young, S. G.; Fielding, C. J.: The ABCs of cholesterol efflux. NatureGenet. 22: 316–318, 1999.
- [41073] 14625.Buchwald, M.: Complementation groups: one or more per gene? NatureGenet. 11: 228–230, 1995.
- [41074] 14626.Stambolian, D.; Ai, Y.; Sidjanin, D.; Nesburn, K.; Sathe, G.;Rosenberg, M.; Bergsma, D. J.: Cloning of the galactokinase cDNAand identification of mutations in two families with cataracts. NatureGenet. 10: 307–312, 1995.
- [41075] 14627.Coppola, G.; De Michele, G.; Cavalcanti, F.; Pianese, L.; Perretti,A.; Santoro, L.; Vita, G.; Toscano, A.; Amboni, M.; Grimaldi, G.;Salvatore, E.; Caruso, G.; Filla, A.: Why do some Friedreich's ataxiapatients retain tendon reflexes? A clinical, neurophysiological andmolecular study. J. Neurol. 246: 353–357, 1999.
- [41076] 14628.Delatycki, M. B.; Knight, M.; Koenig, M.; Cossee, M.; Williamson,R.; Forrest, S. M.: G130V, a common FRDA

point mutation, appearsto have arisen from a common founder. Hum. Genet. 105: 343–346,1999.

- [41077] 14629.Delatycki, M. B.; Paris, D. B. B. P.; Gardner, R. J. M.; Nicholson,G. A.; Nassif, N.; Storey, E.; MacMillan, J. C.; Collins, V.; Williamson,R.; Forrest, S. M.: Clinical and genetic study of Friedreich ataxiain an Australian population. Am. J. Med. Genet. 87: 168–174, 1999.
- [41078] 14630.Durr, A.; Cossee, M.; Agid, Y.; Campuzano, V.; Mignard, C.; Penet,C.; Mandel, J.–L.; Brice, A.; Koenig, M.: Clinical and genetic abnormalitiesin patients with Friedreich's ataxia. New Eng. J. Med. 335: 1169–1175,1996.
- [41079] 14631.Filla, A.; De Michele, G.; Cavalcanti, F.; Pianese, L.; Monticelli,A.; Campanella, G.; Coccozza, S.: The relationship between trinucleotide(GAA) repeat length and clinical features in Friedreich ataxia. Am.J. Hum. Genet. 59: 554–560, 1996.
- [41080] 14632.Fujita, R.; Hanauer, A.; Vincent, A.; Mandel, J.–L.; Koenig, M.: Physical mapping of two loci (D9S5 and D9S15) tightly linked toFriedreich ataxia locus (FRDA) and identification of nearby CpG islandsby pulse–field gel electrophoresis. Genomics 10: 915–920, 1991.
- [41081] 14633.Gray, J. V.; Johnson, K. J.: Waiting for frataxin. Nature Genet. 16:323–325, 1997.

- [41082] 14634.Victoria, T.; Rafi, M. A.; Wenger, D. A.: Cloning of the canineGALC cDNA and identification of the mutation causing globoid cellleukodystrophy in West Highland White and Cairn terriers. *Genomics* 33:457–462, 1996.
- [41083] 14635.Wenger, D. A.; Rafi, M. A.; Luzi, P.: Molecular genetics of Krabbedisease (globoid cell leukodystrophy): diagnostic and clinical implications. *Hum.Mutat.* 10: 268–279, 1997.
- [41084] 14636.Zlotogora, J.; Chakraborty, S.; Knowlton, R. G.; Wenger, D. A.: Krabbe disease locus mapped to chromosome 14 by genetic linkage. *Am.J. Hum. Genet.* 47: 37–44, 1990.
- [41085] 14637.Zlotogora, J.; Regev, R.; Zeigler, M.; Iancu, T. C.; Bach, G.: Krabbe disease: increased incidence in a highly inbred community. *Am.J. Med. Genet.* 21: 765–770, 1985.
- [41086] 14638.Rushton, A. R.; Dawson, G.: Genetic linkage studies of the humanglycosphingolipid beta–galactosidases. *Biochem. Genet.* 15: 1071–1082,1977.
- [41087] 14639.Balciunaite, G.; Keller, M. P.; Balciunaite, E.; Piali, L.; Zuklys,S.; Mathieu, Y. D.; Gill, J.; Boyd, R.; Sussman, D. J.; Hollander,G. A.: Wnt glycoproteins regulate the expression of FoxN1, the genedefective in nude mice. *Nature Immun.* 15Oct, 2002. Note: AdvanceElectronic Publication.

- [41088] 14640. Frank, J.; Pignata, C.; Panteleyev, A. A.; Prowse, D. M.; Baden, H.; Weiner, L.; Gaetaniello, L.; Ahmad, W.; Pozzi, N.; Caerhalmi-Friedman, P. B.; Aita, V. M.; Uyttendaele, H.; Gordon, D.; Ott, J.; Brissette, J. L.; Christiano, A. M.: Exposing the human nude phenotype. *Nature* 398:473–474, 1999.
- [41089] 14641. Nehls, M.; Pfeifer, D.; Schorpp, M.; Hedrich, H.; Boehm, T.: Newmember of the winged-helix protein family disrupted in mouse and rat nude mutations. *Nature* 372: 103–107, 1994.
- [41090] 14642. Pignata, C.; Fiore, M.; Guzzetta, V.; Castaldo, A.; Sebastio, G.; Porta, F.; Guarino, A.: Congenital alopecia and nail dystrophy associated with severe functional T-cell immunodeficiency in two sibs. *Am. J. Med. Genet.* 65: 167–170, 1996.
- [41091] 14643. Schorpp, M.; Hofmann, M.; Dear, T. N.; Boehm, T.: Characterization of mouse and human nude genes. *Immunogenetics* 46: 509–515, 1997.
- [41092] 14644. Segre, J. A.; Nemhauser, J. L.; Taylor, B. A.; Nadeau, J. H.; Lander, E. S.: Positional cloning of the nude locus: genetic, physical, and transcription maps of the region and mutations in the mouse and rat. *Genomics* 28:549–559, 1995.

- [41093] 14645.Delpire, E.; Lu, J.; England, R.; Dull, C.; Thorne, T.: Deafness and imbalance associated with inactivation of the secretory Na-K-2Cl co-transporter. *Nature Genet.* 22: 192-195, 1999.
- [41094] 14646.Evans, R. L.; Park, K.; Turner, R. J.; Watson, G. E.; Nguyen, H.-V.; Dennett, M. R.; Hand, A. R.; Flagella, M.; Shull, G. E.; Melvin, J.E.: Severe impairment of salivation in Na⁺/K⁺/2Cl⁻ cotransporter(NKCC1)-deficient mice. *J. Biol. Chem.* 275: 26720-26726, 2000.
- [41095] 14647.Payne, J. A.; Xu, J.-C.; Haas, M.; Lytle, C. Y.; Ward, D.; Forbush, B., III: Primary structure, functional expression, and chromosomal localization of the bumetanide-sensitive Na-K-Cl cotransporter in human colon. *J. Biol. Chem.* 270: 17977-17985, 1995.
- [41096] 14648.Quaggin, S. E.; Payne, J. A.; Forbush, B., III; Igarashi, P.: Localization of the renal Na-K-Cl cotransporter gene (Slc12a1) on mouse chromosome 2. *Mammalian Genome* 6: 557-561, 1995.
- [41097] 14649.Xu, J.-C.; Lytle, C.; Zhu, T. T.; Payne, J. A.; Benz, E., Jr.; Forbush, B., III: Molecular cloning and functional expression of the bumetanide-sensitive Na-K-Cl cotransporter. *Proc. Nat. Acad.Sci.* 91: 2201-2205, 1994.
- [41098] 14650.Daniels, S. E.; Bhattacharya, S.; James, A.; Leaves,

N. I.; Young,A.; Hill, M. R.; Faux, J. A.; Ryan, G. F.; le Souef, P. N.; Lathrop,G. M.; Musk, A. W.; Cookson, W. O. C. M.: A genome-wide search for quantitative trait loci underlying asthma. *Nature* 383: 247–250,1996.

[41099] 14651.MacDonald, S. M.; Paznekas, W. A.; Jabs, E. W.: Chromosomal localization of tumor protein, translationally-controlled 1 (TPT1) encoding the human histamine releasing factor (HRF) to 13q12–q14. *Cytogenet. Cell Genet.* (in-press), 1999.

[41100] 14652.MacDonald, S. M.; Rafnar, T.; Langdon, J.; Lichtenstein, L. M.: Molecular identification of an IgE-dependent histamine-releasing factor. *Science* 269: 688–690, 1995.

[41101] 14653.Zhang, K.; Saxon, A.; Max, E. E.: Two unusual forms of human immunoglobulin E encoded by alternative RNA splicing of the epsilon heavy chain membrane exons. *J. Exp. Med.* 176: 233–243, 1992.

[41102] 14654.Simister, N. E.; Mostov, K. E.: An Fc receptor structurally related to MHC class I antigens. *Nature* 337: 184–187, 1989.

[41103] 14655.Sedlacek, Z.; Konecki, D. S.; Korn, B.; Klauck, S. M.; Poustka,A.: Evolutionary conservation and genomic organization of XAP-4, an Xq28 located gene coding for a human rab GDP-dissociation inhibitor (GDI). *Mammalian*

Genome 5: 633–639, 1995.

- [41104] 14656.Sedlacek, Z.; Munstermann, E.; Mincheva, A.; Lichter, P.; Poustka,A.: The human rab GDI beta gene with long retroposon-rich introns maps to 10p15 and its pseudogene to 7p11–p13. Mammalian Genome 9:78–80, 1998.
- [41105] 14657.Desseyn, J.–L.; Aubert, J.–P.; van Seuning, I.; Porchet, N.;Laine, A.: Genomic organization of the 3–prime region of the human mucin gene MUC5B. J. Biol. Chem. 272: 16873–16883, 1997.
- [41106] 14658.Desseyn, J.–L.; Guyonnet–Duperat, V.; Porchet, N.; Aubert, J.–P.;Laine, A.: Human mucin gene MUC5B, the 10.7–kb large central exon encodes various alternate subdomains resulting in a super-repeat: structural evidence for a 11p15.5 gene family. J. Biol. Chem. 272:3168–3178, 1997.
- [41107] 14659.Gipson, I. K.; Moccia, R.; Spurr–Michaud, S.; Argueso, P.; Gargiulo,A. R.; Hill, J. A., III; Offner, G. D.; Keutmann, H. T.: The amount of MUC5B in cervical mucus peaks at midcycle. J. Clin. Endocr. Metab. 86:594–600, 2001.
- [41108] 14660.Keates, A. C.; Nunes, D. P.; Afdhal, N. H.; Troxler, R. F.; Offner,G. D.: Molecular cloning of a major human

gall bladder mucin: complete C-terminal sequence and genomic organization of MUC5B. *Biochem. J.* 324:295–303, 1997.

[41109] 14661. Troxler, R. F.; Offner, G. D.; Zhang, F.; Iontcheva, I.; Oppenheim, F. G.: Molecular cloning of a novel high molecular weight mucin (MG1) from human sublingual gland. *Biochem. Biophys. Res. Commun.* 217:1112–1119, 1995.

[41110] 14662. Birck, C.; Poch, O.; Romier, C.; Ruff, M.; Mengus, G.; Lavigne, A.-C.; Davidson, I.; Moras, D.: Human TAFII28 and TAFII18 interact through a histone fold encoded by atypical evolutionary conserved motifs also found in the SPT3 family. *Cell* 94: 239–249, 1998.

[41111] 14663. Mengus, G.; May, M.; Jacq, X.; Staub, A.; Tora, L.; Chambon, P.; Davidson, I.: Cloning and characterization of hTAFII18, hTAFII20 and hTAFII28: three subunits of the human transcription factor TFIID. *EMBO J.* 14: 1520–1531, 1995.

[41112] 14664. Waisfisz, Q.; Saar, K.; Morgan, N. V.; Altay, C.; Leegwater, P. A.; de Winter, J. P.; Komatsu, K.; Evans, G. R.; Wegner, R.-D.; Reis, A.; Joenje, H.; Arwert, F.; Mathew, C. G.; Pronk, J. C.; Digweed, M.: The Fanconi anemia group E gene, FANCE, maps to chromosome 6p. *Am. J. Hum.*

Genet. 64: 1400–1405, 1999.

- [41113] 14665.Wegner, R.–D.; Henrichs, I.; Joenje, H.; Schroeder–Kurth, T.:Fanconi anemia complementation group E: clinical and cytogenetic dataof the first patient. Clin. Genet. 50: 479–482, 1996.
- [41114] 14666.Green, P.; Lipman, D.; Hillier, L.; Waterston, R.; States, D.;Claverie, J.–M.: Ancient conserved regions in new gene sequencesand the protein databases. Science 259: 1711–1716, 1993.
- [41115] 14667.Schwartz, F.; Eisenman, R.; Knoll, J.; Gessler, M.; Bruns, G.:cDNA sequence, genomic organization, and evolutionary conservationof a novel gene from the WAGR region. Genomics 29: 526–532, 1995.
- [41116] 14668.Schwartz, F.; Neve, R.; Eisenman, R.; Gessler, M.; Bruns, G.:A WAGR region gene between PAX–6 and FSHB expressed in fetal brain. Hum.Genet. 94: 658–664, 1994.
- [41117] 14669.Krishnan, B. R.; Jamry, I.; Chaplin, D. D.: Feature mapping ofthe HLA class I region: localization of the POU5F1 and TCF19 genes. Genomics 30:53–58, 1995.
- [41118] 14670.Ku, D.–H.; Chang, C.; Koniecki, J.; Cannizzaro, L. A.; Boghosian–Sell,L.; Alder, H.; Baserga, R.: A new growth–regulated complementaryDNA with the sequence of a putative trans–activating factor. CellGrowth Differ. 2:

179–186, 1991.

- [41119] 14671. Joseph, R. E.; Walker, J.; Norris, F. A.: Assignment of the inositolpolyphosphate 4–phosphatase type I gene (INPP4A) to human chromosome band 2q11.2 by in situ hybridization. *Cytogenet. Cell Genet.* 87:276–277, 1999.
- [41120] 14672. Norris, F. A.; Auethavekiat, V.; Majerus, P. W.: The isolation and characterization of cDNA encoding human and rat brain inositolpolyphosphate 4–phosphatase. *J. Biol. Chem.* 270: 16128–16133, 1995.
- [41121] 14673. van den Maagdenberg, A. M. J. M.; van den Hurk, H. H.; Olde Weghuis, D.; Wieringa, B.; Geurts van Kessel, A.; Hendriks, W. J. A. J.: Assignment of the human protein tyrosine phosphatase epsilon (PTPRE) gene to chromosome 10q26 by fluorescence in situ hybridization. *Genomics* 30:128–129, 1995.
- [41122] 14674. Kim, D. S.; Jung, H.–H.; Park, S.–H.; Chin, H.: Isolation and characterization of the 5–prime–upstream region of the human N–type calcium channel alpha–1B subunit gene: chromosomal localization and promoter analysis. *J. Biol. Chem.* 272: 5098–5104, 1997.
- [41123] 14675. Williams, M. E.; Brust, P. F.; Feldman, D. H.; Patthi, S.; Simerson, S.; Maroufi, A.; McCue, A. F.; Velicelebi, G.; Ellis, S. B.; Harpold, M. M.: Structure and functional ex–

pression of an omega-conotoxin-sensitive human N-type calcium channel. *Science* 257: 389–395, 1992.

[41124] 14676. Azim, A. C.; Knoll, J. H. M.; Marfatia, S. M.; Peel, D. J.; Bryant, P. J.; Chishti, A. H.: DLG1: chromosome location of the closest human homologue of the *Drosophila* discs large tumor suppressor gene. *Genomics* 30:613–616, 1995.

[41125] 14677. Burgess, D. L.; Rafael, J. A.; Meisler, M. H.; Chamberlain, J. S.: Dlgh1, a mouse homolog of the *Drosophila* discs-large gene, is located on chromosome 16. *Mammalian Genome* 7: 623–624, 1996.

[41126] 14678. Hanada, T.; Lin, L.; Chandy, K. G.; Oh, S. S.; Chishti, A. H.: Human homologue of the *Drosophila* discs large tumor suppressor binds to p56lck tyrosine kinase and Shaker type Kv1.3 potassium channel in T lymphocytes. *J. Biol. Chem.* 272: 26899–26904, 1997.

[41127] 14679. Lue, R. A.; Marfatia, S. M.; Branton, D.; Chishti, A. H.: Cloning and characterization of hdlg: the human homologue of the *Drosophila* discs large tumor suppressor binds to protein 4.1. *Proc. Nat. Acad. Sci.* 91: 9818–9822, 1994.

[41128] 14680. Mori, K.; Iwao, K.; Miyoshi, Y.; Nakagawara, A.; Kofu, K.; Akiyama, T.; Arita, N.; Hayakawa, T.; Nakamura,

Y.: Identification of brain-specific splicing variants of the hDLG1 gene and altered splicing in neuroblastoma cell lines. *J. Hum. Genet.* 43: 123–127, 1998.

[41129] 14681. Peters, L. L.; Ciciotte, S. L.; Lin, L.; Chishti, A. H.: The mouse homolog of the *Drosophila* discs large tumor suppressor gene maps to chromosome 16. *Mammalian Genome* 7: 619–620, 1996.

[41130] 14682. Bertaux, F.; Sharp, A. H.; Ross, C. A.; Lehrach, H.; Bates, G. P.; Wanker, E.: HAP1–huntingtin interactions do not contribute to the molecular pathology in Huntington's disease transgenic mice. *FEBS Lett.* 426: 229–232, 1998.

[41131] 14683. Engelender, S.; Sharp, A. H.; Colomer, V.; Tokito, M. K.; Lanahan, A.; Worley, P.; Holzbaur, E. L. F.; Ross, C. A.: Huntingtin-associated protein 1 (HAP1) interacts with the p150(Glued) subunit of dynactin. *Hum. Molec. Genet.* 6: 2205–2212, 1997.

[41132] 14684. Li, S.-H.; Hosseini, S. H.; Gutekunst, C.-A.; Hersch, S. M.; Ferrante, R. J.; Li, X.-J.: A human HAP1 homologue: cloning, expression, and interaction with huntingtin. *J. Biol. Chem.* 273: 19220–19227, 1998.

[41133] 14685. Li, X.-J.; Li, S.-H.; Sharp, A. H.; Nucifora, F. C., Jr.; Schilling, G.; Lanahan, A.; Worley, P.; Snyder, S. H.; Ross, C. A.: A huntingtin-associated protein enriched in brain with

implications for pathology. *Nature* 378:398–402, 1995.

- [41134] 14686.Nasir, J.; Duan, K.; Nichol, K.; Engelender, S.; Ashworth, R.; Colomer, V.; Thomas, S.; Disteché, C. M.; Hayden, M. R.; Ross, C.A.: Gene structure and map location of the murine homolog of theHuntington–associated protein, Hap1. *Mammalian Genome* 9: 565–570,1998.
- [41135] 14687.Nasir, J.; Lafuente, M.–J.; Duan, K.; Colomer, V.; Engelender,S.; Ingersoll, R.; Margolis, R. L.; Ross, C. A.; Hayden, M. R.: Humanhuntingtin–associated protein (HAP–1) gene: genomic organisation andan intragenic polymorphism. *Gene* 254: 181–187, 2000.
- [41136] 14688.Broccoli, D.; Chong, L.; Oelmann, S.; Fernald, A. A.; Marziliano,N.; van Steensel, B.; Kipling, D.; Le Beau, M. M.; de Lange, T.:Comparison of the human and mouse genes encoding the telomeric protein,TRF1: chromosomal localization, expression and conserved protein domains. *Hum.Molec. Genet.* 6: 69–76, 1997.
- [41137] 14689.Chong, L.; van Steensel, B.; Broccoli, D.; Erdjument–Bromage, H.;Hanish, J.; Tempst, P.; de Lange, T.: A human telomeric protein. *Science* 270:1663–1667, 1995.
- [41138] 14690.Fairall, L.; Chapman, L.; Moss, H.; de Lange, T.; Rhodes, D.:Structure of the TRFH dimerization domain of the human telomeric proteinsTRF1 and TRF2. *Molec. Cell*

8: 351–361, 2001.

- [41139] 14691.Griffith, J. D.; Comeau, L.; Rosenfield, S.; Stansel, R. M.; Bianchi,A.; Moss, H.; de Lange, T.: Mammalian telomeres end in a large duplexloop. *Cell* 97: 503–514, 1999.
- [41140] 14692.Kim, S.; Kaminker, P.; Campisi, J.: TIN2, a new regulator of telomere length in human cells. *Nature Genet.* 23: 405–412, 1999.
- [41141] 14693.Marcand, S.; Gilson, E.; Shore, D.: A protein-counting mechanismfor telomere length regulation in yeast. *Science* 275: 986–990, 1997.
- [41142] 14694.Okabe, J.; Eguchi, A.; Masago, A.; Hayakawa, T.; Nakanishi, M.: TRF1 is a critical trans-acting factor required for de novo telomereformation in human cells. *Hum. Molec. Genet.* 9: 2639–2650, 2000.
- [41143] 14695.Young, A. C.; Chavez, M.; Giambernardi, T. A.; Mattern, V.; McGill,J. R.; Harris, J. M.; Sarosdy, M. F.; Patel, P.; Sakaguchi, A. Y.: Organization and expression of human telomere repeat binding factorgenes. *Somat. Cell Molec. Genet.* 23: 275–286, 1997.
- [41144] 14696.Zakian, V. A.: Telomeres: beginning to understand the end. *Science* 270:1601–1607, 1995.
- [41145] 14697.Brodsky, G.; Otterson, G. A.; Parry, B. B.; Hart, I.; Patterson,D.; Kaye, F. J.: Localization of STCH to human

chromosome 21q11.1. Genomics 30:627–628, 1995.

- [41146] 14698.Otterson, G. A.; Flynn, G. C.; Kratzke, R. A.; Coxon, A.; Johnston,P. G.; Kaye, F. J.: Stch encodes the 'ATPase core' of a microsomalstress70 protein. EMBO J. 13: 1216–1225, 1994.
- [41147] 14699.Reeves, R. H.; Rue, E.; Yu, J.; Kao, F.–T.: Stch maps to mousechromosome 16, extending the conserved synteny with human chromosome21. Genomics 49: 156–157, 1998.
- [41148] 14700.Nielsen, P. J.; Rochelle, J. M.; Seldin, M. F.: The functionalgenes for protein synthesis initiation factor 4AI and 4AII map to mouse chromosomes 11 and 16. Mammalian Genome 4: 185–186, 1993.
- [41149] 14701.Nielsen, P. J.; Trachsel, H.: The mouse protein synthesis initiationfactor 4A gene family includes two related functional genes whichare differentially expressed. EMBO J. 7: 2097–2105, 1988.
- [41150] 14702.Sudo, K.; Takahashi, E.; Nakamura, Y.: Isolation and mapping ofthe human EIF4A2 gene homologous to the murine protein synthesis initiationfactor 4A–II gene Eif4a2. Cytogenet. Cell Genet. 71: 385–388, 1995.
- [41151] 14703.Gladyshev, V. N.; Jeang, K.–T.; Stadtman, T. C.: Selenocysteine,identified as the penultimate C–terminal

residue in human T-cell thioredoxin reductase, corresponds to TGA in the human placental gene. *Proc. Nat. Acad. Sci.* 93: 6146–6151, 1996.

- [41152] 14704. Zhang, K.; Lindsberg, P. J.; Tatlisumak, T.; Kaste, M.; Olsen, H. S.; Andersson, L. C.: Stanniocalcin: a molecular guard of neurons during cerebral ischemia. *Proc. Nat. Acad. Sci.* 97: 3637–3642, 2000.
- [41153] 14705. Gillett, G. T.; Fox, M. F.; Rowe, P. S. N.; Casimir, C. M.; Povey, S.: Mapping of human non-muscle type cofilin (CFL1) to chromosome 11q13 and muscle-type cofilin (CFL2) to chromosome 14. *Ann. Hum. Genet.* 60: 201–211, 1996.
- [41154] 14706. Ogawa, K.; Tashima, M.; Yumoto, Y.; Okuda, T.; Sawada, H.; Okuma, M.; Maruyama, Y.: Coding sequence of human placenta cofilin cDNA. *Nuc. Acids Res.* 18: 7169 only, 1990.
- [41155] 14707. Ono, S.; Minami, N.; Abe, H.; Obinata, T.: Characterization of a novel cofilin isoform that is predominantly expressed in mammalian skeletal muscle. *J. Biol. Chem.* 269: 15280–15286, 1994.
- [41156] 14708. Georgakopoulos, T.; Thireos, G.: Two distinct yeast transcriptional activators require the function of the GCN5 protein to promote normal levels of transcription. *EMBO J.*

11: 4145–4152, 1992.

- [41157] 14709. Inoue, M.; Isomura, M.; Ikegawa, S.; Fujiwara, T.; Shin, S.; Moriya, H.; Nakamura, Y.: Isolation and characterization of a human cDNA clone (GCN5L1) homologous to GCN5, a yeast transcription factor. *Cytogenet. Cell Genet.* 73: 134–136, 1996.
- [41158] 14710. Emahazion, T.; Beskow, A.; Gyllenstein, U.; Brookes, A. J.: Intron-based radiation hybrid mapping of 15 complex I genes of the human electron transport chain. *Cytogenet. Cell Genet.* 82: 115–119, 1998.
- [41159] 14711. Gu, J. Z.; Lin, X.; Wells, D. E.: The human B22 subunit of the NADH–ubiquinone oxidoreductase maps to the region of chromosome 8 involved in branchio–oto–renal syndrome. *Genomics* 35: 6–10, 1996.
- [41160] 14712. Lin, X.; Wells, D. E.; Kimberling, W. J.; Kumar, S.: Human NDUF9 gene: genomic organization and a possible candidate gene associated with deafness disorder mapped to chromosome 8q13. *Hum. Hered.* 49: 75–80, 1999.
- [41161] 14713. Wilkinson, K. D.; Tashayev, V. L.; O'Connor, L. B.; Larsen, C. N.; Kasperek, E.; Pickart, C. M.: Metabolism of the polyubiquitin degradation signal: structure, mechanism, and role of isopeptidase T. *Biochemistry* 34:

14535–14546, 1995.

- [41162] 14714. Byrd, P. J.; Cooper, P. R.; Stankovic, T.; Kullar, H. S.; Watts, G. D. J.; Robinson, P. J.; Taylor, M. R.: A gene transcribed from the bidirectional ATM promoter coding for a serine rich protein: amino acid sequence, structure and expression studies. *Hum. Molec. Genet.* 5:1785–1791, 1996.
- [41163] 14715. Chen, X.; Yang, L.; Udar, N.; Liang, T.; Uhrhammer, N.; Xu, S.; Bay, J.-O.; Wang, Z.; Dandekar, S.; Chiplunkar, S.; Klisak, I.; Telatar, M.; Yang, H.; Concannon, P.; Gatti, R. A.: CAND3: a ubiquitously expressed gene immediately adjacent and in opposite transcriptional orientation to the ATM gene at 11q23.1. *Mammalian Genome* 8: 129–133, 1997.
- [41164] 14716. Imai, T.: Personal Communication. Chiba, Japan 9/12/1996.
- [41165] 14717. Imai, T.; Sugawara, T.; Nishiyama, A.; Shimada, R.; Ohki, R.; Seki, N.; Sagara, M.; Ito, H.; Yamauchi, M.; Hori, T.: The structure and organization of the human NPAT gene. *Genomics* 42: 388–392, 1997.
- [41166] 14718. Imai, T.; Yamauchi, M.; Seki, N.; Sugawara, T.; Saito, T.; Matsuda, Y.; Ito, H.; Nagase, T.; Nomura, N.; Hori, T.: Identification and characterization of a new gene physi-

cally linked to the ATM gene. *GenomeRes.* 6: 439–447, 1996.

[41167] 14719. Borowsky, B.; Hoffman, B. J.: Analysis of a gene encoding two glycine transporter variants reveals alternative promoter usage and a novel gene structure. *J. Biol. Chem.* 273: 29077–29085, 1998.

[41168] 14720. Borowsky, B.; Mezey, E.; Hoffman, B. J.: Two glycine transporter variants with distinct localization in the CNS and peripheral tissues are encoded by a common gene. *Neuron* 10: 851–863, 1993.

[41169] 14721. Jones, E. M. C.; Fernald, A.; Bell, G. I.; Le Beau, M. M.: Assignment of SLC6A9 to human chromosome band 1p33 by in situ hybridization. *Cytogenet. Cell Genet.* 71: 211, 1995.

[41170] 14722. Kim, K.-M.; Kingsmore, S. F.; Han, H.; Yang-Feng, T. L.; Godinot, N.; Seldin, M. F.; Caron, M. G.; Giros, B.: Cloning of the human glycine transporter type 1: molecular and pharmacological characterization of novel isoform variants and chromosomal localization of the gene in the human and mouse genomes. *Molec. Pharmacol.* 45: 608–617, 1994.

[41171] 14723. Freeman, G. J.; Gribben, J. G.; Boussiotis, V. A.; Ng, J. W.; Restivo, V. A.; Lombard, L. A.; Gray, G. S.; Nadler, L.

M. Cloning of B7-2:a CTLA-4 counter-receptor that costimulates human T cell proliferation. Science 262:909-911, 1993.

[41172] 14724.Jeannin, P.; Magistrelli, G.; Aubry, J.-P.; Caron, G.; Gauchat,J.-F.; Renno, T.; Herbault, N.; Goetsch, L.; Blaecke, A.; Dietrich,P.-Y.; Bonnefoy, J.-Y.; Delneste, Y.: Soluble CD86 is a costimulatorymolecule for human T lymphocytes. Immunity 13: 303-312, 2000.

[41173] 14725.Jellis, C. L.; Wang, S. S.; Rennert, P.; Borriello, F.; Sharpe,A. H.; Green, N. R.; Gray, G. S.: Genomic organization of the genecoding for the costimulatory human B-lymphocyte antigen B7-2 (CD86). Immunogenetics 42:85-89, 1995.

[41174] 14726.Arai, Y.; Hosoda, F.; Kobayashi, H.; Arai, K.; Hayashi, Y.; Kamada,N.; Kaneko, Y.; Ohki, M.: The inv(11)(p15q22) chromosome translocationof de novo and therapy-related myeloid malignancies results in fusionof the nucleoporin gene, NUP98, with the putative RNA heli-case gene,DDX10. Blood 89: 3936-3944, 1997.

[41175] 14727.Dash, A. B.; Williams, I. R.; Kutok, J. L.; Tomasson, M. H.; Anastasiadou,E.; Lindahl, K.; Li, S.; Van Etten, R. A.; Borrow, J.; Housman, D.;Druker, B.; Gilliland, D. G.: A murine model of CML blast crisisinduced by cooperation

between BCR/ABL and NUP98/HOXA9. *Proc. Nat. Acad. Sci.* 99: 7622–7627, 2002.

- [41176] 14728. Enninga, J.; Levy, D. E.; Blobel, G.; Fontoura, B. M. A.: Role of nucleoporin induction in releasing an mRNA nuclear export block. *Science* 295:1523–1525, 2002.
- [41177] 14729. Fontoura, B. M.; Blobel, G.; Matunis, M. J.: A conserved biogenesis pathway for nucleoporins: proteolytic processing of a 186-kilodalton precursor generates Nup98 and the novel nucleoporin, Nup96. *J. Cell Biol.* 144: 1097–1112, 1999.
- [41178] 14730. Hodel, A. E.; Hodel, M. R.; Griffis, E. R.; Hennig, K. A.; Ratner, G. A.; Xu, S.; Powers, M. A.: The three-dimensional structure of the autoproteolytic, nuclear pore-targeting domain of the human nucleoporin Nup98. *Molec. Cell* 10: 347–358, 2002.
- [41179] 14731. Jaju, R. J.; Fidler, C.; Haas, O. A.; Strickson, A. J.; Watkins, F.; Clark, K.; Cross, N. C. P.; Cheng, J.-F.; Aplan, P. D.; Kearney, L.; Boulton, J.; Wainscoat, J. S.: A novel gene, NSD1, is fused to NUP98 in the t(5;11)(q35;p15.5) in de novo childhood acute myeloid leukemia. *Blood* 98: 1264–1267, 2001.
- [41180] 14732. Jaju, R. J.; Haas, C. A.; Neat, M.; et al: A new recurrent translocation, t(5;11)(q35p15.5), associated with

del(5q) in childhood acute myeloid leukemia. *Blood* 94: 773–780, 1999.

- [41181] 14733. Mizuno, T.; Kaibuchi, K.; Yamamoto, T.; Kawamura, M.; Sakoda, T.; Fujioka, H.; Matsuura, Y.; Takai, Y.: A stimulatory GDP/GTP exchange protein for smg p21 is active on the post-translationally processed form of c-Ki-ras p21 and rhoA p21. *Proc. Nat. Acad. Sci.* 88: 6442–6446, 1991.
- [41182] 14734. Nakamura, T.; Largaespada, D. A.; Lee, M. P.; Johnson, L. A.; Ohyashiki, K.; Toyama, K.; Chen, S. J.; Willman, C. L.; Chen, I.-M.; Feinberg, A. P.; Jenkins, N. A.; Copeland, N. G.; Shaughnessy, J. D., Jr.: Fusion of the nucleoporin gene NUP98 to HOXA9 by the chromosomal translocation t(7;11)(p15;p15) in human myeloid leukaemia. *Nature-Genet.* 12: 154–158, 1996.
- [41183] 14735. Radu, A.; Moore, M. S.; Blobel, G.: The peptide repeat domain of nucleoporin Nup98 functions as a docking site in transport across the nuclear pore complex. *Cell* 81: 215–222, 1995.
- [41184] 14736. Rosati, R.; La Starza, R.; Veronese, A.; Aventin, A.; Schwienbacher, C.; Vallespi, T.; Negrini, M.; Martelli, M. F.; Mecucci, C.: NUP98 is fused to the NSD3 gene in acute myeloid leukemia associated with t(8;11)(p11.2;p15). *Blood* 99: 3857–3860, 2002.

- [41185] 14737.von Kobbe, C.; van Deursen, J. M. A.; Rodrigues, J. P.; Sitterlin,D.; Bachi, A.; Wu, X.; Wilm, M.; Carmo-Fonseca, M.; Izaurralde, E.: Vesicular stomatitis virus matrix protein inhibits host cell geneexpression by targeting the nucleoporin Nup98. *Molec. Cell* 6: 1243–1252,2000.
- [41186] 14738.Cheng, Y.; Austin, S. C.; Rocca, B.; Koller, B. H.; Coffman, T.M.; Grosser, T.; Lawson, J. A.; FitzGerald, G. A.: Role of prostacyclinin the cardiovascular response to thromboxane A2. *Science* 296: 539–541,2002.
- [41187] 14739.Fuse, I.; Mito, M.; Hattori, A.; Higuchi, W.; Shibata, A.; Ushikubi,F.; Okuma, M.; Yahata, K.: Defective signal transduction inducedby thromboxane A2 in a patient with a mild bleeding disorder: impairedphospholipase C activation despite normal phospholipase A2 activation. *Blood* 81:994–1000, 1993.
- [41188] 14740.Hirata, M.; Hayashi, Y.; Ushikubi, F.; Yokota, Y.; Kageyama, R.;Nakanishi, S.; Narumiya, S.: Cloning and expression of cDNA for ahuman thromboxane A2 receptor. *Nature* 349: 617–620, 1991.
- [41189] 14741.Hirata, T.; Kakizuka, A.; Ushikubi, F.; Fuse, I.; Okuma, M.; Narumiya,S.: Arg60-to-leu mutation of the human thromboxane A2 receptor ina dominantly inherited bleeding disorder. *J. Clin. Invest.* 94: 1662–1667,1994.

- [41190] 14742.Watanabe, O.; Maruyama, I.; Arimura, K.; Kitajima, I.; Arimura,H.; Hanatani, M.; Matsuo, K.; Arisato, T.; Osame, M.: Overproductionof vascular endothelial growth factor/vascular permeability factoris causative in Crow-Fukase (POEMS) syndrome. Muscle Nerve 21: 1390–1397,1998.
- [41191] 14743.Wei, M.–H.; Popescu, N. C.; Lerman, M. I.; Merrill, M. J.; Zimonjic,D. B.: Localization of the human vascular endothelial growth factorgene, VEGF, at chromosome 6p12. Hum. Genet. 97: 794–797, 1996.
- [41192] 14744.Wong, A. K.; Alfert, M.; Castrillon, D. H.; Shen, Q.; Holash,J.; Yancopoulos, G. D.; Chin, L.: Excessive tumor-elaborated VEGFand its neutralization define a lethal para-neoplastic syndrome. Proc.Nat. Acad. Sci. 98: 7481–7486, 2001.
- [41193] 14745.Wulff, C.; Wilson, H.; Largue, P.; Duncan, W. C.; Armstrong, D.G.; Fraser, H. M.: Angiogenesis in the human corpus luteum: localizationand changes in angiopoi-etins, Tie-2, and vascular endothelial growthfactor messenger ribonucleic acid. J. Clin. Endocr. Metab. 85: 4302–4309,2000.
- [41194] 14746.Ylikorkala, A.; Rossi, D. J.; Korsisaari, N.; Luukko, K.; Alitalo,K.; Henkemeyer, M.; Makela, T. P.: Vascular ab-

normalities and deregulation of VEGF in Lkb1-deficient mice. *Science* 293: 1323–1326, 2001.

- [41195] 14747. Bodner, M.; Fridkin, M.; Gozes, I.: Coding sequences for vasoactive intestinal peptide and PHM-27 peptide are located on two adjacent exons in the human genome. *Proc. Nat. Acad. Sci.* 82: 3548–3551, 1985.
- [41196] 14748. Delgado, M.; Abad, C.; Martinez, C.; Leceta, J.; Gomariz, R. P.: Vasoactive intestinal peptide prevents experimental arthritis by downregulating both autoimmune and inflammatory components of the disease. *Nature Med.* 7: 563–568, 2001.
- [41197] 14749. Gotoh, E.; Yamagami, T.; Yamamoto, H.; Okamoto, H.: Chromosomal assignment of human VIP/PHM-27 gene to 6q26–q27 region by spot blot hybridization and in situ hybridization. *Biochem. Int.* 17: 555–562, 1988.
- [41198] 14750. Gozes, I.; Avidor, R.; Yahav, Y.; Katznelson, D.; Croce, C. M.; Huebner, K.: The gene encoding vasoactive intestinal peptide is located on human chromosome 6p21–6qter. *Hum. Genet.* 75: 41–44, 1987.
- [41199] 14751. Gozes, I.; Nakai, H.; Byers, M.; Avidor, R.; Weinstein, Y.; Shani, Y.; Shows, T. B.: Sequential expression in the nervous system of C-MYB and VIP genes, located in human chromosomal region 6q24. *Somat. Cell Molec.*

Genet. 13: 305–313, 1987.

- [41200] 14752.Heinz–Erian, P.; Dey, R. D.; Flux, M.; Said, S. I.: De-
ficientvasoactive intestinal peptide innervation in sweat
glands of cysticfibrosis patients. Science 229: 1407–1408,
1985.
- [41201] 14753.Itoh, N.; Obata, K.; Yanaihara, N.; Okamoto, H.:
Human preprovasoactiveintestinal polypeptide contains a
novel PHI–27–like peptide, PHM–27. Nature 304:547–549,
1983.
- [41202] 14754.Linder, S.; Barkhem, T.; Norberg, A.; Persson, H.;
Schalling, M.;Hokfelt, T.; Magnusson, G.: Structure and
expression of the geneencoding the vasoactive intestinal
peptide precursor. Proc. Nat.Acad. Sci. 84: 605–609,
1987.
- [41203] 14755.Omary, M. B.; Kagnoff, M. F.: Identification of nu-
clear receptorsfor VIP on a human colonic adenocarci-
noma cell line. Science 238:1578–1581, 1987.
- [41204] 14756.Couvineau, A.; Rouyer–Fessard, C.; Darmoul, D.;
Maoret, J.–J.;Carrero, I.; Ogier–Denis, E.; Laburthe, M.: Hu-
man intestinal VIPreceptor: cloning and functional expres-
sion of two cDNA encoding proteinswith different N-
terminal domains. Biochem. Biophys. Res. Commun.
200:769–776, 1994.

- [41205] 14757.Hashimoto, H.; Nishino, A.; Shintani, N.; Hagiwara, N.; Copeland, N. G.; Jenkins, N. A.; Yamamoto, K.; Matsuda, T.; Ishihara, T.; Nagata, S.; Baba, A.: Genomic organization and chromosomal location of the mouse vasoactive intestinal polypeptide 1 (VIPAC-1) receptor. *Genomics* 58:90–93, 1999.
- [41206] 14758.Sreedharan, S. P.; Huang, J.-X.; Cheung, M.-C.; Goetzl, E. J.: Structure, expression, and chromosomal localization of the type I human vasoactive intestinal peptide receptor gene. *Proc. Nat. Acad. Sci.* 92: 2939–2943, 1995.
- [41207] 14759.Sreedharan, S. P.; Patel, D. R.; Huang, J.-X.; Goetzl, E. J.: Cloning and functional expression of a human neuroendocrine vasoactive intestinal peptide receptor. *Biochem. Biophys. Res. Commun.* 193:546–553, 1993.
- [41208] 14760.Sreedharan, S. P.; Robichon, A.; Peterson, K. E.; Goetzl, E. J.: Cloning and expression of the human vasoactive intestinal peptide receptor. *Proc. Nat. Acad. Sci.* 88: 4986–4990, 1991.
- [41209] 14761.Ueno, S.; Maruki, Y.; Nakamura, M.; Tomemori, Y.; Kamae, K.; Tanabe, H.; Yamashita, Y.; Matsuda, S.; Kaneko, S.; Sano, A.: The gene encoding a newly discovered protein, chorein, is mutated in chorea-acanthocytosis. *Nature Genet.* 28: 121–122, 2001.

- [41210] 14762.Hirata, T.; Ushikubi, F.; Kakizuka, A.; Okuma, M.; Narumiya, S.: Two thromboxane A(2) receptor isoforms in human platelets: opposite coupling to adenylyl cyclase with different sensitivity to arg60-to-leu mutation. *J. Clin. Invest.* 97: 949–956, 1996.
- [41211] 14763.Nusing, R. M.; Hirata, M.; Kakizuka, A.; Eki, T.; Ozawa, K.; Narumiya, S.: Characterization and chromosomal mapping of the human thromboxane A2 receptor gene. *J. Biol. Chem.* 268: 25253–25259, 1993.
- [41212] 14764.Schwengel, D. A.; Nouri, N.; Meyers, D. A.; Levitt, R. C.: Linkage mapping of the human thromboxane A2 receptor (TBXA2R) to chromosome 19p13.3 using transcribed 3-prime untranslated DNA sequence polymorphisms. *Genomics* 18:212–215, 1993.
- [41213] 14765.Thomas, D. W.; Mannon, R. B.; Mannon, P. J.; Lattour, A.; Oliver, J. A.; Hoffman, M.; Smithies, O.; Koller, B. H.; Coffman, T. M.: Coagulation defects and altered hemodynamic responses in mice lacking receptors for thromboxane A(2). *J. Clin. Invest.* 102: 1994–2001, 1998.
- [41214] 14766.Unoki, M.; Furuta, S.; Onouchi, Y.; Watanabe, O.; Doi, S.; Fujiwara, H.; Miyatake, A.; Fujita, K.; Tamari, M.; Nakamura, Y.: Association studies of 33 single nucleotide polymorphisms (SNPs) in 29 candidate genes for bronchial

asthma: positive association a T924C polymorphism in the thromboxane A₂ receptor gene. Hum. Genet. 106: 440–446, 2000.

- [41215] 14767. Ushikubi, F.; Nakajima, M.; Hirata, M.; Okuma, M.; Fujiwara, M.; Narumiya, S.: Purification of the thromboxane A₂/prostaglandin H₂receptor from human blood platelets.. J. Biol. Chem. 264: 16496–16501, 1989.
- [41216] 14768. Ushikubi, F.; Okuma, M.; Kanaji, K.; Sugiyama, T.; Ogorochi, T.; Narumiya, S.; Uchino, H.: Hemorrhagic thrombocytopathy with platelet thromboxane A₂ abnormality: defective signal transduction with normal binding activity. Thromb. Haemost. 57: 158–164, 1987.
- [41217] 14769. Ades, E. W.; Zwerner, R. K.; Acton, R. T.; Balch, C. M.: Isolation and partial characterisation of the human homologue of Thy-1. J. Exp. Med. 151: 400–406, 1980.
- [41218] 14770. Bonewald, L.; Ades, E. W.; Tung, E.; Marchalonis, J. J.; Wang, A. C.: Biochemical characterization of human Thy-1. J. Immunogenet. 11: 283–296, 1984.
- [41219] 14771. Gatti, R. A.; Lathrop, G. M.; Salser, W.; Silver, J.; Lalouel, J. M.; White, R.: Location of Thy-1 with respect to a primary linkage map of chromosome 11q. (Abstract) Cytogenet. Cell Genet. 46: 618 only, 1987.
- [41220] 14772. Gatti, R. A.; Shaked, R.; Wei, S.; Koyama, M.; Salser,

W.; Silver, J.: DNA polymorphism in the human Thy-1 gene. Hum. Immun. 22:145–150, 1988.

[41221] 14773. Greenspan, R. J.; O'Brien, M. C.: Genetic evidence for the role of Thy-1 in neurite outgrowth in the mouse. J. Neurogenet. 5: 25–36, 1989.

[41222] 14774. Grzeschik, K.-H.; Kazazian, H. H.: Report of the committee on the genetic constitution of chromosomes 10, 11, and 12. Cytogenet. Cell Genet. 40: 179–205, 1985.

[41223] 14775. Letarte-Muirhead, M.; Barclay, A. N.; Williams, A. F.: Purification of the Thy-1 molecule, a major cell surface glycoprotein of rat thymocytes. Biochem. J. 151: 685–697, 1975.

[41224] 14776. Mansour, M. H.; Negm, H. I.; Cooper, E. L.: Thy-1 evolution. Dev. Comp. Immun. 11: 3–15, 1987.

[41225] 14777. McKenzie, J. L.; Fabre, J. W.: Human Thy-1: unusual localization and possible functional significance in lymphoid tissues. J. Immun. 126: 843–850, 1981.

[41226] 14778. Morris, R.: Thy-1 in developing nervous tissue. Dev. Neurosci. 7: 133–160, 1985.

[41227] 14779. Raff, M. C.: Surface antigenic markers for distinguishing T and B lymphocytes in mice. Transplant. Rev. 6: 52–80, 1971.

[41228] 14780. Rettig, W. J.; Dracopoli, N. C.; Chesa, P. G.; Spen-

gler, B. A.; Beresford, H. R.; Davies, P.; Biedler, J. L.; Old, L. J.: Role of human chromosome 11 in determining surface antigenic phenotype of normal and malignant cells: somatic cell genetic analysis of eight antigens, including putative human Thy-1. *J. Exp. Med.* 162: 1603–1619, 1985.

[41229] 14781. Rettig, W. J.; Dracopoli, N. C.; Silver, J.; Old, L. J.: Human THY-1: regional mapping on chromosome 11 and comparison with other chromosome 11-encoded cell surface glycoproteins. (Abstract) *Cytogenet. Cell Genet.* 40: 731 only, 1985.

[41230] 14782. Rettig, W. J.; Dracopoli, N. C.; Spengler, B. A.; Biedler, J. L.; Old, L. J.: Somatic cell genetic analysis of human cell surface antigens, including putative human Thy-1: eight distinct antigenic systems controlled by chromosome 11. (Abstract) *Cytogenet. Cell Genet.* 40: 732 only, 1985.

[41231] 14783. Seki, T.; Spurr, N.; Obata, F.; Goyert, S.; Goodfellow, P.; Silver, J.: The human Thy-1 gene: structure and chromosomal location. *Proc. Nat. Acad. Sci.* 82: 6657–6661, 1985.

[41232] 14784. Tse, A. G. D.; Barclay, A. N.; Watts, A.; Williams, A. F.: A glycosphospholipid tail at the carboxyl terminus of the Thy-1 glycoprotein of neurons and thymocytes. *Science*

230: 1003–1008, 1985.

- [41233] 14785.Chen, Z.; Friedrich, G. A.; Soriano, P.: Transcriptional enhancerfactor 1 disruption by a retroviral gene trap leads to heart defects and embryonic lethality in mice. *Genes Dev.* 8: 2293–2301, 1994.
- [41234] 14786.Aso, T.; Tsai, P.; Kawaguchi, T.; Menninger, J. C.; Kitajima, S.; Yasukochi, Y.; Ward, D. C.; Weissman, S. M.: Assignment of the humanGTF2F1 gene to chromosome 19p13.3. *Genomics* 16: 252–253, 1993.
- [41235] 14787.Aso, T.; Vasavada, H. A.; Kawaguchi, T.; Germino, F. J.; Ganguly, S.; Kitajima, S.; Weissman, S. M.; Yasukochi, Y.: Characterization of cDNA for the large subunit of the transcription initiation factorTFIIF. *Nature* 355: 461–464, 1992.
- [41236] 14788.Finkelstein, A.; Kostrub, C. F.; Li, J.; Chavez, D. P.; Wang, B.Q.; Fang, S. M.; Greenblatt, J.; Burton, Z. F.: A cDNA encoding RAP74, a general initiation factor for transcription by RNA polymerase II. *Nature* 355:464–467, 1992.
- [41237] 14789.Joliot, V.; Demma, M.; Prywes, R.: Interaction with RAP74 subunit of TFIIF is required for transcriptional activation by serum response factor. *Nature* 373: 632–635, 1995.

- [41238] 14790. Spt, M.; Burton, Z. F.; Greenblatt, J.: Structure and associated DNA-helicase activity of a general transcription initiation factor that binds to RNA polymerase II. *Nature* 341: 410–414, 1989.
- [41239] 14791. Field, S. J.; Tsai, F.-Y.; Kuo, F.; Zubiaga, A. M.; Kaelin, W.G., Jr.; Livingston, D. M.; Orkin, S. H.; Greenberg, M. E.: E2F-1 functions in mice to promote apoptosis and suppress proliferation. *Cell* 85:549–561, 1996.
- [41240] 14792. Helin, K.; Lees, J. A.; Vidal, M.; Dyson, N.; Harlow, E.; Fattaey, A.: A cDNA encoding a pRB-binding protein with properties of the transcription factor E2F. *Cell* 70: 337–350, 1992.
- [41241] 14793. Irwin, M.; Marin, M. C.; Phillips, A. C.; Seelan, R. S.; Smith, D. I.; Liu, W.; Flores, E. R.; Tsai, K. Y.; Jacks, T.; Vousden, K.H.; Kaelin, W. G., Jr.: Role for the p53 homologue p73 in E2F-1-induced apoptosis. *Nature* 407: 645–648, 2000.
- [41242] 14794. Jacks, T.; Fazeli, A.; Schmitt, E. M.; Bronson, R. T.; Goodell, M. A.; Weinberg, R. A.: Effects of an Rb mutation in the mouse. *Nature* 359:295–300, 1992.
- [41243] 14795. Lees, J. A.; Saito, M.; Valentine, M.; Look, T.; Harlow, E.; Dyson, N.; Helin, K.: The retinoblastoma protein binds to a family of E2F transcription factors. *Molec. Cell.*

Biol. 13: 7813–7825, 1993.

- [41244] 14796. Leone, G.; Sears, R.; Huang, E.; Rempel, R.; Nuckolls, F.; Park, C.-H.; Giangrande, P.; Wu, L.; Saavedra, H. I.; Field, S. J.; Thompson, M. A.; Yang, H.; Fujiwara, Y.; Greenberg, M. E.; Orkin, S.; Smith, C.; Nevins, J. R.: Myc requires distinct E2F activities to induce S phase and apoptosis. Molec. Cell 8: 105–113, 2001.
- [41245] 14797. Lissy, N. A.; Davis, P. K.; Irwin, M.; Kaelin, W. G.; Dowdy, S. F.: A common E2F-1 and p73 pathway mediates cell death induced by TCR activation. Nature 407: 642–645, 2000.
- [41246] 14798. Neuman, E.; Sellers, W. R.; McNeil, J. A.; Lawrence, J. B.; Kaelin, W. G., Jr.: Structure and partial genomic sequence of the human E2F1 gene. Gene 173: 163–169, 1996.
- [41247] 14799. Nevins, J. R.: The Rb/E2F pathway and cancer. Hum. Molec. Genet. 10: 699–703, 2001.
- [41248] 14800. Nevins, J. R.: E2F: a link between the Rb tumor suppressor protein and viral oncoproteins. Science 258: 424–429, 1992.
- [41249] 14801. Ohtani, K.; DeGregori, J.; Nevins, J. R.: Regulation of the cyclin E gene by transcription factor E2F1. Proc. Nat. Acad. Sci. 92: 12146–12150, 1995.

- [41250] 14802.Arden, K. C.; Boutin, J.-M.; Djiane, J.; Kelly, P. A.; Cavenee, W. K.: The receptors for prolactin and growth hormone are localized in the same region of human chromosome 5. *Cytogenet. Cell Genet.* 53:161–165, 1990.
- [41251] 14803.Arden, K. C.; Cavenee, W. K.; Boutin, J.-M.; Kelly, P. A.: The genes encoding the receptors for prolactin and growth hormone map to human chromosome 5. (Abstract) *Am. J. Hum. Genet.* 45 (suppl.):A129 only, 1989.
- [41252] 14804.Boutin, J.-M.; Edery, M.; Shirota, M.; Jolicoeur, C.; Lesueur, L.; Ali, S.; Gould, D.; Djiane, J.; Kelly, P. A.: Identification of a cDNA encoding a long form of prolactin receptor in human hepatoma and breast cancer cells. *Molec. Endocr.* 3: 1455–1461, 1989.
- [41253] 14805.Cunningham, B. C.; Bass, S.; Fuh, G.; Wells, J. A.: Zinc mediation of the binding of human growth hormone to the human prolactin receptor. *Science* 250:1709–1712, 1990.
- [41254] 14806.Glasow, A.; Horn, L.-C.; Taymans, S. E.; Stratakis, C. A.; Kelly, P. A.; Kohler, U.; Gillespie, J.; Vonderhaar, B. K.; Bornstein, S.R.: Mutational analysis of the PRL receptor gene in human breast tumors with differential PRL receptor protein expression. *J. Clin. Endocr. Metab.* 86: 3826–3832, 2001.

- [41255] 14807.Hu, Z.-Z.; Zhuang, L.; Meng, J.; Leondires, M.; Dufau, M. L.:The human prolactin receptor gene structure and alternative promoter utilization: the generic promoter hPIII and a novel human promoter hP(N). *J. Clin. Endocr. Metab.* 84: 1153–1156, 1999.
- [41256] 14808.Ormandy, C. J.; Camus, A.; Barra, J.; Damotte, D.; Lucas, B.; Buteau, H.; Edery, M.; Brousse, N.; Babinet, C.; Binart, N.; Kelly, P. A.: Null mutation of the prolactin receptor gene produces multiple reproductive defects in the mouse. *Genes Dev.* 11: 167–178, 1997.
- [41257] 14809.Perrot–Applanat, M.; Gualillo, O.; Pezet, A.; Vincent, V.; Edery, M.; Kelly, P. A.: Dominant negative and cooperative effects of mutant forms of prolactin receptor. *Molec. Endocr.* 11: 1020–1032, 1997.
- [41258] 14810.Tan, F.; Morris, P. W.; Skidgel, R. A.; Erdos, E. G.: Sequencing and cloning of human prolylcarboxypeptidase (angiotensinase C): similarity to both serine carboxypeptidase and prolylendopeptidase families. *J. Biol. Chem.* 268: 16631–16638, 1993.
- [41259] 14811.Watson, B., Jr.; Nowak, N. J.; Myracle, A. D.; Shows, T. B.; Warnock, D. G.: The human angiotensinase C gene (HUMPCP) maps to 11q14 within 700 kb of D11S901: a candidate gene for essential hypertension. *Genomics*

44:365–367, 1997.

- [41260] 14812.Cheng, S. Y.; Gong, Q. H.; Parkinson, C.; Robinson, E. A.; Appella,E.; Merlino, G. T.; Pastan, I.: The nucleotide sequence of a humancellular thyroid hormone–binding protein present in endoplasmic reticulum. J.Biol. Chem. 262: 11221–11227, 1987.
- [41261] 14813.Koivu, J.; Myllyla, R.; Halaakoski, T.; Pihlajaniemi, T.; Tasanen,K.; Kivirikko, K. I.: A single polypeptide acts both as the betasubunit of prolyl 4–hydroxylase and as a protein disulfide–isomerase. J.Biol. Chem. 262: 6447–6449, 1987.
- [41262] 14814.Morris, J. I.; Varandani, P. T.: Characterization of a cDNA forhuman glutathione–insulin transhydrogenase (protein–disulfide isomerase/oxidoreductase). Biochim.Biophys. Acta 949: 169–180, 1988.
- [41263] 14815.Noiva, R.; Lennarz, W. J.: Protein disulfide isomerase: a multifunctionalprotein resident in the lumen of the endoplasmic reticulum. J. Biol.Chem. 267: 3553–3556, 1992.
- [41264] 14816.Pajunen, L.; Hoyhtya, M.; Tryggvason, K.; Kivirikko, K. I.; Myllyla,R.: Species–specific antibodies in the assignment of the gene forthe beta–subunit of human prolyl 4–hydroxylase. (Abstract) Cytogenet.Cell Genet. 40: 719

only, 1985.

- [41265] 14817.Pajunen, L.; Jones, T. A.; Goddard, A.; Sheer, D.; Solomon, E.; Pihlajaniemi, T.; Kivirikko, K. I.: Regional assignment of the humangene coding for a multifunctional polypeptide (P4HB) acting as the beta-subunit of prolyl 4-hydroxylase and the enzyme protein disulfide isomerase to 17q25. Cytogenet. Cell Genet. 56: 165–168, 1991.
- [41266] 14818.Pajunen, L.; Myllyla, R.; Helaakoski, T.; Pihlajaniemi, T.; Tasanen, K.; Hoyhtya, M.; Tryggvason, K.; Solomon, E.; Kivirikko, K. I.: Assignment of the gene coding for both the beta-subunit of prolyl 4-hydroxylase and protein disulphide isomerase to human chromosome region 17q23–25. (Abstract) Cytogenet. Cell Genet. 46: 674 only, 1987.
- [41267] 14819.Pajunen, L.; Myllyla, R.; Helaakoski, T.; Pihlajaniemi, T.; Tasanen, K.; Hoyhtya, M.; Tryggvason, K.; Solomon, E.; Kivirikko, K. I.: Assignment of the gene coding for both the beta-subunit of prolyl 4-hydroxylase and the enzyme disulfide isomerase to human chromosome region 17p11–qter. Cytogenet. Cell Genet. 47: 37–41, 1988.
- [41268] 14820.Pihlajaniemi, T.; Helaakoski, T.; Tasanen, K.; Myllyla, R.; Huhtala, M.-L.; Koivu, J.; Kivirikko, K. I.: Molecular

cloning of the beta-subunit of human prolyl 4-hydroxylase: this subunit and protein disulphide isomerase are products of the same gene. *EMBO J.* 6: 643–649, 1987.

- [41269] 14821. Popescu, N. C.; Cheng, S.; Pastan, I.: Chromosomal localization of the gene for a human thyroid hormone-binding protein. *Am. J. Hum. Genet.* 42: 560–564, 1988.
- [41270] 14822. Tasanen, K.; Parkkonen, T.; Chow, L. T.; Kivirikko, K. I.; Pihlajaniemi, T.: Characterization of the human gene for a polypeptide that acts both as the beta-subunit of prolyl 4-hydroxylase and as protein disulfide isomerase. *J. Biol. Chem.* 263: 16218–16224, 1988.
- [41271] 14823. Kelly, A.; Powis, S. H.; Glynn, R.; Radley, E.; Beck, S.; Trowsdale, J.: Second proteasome-related gene in the human MHC class II region. *Nature* 353: 667–668, 1991.
- [41272] 14824. Martinez, C. K.; Monaco, J. J.: Homology of proteasome subunits to a major histocompatibility complex-linked LMP gene. *Nature* 353: 664–667, 1991.
- [41273] 14825. Petes, T. D.: Meiotic recombination hot spots and cold spots. *Nature Rev. Genet.* 2: 360–369, 2001.
- [41274] 14826. Van Kaer, L.; Ashton-Rickardt, P. G.; Eichelberger, M.; Gaczynska, M.; Nagashima, K.; Rock, K. L.; Goldberg, A. L.; Doherty, P. C.; Tonegawa, S.: Altered peptidase and

viral-specific T cell response in LMP2mutant mice. *Immunity* 1: 533–541, 1994.

[41275] 14827.Zhou, P.; Zanelli, E.; Smart, M.; David, C.: Genomic organizationand tissue expression of mouse proteasome gene Lmp-2. *Genomics* 16:664–668, 1993.

[41276] 14828.Bodmer, J. G.; Marsh, S. G. E.; Albert, E. D.; Bodmer, W. F.; Dupont,B.; Erlich, H. A.; Mach, B.; Mayr, W. R.; Parham, P.; Sasazuki, T.;Schreuder, G. M. T.; Strominger, J. L.; Svejgaard, A.; Terasaki, P.I.: Nomenclature for factors of the HLA system, 1991. *Tissue Antigens* 39:161–173, 1992.

[41277] 14829.Deng, G. Y.; Muir, A.; Maclaren, N. K.; She, J.-X.: Associationof LMP2 and LMP7 genes within the major histocompatibility complexwith insulin-dependent diabetes mellitus: population and family studies. *Am.J. Hum. Genet.* 56: 528–534, 1995.

[41278] 14830.Fehling, H. J.; Swat, W.; Laplace, C.; Kuhn, R.; Rajewsky, K.;Muller, U.; von Boehmer, H.: MHC class I expression in mice lackingthe proteasome subunit LMP-7. *Science* 265: 1234–1237, 1994.

[41279] 14831.Doolittle, R. F.; Hunkapiller, M. W.; Hood, L. E.; Devare, S.G.; Robbins, K. C.; Aaronson, S. A.; Antoniades, H. N.: Simian sarcomavirus onc gene, v-sis, is derived from

the gene (or genes) encoding a platelet-derived growth factor. *Science* 221: 275–277, 1983.

- [41280] 14832. Frolova, L. Y.; Sudomoina, M. A.; Grigorieva, A. Y.; Zinovieva, O. L.; Kisselev, L. L.: Cloning and nucleotide sequence of the structural gene encoding for human tryptophanyl-tRNA synthetase. *Gene* 109:291–296, 1991.
- [41281] 14833. Vassart, G.: Personal Communication. Brussels, Belgium 1/15/1992.
- [41282] 14834. Wenger, G. D.: Personal Communication. Columbus, Ohio 8/3/1993.
- [41283] 14835. Gardner, T. L.; Elston, D. M.; Wotowic, P. J.: A familial dermatofibrosarcoma protuberans. *J. Am. Acad. Derm.* 39: 504–505, 1998.
- [41284] 14836. Groffen, J.; Heisterkamp, N.; Stephenson, J. R.; Geurts van Kessel, A.; de Klein, A.; Grosveld, G.; Bootsma, D.: c-sis is translocated from chromosome 22 to chromosome 9 in chronic myelocytic leukemia. *J. Exp. Med.* 158: 9–15, 1983.
- [41285] 14837. Hermansson, M.; Nister, M.; Betsholtz, C.; Heldin, C.-H.; Westermarck, B.; Funai, K.: Endothelial cell hyperplasia in human glioblastoma: coexpression of mRNA for platelet-derived growth factor (PDGF) B chain and PDGF receptor suggests autocrine growth stimulation. *Proc.*

Nat.Acad. Sci. 85: 7748–7752, 1988.

- [41286] 14838. Josephs, S. F.; Dalla-Favera, R.; Gelmann, E. P.; Gallo, R. C.; Wong-Staal, F.: 5-prime viral and human cellular sequences corresponding to the transforming gene of simian sarcoma virus. *Science* 219: 503–505, 1983.
- [41287] 14839. Josephs, S. F.; Guo, C.; Ratner, L.; Wong-Staal, F.: Human proto-oncogene nucleotide sequences corresponding to the transforming region of simian sarcoma virus. *Science* 223: 487–491, 1984.
- [41288] 14840. Josephs, S. F.; Ratner, L.; Clarke, M. F.; Westin, E. H.; Reitz, M. S.; Wong-Staal, F.: Transforming potential of human c-sis nucleotide sequences encoding platelet-derived growth factor. *Science* 225: 636–639, 1984.
- [41289] 14841. Kelly, J. D.; Raines, E. W.; Ross, R.; Murray, M. J.: The B chain of PDGF alone is sufficient for mitogenesis. *EMBO J.* 4: 3399–3405, 1985.
- [41290] 14842. Kiuru-Kuhlefelt, S.; El-Rifai, W.; Fanburg-Smith, J.; Kere, J.; Miettinen, M.; Knuutila, S.: Concomitant DNA copy number amplification at 17q and 22q in dermatofibrosarcoma protuberans. *Cytogenet. Cell Genet.* 92: 192–195, 2001.
- [41291] 14843. Kozak, C. A.; Sears, J. F.; Hoggan, M. D.: Genetic mapping of the mouse proto-oncogene c-sis to chromo-

some 15. Science 221: 867–869,1983.

- [41292] 14844.Lindahl, P.; Johansson, B. R.; Leveen, P.; Betsholtz, C.: Pericyteloss and microaneurysm formation in PDGF–B–deficient mice. Science 277:242–245, 1997.
- [41293] 14845.Owen, A. J.; Pantazis, P.; Antoniades, H. N.: Simian sarcomavirus–transformed cells secrete a mitogen identical to platelet–derivedgrowth factor. Science 225: 54–56, 1984.
- [41294] 14846.Rao, C. D.; Igarashi, H.; Chiu, I.–M.; Robbins, K. C.; Aaronson,S. A.: Structure and sequence of the human c–sis/platelet–derivedgrowth factor 2 (SIS/PDGF2) transcriptional unit. Proc. Nat. Acad.Sci. 83: 2392–2396, 1986.
- [41295] 14847.Robbins, K. C.; Antoniades, H. N.; Devare, S. G.; Hunkapiller,M. W.; Aaronson, S. A.: Structural and immunological similaritiesbetween simian sarcoma virus gene product(s) and human platelet–derivedgrowth factor. Nature 305: 605–608, 1983.
- [41296] 14848.Robbins, K. C.; Devare, S. G.; Reddy, E. P.; Aaronson, S. A.:In vivo identification of the transforming gene product of simiansarcoma virus. Science 218: 1131–1133, 1982.
- [41297] 14849.Simon, M.–P.; Navarro, M.; Roux, D.; Pouyssegur, J.: Structuraland functional analysis of a chimeric protein

COL1A1–PDGFB generated by the translocation t(17;22)(q22;q13.1) in dermatofibrosarcoma protuberans(DP). *Oncogene* 20: 2965–2975, 2001.

- [41298] 14850. Smidt, M.; Kirsch, I.; Ratner, L.: Deletion of Alu sequences in the fifth c-sis intron in individuals with meningiomas. *J. Clin. Invest.* 86: 1151–1157, 1990.
- [41299] 14851. Turc-Carel, C.; Philip, I.; Berger, M. P.; Philip, T.; Lenoir, G. M.: Chromosomal translocations in Ewing's sarcoma. (Letter) *New Eng. J. Med.* 309: 497–498, 1983.
- [41300] 14852. Shimizu, A.; O'Brien, K. P.; Sjoblom, T.; Pietras, K.; Buchdunger, E.; Collins, V. P.; Heldin, C.-H.; Dumanski, J. P.; Ostman, A.: The dermatofibrosarcoma protuberans-associated collagen type I-alpha-1/platelet-derived growth factor (PDGF) beta-chain fusion gene generates a transforming protein that is processed to functional PDGF-BB. *Cancer Res.* 59:3719–3723, 1999.
- [41301] 14853. Simon, M.-P.; Pedeutour, F.; Sirvent, N.; Grosgeorge, J.; Minoletti, F.; Coindre, J.-M.; Terrier-Lacombe, M.-J.; Mandahl, N.; Craver, R.D.; Blin, N.; Sozzi, G.; Turc-Carel, C.; O'Brien, K. P.; Kedra, D.; Fransson, I.; Guilbaud, C.; Dumanski, J. P.: Deregulation of the platelet-derived growth factor B-chain gene via fusion with collagen gene COL1A1 in dermatofibrosarcoma protuberans and giant-

cell fibroblastoma. *NatureGenet.* 15: 95–98, 1997.

- [41302] 14854. Waterfield, M. D.; Scrace, G. T.; Whittle, N.; Stroobant, P.; Johnsson, A.; Wasteson, A.; Westermark, B.; Heldin, C.-H.; Huang, J. S.; Deuel, T. F.: Platelet-derived growth factor is structurally related to the putative transforming protein p28(sis) of simian sarcomavirus. *Nature* 304: 35–39, 1983.
- [41303] 14855. Graphodatsky, A.; Frolova, L.; Biltueva, L.; Eremina, V.; Lushnikova, T.; Sudomoina, M.; Zinovieva, O.; Kisselev, L.: Localization of the tryptophanyl tRNA synthetase gene (WARS) on human and bovine chromosomes by in situ hybridization. *Mammalian Genome* 4: 183–184, 1993.
- [41304] 14856. Jensen, L. L.; Nielsen, M. M.; Justesen, J.; Hansen, L. L.: Assignment of human NADH dehydrogenase (ubiquinone) 1 beta subcomplex 3 (NDUFB3) and of its four pseudogenes to human chromosomes 2q31.3, 1p13.3–p13.1, 9q32–q34.1, 14q22.3–q23.1 and 14q32.2 by radiation hybrid mapping. *Cytogenet. Cell Genet.* 93: 147–150, 2001.
- [41305] 14857. Otani, A.; Slike, B. M.; Dorrell, M. I.; Hood, J.; Kinder, K.; Ewalt, K. L.; Cheresh, D.; Schimmel, P.; Friedlander, M.: A fragment of human TrpRS as a potent antagonist of ocular angiogenesis. *Proc. Nat. Acad. Sci.* 99:

178–183, 2002.

- [41306] 14858. Shimizu, N.; Kucherlapati, R. S.; Ruddle, F. H.: Assignment of a human gene for tryptophanyl-tRNA synthetase to chromosome 14 using human-mouse somatic cell hybrids. *Somat. Cell Genet.* 2: 345–357, 1976.
- [41307] 14859. Tolstrup, A. B.; Bejder, A.; Fleckner, J.; Justesen, J.: Transcriptional regulation of the interferon-gamma-inducible tryptophanyl-tRNA synthetase includes alternative splicing. *J. Biol. Chem.* 270: 397–403, 1995.
- [41308] 14860. Turpaev, K. T.; Zakhariev, V. M.; Sokolova, I. V.; Narovlyansky, A. N.; Amchenkova, A. M.; Justesen, J.; Frolova, L. Y.: Alternative processing of the tryptophanyl-tRNA synthetase mRNA from interferon-treated human cells. *Europ. J. Biochem.* 240: 732–737, 1996.
- [41309] 14861. Wakasugi, K.; Slike, B. M.; Hood, J.; Otani, A.; Ewalt, K. L.; Friedlander, M.; Cheresch, D. A.; Schimmel, P.: A human aminoacyl-tRNA synthetase as a regulator of angiogenesis. *Proc. Nat. Acad. Sci.* 99: 173–177, 2002.
- [41310] 14862. Miller, J. S.; Moxley, G.; Schwartz, L. B.: Cloning and characterization of a second complementary DNA for human tryptase. *J. Clin. Invest.* 86: 864–870, 1990.
- [41311] 14863. Miller, J. S.; Westin, E. H.; Schwartz, L. B.: Cloning and characterization of complementary DNA for human

tryptase. J. Clin. Invest. 84: 1188–1195,1989.

- [41312] 14864.Vanderslice, P.; Ballinger, S. M.; Tam, E. K.; Goldstein, S. M.; Craik, C. S.; Caughey, G. H.: Human mast cell tryptase: multiple cDNAs and genes reveal a multigene serine protease family. Proc.Nat. Acad. Sci. 87: 3811–3815, 1990.
- [41313] 14865.Mach, B.; Steimle, V.; Reith, W.: MHC class II-deficient combined immunodeficiency: a disease of gene regulation. Immun. Rev. 138:207–221, 1994.
- [41314] 14866.Babcock, M.; de Silva, D.; Oaks, R.; Davis-Kaplan, S.; Jiralerspong, S.; Montermini, L.; Pandolfo, M.; Kaplan, J.: Regulation of mitochondrial iron accumulation by Yfh1p, a putative homolog of frataxin. Science 276:1709–1712, 1997.
- [41315] 14867.Bidichandani, S. I.; Ashizawa, T.; Patel, P. I.: The GAA triplet-repeat expansion in Friedreich ataxia interferes with transcription and maybe associated with an unusual DNA structure. Am. J. Hum. Genet. 62:111–121, 1998.
- [41316] 14868.Bidichandani, S. I.; Ashizawa, T.; Patel, P. I.: Atypical Friedreich ataxia caused by compound heterozygosity for a novel missense mutation and the GAA triplet-repeat expansion. (Letter) Am. J. Hum. Genet. 60:1251–1256,

1997.

- [41317] 14869.Cavadini, P.; Gellera, C.; Patel, P. I.; Isaya, G.: Human frataxin maintains mitochondrial iron homeostasis in *Saccharomyces cerevisiae*. *Hum.Molec. Genet.* 9: 2523–2530, 2000.
- [41318] 14870.Sukhatme, V. P.; Vollmer, A. C.; Erikson, J.; Isobe, M.; Croce, C.; Parnes, J. R.: Gene for the human T cell differentiation antigen Leu-2/T8 is closely linked to the kappa light chain locus on chromosome 2. *J. Exp. Med.* 161: 429–434, 1985.
- [41319] 14871.Traver, D.; Akashi, K.; Manz, M.; Merad, M.; Miyamoto, T.; Engleman, E. G.; Weissman, I. L.: Development of CD8-alpha-positive dendritic cells from a common myeloid progenitor. *Science* 290: 2152–2154, 2000.
- [41320] 14872.Weichhold, G. M.; Huber, C.; Parnes, J. R.; Zachau, H. G.: The CD8-alpha locus is located on the telomere side of the immunoglobulin-kappa locus at a distance of 2 Mb. *Genomics* 16: 512–514, 1993.
- [41321] 14873.Balk, S. P.; Burke, S.; Polischuk, J. E.; Frantz, M. E.; Yang, L.; Porcelli, S.; Colgan, S. P.; Blumberg, R. S.: Beta-2-microglobulin-independent MHC class Ib molecule expressed by human intestinal epithelium. *Science* 265:259–262, 1994.

- [41322] 14874.Benlagha, K.; Kyin, T.; Beavis, A.; Teyton, L.; Bendelac, A.:A thymic precursor to NK T cell lineage. Science 296: 553–555, 2002.
- [41323] 14875.Bradbury, A.; Milstein, C.; Kozak, C. A.: Chromosomal localization of Cd1d genes in the mouse. Somat. Cell Molec. Genet. 17: 93–96,1991.
- [41324] 14876.Honey, K.; Benlagha, K.; Beers, C.; Forbush, K.; Teyton, L.; Kleijmeer, M. J.; Rudensky, A. Y.; Bendelac, A.: Thymocyte expression of cathepsinL is essential for NKT cell development. Nature Immun. 7Oct, 2002.Note: Advance Electronic Publication.
- [41325] 14877.MacDonald, H. R.: T before NK. Science 296: 481–482, 2002.
- [41326] 14878.Pellicci, D. G.; Hammond, K. J. L.; Uldrich, A. P.; Baxter, A.G.; Smyth, M. J.; Godfrey, D. I.: A natural killer T (NKT) cell developmental pathway involving a thymus-dependent NK1.1(–)CD4(+) CD1d-dependent precursor stage. J. Exp. Med. 195: 835–844, 2002.
- [41327] 14879.Prigozy, T. I.; Naidenko, O.; Qasba, P.; Elewaut, D.; Brossay, L.; Khurana, A.; Natori, T.; Koezuka, Y.; Kulkarni, A.; Kronenberg, M.: Glycolipid antigen processing for presentation by CD1d molecules. Science 291:664–667, 2001.

- [41328] 14880. Shi, F.-D.; Flodstrom, M.; Balasa, B.; Kim, S. H.; Van Gunst, K.; Strominger, J. L.; Wilson, S. B.; Sarvetnick, N.: Germ line deletion of the CD1 locus exacerbates diabetes in the NOD mouse. *Proc. Nat. Acad. Sci.* 98: 6777–6782, 2001.
- [41329] 14881. Hashimoto, K.; Hirai, M.; Kurosawa, Y.: A gene outside the human MHC related to classical HLA class I genes. *Science* 269: 693–695, 1995.
- [41330] 14882. Shilo, B. Z.; Weinberg, R. A.: DNA sequences homologous to vertebrate oncogenes are conserved in *Drosophila melanogaster*. *Proc. Nat. Acad. Sci.* 78: 6789–6792, 1981.
- [41331] 14883. Takahashi, E.; Hori, T.; O'Connell, P.; Leppert, M.; White, R.: Mapping of the MYC gene to band 8q24.12–q24.13 by R-banding and distal to fra(8)(q24.11), FRA8E, by fluorescence in situ hybridization. *Cytogenet. Cell Genet.* 57: 109–111, 1991.
- [41332] 14884. Taub, R.; Kirsch, I.; Morton, C.; Lenoir, G.; Swan, D.; Tronick, S.; Aaronson, S.; Leder, P.: Translocation of the c-myc gene into the immunoglobulin heavy chain locus in human Burkitt lymphoma and murine plasmacytoma cells. *Proc. Nat. Acad. Sci.* 79: 7837–7841, 1982.
- [41333] 14885. Trumpp, A.; Refaeli, Y.; Oskarsson, T.; Gasser, S.;

Murphy, M.; Martin, G. R.; Bishop, J. M.: c-Myc regulates mammalian body size by controlling cell number but not cell size. *Nature* 414: 768–773, 2001.

[41334] 14886. Vafa, O.; Wade, M.; Kern, S.; Beeche, M.; Pandita, T. K.; Hampton, G. M.; Wahl, G. M.: c-Myc can induce DNA damage, increase reactive oxygen species, and mitigate p53 function: a mechanism for oncogene-induced genetic instability. *Molec. Cell* 9: 1031–1044, 2002.

[41335] 14887. Watt, R.; Nishikura, K.; Sorrentino, J.; ar-Rushdi, A.; Croce, C. M.; Rovera, G.: The structure and nucleotide sequence of the 5'-prime end of the human c-myc oncogene. *Proc. Nat. Acad. Sci.* 80: 6307–6311, 1983.

[41336] 14888. Watt, R.; Stanton, L. W.; Marcu, K. B.; Gallo, R. C.; Croce, C. M.; Rovera, G.: Nucleotide sequence of cloned cDNA of human c-myc oncogene. *Nature* 303: 725–728, 1983.

[41337] 14889. Azarnia, R.; Reddy, S.; Kmiecik, T. E.; Shalloway, D.; Loewenstein, W. R.: The cellular src gene product regulates junctional cell-to-cell communication. *Science* 239: 398–401, 1988.

[41338] 14890. Czernilofsky, A. P.; Levinson, A. D.; Varmus, H. E.; Bishop, J. M.; Tischler, E.; Goodman, H.: Correction to the nucleotide sequence of the src gene of Rous sarcoma

virus. *Nature* 301: 736–738, 1983.

- [41339] 14891. Gibbs, C. P.; Tanaka, A.; Anderson, S. K.; Radul, J.; Baar, J.; Ridgway, A.; Kung, H.-J.; Fujita, D. J.: Isolation and structural mapping of a human c-src gene homologous to the transforming gene (v-src) of Rous sarcoma virus. *J. Virol.* 53: 19–24, 1985.
- [41340] 14892. Irby, R. B.; Mao, W.; Coppola, D.; Kang, J.; Loubeau, J. M.; Trudeau, W.; Karl, R.; Fujita, D. J.; Jove, R.; Yeatman, T. J.: Activating SRC mutation in a subset of advanced human colon cancers. *Nature Genet.* 21: 187–190, 1999.
- [41341] 14893. Le Beau, M. M.; Westbrook, C. A.; Diaz, M. O.; Rowley, J. D.: c-src is consistently conserved in the chromosomal deletion (20q) observed in myeloid disorders. *Proc. Nat. Acad. Sci.* 82: 6692–6696, 1985.
- [41342] 14894. Lowe, C.; Yoneda, T.; Boyce, B. F.; Chen, H.; Mundy, G. R.; Soriano, P.: Osteopetrosis in Src-deficient mice is due to an autonomous defect of osteoclasts. *Proc. Nat. Acad. Sci.* 90: 4485–4489, 1993.
- [41343] 14895. Morris, C. M.; Honeybone, L. M.; Hollings, P. E.; Fitzgerald, P. H.: Localization of the SRC oncogene to chromosome band 20q11.2 and loss of this gene with deletion (20q) in two leukemic patients. *Blood* 74: 1768–1773, 1989.

- [41344] 14896.Sakaguchi, A. Y.; Mohandas, T.; Naylor, S. L.: A human c-src gene resides on the proximal long arm of chromosome 20 (cen-q13.1). *CancerGenet. Cytogenet.* 18: 123-129, 1985.
- [41345] 14897.Sakaguchi, A. Y.; Naylor, S. L.; Weinberg, R. A.; Shows, T. B.: Organization of human proto-oncogenes. (Abstract) *Am. J. Hum. Genet.* 34:175A, 1982.
- [41346] 14898.Sakaguchi, A. Y.; Zabel, B. U.; Grzeschik, K. H.; Law, M. L.; Naylor, S. L.: Human proto-oncogene assignments. (Abstract) *Cytogenet. Cell Genet.* 37: 572-573, 1984.
- [41347] 14899.Wong, B. R.; Besser, D.; Kim, N.; Arron, J. R.; Volodyskaia, M.; Hanafusa, H.; Choi, Y.: TRANCE, a TNF family member, activates Akt/PKB through a signaling complex involving TRAF6 and c-Src. *Molec. Cell* 4: 1041-1049, 1999.
- [41348] 14900.Mangelsdorf, D. J.; Borgmeyer, U.; Heyman, R. A.; Zhou, J. Y.; Ong, E. S.; Oro, A. E.; Kakizuka, A.; Evans, R. M.: Characterization of three RXR genes that mediate the action of 9-cis retinoic acid. *Genes Dev.* 6: 329-344, 1992.
- [41349] 14901.Attard-Montalto, S.; Evans, N.; Sherwood, R. A.: Carotenemia with low vitamin A levels and low retinol-

- binding protein. *J. Inherit. Metab. Dis.* 15: 929–930, 1992.
- [41350] 14902.Chainani, M.; Sampsell, B.; Elliott, R. W.: Localization of the gene for plasma retinol binding protein to the distal half of mouse chromosome 19. *Genomics* 9: 376–379, 1991.
- [41351] 14903.Matsuo, T.: Familial retinol-binding-protein deficiency? (Letter) *Lancet* II:910 only, 1987.
- [41352] 14904.Matsuo, T.; Matsuo, N.; Shiraga, F.; Koide, N.: Familial retinol-binding-protein deficiency. (Letter) *Lancet* II: 402–403, 1987.
- [41353] 14905.Morgan, F. F.; Canfield, R. E.; Goodman, D. S.: The partial structure of human plasma prealbumin and retinol-binding protein. *Biochim. Biophys. Acta* 236: 798–801, 1971.
- [41354] 14906.Rask, L.; Anundi, H.; Fohlman, J.; Peterson, P. A.: The complete amino acid sequence of human serum retinol-binding protein. *Uppsala J. Med. Sci.* 92: 115–146, 1987.
- [41355] 14907.Rask, L.; Vahlquist, A.; Peterson, P. A.: Studies on two physiological forms of the human retinol-binding protein differing in vitamin A and arginine content. *J. Biol. Chem.* 246: 6638–6646, 1971.
- [41356] 14908.Rocchi, M.; Covone, A.; Romeo, G.; Faraonio, R.;

Colantuoni, V.: Regional mapping of RBP4 to 10q23–q24 and RBP1 to 3q21–q22 in man. *Somat.Cell Molec. Genet.* 15: 185–190, 1989.

[41357] 14909.Seeliger, M. W.; Biesalski, H. K.; Wissinger, B.; Gollnick, H.;Gielen, S.; Frank, J.; Beck, S.; Zrenner, E.: Phenotype in retinoldeficiency due to a hereditary defect in retinol binding protein synthesis. *Invest.Ophthal. Vis. Sci.* 40: 3–11, 1999.

[41358] 14910.Thuluvath, P. J.: Familial retinol–binding–protein deficiency?(Letter) *Lancet* II: 910 only, 1987.

[41359] 14911.Colantuoni, V.; Cortese, R.; Nilsson, M.; Lundvall, J.; Bavik,C. O.; Eriksson, U.; Peterson, P. A.; Sundelin, J.: Cloning and sequencingof a full length cDNA corresponding to human cellular retinol–bindingprotein. *Biochem. Biophys. Res. Commun.* 130: 431–439, 1985.

[41360] 14912.Colantuoni, V.; Rocchi, M.; Roncuzzi, L.; Romeo, G.: Mapping ofhuman cellular retinol–binding protein to chromosome 3. *Cytogenet.Cell Genet.* 43: 221–222, 1986.

[41361] 14913.De Baere, E.; Speleman, F.; Van Roy, N.; De Paepe, A.; Messiaen,L.: Assignment of the cellular retinol–binding protein 1 gene (RBP1)and of the coatomer beta–prime subunit gene (COPB2) to human chromosomeband 3q23 by in situ hybridization. *Cytogenet. Cell Genet.* 82:

226–227,1998.

- [41362] 14914.Nilsson, M. H. L.; Spurr, N. K.; Lundvall, J.; Rask, L.; Peterson,P. A.: Human cellular retinol–binding protein gene organization andchromosomal location. *Europ. J. Biochem.* 173: 35–44, 1988.
- [41363] 14915.Carson, N. L.; Simpson, N. E.: A physical map of 13 markers onchromosome 10 from dosage studies on abnormal cell lines. (Abstract) *Cytogenet.Cell Genet.* 51: 974–975, 1989.
- [41364] 14916.Chin, K. S.; Mathew, C. G. P.; Fong, S. L.; Bridges, C. D. B.;Ponder, B. A. J.: Styl RFLP recognised by a human IRBP cDNA localisedto chromosome 10. *Nucleic Acids Res.* 16: 1645 only, 1988.
- [41365] 14917.Danciger, M.; Kozak, C. A.; Nickerson, J.; Redmond, T. M.; Farber,D. B.: Localization of the gene for interphotoreceptor retinoid–bindingprotein to mouse chromosome 14 near Np–1. *Genomics* 8: 727–731, 1990.
- [41366] 14918.Farrer, L. A.; Castiglione, C. M.; Kidd, J. R.; Myers, S.; Carson,N.; Simpson, N. E.; Kidd, K. K.: A linkage group of five DNA markerson human chromosome 10. *Genomics* 3: 72–77, 1988.
- [41367] 14919.Fong, S.–L.; Liou, G. I.; Landers, R. A.; Alvarez, R. A.; Gonzalez–Fernandez,F.; Glazebrook, P. A.; Lam, D. M.

K.; Bridges, C. D. B.: Characterization, localization, and biosynthesis of an interstitial retinol-binding glycoprotein in the human eye. *J. Neurochem.* 42: 1667–1676, 1984.

[41368] 14920. Liou, G. I.; Fong, S.-L.; Beattie, W. G.; Cook, R. G.; Leone, J.; Landers, R. A.; Alvarez, R. A.; Wang, C.; Li, Y.; Bridges, C. D. B.: Bovine interstitial retinol-binding protein (IRBP)—isolation and sequence analysis of cDNA clones, characterization and in vitro translation of mRNA. *Vision Res.* 26: 1645–1653, 1986.

[41369] 14921. Liou, G. I.; Fong, S.-L.; Gosden, J.; van Tuinen, P.; Ledbetter, D. H.; Christie, S.; Rout, D.; Bhattacharya, S.; Cook, R. G.; Li, Y.; Wang, C.; Bridges, C. D. B.: Human interstitial retinol-binding protein (IRBP): cloning, partial sequence, and chromosomal localization. *Somat. Cell Molec. Genet.* 13: 315–323, 1987.

[41370] 14922. Liou, G. I.; Li, Y.; Wang, C.; Fong, S.-L.; Bhattacharya, F. S.; Bridges, C. D. B.: Bgl II RFLP recognized by a human IRBP cDNA localized to chromosome 10. *Nucleic Acids Res.* 15: 3196 only, 1987.

[41371] 14923. McGuire, W.; Hill, A. V. S.; Allsopp, C. E. M.; Greenwood, B. M.; Kwiatkowski, D.: Variation in the TNF- α promoter region associated with susceptibility to cerebral malaria. *Nature* 371: 508–511, 1994.

- [41372] 14924. Benbrook, D.; Pfahl, M.: A novel thyroid hormone receptor encoded by a cDNA clone from a human testis library. *Science* 238: 788–791, 1987.
- [41373] 14925. Bernal, J.; Refetoff, S.; DeGroot, L. J.: Abnormalities of triiodothyronine binding to lymphocyte and fibroblast nuclei from a patient with peripheral tissue resistance to thyroid hormone action. *J. Clin. Endocr. Metab.* 47:1266–1272, 1978.
- [41374] 14926. Dayton, A. I.; Selden, J. R.; Laws, G.; Dorney, D. J.; Finan, J.; Tripputi, P.; Emanuel, B. S.; Rovera, G.; Nowell, P. C.; Croce, C. M.: A human c-erbA oncogene homologue is closely proximal to the chromosome 17 breakpoint in acute promyelocytic leukemia. *Proc. Nat. Acad. Sci.* 81: 4495–4499, 1984.
- [41375] 14927. Debuire, B.; Henry, C.; Benaissa, M.; Biserte, G.; Claverie, J. M.; Saule, S.; Martin, P.; Stehelin, D.: Sequencing the erbA gene of avian erythroblastosis virus reveals a new type of oncogene. *Science* 224:1456–1459, 1984.
- [41376] 14928. Ferro, M. T.; San Roman, C.: Constitutional t(15;17). *Cancer Genet. Cytogenet.* 4: 89–91, 1981.
- [41377] 14929. Gullberg, H.; Rudling, M.; Forrest, D.; Angelin, B.; Vennstrom, B.: Thyroid hormone receptor beta-deficient mice show complete loss of the normal cholesterol

7- α -hydroxylase (CYP7A) response to thyroid hormone but display enhanced resistance to dietary cholesterol. *Molec. Endocr.* 14: 1739–1749, 2000.

- [41378] 14930. Ichikawa, K.; Hughes, I. A.; Horwitz, A. L.; DeGroot, L. J.: Characterization of nuclear thyroid hormone receptors of cultured skin fibroblasts from patients with resistance to thyroid hormone. *Metabolism* 36:392–399, 1987.
- [41379] 14931. Iskaros, J.; Pickard, M.; Evans, I.; Sinha, A.; Hardiman, P.; Ekins, R.: Thyroid hormone receptor gene expression in first trimester human fetal brain. *J. Clin. Endocr. Metab.* 85: 2620–2623, 2000.
- [41380] 14932. Jansson, M.; Philipson, L.; Vennstrom, B.: Isolation and characterization of multiple human genes homologous to the oncogenes of avian erythroblastosis virus. *EMBO J.* 2: 561–565, 1983.
- [41381] 14933. Jhanwar, S. C.; Chaganti, R. S. K.; Croce, C. M.: Germ-line chromosomal localization of human c-erb-A oncogene. *Somat. Cell Molec. Genet.* 11:99–102, 1985.
- [41382] 14934. Kaneshige, M.; Suzuki, H.; Kaneshige, K.; Cheng, J.; Wimbrow, H.; Barlow, C.; Willingham, M. C.; Cheng, S.: A targeted dominant negative mutation of the thyroid hormone α -1 receptor causes increased mortality, infertility, and dwarfism in mice. *Proc. Nat. Acad. Sci.*

98:15095–15100, 2001.

- [41383] 14935.Laudet, V.; Begue, A.; Henry–Duthoit, C.; Joubel, A.; Martin,P.; Stehelin, D.; Saule, S.: Genomic organization of the human thyroidhormone receptor alpha (c–erbA–1) gene. *Nucleic Acids Res.* 19: 1105–1112,1991.
- [41384] 14936.Le Beau, M. M.; Westbrook, C. A.; Diaz, M. O.; Rowley, J. D.;Oren, M.: Translocation of the p53 gene in t(15;17) in acute promyelocyticleukaemia. *Nature* 316: 826–828, 1985.
- [41385] 14937.Mathieu–Mahul, D.; Xu, D. Q.; Saule, S.; Lidereau, R.; Galibert,F.; Berger, R.; Mauchauffe, M.; Larsen, C. J.: An EcoRI restrictionfragment length polymorphism (RFLP) in the human c–erb A locus. *Hum.Genet.* 71: 41–44, 1985.
- [41386] 14938.McCabe, C. J.; Gittoes, N. J.; Sheppard, M. C.; Franklyn, J. A.: Thyroid receptor alpha–1 and alpha–2 mutations in nonfunctioningpituitary tumors. *J. Clin. Endocr. Metab.* 84: 649–653, 1999.
- [41387] 14939.Menezes–Ferreira, M. M.; Eil, C.; Wortsman, J.; Weintraub, B.D.: Decreased nuclear uptake of [125–I]triiodo–L–thyronine in fibroblastsfrom patients with peripheral thyroid hormone resistance. *J. Clin.Endocr. Metab.* 59: 1081–1087, 1984.
- [41388] 14940.Mitelman, F.; Manolov, G.; Manolova, Y.; Billstrom,

R.; Heim, S.; Kristoffersson, U.; Mandahl, N.; Ferro, M. T.; San Roman, C.: High resolution chromosome analysis of constitutional and acquired t(15;17) maps c-erbA to sub-band 17q11.2. *Cancer Genet. Cytogenet.* 22:95–98, 1986.

[41389] 14941. Miyajima, N.; Horiuchi, R.; Shibuya, Y.; Fukushige, S.; Matsubara, K.; Toyoshima, K.; Yamamoto, T.: Two erbA homologs encoding proteins with different T(3) binding capacities are transcribed from opposite DNA strands of the same genetic locus. *Cell* 57: 31–39, 1989.

[41390] 14942. Nagaya, T.; Nomura, Y.; Fujieda, M.; Seo, H.: Heterodimerization preferences of thyroid hormone receptor alpha isoforms. *Biochem. Biophys. Res. Commun.* 226: 426–430, 1996.

[41391] 14943. Nakai, A.; Seino, S.; Sakurai, A.; Szilak, I.; Bell, G. I.; DeGroot, L. J.: Characterization of a thyroid hormone receptor expressed in human kidney and other tissues. *Proc. Nat. Acad. Sci.* 85: 2781–2785, 1988.

[41392] 14944. Ng, L.; Rusch, A.; Amma, L. L.; Nordstrom, K.; Erway, L. C.; Vennstrom, B.; Forrest, D.: Suppression of the deafness and thyroid dysfunction in Thrb-null mice by an independent mutation in the Thra thyroid hormone receptor gene. *Hum. Molec. Genet.* 10: 2701–2708, 2001.

[41393] 14945. Puzianowska-Kuznicka, M.; Krystyniak, A.; Madej,

A.; Cheng, S.-Y.;Nauman, J.: Functionally impaired TR mutants are present in thyroidpapillary cancer. J. Clin. Endocr. Metab. 87: 1120–1128, 2002.

- [41394] 14946.Refetoff, S.; DeGroot, L. J.; Benard, B.; DeWind, L. T.: Studiesof a sibship with apparent hereditary resistance to the intracellularaction of thyroid hormone. Metabolism 21: 723–756, 1972.
- [41395] 14947.Rider, S. H.; Bailey, C. J.; Voss, R.; Sheer, D.; Hiorns, L. R.;Solomon, E.: RFLP for the human erb-A1 gene. Nucleic Acids Res. 15:863 only, 1987.
- [41396] 14948.Sakurai, A.; Nakai, A.; DeGroot, L. J.: Expression of three formsof thyroid hormone receptor in human tissues. Molec. Endocr. 3:392–399, 1989.
- [41397] 14949.Cerretti, D. P.; Lyman, S. D.; Kozlosky, C. J.; Copeland, N. G.;Gilbert, D. J.; Jenkins, N. A.; Valentine, V.; Kirstein, M. N.; Shapiro,D. N.; Morris, S. W.: The genes encoding the Eph-related receptortyrosine kinase ligands LERK–1 (EPLG1, Epl1), LERK–3 (EPLG3, Epl3),and LERK–4 (EPLG4, Epl4) are clustered on human chromosome 1 and mousechromosome 3. Genomics 33: 277–282, 1996.
- [41398] 14950.Holzman, L. B.; Marks, R. M.; Dixit, V. M.: A novel immediate–earlyresponse gene of endothelium is induced by cytokines and encodes asecreted protein. Molec. Cell.

Biol. 10: 5830–5838, 1990.

- [41399] 14951. Pandey, A.; Lindberg, R. A.; Dixit, V. M.: Receptor orphans find a family. *Curr. Biol.* 5: 986–989, 1995.
- [41400] 14952. Bunz, F.; Dutriaux, A.; Lengauer, C.; Waldman, T.; Zhou, S.; Brown, J. P.; Sedivy, J. M.; Kinzler, K. W.; Vogelstein, B.: Requirement for p53 and p21 to sustain G2 arrest after DNA damage. *Science* 282:1497–1501, 1998.
- [41401] 14953. Colucci-Guyon, E.; Portier, M.-M.; Dunia, I.; Paulin, D.; Pournin, S.; Babinet, C.: Mice lacking vimentin develop and reproduce without an obvious phenotype. *Cell* 79: 679–694, 1994.
- [41402] 14954. Ferrari, S.; Battini, R.; Kaczmarek, L.; Rittling, S.; Calabretta, B.; de Riel, J. K.; Philiponis, V.; Weil, J.-F.; Baserga, R.: Coding sequence and growth regulation of the human vimentin gene. *Molec. Cell. Biol.* 6: 3614–3620, 1986.
- [41403] 14955. Gieser, L.; Swaroop, A.: Expressed sequence tags and chromosomal localization of cDNA clones from a subtracted retinal pigment epithelium library. *Genomics* 13: 873–876, 1992.
- [41404] 14956. Lilienbaum, A.; Legagneux, V.; Portier, M.-M.; Delagi, K.; Paulin, D.: Vimentin gene: expression in human lymphocytes and in Burkitt's lymphoma cells. *EMBO J.* 5:

2809–2814, 1986.

- [41405] 14957. Marcus, E. M.; Smith, B. A.; Telenius, H.; Lands-
vater, R. M.; Buys, C. H. C. M.; Ferrari, S.; Ponder, B. A. J.;
Mathew, C. G. P.: BclIRFLP for the human vimentin gene.
Nucleic Acids Res. 16: 9068 only, 1988.
- [41406] 14958. Mathew, C. G.; Wakeling, W.; Jones, E.; Easton, D.;
Fisher, R.; Strong, C.; Smith, B.; Chin, K.; Little, P.; Naka-
mura, Y.; Shows, T. B.; Jones, C.; Goodfellow, P. J.; Povey,
S.; Ponder, B. A. J.: Regional localization of polymorphic
markers on chromosome 10 by physical and genetic map-
ping. Ann. Hum. Genet. 54: 121–129, 1990.
- [41407] 14959. Perreau, J.; Lilienbaum, A.; Vasseur, M.; Paulin, D.:
Nucleotide sequence of the human vimentin gene and reg-
ulation of its transcription in tissues and cultured cells.
Gene 62: 7–16, 1988.
- [41408] 14960. Gimona, M.; Small, J. V.; Moeremans, M.; Van
Damme, J.; Puype, M.; Vandekerckhove, J.: Porcine vinculin
and metavinculin differ by a 68-residue insert located
close to the carboxy-terminal part of the molecule. EMBO
J. 7: 2329–2334, 1988.
- [41409] 14961. Burglen, L.; Amiel, J.; Viollet, L.; Lefebvre, S.; Burlet,
P.; Clermont, O.; Raclin, V.; Landrieu, P.; Verloes, A.;
Munnich, A.; Melki, J.: Survival motor neuron gene deletion

in the arthrogryposismultiplex congenita–spinal muscular atrophy association. *J. Clin. Invest.* 98: 1130–1132, 1996.

[41410] 14962. Gabreels, B. A. T. F.; Swaab, D. F.; de Kleijn, D. P. V.; Seidah, N. G.; Van de Loo, J.–W.; Van de Ven, W. J. M.; Martens, G. J. M.; van Leeuwen, F. W.: Attenuation of the polypeptide 7B2, prohormoneconvertase PC2, and vasopressin in the hypothalamus of some Prader–Willi patients: indications for a processing defect. *J. Clin. Endocr. Metab.* 83:591–599, 1998.

[41411] 14963. Martens, G. J. M.: Cloning and sequence analysis of human pituitary cDNA encoding the novel polypeptide 7B2. *FEBS Lett.* 234: 160–164, 1988.

[41412] 14964. Mattei, M. G.; Mbikay, M.; Sylla, B. S.; Lenoir, G.; Mattei, J. F.; Seidah, N. G.; Chretien, M.: Assignment of the gene for neuroendocrine protein 7B2 (SGNE1 locus) to mouse chromosome region 2[E3–F3] and to human chromosome region 15q11–q15. *Genomics* 6: 436–440, 1990.

[41413] 14965. Mbikay, M.; Seidah, N. G.; Chretien, M.: Neuroendocrine secretory protein 7B2: structure, expression and functions. *Biochem. J.* 357:329–342, 2001.

[41414] 14966. Roebroek, A. J. M.; Dehaen, M. R. M.; van Bokhoven, A.; Martens, G. J. M.; Marynen, P.; van den Berghe, H.; Van de Ven, W. J. M.: Regional mapping of the

human gene encoding the novel pituitary polypeptide 7B2 to chromosome 15q13–q14 by in situ hybridization. *Cytogenet. Cell Genet.* 50: 158–160, 1989.

- [41415] 14967. Westphal, C. H.; Muller, L.; Zhou, A.; Zhu, X.; Bonner-Weir, S.; Schambelan, M.; Steiner, D. F.; Lindberg, I.; Leder, P.: The neuroendocrine protein 7B2 is required for peptide hormone processing in vivo and provides a novel mechanism for pituitary Cushing's disease. *Cell* 96:689–700, 1999.
- [41416] 14968. Bell, K.; Hopper, K. E.; McKenzie, H. A.; Murphy, W. H.; Shaw, D. C.: A comparison of bovine alpha-lactalbumin A and B of Droughtmaster. *Biochim. Biophys. Acta* 214: 437–444, 1970.
- [41417] 14969. Bell, K.; McKenzie, H. A.; Murphy, W. H.; Shaw, D. C.: Beta-lactoglobulin (Droughtmaster): a unique protein variant. *Biochim. Biophys. Acta* 214:427–436, 1970.
- [41418] 14970. Julkunen, M.; Seppala, M.; Janne, O. A.: Complete amino acid sequence of human placental protein 14: a progesterone-regulated uterine protein homologous to beta-lactoglobulins. *Proc. Nat. Acad. Sci.* 85: 8845–8849, 1988.
- [41419] 14971. Morris, H. R.; Dell, A.; Easton, R. L.; Panico, M.; Koistinen, H.; Koistinen, R.; Oehninger, S.; Patankar, M. S.;

Seppala, M.; Clark, G. F.: Gender-specific glycosylation of human glycodelin affects its contraceptive activity. *J. Biol. Chem.* 271: 32159–32167, 1996.

- [41420] 14972. Song, M.; Ramaswamy, S.; Ramachandran, S.; Flowers, L. C.; Horowitz, I. R.; Rock, J. A.; Parthasarathy, S.: Angiogenic role for glycodelin in tumorigenesis. *Proc. Nat. Acad. Sci.* 98: 9265–9270, 2001.
- [41421] 14973. Van Cong, N.; Vaisse, C.; Gross, M.-S.; Slim, R.; Milgrom, E.; Bernheim, A.: The human placental protein 14 (PP14) gene is localized on chromosome 9q34. *Hum. Genet.* 86: 515–518, 1991.
- [41422] 14974. Lee, F. S.; Fox, E. A.; Zhou, H.-M.; Strydom, D. J.; Vallee, B. L.: Primary structure of human placental ribonuclease inhibitor. *Biochemistry* 27: 8545–8553, 1988.
- [41423] 14975. Weremowicz, S.; Fox, E. A.; Morton, C. C.; Vallee, B. L.: The placental ribonuclease inhibitor (RNH) gene is located on chromosome subband 11p15.5. *Genomics* 8: 717–721, 1990.
- [41424] 14976. Zneimer, S. M.; Crawford, D.; Schneider, N. R.; Beutler, B.: Mapping of the human ribonuclease inhibitor gene (RNH) to chromosome 11p15 by in situ hybridization. *Genomics* 8: 175–178, 1990.
- [41425] 14977. Coughlin, P.; Nicholl, J.; Sun, J.; Salem, H.; Bird, P.;

Sutherland,G. R.: Chromosomal mapping of the human proteinase inhibitor 6 (PI6)gene to 6p25 by fluorescence in situ hybridization. *Genomics* 26:431–433, 1995.

[41426] 14978.Coughlin, P.; Sun, J.; Cerruti, L.; Salem, H. H.; Bird, P.: Cloningand molecular characterization of a human intracellular serine proteinaseinhibitor. *Proc. Nat. Acad. Sci.* 90: 9417–9421, 1993.

[41427] 14979.Siu, G.; Wurster, A. L.; Duncan, D. D.; Soliman, T. M.; Hedrick,S. M.: A transcriptional silencer controls the developmental expressionof the CD4 gene. *EMBO J.* 13: 3570–3579, 1994.

[41428] 14980.Zhang, K.; Westberg, J. A.; Paetau, A.; von Boguslawsky, K.; Lindsberg,P.; Erlander, M.; Guo, H.; Su, J.; Olsen, H. S.; Andersson, L. C.: High expression of stannio-calcin in differentiated brain neurons. *Am.J. Path.* 153: 439–445, 1998.

[41429] 14981.Hamilton, S. E.; Hurley, J. B.: A phosphodiesterase inhibitorspecific to a subset of bovine retinal cones. *J. Biol. Chem.* 265:11259–11264, 1990.

[41430] 14982.Shimizu–Matsumoto, A.; Itoh, K.; Inazawa, J.; Nishida, K.; Matsumoto,Y.; Kinoshita, S.; Matsubara, K.; Okubo, K.: Isolation and chromosomallocalization of the human cone cGMP phosphodiesterase gamma cDNA

(PDE6H). Genomics 32:121–124, 1996.

- [41431] 14983.Chen, J.; Engle, S. J.; Seilhamer, J. J.; Tischfield, J. A.: Cloning and recombinant expression of a novel human low molecular weight Ca^{2+} –dependent phospholipase A2. J. Biol. Chem. 269: 2365–2368, 1994.
- [41432] 14984.Tischfield, J. A.; Xia, Y.–R.; Shih, D. M.; Klisak, I.; Chen, J.;Engle, S. J.; Siakotos, A. N.; Winstead, M. V.; Seilhamer, J. J.;Allamand, V.; Gyapay, G.; Lusk, A. J.: Low molecular weight, calcium–dependent phospholipase A(2) genes are linked and map to homologous chromosomal regions in mouse and human. Genomics 32: 328–333, 1996.
- [41433] 14985.Alexopoulou, L.; Thomas, V.; Schnare, M.; Lobet, Y.; Anguita, J.;Schoen, R. T.; Medzhitov, R.; Fikrig, E.; Flavell, R. A.: Hyporesponsiveness to vaccination with *Borrelia burgdorferi* OspA in humans and in TLR1– and TLR2–deficient mice. Nature Med. 8: 878–884, 2002.
- [41434] 14986.Gay, N. J.; Keith, F. J.: Drosophila Toll and IL–1 receptor. (Letter) Nature 351:355–356, 1991.
- [41435] 14987.Muzio, M.; Bosisio, D.; Polentarutti, N.; D'amico, G.; Stoppacciaro, A.; Mancinelli, R.; van't Veer, C.; Penton–Rol, G.; Ruco, L. P.; Allavena, P.; Mantovani, A.: Differential expression and regulation of Toll–like receptors (TLR) in hu–

man leukocytes: selective expression of TLR3 in dendritic cells. *J. Immun.* 164: 5998–6004, 2000.

- [41436] 14988. Rock, F. L.; Hardiman, G.; Timans, J. C.; Kastelein, R. A.; Bazan, J. F.: A family of human receptors structurally related to *Drosophila* Toll. *Proc. Nat. Acad. Sci.* 95: 588–593, 1998.
- [41437] 14989. Taguchi, T.; Mitcham, J. L.; Dower, S. K.; Sims, J. E.; Testa, J. R.: Chromosomal localization of TIL, a gene encoding a protein related to the *Drosophila* transmembrane receptor Toll, to human chromosome 4p14. *Genomics* 32: 486–488, 1996.
- [41438] 14990. Xu, Y.; Tao, X.; Shen, B.; Horng, T.; Medzhitov, R.; Manley, J. L.; Tong, L.: Structural basis for signal transduction by the Toll/interleukin-1 receptor domains. *Nature* 408: 111–115, 2000.
- [41439] 14991. Perrimon, N.; Mahowald, A. P.: Multiple functions of segment polarity genes in *Drosophila*. *Dev. Biol.* 119: 587–600, 1987.
- [41440] 14992. Pizzuti, A.; Novelli, G.; Mari, A.; Ratti, A.; Colosimo, A.; Amati, F.; Penso, D.; Sangiuolo, F.; Calabrese, G.; Palka, G.; Silani, V.; Gennarelli, M.; Mingarelli, R.; Scarlato, G.; Scambler, P.; Dallapiccola, B.: Human homologue sequences to the *Drosophila* dishevelled segment-polar-

itygene are deleted in the DiGeorge syndrome. *Am. J. Hum. Genet.* 58:722–729, 1996.

- [41441] 14993.Brown, E. J.; Albers, M. W.; Shin, T. B.; Ichikawa, K.; Keith, C. T.; Lane, W. S.; Schreiber, S. L.: A mammalian protein targeted by G1-arresting rapamycin-receptor complex. *Nature* 369: 756–758, 1994.
- [41442] 14994.Dennis, P. B.; Jaeschke, A.; Saitoh, M.; Fowler, B.; Kozma, S.C.; Thomas, G.: Mammalian TOR: a homeostatic ATP sensor. *Science* 294:1102–1105, 2001.
- [41443] 14995.Fang, Y.; Vilella-Bach, M.; Bachmann, R.; Flanigan, A.; Chen, J.: Phosphatidic acid-mediated mitogenic activation of mTOR signaling. *Science* 294:1942–1945, 2001.
- [41444] 14996.Hara, K.; Maruki, Y.; Long, X.; Yoshino, K.; Oshiro, N.; Hidayat, S.; Tokunaga, C.; Avruch, J.; Yonezawa, K.: Raptor, a binding partner of target of rapamycin, mediates TOR action. *Cell* 110: 177–189, 2002.
- [41445] 14997.Kim, D.-H.; Sarbassov, D. D.; Ali, S. M.; King, J. E.; Latek, R.R.; Erdjument-Bromage, H.; Tempst, P.; Sabatini, D. M.: mTOR interacts with raptor to form a nutrient-sensitive complex that signals to the cell growth machinery. *Cell* 110: 163–175, 2002.
- [41446] 14998.Lench, N. J.; Macadam, R.; Markham, A. F.: The human gene encoding FKBP-rapamycin associated protein

(FRAP) maps to chromosomal band 1p36.2. Hum. Genet. 99: 547–549, 1997.

- [41447] 14999. Moore, P. A.; Rosen, C. A.; Carter, K. C.: Assignment of the human FKBP12–rapamycin–associated protein (FRAP) gene to chromosome 1p36 by fluorescence in situ hybridization. Genomics 33: 331–332, 1996.
- [41448] 15000. Onyango, P.; Lubyova, B.; Gardellin, P.; Kurzbauer, R.; Weith, A.: Molecular cloning and expression analysis of five novel genes in chromosome 1p36. Genomics 50: 187–198, 1998.
- [41449] 15001. Sabatini, D. M.; Erdjument–Bromage, H.; Lui, M.; Tempst, P.; Snyder, S. H.: RAFT1: a mammalian protein that binds to FKBP12 in a rapamycin–dependent fashion and is homologous to yeast TORs. Cell 78: 35–43, 1994.
- [41450] 15002. Barbier, M.; Attoub, S.; Calvez, R.; Laffargue, M.; Jarry, A.; Mareel, M.; Altruda, F.; Gespach, C.; Wu, D.; Lu, B.; Hirsch, E.; Wymann, M. P.: Weakening link to colorectal cancer? Nature 413: 796 only, 2001.
- [41451] 15003. Crackower, M. A.; Oudit, G. Y.; Kozieradzki, I.; Sarao, R.; Sun, H.; Sasaki, T.; Hirsch, E.; Suzuki, A.; Shioi, T.; Irie–Sasaki, J.; Sah, R.; Cheng, H.–Y. M.; and 13 others: Regulation of myocardial contractility and cell size by distinct PI3K–PTEN signaling pathways. Cell 110: 737–749,

2002.

- [41452] 15004.Hirsch, E.; Katanaev, V. L.; Garlanda, C.; Azzolino, O.; Pirola, L.; Silengo, L.; Sozzani, S.; Mantovani, A.; Altruda, F.; Wymann, M. P.: Central role for G protein-coupled phosphoinositide 3-kinase γ in inflammation. *Science* 287: 1049–1053, 2000.
- [41453] 15005.Jiang, K.; Zhong, B.; Gilvary, D. L.; Corliss, B. C. Hong-Geller, E.; Wei, S; Djeu, J. Y.: Pivotal role of phosphoinositide-3 kinase in regulation of cytotoxicity in natural killer cells. *Nature Immun.* 1:419–425, 2000.
- [41454] 15006.Kikuta, Y.; Kato, M.; Yamashita, Y.; Miyauchi, Y.; Tanaka, K.; Kamada, N.; Kusunose, M.: Human leukotriene B₄ omega-hydroxylase (CYP4F3) gene: molecular cloning and chromosomal localization. *DNACell Biol.* 17: 221–230, 1998.
- [41455] 15007.Kikuta, Y.; Kusunose, E.; Endo, K.; Yamamoto, S.; Sogawa, K.; Fujii-Kuriyama, Y.; Kusunose, M.: A novel form of cytochrome P-450 family 4 in human polymorphonuclear leukocytes: cDNA cloning and expression of leukotriene B₄ omega-hydroxylase. *J. Biol. Chem.* 268: 9376–9380, 1993.
- [41456] 15008.Klingensmith, J.; Nusse, R.; Perrimon, N.: The *Drosophila* segment polarity gene *dishevelled* encodes a

novel protein required for response to wingless signal.

Genes Dev. 8: 118–130, 1994.

[41457] 15009. Holmes, S. E.; Riaz, M. A.; Gong, W.; McDermid, H. E.; Sellinger, B. T.; Hua, A.; Chen, F.; Wang, Z.; Zhang, G.; Roe, B.; Gonzalez, I.; McDonald-McGinn, D. M.; Zackai, E.; Emanuel, B. S.; Budarf, M. L.: Disruption of the clathrin heavy chain-like gene (CLTCL) associated with features of DGS/VCFS: a balanced (21;22)(p12;q11) translocation. Hum. Molec. Genet. 6: 357–367, 1997.

[41458] 15010. Kedra, D.; Peyrard, M.; Fransson, I.; Collins, J. E.; Dunham, I.; Roe, B. A.; Dumanski, J. P.: Characterization of a second human clathrin heavy chain polypeptide gene (CLH-22) from chromosome 22q11. Hum. Molec. Genet. 5: 625–631, 1996.

[41459] 15011. Long, K. R.; Trofatter, J. A.; Ramesh, V.; McCormick, M. K.; Buckler, A. J.: Cloning and characterization of a novel human clathrin heavy chain gene (CLTCL). Genomics 35: 466–472, 1996.

[41460] 15012. Lagenaur, C.; Kunemund, V.; Fischer, G.; Fushiki, S.; Schachner, M.: Monoclonal M6 antibody interferes with neurite extension of cultured neurons. J. Neurobiol. 23: 71–88, 1992.

[41461] 15013. Shimizu, F.; Watanabe, T. K.; Fujiwara, T.; Taka-

hashi, E.; Nakamura, Y.; Maekawa, H.: Isolation and mapping of the human glycoprotein M6 gene (GPM6A) to 4q33-to-q34. *Cytogenet. Cell Genet.* 74: 138–139, 1996.

[41462] 15014. Bui, T. D.; Beier, D. R.; Jonssen, M.; Smith, K.; Dorrington, S. M.; Kaklamanis, L.; Kearney, L.; Regan, R.; Sussman, D. J.; Harris, A. L.: cDNA cloning of a human dishevelled DVL-3 gene, mapping to 3q27, and expression in human breast and colon carcinomas. *Biochem. Biophys. Res. Commun.* 239: 510–516, 1997.

[41463] 15015. Cheng, Y.-S. E.; Patterson, C. E.; Staeheli, P.: Interferon-induced guanylate-binding proteins lack an N(T)KXD consensus motif and bind GMP in addition to GDP and GTP. *Molec. Cell. Biol.* 11: 4717–4725, 1991.

[41464] 15016. Kumar, S.; Li, Q.; Dua, A.; Ying, Y.-K.; Bagchi, M. K.; Bagchi, I. C.: Messenger ribonucleic acid encoding interferon-inducible guanylate-binding protein 1 is induced in human endometrium within the putative window of implantation. *J. Clin. Endocr. Metab.* 86: 2420–2427, 2001.

[41465] 15017. Prochazka, M.; Staeheli, P.; Holmes, R. S.; Haller, O.: Interferon-induced guanylate-binding proteins: mapping of the murine Gbp-1 locus to chromosome 3. *Virology* 145: 273–279, 1985.

[41466] 15018. Strehlow, I.; Lohmann-Matthes, M. L.; Decker, T.:

The interferon-inducibleGBP1 gene: structure and mapping to human chromosome 1. *Gene* 144:295–299, 1994.

- [41467] 15019. Avraham, K. B.; Prezioso, V. R.; Chen, W. S.; Lai, E.; Sladek, F. M.; Zhong, W.; Darnell, J. E., Jr.; Jenkins, N. A.; Copeland, N.G.: Murine chromosomal location of four hepatocyte-enriched transcription factors: HNF-3-alpha, HNF3-beta, HNF-3-gamma, and HFN-4. *Genomics* 13:264–268, 1992.
- [41468] 15020. Chartier, F. L.; Bossu, J.-P.; Laudet, V.; Fruchart, J.-C.: Cloning and sequencing of cDNAs encoding the human hepatocyte nuclear factor 4 indicate the presence of two isoforms in human liver. *Gene* 147:269–272, 1994.
- [41469] 15021. Eeckhoute, J.; Formstecher, P.; Laine, B.: Maturity-onset diabetes of the young type 1 (MODY1)-associated mutations R154X and E276Q in hepatocyte nuclear factor 4-alpha (HNF4-alpha) gene impair recruitment of p300, a key transcriptional coactivator. *Molec. Endocr.* 15: 1200–1210, 2001.
- [41470] 15022. Furuta, H.; Iwasaki, N.; Oda, N.; Hinokio, Y.; Horikawa, Y.; Yamagata, K.; Yano, N.; Sugahiro, J.; Ogata, M.; Ohgawara, H.; Omori, Y.; Iwamoto, Y.; Bell, G. I.: Organization and partial sequence of the hepatocyte nuclear factor-4-alpha/MODY1 gene and identification of a mis-

sense mutation, R127W, in a Japanese family with MODY.
Diabetes 46: 1652–1657, 1997.

- [41471] 15023. Hani, E. H.; Suaud, L.; Boutin, P.; Chevre, J.-C.; Durand, E.; Philippi, A.; Demenais, F.; Vionnet, N.; Furuta, H.; Velho, G.; Bell, G. I.; Laine, B.; Froguel, P.: A missense mutation in hepatocyte nuclear factor-4- α , resulting in a reduced transactivation activity, in human late-onset non-insulin-dependent diabetes mellitus. J. Clin. Invest. 101: 521–526, 1998.
- [41472] 15024. Lausen, J.; Thomas, H.; Lemm, I.; Bulman, M.; Borgschulze, M.; Lingott, A.; Hattersley, A. T.; Ryffel, G. U.: Naturally occurring mutations in the human HNF4- α gene impair the function of the transcription factor to a varying degree. Nucleic Acids Res. 28: 430–437, 2000.
- [41473] 15025. Li, J.; Ning, G.; Duncan, S. A.: Mammalian hepatocyte differentiation requires the transcription factor HNF-4- α . Genes Dev. 14: 464–474, 2000.
- [41474] 15026. Stoffel, M.; Duncan, S. A.: The maturity-onset diabetes of the young (MODY1) transcription factor HNF4- α regulates expression of genes required for glucose transport and metabolism. Proc. Nat. Acad. Sci. 94: 13209–13214, 1997.
- [41475] 15027. Yamagata, K.; Furuta, H.; Oda, N.; Kaisaki, P. J.;

Menzel, S.;Cox, N. J.; Fajans, S. S.; Signorini, S.; Stoffel, M.; Bell, G. I.: Mutations in the hepatocyte nuclear factor-4-alpha gene in maturity-onsetdiabetes of the young (MODY1). *Nature* 384: 458-460, 1996.

[41476] 15028.Zouali, H.; Hani, E. H.; Philippi, A.; Vionnet, N.; Beckmann,J. S.; Demenais, F.; Froguel, P.: A susceptibility locus for early-onsetnon-insulin dependent (type 2) diabetes mellitus maps to chromosome20q, proximal to the phosphoenolpyruvate carboxykinase gene. *Hum.Molec. Genet.* 6: 1401-1408, 1997.

[41477] 15029.Frolova, L.; Le Goff, X.; Rasmussen, H. H.; Cheperegin, S.; Drugeon,G.; Kress, M.; Arman, I.; Haenni, A.-L.; Celis, J. E.; Philippe, M.;Justesen, J.; Kisselev, L.: A highly conserved eukaryotic proteinfamily possessing properties of polypeptide chain release factor. *Nature* 372:701-703, 1994.

[41478] 15030.Guenet, L.; Henry, C.; Toutain, B.; Dubourg, C.; Le Gall, J. Y.;David, V.; Le Treut, A.: Eukaryotic translation termination factorgene (ETF1/eRF1) maps at D5S500 in a commonly deleted region of chromosome5q31 in malignant myeloid diseases. *Cytogenet. Cell Genet.* 88: 82-86,2000.

[41479] 15031.Hansen, L. L.; Jakobsen, C. G.; Justesen, J.: Assign-

ment of the human translation termination factor 1 (ETF1) to 5q31.1 and of the proximal marker D5S1995 by radiation hybrid mapping. *Cytogenet. Cell Genet.* 87: 256–257, 1999.

[41480] 15032. Le Goff, X.; Philippe, M.; Jean-Jean, O.: Overexpression of human release factor 1 alone has an antisuppressor effect in human cells. *Molec. Cell Biol.* 17: 3164–3172, 1997.

[41481] 15033. Song, H.; Mugnier, P.; Das, A. K.; Webb, H. M.; Evans, D. R.; Tuite, M. F.; Hemmings, B. A.; Barford, D.: The crystal structure of human eukaryotic release factor eRF1—mechanism of stop codon recognition and peptidyl-tRNA hydrolysis. *Cell* 100: 311–321, 2000.

[41482] 15034. DiDonato, C. J.; Chen, X.-N.; Noya, D.; Korenberg, J. R.; Nadeau, J. H.; Simard, L. R.: Cloning, characterization, and copy number of the murine survival motor neuron gene: homolog of the spinal muscular atrophy-determining gene. *Genome Res.* 7: 339–352, 1997.

[41483] 15035. DiDonato, C. J.; Morgan, K.; Carpten, J. D.; Fuerst, P.; Ingraham, S. E.; Prescott, G.; McPherson, J. D.; Wirth, B.; Zerres, K.; Hurko, O.; Wasmuth, J. J.; Mendell, J. R.; Burghes, A. H. M.; Simard, L. R.: Association between Ag1-CA alleles and severity of autosomal recessive proxi-

mal spinal muscular atrophy. Am. J. Hum. Genet.
55:1218–1229, 1994.

- [41484] 15036.Feldkotter, M.; Schwarzer, V.; Wirth, R.; Wienker, T. F.; Wirth,B.: Quantitative analyses of SMN1 and SMN2 based on real-time lightCyclerPCR: fast and highly reliable carrier testing and prediction of severityof spinal muscular atrophy. Am. J. Hum. Genet. 70: 358–368, 2002.
- [41485] 15037.Friesen, W. J.; Massenet, S.; Paushkin, S.; Wyce, A.; Dreyfuss,G.: SMN, the product of the spinal muscular atrophy gene, binds preferentiallyto dimethylarginine-containing protein targets. Molec. Cell 7: 1111–1117,2001.
- [41486] 15038.Frugier, T.; Tiziano, F. D.; Cifuentes–Diaz, C.; Min–iou, P.; Roblot,N.; Dierich, A.; Le Meur, M.; Melki, J.: Nuclear targeting defectof SMN lacking the C–terminus in a mouse model of spinal muscularatrophy. Hum. Molec. Genet. 9: 849–858, 2000.
- [41487] 15039.Gambardella, A.; Mazzei, R.; Toscano, A.; Annesi, G.; Pasqua,A.; Annesi, F.; Quattrone, F.; Oliveri, R. L.; Valentino, P.; Bono,F.; Aguglia, U.; Zappia, M.; Vita, G.; Quattrone, A.: Spinal muscularatrophy due to an isolated deletion of exon 8 of the telomeric survivalmotor neuron gene. Ann. Neurol. 44: 836–839, 1998.
- [41488] 15040.Hahnen, E.; Schonling, J.; Rudnik–Schoneborn, S.;

Raschke, H.;Zerres, K.; Wirth, B.: Missense mutations in exon 6 of the survivalmotor neuron gene in patients with spinal muscular atrophy (SMA). Hum.Molec. Genet. 6: 821–825, 1997.

[41489] 15041.Hannus, S.; Buhler, D.; Romano, M.; Seraphin, B.; Fischer, U.: The Schizosaccharomyces pombe protein Yab8p and a novel factor,Yip1p, share structural and functional similarity with the spinalmuscular atrophy-associated proteins SMN and SIP1. Hum. Molec. Genet. 9:663–674, 2000.

[41490] 15042.Hsieh–Li, H. M.; Chang, J.–G.; Jong, Y.–J.; Wu, M.–H.; Wang, N.M.; Tsai, C. H.; Li, H.: A mouse model for spinal muscular atrophy. NatureGenet. 24: 66–70, 2000.

[41491] 15043.Jablonka, S.; Bandilla, M.; Wiese, S.; Buhler, D.; Wirth, B.;Sendtner, M.; Fischer, U.: Co–regulation of survival of motor neuron(SMN) protein and its interactor SIP1 during development and in spinalmuscular atrophy. Hum. Molec. Genet. 10: 497–505, 2001.

[41492] 15044.Liu, Q.; Dreyfuss, G.: A novel nuclear structure containing thesurvival of motor neurons protein. EMBO J. 15: 3555–3565, 1996.

[41493] 15045.Liu, Q.; Fischer, U.; Wang, F.; Dreyfuss, G.: The spinal muscularatrophy disease gene product, SMN, and

its associated protein SIP1 are in a complex with spliceosomal snRNP proteins. *Cell* 90: 1013–1021, 1997.

[41494] 15046. Lorson, C. L.; Androphy, E. J.: The domain encoded by exon 2 of the survival motor neuron protein mediates nucleic acid binding. *Hum. Molec. Genet.* 7: 1269–1275, 1998.

[41495] 15047. Lorson, C. L.; Hahnen, E.; Androphy, E. J.; Wirth, B.: A single nucleotide in the SMN gene regulates splicing and is responsible for spinal muscular atrophy. *Proc. Nat. Acad. Sci.* 96: 6307–6311, 1999.

[41496] 15048. Lorson, C. L.; Strasswimmer, J.; Yao, J.-M.; Baleja, J. D.; Hahnen, E.; Wirth, B.; Le, T.; Burghes, A. H. M.; Androphy, E. J.: SMN oligomerization defect correlates with spinal muscular atrophy severity. *Nature Genet.* 19: 63–66, 1998.

[41497] 15049. McAndrew, P. E.; Parsons, D. W.; Simard, L. R.; Rochette, C.; Ray, P. N.; Mendell, J. R.; Prior, T. W.; Burghes, A. H.: Identification of proximal spinal muscular atrophy carriers and patients by analysis of SMNT and SMNC gene copy number. *Am. J. Hum. Genet.* 60: 1411–1422, 1997.

[41498] 15050. Meister, G.; Buhler, D.; Lagerbauer, B.; Zobawa, M.; Lottspeich, F.; Fischer, U.: Characterization of a nuclear 20S complex containing the survival of motor neurons

(SMN) protein and a specific subset of spliceosomal Sm proteins. Hum. Molec. Genet. 9: 1977–1986, 2000.

- [41499] 15051. Mohaghegh, P.; Rodrigues, N. R.; Owen, N.; Ponting, C. P.; Le, T. T.; Burghes, A. H. M.; Davies, K. E.: Analysis of mutations in the tudor domain of the survival motor neuron protein SMN. Europ. J. Hum. Genet. 7: 519–525, 1999.
- [41500] 15052. Monani, U. R.; Lorson, C. L.; Parsons, D. W.; Prior, T. W.; Androphy, E. J.; Burghes, A. H. M.; McPherson, J. D.: A single nucleotide difference that alters splicing patterns distinguishes the SMA gene SMN1 from the copy gene SMN2. Hum. Molec. Genet. 8: 1177–1183, 1999.
- [41501] 15053. Mourelatos, Z.; Abel, L.; Yong, J.; Kataoka, N.; Dreyfuss, G.: SMN interacts with a novel family of hnRNP and spliceosomal proteins. EMBO J. 20: 5443–5452, 2001.
- [41502] 15054. Owen, N.; Doe, C. L.; Mellor, J.; Davies, K. E.: Characterization of the Schizosaccharomyces pombe orthologue of the human survival motor neuron (SMN) protein. Hum. Molec. Genet. 9: 675–684, 2000.
- [41503] 15055. Pagliardini, S.; Giavazzi, A.; Setola, V.; Lizier, C.; Di Luca, M.; DeBiasi, S.; Battaglia, G.: Subcellular localization and axonal transport of the survival motor neuron (SMN) protein in the developing rat spinal cord. Hum. Molec.

Genet. 9: 47–56, 2000.

- [41504] 15056.Parsons, D. W.; McAndrew, P. E.; Iannaccone, S. T.; Mendell, J.R.; Burghes, A. H. M.; Prior, T. W.: Intragenic telSMN mutations:frequency, distribution, evidence of a founder effect, and modification of the spinal muscular atrophy phenotype by cenSMN copy number. Am.J. Hum. Genet. 63: 1712–1723, 1998.
- [41505] 15057.Parsons, D. W.; McAndrew, P. E.; Monani, U. R.; Mendell, J. R.;Burghes, A. H. M.; Prior, T. W.: An 11 base pair duplication in exon6 of the SMN gene produces a type I spinal muscular atrophy (SMA)phenotype: further evidence for SMN as the primary SMA–determininggene. Hum. Molec. Genet. 5: 1727–1732, 1996.
- [41506] 15058.Lewandoski, M.; Sun, X.; Martin, G. R.: Fgf8 signalling from theAER is essential for normal limb development. Nature Genet. 26:460–463, 2000.
- [41507] 15059.Lorenzi, M. V.; Long, J. E.; Miki, T.; Aaronson, S. A.: Expressioncloning, developmental expression and chromosomal localization of fibroblast growth factor–8. Oncogene 10: 2051–2055, 1995.
- [41508] 15060.Meyers, E. N.; Martin, G. R.: Differences in left–right axis pathways in mouse and chick: functions of FGF8 and SHH. Science 285:403–406, 1999.

- [41509] 15061.Moon, A. M.; Capecchi, M. R.: Fgf8 is required for outgrowth and patterning of the limbs. *Nature Genet.* 26: 455–459, 2000.
- [41510] 15062.Payson, R. A.; Wu, J.; Liu, Y.; Chiu, I.-M.: The human FGF-8 gene localizes on chromosome 10q24 and is subjected to induction by androgen in breast cancer cells. *Oncogene* 13: 47–53, 1996.
- [41511] 15063.Streit, A.; Berliner, A. J.; Papanayotou, C.; Sirulnik, A.; Stern, C. D.: Initiation of neural induction by FGF signalling before gastrulation. *Nature* 406:74–78, 2000.
- [41512] 15064.Sun, X.; Meyers, E. N.; Lewandoski, M.; Martin, G. R.: Targeted disruption of Fgf8 causes failure of cell migration in the gastrulating mouse embryo. *Genes Dev.* 13: 1834–1846, 1999.
- [41513] 15065.White, R. A.; Dowler, L. L.; Angeloni, S. V.; Pasztor, L. M.; MacArthur, C. A.: Assignment of FGF8 to human chromosome 10q25–q26: mutations in FGF8 may be responsible for some types of acrocephalosyndactyly linked to this region. *Genomics* 30: 109–111, 1995.
- [41514] 15066.Yoshiura, K.; Leysens, N. J.; Chang, J.; Ward, D.; Murray, J.C.; Muenke, M.: Genomic structure, sequence, and mapping of human FGF8 with no evidence for its role in craniosynostosis/limb defects syndromes. *Am. J. Med.*

Genet. 72: 354–362, 1997.

- [41515] 15067. Zammit, C.; Coope, R.; Gomm, J. J.; Shousha, S.; Johnston, C. L.; Coombes, R. C.: Fibroblast growth factor 8 is expressed at higher levels in lactating human breast and in breast cancer. *Brit. J. Cancer* 86:1097–1103, 2002.
- [41516] 15068. Cao, H.; Mok, A.; Miskie, B.; Hegele, R. A.: Single-nucleotide polymorphisms of the proprotein convertase subtilisin/kexin type 5 (PCSK5) gene. *J. Hum. Genet.* 46: 730–732, 2001.
- [41517] 15069. van de Loo, J.-W. H. P.; Creemers, J. W. M.; Kas, K.; Roebroek, A. J. M.; Van de Ven, W. J. M.: Assignment of the human proprotein convertase gene PCSK5 to chromosome 9q21.3. *Cytogenet. Cell Genet.* 75:227–229, 1996.
- [41518] 15070. Aramburu, J.; Garcia-Cozar, F.; Raghavan, A.; Okamura, H.; Rao, A.; Hogan, P. G.: Selective inhibition of NFAT activation by a peptide spanning the calcineurin targeting site of NFAT. *Molec. Cell* 1:627–637, 1998.
- [41519] 15071. Castigli, E.; Pahwa, R.; Good, R. A.; Geha, R. S.; Chatila, T. A.: Molecular basis of a multiple lymphokine deficiency in a patient with severe combined immunodeficiency. *Proc. Nat. Acad. Sci.* 90:4728–4732, 1993.
- [41520] 15072. Chatila, T.; Castigli, E.; Pahwa, R.; Pahwa, S.; Chirmule, N.; Oyaizu, N.; Good, R. A.; Geha, R. S.: Primary

combined immunodeficiency resulting from defective transcription of multiple T-cell lymphokine genes. *Proc. Nat. Acad. Sci.* 87: 10033–10037, 1990.

[41521] 15073. Horsley, V.; Pavlath, G. K.: NFAT: ubiquitous regulator of cell differentiation and adaptation. *J. Cell Biol.* 156: 771–774, 2002.

[41522] 15074. Jauliac, S.; Lopez-Rodriguez, C.; Shaw, L. M.; Brown, L. F.; Rao, A.; Toker, A.: The role of NFAT transcription factors in integrin-mediated carcinoma invasion. *Nature Cell Biol.* 4: 540–544, 2002.

[41523] 15075. Li, X.; Ho, S. N.; Luna, J.; Giacalone, J.; Thomas, D. J.; Timmerman, L. A.; Crabtree, G. R.; Francke, U.: Cloning and chromosomal localization of the human and murine genes for the T-cell transcription factors NFATc and NFATp. *Cytogenet. Cell Genet.* 68: 185–191, 1995.

[41524] 15076. Northrop, J. P.; Ho, S. N.; Chen, L.; Thomas, D. J.; Timmerman, L. A.; Nolan, G. P.; Admon, A.; Crabtree, G. R.: NF-AT components define a family of transcription factors targeted in T-cell activation. *Nature* 369: 497–502, 1994.

[41525] 15077. Okamura, H.; Aramburu, J.; Garcia-Rodriguez, C.; Viola, J. P. B.; Raghavan, A.; Tahiliani, M.; Zhang, X.; Qin, J.; Hogan, P. G.; Rao, A.: Concerted dephosphorylation of the transcription factor NFAT1 induces a conformational

switch that regulates transcriptional activity. *Molec.Cell* 6: 539–550, 2000.

[41526] 15078.Pahwa, R.; Chatila, T.; Pahwa, S.; Paradise, C.; Day, N. K.; Geha, R.; Schwartz, S. A.; Slade, H.; Oyaizu, N.; Good, R. A.: Recombinant interleukin 2 therapy in severe combined immunodeficiency disease. *Proc.Nat. Acad. Sci.* 86: 5069–5073, 1989.

[41527] 15079.Park, J.; Takeuchi, A.; Sharma, S.: Characterization of a new isoform of the NFAT (nuclear factor of activated T cells) gene family member NFATc. *J. Biol. Chem.* 271: 20914–20921, 1996.

[41528] 15080.Peng, S. L.; Gerth, A. J.; Ranger, A. M.; Glimcher, L. H.: NFATc1 and NFATc2 together control both T and B cell activation and differentiation. *Immunity* 14:13–20, 2001.

[41529] 15081.Aoki, K.; Suzuki, K.; Sugano, T.; Tasaka, T.; Nakahara, K.; Kuge, O.; Omori, A.; Kasai, M.: A novel gene, 'Translin,' encodes a recombination hotspot binding protein associated with chromosomal translocations. *Nature-Genet.* 10: 167–174, 1995.

[41530] 15082.Badge, R. M.; Yardley, J.; Jeffreys, A. J.; Armour, J. A. L.: Crossover breakpoint mapping identifies a sub-telomeric hotspot for meiotic recombination. *Hum. Molec. Genet.* 9: 1239–1244, 2000.

- [41531] 15083.Hosaka, T.; Kanoe, H.; Nakayama, T.; Murakami, H.; Yamamoto, H.;Nakamata, T.; Tsuboyama, T.; Oka, M.; Kasai, M.; Sasaki, M. S.; Nakamura,T.; Toguchida, J.: Translin binds to the sequences adjacent to thebreak-points of the TLS and CHOP genes in liposarcomas with translocationt(12;16). *Oncogene* 19: 5821–5825, 2000.
- [41532] 15084.Kasai, M.; Aoki, K.; Matsuo, Y.; Minowada, J.; Maziarz, R. T.;Strominger, J. L.: Recombination hotspot associated factors specificallyrecognize novel target sequences at the site of interchromosomal rearrange-mentsin T-ALL patients with t(8;14)(q24;q11)and (t(1;14)(p32;q11). *Int.Immun.* 6: 1017–1025, 1994.
- [41533] 15085.Arceci, R. J.; King, A. A.; Simon, M. C.; Orkin, S. H.; Wilson,D. B.: Mouse GATA-4: a retinoic acid-inducible GATA-binding transcriptionfactor expressed in endoder-mally derived tissues and heart. *Molec.Cell. Biol.* 13: 2235–2246, 1993.
- [41534] 15086.Cirillo, L. A.; Lin, F. R.; Cuesta, I.; Friedman, D.; Jarnik, M.;Zaret, K. S.: Opening of compacted chromatin by early developmentaltranscription factors HNF3(FoxA) and GATA-4. *Molec. Cell* 9: 279–289,2002.
- [41535] 15087.Crispino, J. D.; Lodish, M. B.; Thurberg, B. L.; Litovsky, S. H.;Collins, T.; Molkentin, J. D.; Orkin, S. H.:

Proper coronary vascular development and heart morphogenesis depend on interaction of GATA-4 with FOG cofactors. *Genes Dev.* 15: 839–844, 2001.

[41536] 15088. Hasegawa, K.; Lee, S. J.; Jobe, S. M.; Markham, B. E.; Kitsis, R. N.: cis-acting sequences that mediate induction of beta-myosin heavy chain gene expression during left ventricular hypertrophy due to aortic constriction. *Circulation* 96: 3943–3953, 1997.

[41537] 15089. Huang, W.-Y.; Cukerman, E.; Liew, C.-C.: Identification of a GATA motif in the cardiac alpha-myosin heavy-chain-encoding gene and isolation of a human GATA-4 cDNA. *Gene* 155: 219–223, 1995.

[41538] 15090. Huang, W.-Y.; Heng, H. H. Q.; Liew, C.-C.: Assignment of the human GATA4 gene to 8p23.1–p22 using fluorescence in situ hybridization analysis. *Cytogenet. Cell Genet.* 72: 217–218, 1996.

[41539] 15091. Kennedy, S. J.; Teebi, A. S.; Adatia, I.; Teshima, I.: Inherited duplication, dup(8)(p23.1p23.1)Pat, in a father and daughter with congenital heart defects. (Letter) *Am. J. Med. Genet.* 104: 79–80, 2001.

[41540] 15092. Ketola, I.; Pentikainen, V.; Vaskivuo, T.; Ilvesmaki, V.; Herva, R.; Dunkel, L.; Tapanainen, J. S.; Toppari, J.; Heikinheimo, M.: Expression of transcription factor GATA-

4 during human testicular development and disease. J.

Clin. Endocr. Metab. 85: 3925–3931, 2000.

- [41541] 15093. Laitinen, M. P. E.; Anttonen, M.; Ketola, I.; Wilson, D. B.; Ritvos, O.; Butzow, R.; Heikinheimo, M.: Transcription factors GATA-4 and GATA-6 and a GATA family co-factor, FOG-2, are expressed in human ovary and sex cord-derived ovarian tumors. J. Clin. Endocr. Metab. 85:3476–3483, 2000.
- [41542] 15094. Molkentin, J. D.; Kalvakolanu, D. V.; Markham, B. E.: Transcription factor GATA-4 regulates cardiac muscle-specific expression of the α -myosin heavy-chain gene. Molec. Cell. Biol. 14: 4947–4957, 1994.
- [41543] 15095. Pehlivan, T.; Pober, B. R.; Brueckner, M.; Garrett, S.; Slauch, R.; Van Rheaden, R.; Wilson, D. B.; Watson, M. S.; Hing, A. V.: GATA4 haploinsufficiency in patients with interstitial deletion of chromosome region 8p23.1 and congenital heart disease. Am. J. Med. Genet. 83:201–206, 1999.
- [41544] 15096. White, R. A.; Dowler, L. L.; Pasztor, L. M.; Gatson, L. L.; Adkison, L. R.; Angeloni, S. V.; Wilson, D. B.: Assignment of the transcription factor GATA4 gene to human chromosome 8 and mouse chromosome 14: Gata4 is a candidate gene for Ds (disorganization). Genomics 27:

20–26,1995.

- [41545] 15097. Brenneman, M. A.; Wagener, B. M.; Miller, C. A.; Allen, C.; Nickoloff, J. A.: XRCC3 controls the fidelity of homologous recombination: roles for XRCC3 in late stages of recombination. *Molec. Cell* 10: 387–395, 2002.
- [41546] 15098. Liu, N.; Lamerdin, J. E.; Tebbs, R. S.; Schild, D.; Tucker, J. D.; Shen, M. R.; Brookman, K. W.; Siciliano, M. J.; Walter, C. A.; Fan, W.; Narayana, L. S.; Zhou, Z.-Q.; Adamson, A. W.; Sorensen, K. J.; Chen, D. J.; Jones, N. J.; Thompson, L. H.: XRCC2 and XRCC3, new human Rad51-family members, promote chromosome stability and protect against DNA cross-links and other damages. *Molec. Cell* 1: 783–793, 1998.
- [41547] 15099. Tebbs, R. S.; Zhao, Y.; Tucker, J. D.; Scheerer, J. B.; Siciliano, M. J.; Hwang, M.; Liu, N.; Legerski, R. J.; Thompson, L. H.: Correction of chromosomal instability and sensitivity to diverse mutagens by a cloned cDNA of the XRCC3 DNA repair gene. *Proc. Nat. Acad. Sci.* 92:6354–6358, 1995.
- [41548] 15100. Winsey, S. L.; Haldar, N. A.; Marsh, H. P.; Bunce, M.; Marshall, S. E.; Harris, A. L.; Wojnarowska, F.; Welsh, K. I.: A variant within the DNA repair gene XRCC3 is associated with the development of melanoma skin cancer. *Cancer*

Res. 60: 5612–5616, 2000.

- [41549] 15101. Hayashida, S.; Yamasaki, K.; Asada, Y.; Soeda, E.; Niikawa, N.; Kishino, T.: Construction of a physical and transcript map flanking the imprinted MEST/PEG1 region at 7q32. *Genomics* 66: 221–225, 2000.
- [41550] 15102. de Wind, N.; Dekker, M.; Claij, N.; Jansen, L.; van Klink, Y.; Radman, M.; Riggins, G.; van der Valk, M.; van't Wout, K.; te Riele, H.: HNPCC-like cancer predisposition in mice through simultaneous loss of Msh3 and Msh6 mismatch-repair protein functions. *Nature Genet.* 23:359–362, 1999.
- [41551] 15103. Braun-Dullaeus, R. C.; Mann, M. J.; Ziegler, A.; von der Leyen, H. E.; Dzau, V. J.: A novel role for the cyclin-dependent kinase inhibitor p27(Kip1) in angiotensin II-stimulated vascular smooth muscle cell hypertrophy. *J. Clin. Invest.* 104: 815–823, 1999.
- [41552] 15104. Carrano, A. C.; Eytan, E.; Hershko, A.; Pagano, M.: SKP2 is required for ubiquitin-mediated degradation of the CDK inhibitor p27. *Nature Cell Biol.* 1: 193–199, 1999.
- [41553] 15105. Di Cristofano, A.; De Acetis, M.; Koff, A.; Cordon-Cardo, C.; Pandolfi, P. P.: Pten and p27(KIP1) cooperate in prostate cancer tumor suppression in the mouse. *Nature Genet.* 27: 222–224, 2001.

- [41554] 15106.Fero, M. L.; Rivkin, M.; Tasch, M.; Porter, P.; Carow, C. E.; Firpo,E.; Polyak, K.; Tsai, L.-H.; Broudy, V.; Perlmutter, R. M.; Kaushansky,K.; Roberts, J. M.: A syndrome of multiorgan hyperplasia with featuresof gigantism, tumorigenesis, and female sterility in p27(Kip1)-deficientmice. *Cell* 85: 733–744, 1996.
- [41555] 15107.Albertella, M. R.; Campbell, R. D.: Characterization of a novelgene in the human major histocompatibility complex that encodes apotential new member of the I kappa B family of proteins. *Hum. Molec.Genet.* 3: 793–799, 1994.
- [41556] 15108.Buyse, I. M.; Shao, G.; Huang, S.: The retinoblastoma proteinbinds to RIZ, a zinc-finger protein that shares an epitope with theadenovirus E1A protein. *Proc. Nat. Acad. Sci.* 92: 4467–4471, 1995.
- [41557] 15109.Buyse, I. M.; Takahashi, E.; Huang, S.: Physical mapping of theretinoblastoma interacting zinc finger gene RIZ to D1S228 on chromosome1p36. *Genomics* 34: 119–121, 1996.
- [41558] 15110.Chadwick, R. B.; Jiang, G.-L.; Bennington, G. A.; Yuan, B.; Johnson,C. K.; Stevens, M. W.; Niemann, T. H.; Peltomaki, P.; Huang, S.; dela Chapelle, A.: Candidate tumor suppressor RIZ is frequently involvedin colorectal

carcinogenesis. Proc. Nat. Acad. Sci. 97: 2662–2667,2000.

[41559] 15111.Mock, B. A.; Coleman, M. P.; Huang, S.: Riz maps to distal chromosome4 near genes involved in tumorigenesis and nerve degeneration. MammalianGenome 7: 637 only, 1996.

[41560] 15112.Poetsch, M.; Dittberner, T.; Woenckhaus, C.: Frameshift mutations of RIZ, but no point mutations in RIZ1 exons in malignant melanomas with deletions in 1p36. Oncogene 21: 3038–3042, 2002.

[41561] 15113.Malek, N. P.; Sundberg, H.; McGrew, S.; Nakayama, K.; Kyriakidis, T. R.; Roberts, J. M.: A mouse knock-in model exposes sequential proteolytic pathways that regulate p27(Kip1) in G1 and S phase. Nature 413:323–327, 2001.

[41562] 15114.Martin, E.; Cacheux, V.; Cave, H.; Lapierre, J. M.; Le Paslier, D.; Grandchamp, B.: Localization of the CDKN4/p27(Kip1) gene to human chromosome 12p12.3. Hum. Genet. 96: 668–670, 1995.

[41563] 15115.Mitsuhashi, T.; Aoki, Y.; Eksioglu, Y. Z.; Takahashi, T.; Bhide, P. G.; Reeves, S. A.; Caviness, V. S., Jr.: Overexpression of p27(Kip1)lengthens the G1 phase in a mouse model that targets inducible gene expression to central nervous system progenitor cells. Proc. Nat. Acad. Sci. 98:

6435–6440, 2001.

- [41564] 15116. Polyak, K.; Lee, M.-H.; Erdjument-Bromage, H.; Koff, A.; Roberts, J. M.; Tempst, P.; Massague, J.: Cloning of p27(Kip1), a cyclin-dependent kinase inhibitor and a potential mediator of extracellular antimitogenic signals. *Cell* 78: 59–66, 1994.
- [41565] 15117. Saito, T.; Seki, N.; Hattori, A.; Hayashi, A.; Abe, M.; Araki, R.; Fujimori, A.; Fukumura, R.; Kozuma, S.; Matsuda, Y.: Structure, expression profile, and chromosomal location of a mouse gene homologous to human DNA-PK(cs) interacting protein (KIP) gene. *Mammalian Genome* 10: 315–317, 1999.
- [41566] 15118. Sherr, C. J.; Roberts, J. M.: Inhibitors of mammalian G1 cyclin-dependent kinases. *Genes Dev.* 9: 1149–1163, 1995.
- [41567] 15119. Toyoshima, H.; Hunter, T.: p27, a novel inhibitor of G1 cyclin-Cdk protein kinase activity, is related to p21. *Cell* 78: 67–74, 1994.
- [41568] 15120. Hudson, A. W.; Birnbaum, M. J.: Identification of a nonneuronal isoform of synaptotagmin. *Proc. Nat. Acad. Sci.* 92: 5895–5899, 1995.
- [41569] 15121. Chen, C.-C.; Akopian, A. N.; Sivilotti, L.; Colquhoun, D.; Burnstock, G.; Wood, J. N.: A P2X

purinoceptor expressed by a subset of sensory neurons.

Nature 377: 428–430, 1995.

- [41570] 15122. Cockayne, D. A.; Hamilton, S. G.; Zhu, Q.-M.; Dunn, P. M.; Zhong, Y.; Novakovic, S.; Malmberg, A. B.; Cain, G.; Berson, A.; Kassotakis, L.; Hedley, L.; Lachnit, W. G.; Burnstock, G.; McMahon, S. B.; Ford, A. P. D. W.: Urinary bladder hyporeflexia and reduced pain-related behaviour in P2X3-deficient mice. Nature 407: 1011–1015, 2000.
- [41571] 15123. Cook, S. P.; McCleskey, E. W.: ATP, pain and a full bladder. Nature 407: 951–952, 2000.
- [41572] 15124. Garcia-Guzman, M.; Stuhmer, W.; Soto, F.: Molecular characterization and pharmacological properties of the human P2X3 purinoceptor. Molec. Brain Res. 47: 59–66, 1997.
- [41573] 15125. Kennedy, C.; Leff, P.: Painful connection for ATP. Nature 377: 385–386, 1995.
- [41574] 15126. Lewis, C.; Neldhart, S.; Holy, C.; North, R. A.; Buell, G.; Surprenant, A.: Coexpression of P2X2 and P2X3 receptor subunits can account for ATP-gated currents in sensory neurons. Nature 377: 432–435, 1995.
- [41575] 15127. Souslova, V.; Cesare, P.; Ding, Y.; Akopian, A. N.; Stanfa, L.; Suzuki, R.; Carpenter, K.; Dickenson, A.; Boyce, S.; Hill, R.; Nebenius-Oosthuizen, D.; Smith, A. J. H.; Kidd,

E. J.; Wood, J. N.: Warm-coding deficits and aberrant inflammatory pain in mice lacking P2X3 receptors. *Nature* 407:1015–1017, 2000.

[41576] 15128. Souslova, V.; Ravenall, S.; Fox, M.; Wells, D.; Wood, J. N.; Akopian, A. N.: Structure and chromosomal mapping of the mouse P2X(3) gene. *Gene* 195:101–111, 1997.

[41577] 15129. Khakh, B. S.; Zhou, X.; Sydes, J.; Galligan, J. J.; Lester, H. A.: State-dependent cross-inhibition between transmitter-gated cation channels. *Nature* 406: 405–410, 2000.

[41578] 15130. Buell, G.; Lewis, C.; Collo, G.; North, R. A.; Surprenant, A.: An antagonist-insensitive P2X receptor expressed in epithelia and brain. *EMBO J.* 15: 55–62, 1996.

[41579] 15131. Buell, G.; Lewis, C.; Collo, G.; North, R. A.; Surprenant, A.: Cloning and expression of a family of ATP-gated ion channels. (Abstract) *Soc. Neurosci.* 21: 1262, 1995.

[41580] 15132. Garcia-Guzman, M.; Soto, F.; Gomez-Hernandez, J. M.; Lund, P. E.; Stuhmer, W.: Characterization of the recombinant human P2X4 receptor reveals pharmacological differences to the rat homologue. *Molec. Pharm.* 51: 109–118, 1997.

[41581] 15133. Chen, J. D.; Evans, R. M.: A transcriptional co-

repressor that interacts with nuclear hormone receptors.

Nature 377: 454–457, 1995.

[41582] 15134. Fischle, W.; Dequiedt, F.; Hendzel, M. J.; Guenther, M. G.; Lazar, M. A.; Voelter, W.; Verdin, E.: Enzymatic activity associated with class II HDACs is dependent on a multi-protein complex containing HDAC3 and SMRT/N-CoR.

Molec. Cell 9: 45–57, 2002.

[41583] 15135. Horlein, A. J.; Naar, A. M.; Heinzel, T.; Torchia, J.; Gloss, B.; Kurokawa, R.; Ryan, A.; Kamel, Y.; Soderstrom, M.; Glass, C. K.; Rosenfeld, M. G.: Ligand-independent repression by the thyroid hormone receptor mediated by a nuclear receptor co-repressor. Nature 377:

397–403, 1995.

[41584] 15136. Hu, X.; Lazar, M. A.: The CoRNR motif controls the recruitment of corepressors by nuclear hormone receptors. Nature 402: 93–96, 1999.

[41585] 15137. Jiang, Q.; Galiegue-Zouitina, S.; Roumier, C.; Hildebrand, M. P.; Thomas, S.; Coignet, L. J.: Genomic organization and refined mapping of the human nuclear corepressor 2 (NCOR2)/ silencing mediator of retinoid and thyroid hormone receptor (SMRT) gene on chromosome 12q24.3. Cytogenet. Cell Genet. 92: 217–220, 2001.

[41586] 15138. Ordentlich, P.; Downes, M.; Xie, W.; Genin, A.;

Spinner, N. B.; Evans, R. M.: Unique forms of human and mouse nuclear receptor corepressor SMRT. *Proc. Nat. Acad. Sci.* 96: 2639–2644, 1999.

- [41587] 15139. Colvin, J. S.; Green, R. P.; Schmahl, J.; Capel, B.; Ornitz, D.M.: Male-to-female sex reversal in mice lacking fibroblast growth factor 9. *Cell* 104: 875–889, 2001.
- [41588] 15140. Mattei, M.-G.; Penault-Llorca, F.; Coulier, F.; Birnbaum, D.: The human FGF9 gene maps to chromosomal region 13q11–q12. *Genomics* 29: 811–812, 1995.
- [41589] 15141. Miyamoto, M.; Naruo, K.-I.; Seko, C.; Matsumoto, S.; Kondo, T.; Kurokawa, T.: Molecular cloning of a novel cytokine cDNA encoding the ninth member of the fibroblast growth factor family, which has a unique secretion property. *Molec. Cell. Biol.* 13: 4251–4259, 1993.
- [41590] 15142. Garcia, J. G.; Lazar, V.; Gilbert-McClain, L. I.; Gallagher, P.J.; Verin, A. D.: Myosin light chain kinase in endothelium: molecular cloning and regulation. *Am. J. Resp. Cell Molec. Biol.* 16: 489–494, 1997.
- [41591] 15143. Giorgi, D.; Brand-Arpon, V.; Rouquier, S.: The functional myosin light chain kinase (MYLK) gene localizes with marker D3S3552 on human chromosome 3q21 in a greater than 5-Mb yeast artificial chromosomal region and is not linked to olfactory receptor genes. *Cytogenet. Cell*

Genet. 92: 85–88, 2001.

- [41592] 15144.Lazar, V.; Garcia, J. G. N.: A single human myosin light chainkinase gene (MLCK; MYLK) transcribes multiple nonmuscle isoforms. Genomics 57:256–267, 1999.
- [41593] 15145.Potier, M.–C.; Chelot, E.; Pekarsky, Y.; Gardiner, K.; Rossier,J.; Turnell, W. G.: The human myosin light chain kinase (MLCK) fromhippocampus: cloning, sequencing, expression, and localization to3cen–q21. Genomics 29: 562–570, 1995.
- [41594] 15146.Walker, L. A.; MacDonald, J. A.; Liu, X.; Nakamoto, R. K.; Haystead,T. A. J.; Somlyo, A. V.; Somlyo, A. P.: Site–specific phosphorylationand point mutations of telokin modulate its Ca(2+)–desensitizing effectin smooth muscle. J. Biol. Chem. 276: 24519–24524, 2001.
- [41595] 15147.Watterson, D. M.; Schavocky, J. P.; Guo, L.; Weiss, C.; Chlenski,A.; Shirinsky, V. P.; Van Eldik, L. J.; Haiech, J.: Analysis of thekinase–related protein gene found at human chromosome 3q21 in a multi–genecluster: organization, expression, alternative splicing, and polymorphic–marker. J. Cell. Biochem. 75: 481–491, 1999.
- [41596] 15148.Allcock, R. J. N.; Christiansen, F. T.; Price, P.: The centralMHC gene IKBL carries a structural polymorphism that is associatedwith HLA–A3,B7,DR15. Immunogenetics

49: 660–665, 1999.

- [41597] 15149. Druck, T.; Gu, Y.; Prabhala, G.; Cannizzaro, L. A.; Park, S.-H.; Huebner, K.; Keen, J. H.: Chromosome localization of human genes for clathrin adaptor polypeptides AP2-beta and AP50 and the clathrin-binding protein, VCP. *Genomics* 30: 94–97, 1995.
- [41598] 15150. Egerton, M.; Ashe, O. R.; Chen, D.; Druker, B. J.; Burgess, W. H.; Samelson, L. E.: VCP, the mammalian homolog of cdc48, is tyrosine phosphorylated in response to T cell antigen receptor activation. *EMBO J.* 11: 3533–3540, 1992.
- [41599] 15151. Hoyle, J.; Tan, K. H.; Fisher, E. M. C.: Mapping the valosin-containing protein (VCP) gene on human chromosome 9 and mouse chromosome 4, and a likely pseudogene on the mouse X chromosome. *Mammalian Genome* 8: 778–780, 1997.
- [41600] 15152. Koller, K. J.; Brownstein, M. J.: Use of a cDNA clone to identify a supposed precursor protein containing valosin. *Nature* 325: 542–545, 1987.
- [41601] 15153. Pleasure, I. T.; Black, M. M.; Keen, J. H.: Valosin-containing protein, VCP, is a ubiquitous clathrin-binding protein. *Nature* 365: 459–462, 1993.
- [41602] 15154. Ye, Y.; Meyer, H. H.; Rapoport, T. A.: The AAA AT-

Pase Cdc48/p97 and its partners transport proteins from the ER into the cytosol. *Nature* 414:652–656, 2001.

- [41603] 15155. Liu, Q.; Feng, Y.; Forgac, M.: Activity and in vitro reassembly of the coated vesicle (H⁺)-ATPase requires the 50-kDa subunit of the clathrin assembly complex AP-2. *J. Biol. Chem.* 269: 31592–31597, 1994.
- [41604] 15156. Thuriereau, C.; Brosius, J.; Burne, C.; Jolles, P.; Keen, J. H.; Mattaliano, R. J.; Chow, E. P.; Ramachandran, K. L.; Kirchhausen, T.: Molecular cloning and complete amino acid sequence of AP50, an assembly protein associated with clathrin-coated vesicles. *Cell* 7:663–669, 1988.
- [41605] 15157. Ponnambalam, S.; Robinson, M. S.; Jackson, A. P.; Peiperl, L.; Parham, P.: Conservation and diversity in families of coated vesicle adaptins. *J. Biol. Chem.* 265: 4814–4820, 1990.
- [41606] 15158. Robinson, M. S.: Cloning of cDNAs encoding two related 100-kDa coated vesicle proteins (alpha-adaptins). *J. Cell Biol.* 108: 833–42, 1989.
- [41607] 15159. Waelter, S.; Scherzinger, E.; Hasenbank, R.; Nordhoff, E.; Lurz, R.; Goehler, H.; Gauss, C.; Sathasivam, K.; Bates, G. P.; Lehrach, H.; Wanker, E. E.: The huntingtin interacting protein HIP1 is a clathrin and alpha-adaptin-binding protein involved in receptor-mediated

endocytosis. Hum.Molec. Genet. 10: 1807–1817, 2001.

[41608] 15160.Kaneko–Ishino, T.; Kuroiwa, Y.; Miyoshi, N.; Kohda, T.; Suzuki,R.; Yokoyama, M.; Viville, S.; Barton, S. C.; Ishino, F.; Surani,M. A.: Peg1/Mest imprinted gene on chromosome 6 identified by cDNAsubtraction hybridization. Nature Genet. 11: 52–59, 1995.

[41609] 15161.Kobayashi, S.; Kohda, T.; Miyoshi, N.; Kuroiwa, Y.; Aisaka, K.;Tsutsumi, O.; Kaneko–Ishino, T.; Ishino, F.: Human PEG1/MEST, an imprinted gene on chromosome 7. Hum. Molec. Genet. 6: 781–786, 1997.

[41610] 15162.Kosaki, K.; Kosaki, R.; Craigen, W. J.; Matsuo, N.: Isoform–specific imprinting of the human PEG1/MEST gene. (Letter) Am. J. Hum. Genet. 66:309–312, 2000.

[41611] 15163.Kotzot, D.; Schmitt, S.; Bernasconi, F.; Robinson, W. P.; Lurie,I. W.; Ilyina, H.; Mehes, K.; Hamel, B. C. J.; Otten, B. J.; Hergersberg,M.; Werder, E.; Shoenle, E.; Schinzel, A.: Uniparental disomy 7 in Silver–Russell syndrome and primordial growth retardation. Hum. Molec.Genet. 4: 583–587, 1995.

[41612] 15164.Lefebvre, L.; Viville, S.; Barton, S. C.; Ishino, F.; Keverne,E. B.; Surani, M. A.: Abnormal maternal behaviour and growth retardation associated with loss of the imprinted gene Mest. Nature Genet. 20:163–169, 1998.

- [41613] 15165.Lefebvre, L.; Viville, S.; Barton, S. C.; Ishino, F.; Surani, M.A.: Genomic structure and parent-of-origin-specific methylation of *Peg1*. *Hum. Molec. Genet.* 6: 1907–1915, 1997.
- [41614] 15166.Seki, T.; Tada, S.; Katada, T.; Enomoto, T.: Cloning of a cDNA encoding a novel importin- α homologue, *Qip1*: discrimination of *Qip1* and *Rch1* from *hSrp1* by their ability to interact with DNA helicase *Q1/RecQL*. *Biochem. Biophys. Res. Commun.* 234: 48–53, 1997.
- [41615] 15167.Ayala-Madriral, M. L.; Doerr, S.; Ramirez-Duenas, M. L.; Hansmann, I.: Assignment of karyopherin α 1 (*KPNA1*) to human chromosome band 3q21 by in situ hybridization. *Cytogenet. Cell Genet.* 90: 58–59, 2000.
- [41616] 15168.Conti, E.; Uy, M.; Leighton, L.; Blobel, G.; Kuriyan, J.: Crystallographic analysis of the recognition of a nuclear localization signal by the nuclear import factor karyopherin α . *Cell* 94: 193–204, 1998.
- [41617] 15169.Cortes, P.; Ye, Z.-S.; Baltimore, D.: RAG-1 interacts with the repeated amino acid motif of the human homologue of the yeast protein *SRP1*. *Proc. Nat. Acad. Sci.* 91: 7633–7637, 1994.
- [41618] 15170.Chen, H.; Antonarakis, S. E.: Localization of a human homolog of the mouse *Tiam-1* gene to chromosome

21q22.1. Genomics 30: 123–127,1995.

- [41619] 15171.Habets, G. G. M.; Scholtes, E. H. M.; Zuydgeest, D.; van der Kammen,R. A.; Stam, J. C.; Berns, A.; Collard, J. G.: Identification ofan invasion–inducing gene, Tiam–1, that encodes a protein with homologyto GDP–GTP exchangers for Rho–like proteins. Cell 77: 537–549, 1994.
- [41620] 15172.Habets, G. G. M.; van der Kammen, R. A.; Jenkins, N. A.; Gilbert,D. J.; Copeland, N. G.; Hagemeijer, A.; Collard, J. G.: The invasion–inducingTIAM1 gene maps to human chromosome band 21q22 and mouse chromosome16. Cytogenet. Cell. Genet. 70: 48–51, 1995.
- [41621] 15173.Habets, G. G. M.; van der Kammen, R. A.; Stam, J. C.; Michiels,F.; Collard, J. G.: Sequence of the human invasion–inducing TIAM1gene, its conservation in evolution and its expression in tumor celllines of different tissue origin. Oncogene 10: 1371–1376, 1995.
- [41622] 15174.Malliri, A.; van der Kammen, R. A.; Clark, K.; van der Valk, M.;Michiels, F.; Collard, J. G.: Mice deficient in the Rac activatorTiam1 are resistant to Ras–induced skin tumours. Nature 417: 867–871,2002.
- [41623] 15175.Ghetti, A.; Pinol–Roma, S.; Michael, W. M.; Morandi, C.; Dreyfuss,G.: HNRNP I, the polypyrimidine tract–binding protein: distinct nuclearlocalization and association with

hnRNAs. *Nucleic Acids Res.* 20:3671–3678, 1992.

[41624] 15176. Raimondi, E.; Romanelli, M. G.; Moralli, D.; Gamberi, C.; Russo, M. P.; Morandi, C.: Assignment of the human gene encoding heterogeneous nuclear RNA ribonucleoprotein I (PTB) to chromosome 14q23–q24.1. *Genomics* 27:553–555, 1995.

[41625] 15177. Romanelli, M. G.; Lorenzi, P.; Morandi, C.: Organization of the human gene encoding heterogeneous nuclear ribonucleoprotein type I (hnRNP I) and characterization of hnRNP I related pseudogene. *Gene* 255:267–272, 2000.

[41626] 15178. Hoogenraad, C. C.; Koekkoek, B.; Akhmanova, A.; Krugers, H.; Dortland, B.; Miedema, M.; van Alphen, A.; Kistler, W. M.; Jaegle, M.; Koutsourakis, M.; Van Camp, N.; Verhoye, M.; van der Linden, A.; Kaverina, I.; Grosveld, F.; De Zeeuw, C. I.; Galjart, N.: Targeted mutation of *Cyln2* in the Williams syndrome critical region links CLIP-115 haploinsufficiency to neurodevelopmental abnormalities in mice. *Nature Genet.* 32: 116–127, 2002. Note: Erratum: *Nature Genet.* 32: 331 only, 2002.

[41627] 15179. Gollob, M. H.; Green, M. S.; Roberts, R.: A gene responsible for familial Wolff–Parkinson–White syndrome. (Letter) *New Eng. J. Med.* 345:1063–1064, 2001.

[41628] 15180. L'Allemand, D.; Tardy, V.; Gruters, A.; Schnabel, D.;

Krude, H.;Morel, Y.: How a patient homozygous for a 30-kb deletion of the C4-CYP21 genomic region can have a nonclassic form of 21-hydroxylase deficiency. J.Clin. Endocr. Metab. 85: 4562-4567, 2000.

[41629] 15181.Lako, M.; Ramsden, S.; Campbell, R. D.; Strachan, T.: Mutationscreening in British 21-hydroxylase deficiency families and developmentof novel microsatellite based approaches to prenatal diagnosis. J.Med. Genet. 36: 119-124, 1999.

[41630] 15182.Lau, I. F.; Soardi, F. C.; Lemos-Marini, S. H. V.; Guerra, G.,Jr.; Baptista, M. T. M.; De Mello, M. P.: H28+C insertion in theCYP21 gene: a novel frameshift mutation in a Brazilian patient withthe classical form of 21-hydroxylase deficiency. J. Clin. Endocr.Metab. 86: 5877-5880, 2001.

[41631] 15183.Laue, L.; Merke, D. P.; Jones, J. V.; Barnes, K. M.; Hill, S.;Cutler, G. B.: A preliminary study of flutamide, testolactone, andreduced hydrocortisone dose in the treatment of congenital adrenalhyperplasia. J. Clin. Endocr. Metab. 81: 3535-3539, 1996.

[41632] 15184.Layrisse, A.; White, C.; Gunczler, P.; Gafaro Valera, L.; Arias,S.; Yunis, E. J.; Alper, C. A.; Awdeh, Z. L.: Sharing of MHC haplotypesamong apparently unrelated patients

with congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *Immunogenetics* 25: 99–103, 1987.

[41633] 15185. Lee, H.-H.; Chao, H.-T.; Ng, H.-T.; Choo, K.-B.: Direct molecular diagnosis of CYP21 mutations in congenital adrenal hyperplasia. *J. Med. Genet.* 33: 371–375, 1996.

[41634] 15186. Levine, L. S.; Dupont, B.; Lorenzen, F.; Pang, S.; Pollack, M.; Oberfield, S.; Kohn, B.; Lerner, A.; Cacciari, E.; Mantero, F.; Cassio, A.; Scaroni, C.; Chiumello, G.; Rondonani, G. F.; Gargantini, L.; Giovannelli, G.; Viridis, R.; Bartolotta, E.; Migliori, C.; Pintor, C.; Tato, L.; Barboni, F.; New, M. I.: Cryptic 21-hydroxylase deficiency in families of patients with classical congenital adrenal hyperplasia. *J. Clin. Endocr. Metab.* 51: 1316–1324, 1980.

[41635] 15187. Levine, L. S.; Dupont, B.; Lorenzen, F.; Pang, S.; Pollack, M.; Oberfield, S. E.; Kohn, B.; Lerner, A.; Cacciari, E.; Mantero, F.; Cassio, A.; Scaroni, C.; Chiumello, G.; Rondonani, G. F.; Gargantini, L.; Giovannelli, G.; Viridis, R.; Bartolotta, E.; Migliori, C.; Pintor, C.; Tato, L.; Barboni, F.; New, M. I.: Genetic and hormonal characterization of cryptic 21-hydroxylase deficiency. *J. Clin. Endocr. Metab.* 53: 1193–1198, 1981.

[41636] 15188. Levine, L. S.; Pang, S.; Dupont, B.; Pollack, M.; Lorenzen, F.; New, M. I.: Detection of heterozygote of

21-hydroxylase deficiency.(Letter) Lancet I: 603–604, 1980.

[41637] 15189.Levine, L. S.; Zachmann, M.; New, M. I.; Prader, A.; Pollack,M. S.; O'Neill, G. J.; Yang, S. Y.; Oberfield, S. E.; Dupont, B.:Genetic mapping of the 21-hydroxylase–deficiency gene within the HLAlinkage group. New Eng. J. Med. 299: 911–915, 1978.

[41638] 15190.Levo, A.; Partanen, J.: Mutation–haplotype analysis of steroid21-hydroxylase (CYP21) deficiency in Finland. Implications for thepopulation history of defective alleles. Hum. Genet. 99: 488–497,1997.

[41639] 15191.Lewis, V. G.; Money, J.; Epstein, R.: Concordance of verbal andnonverbal ability in the adrenogenital syn–drome. Johns Hopkins Med.J. 122: 192–195, 1968.

[41640] 15192.Libber, S. M.; Migeon, C. J.; Bias, W. B.: Ascertain–ment of 21-hydroxylasedeficiency in individuals with HLA–B14 haplotype. J. Clin. Endocr.Metab. 60: 727–730, 1985.

[41641] 15193.Lo, J. C.; Schwitzgebel, V. M.; Tyrrell, J. B.; Fitzger–ald, P.A.; Kaplan, S. L.; Conte, F. A.; Grumbach, M. M.: Normal female infantsborn of mothers with classic con–genital adrenal hyperplasia due to21-hydroxylase defi–ciency. J. Clin. Endocr. Metab. 84: 930–936,1999.

- [41642] 15194.Lopez-Gutierrez, A. U.; Riba, L.; Ordonez-Sanchez, M. L.; Ramirez-Jimenez,S.; Cerrillo-Hinojosa, M.; Tusie-Luna, M. T.: Uniparental disomyfor chromosome 6 results in steroid 21-hydroxylase deficiency: evidenceof different genetic mechanisms involved in the production of thedis-ease. *J. Med. Genet.* 35: 1014-1019, 1998.
- [41643] 15195.Lorenzen, F.; Pang, S.; New, M. I.; Dupont, B.; Pol-lack, M.; Chow,D. M.; Levine, L. S.: Hormonal phenotype and HLA-genotype in familiesof patients with congenital adrenal hyperplasia (21-hydroxylase deficiency). *Pe-diat.Res.* 13: 1356-1360, 1979.
- [41644] 15196.Lorenzen, F.; Pang, S.; New, M. I.; Oberfield, S.; Dupont, B.;Chow, D.; Schneider, B.; Levine, L.: Studies of the C-21 and C-19steroids and HLA genotyping in sib-lings and parents of patients withcongenital adrenal hy-perplasia due to 21-hydroxylase deficiency. *J.Clin. Endocr. Metab.* 50: 572-577, 1980.
- [41645] 15197.Lubani, M. M.; Issa, A.-R. A.; Bushnaq, R.; Al-Saleh, Q. A.; Dudin,K. I.; Reavey, P. C.; El-Khalifa, M. Y.; Manand-har, D. S.; Abdul Al,Y. K.; Ismail, E. A.; Teebi, A. S.: Preva-lence of congenital adrenalhyperplasia in Kuwait. *Europ. J. Pediat.* 149: 391-392, 1990.
- [41646] 15198.Matteson, K. J.; Phillips, J. A., III; Miller, W. L.;

Chung, B.-C.;Orlando, P. J.; Frisch, H.; Ferrandez, A.; Burr, I. M.: P450XXI (steroid21-hydroxylase) gene deletions are not found in family studies ofcongenital adrenal hyperplasia. Proc. Nat. Acad. Sci. 84: 5858-5862,1987.

[41647] 15199.McAlpine, P. J.: Personal Communication. Winnipeg, Manitoba,Canada 2/26/1988.

[41648] 15200.McGuire, L. S.; Omenn, G. S.: Congenital adrenal hyperplasia.I. Family studies of IQ. Behav. Genet. 5: 165-173, 1975.

[41649] 15201.Merkatz, I. R.; New, M. I.; Peterson, R. E.; Seaman, M. P.: Prenataldiagnosis of adrenogenital syndrome by amniocentesis. J. Pediat. 75:977-982, 1969.

[41650] 15202.Merke, D. P.; Chrousos, G. P.; Eisenhofer, G.; Weise, M.; Keil,M. F.; Rogol, A. D.; Van Wyk, J. J.; Bornstein, S. R.: Adrenomedullarydysplasia and hypofunction in patients with classic 21-hydroxylasedeficiency. New Eng. J. Med. 343: 1362-1368, 2000.

[41651] 15203.Merke, D. P.; Keil, M. F.; Jones, J. V.; Fields, J.; Hill, S.;Cutler, G. B., Jr.: Flutamide, testolactone, and reduced hydrocortisonedose maintain normal growth velocity and bone maturation despite elevatedandrogen levels in children with congenital adrenal hyperplasia. J.Clin. Endocr. Metab. 85: 1114-1120, 2000.

- [41652] 15204.Meyer-Bahlburg, H. F. L.: What causes low rates of child-bearing in congenital adrenal hyperplasia? J. Clin. Endocr. Metab. 84: 1844–1847, 1999.
- [41653] 15205.Miller, W. L.: Gene conversions, deletions and polymorphisms in congenital adrenal hyperplasia. Am. J. Hum. Genet. 42: 4–7, 1988.
- [41654] 15206.Miller, W. L.: Personal Communication. San Francisco, Calif. 1/3/1996.
- [41655] 15207.Bannerman, R. M.; Edwards, J. A.; Kreimer-Birnbaum, M.; McFarland, E.; Russell, E. S.: Hereditary microcytic anaemia in the mouse; studies in iron distribution and metabolism. Brit. J. Haemat. 23: 235–245, 1972.
- [41656] 15208.Vincent, J. B.; Herbrick, J.-A.; Gurling, H. M. D.; Bolton, P.F.; Roberts, W.; Scherer, S. W.: Identification of a novel gene on chromosome 7q31 that is interrupted by a translocation breakpoint in an autistic individual. Am. J. Hum. Genet. 67: 510–514, 2000.
- [41657] 15209.Barichard, F.; Joulin, V.; Henry, I.; Garel, M.-C.; Valentin, C.; Rosa, R.; Cohen-Solal, M.; Junien, C.: Chromosomal assignment of the human 2,3-bisphosphoglycerate mutase gene (BPGM) to region 7q34–7q22. Hum. Genet. 77: 283–285, 1987.
- [41658] 15210.Bowdler, A. J.; Prankerd, T. A. J.: Studies in congen-

ital non-spherocythaemolytic anaemias with specific enzyme defects. *Acta Haemat.* 31:65–78, 1964.

- [41659] 15211.Chen, S.-H.; Anderson, J. E.; Giblett, E. R.: 2,3-diphosphoglyceratemutase: its demonstration by electrophoresis and the detection of a genetic variant. *Biochem. Genet.* 5: 481–486, 1971.
- [41660] 15212.Galacteros, F.; Rosa, R.; Prehu, M. O.; Najean, Y.; Calvin, M.C.: Deficit en diphosphoglycerate mutase: nouveaux cas associes a une polyglobulie. *Nouv. Rev. Franc. Hemat.* 26: 69–74, 1984.
- [41661] 15213.Joulin, V.; Barichard, F.; Henry, I.; Garel, M. C.; Valentin, C.; Rosa, R.; Cohen-Solal, M.; Junien, C.: Chromosomal assignment of the human 2,3-bisphosphoglycerate mutase gene (BPGM) to region 7q22–7q34.(Abstract) *Cytogenet. Cell Genet.* 46: 635 only, 1987.
- [41662] 15214.Joulin, V.; Garel, M.-C.; Le Boulch, P.; Valentin, C.; Rosa, R.; Rosa, J.; Cohen-Solal, M.: Isolation and characterization of the human 2,3-bisphosphoglycerate mutase gene. *J. Biol. Chem.* 263: 15785–15790, 1988.
- [41663] 15215.Joulin, V.; Peduzzi, J.; Romeo, P.-H.; Rosa, R.; Valentin, C.; Dubart, A.; Lapeyre, B.; Blouquit, Y.; Garel, M.-C.; Goossens, M.; Rosa, J.; Cohen-Solal, M.: Molecular

cloning and sequencing of the human erythrocyte

2,3-bisphosphoglycerate mutase cDNA: revised amino acid sequence. EMBO J. 5: 2275–2283, 1986.

[41664] 15216. Labie, D.; Leroux, J.-P.; Najman, A.; Reyrolle, C.:

Familial diphosphoglycerate mutase deficiency: influence on the oxygen affinity curves of hemoglobin. FEBS Lett. 9: 37–40, 1970.

[41665] 15217. Lemarchandel, V.; Joulin, V.; Valentin, C.; Rosa, R.;

Galacteros, F.; Rosa, J.; Cohen-Solal, M.: Compound heterozygosity in a complete erythrocyte bisphosphoglycerate mutase deficiency. Blood 80: 2643–2649, 1992.

[41666] 15218. Rosa, R.; Audit, I.; Rosa, J.: Diphosphoglycerate

mutase and 2,3-diphosphoglycerate phosphatase activities of red cells: comparative electrophoretic study. Biochem. Biophys. Res. Commun. 51: 536–542, 1973.

[41667] 15219. Rosa, R.; Blouquit, Y.; Calvin, M.-C.; Prome, D.;

Prome, J.-C.; Rosa, J.: Isolation, characterization, and structure of a mutant 89 arg-to-cys bisphosphoglycerate mutase: implication of the active site in the mutation. J. Biol. Chem. 264: 7837–7843, 1989.

[41668] 15220. Rosa, R.; Prehu, M.-O.; Beuzard, Y.; Rosa, J.: The

first case of a complete deficiency of diphosphoglycerate mutase in human erythrocytes. J. Clin. Invest. 62:

907–915, 1978.

- [41669] 15221.Sasaki, R.; Ikura, K.; Sugimoto, E.; Chiba, H.: Purification of bisphosphoglyceromutase, 2,3-bisphosphoglycerate phosphatase and phosphoglyceromutase from human erythrocytes. *Europ. J. Biochem.* 50:581–593, 1975.
- [41670] 15222.Schroter, W.: Kongenitale nichtsphaerocytaere haemolytische Anaemie bei 2,3-Diphosphoglyceratmutasemangel der Erythrocyten im fruehen Sauglingsalter. *Klin. Wschr.* 43: 1147–1153, 1965.
- [41671] 15223.Scott, E. M.; Wright, R. C.: An alternate method for demonstration of bisphosphoglyceromutase (DPGM) on starch gels. *Am. J. Hum. Genet.* 34:1013–1015, 1982.
- [41672] 15224.Yanagawa, S.; Hitomi, K.; Sasaki, R.; Chiba, H.: Isolation and characterization of cDNA encoding rabbit reticulocyte 2,3-bisphosphoglycerate synthase. *Gene* 44: 185–191, 1986.
- [41673] 15225.Cali, J. J.; Hsieh, C.-L.; Francke, U.; Russell, D. W.: Mutations in the bile acid biosynthetic enzyme sterol 27-hydroxylase underlie cerebrotendinous xanthomatosis. *J. Biol. Chem.* 266: 7779–7783, 1991.
- [41674] 15226.Cali, J. J.; Russell, D. W.: Characterization of human

sterol27-hydroxylase: a mitochondrial cytochrome P-450 that catalyzes multipleoxidation reactions in bile acid biosynthesis. J. Biol. Chem. 266:7774-7778, 1991.

[41675] 15227.Jaspers, M.; Zhang, Z.; Marynen, P.; Vekemans, S.; Aly, M. S.;Cuppens, H.; Hillicker, C.; Cassiman, J.-J.: Localization of thegenes encoding the alpha-2 and alpha-4 subunits of the human VLA-receptorsto chromosome 5q23-31 and 2q31-32 respectively. (Abstract) Cyto-genet.Cell Genet. 58: 1870 only, 1991.

[41676] 15228.Kaplan, C.; Morel-Kopp, M. C.; Kroll, H.; Kiefel, V.; Schlegel,N.; Chesnel, N.; Mueller-Eckhardt, C.: HPA-5b (Br-a) neonatal alloimmunethrombocytopenia: clinical and immunological analysis of 39 cases. Brit.J. Haemat. 78: 425-429, 1991.

[41677] 15229.Kiefel, V.; Santoso, S.; Katzmann, B.; Mueller-Eckhardt, C.: Anew platelet-specific alloantigen Br(a): report of 4 cases with neonatalalloimmune thrombocytopenia. Vox Sang. 54: 101-106, 1988.

[41678] 15230.Kiefel, V.; Shechter, Y.; Atias, D.; Kroll, H.; Santoso, S.; Mueller-Eckhardt,C.: Neonatal alloimmune thrombocytopenia due to anti-Br(b) (HPA-5a):report of three cases in two families. Vox Sang. 60: 244-245, 1991.

[41679] 15231.Kritzik, M.; Savage, B.; Nugent, D. J.; Santoso, S.;

Ruggeri, Z.M.; Kunicki, T. J.: Nucleotide polymorphisms in the alpha-2 gene define multiple alleles that are associated with differences in platelet alpha-2/beta-1 density. *Blood* 92: 2382–2388, 1998.

[41680] 15232. Kunicki, T. J.; Kritzik, M.; Annis, D. S.; Nugent, D. J.: Hereditary variation in platelet integrin alpha-2-beta-1 density is associated with two silent polymorphisms in the alpha-2 gene coding sequence. *Blood* 89:1939–1943, 1997.

[41681] 15233. Mueller-Eckhardt, C.; Kiefel, V.; Grubert, A.; Kroll, H.; Weisheit, M.; Schmidt, S.; Mueller-Eckhardt, G.; Santos, S.: 348 cases of suspected neonatal alloimmune thrombocytopenia. *Lancet* I: 363–366, 1989.

[41682] 15234. Nieuwenhuis, H. K.; Akkerman, J. W. N.; Houdijk, W. P.; Sixma, J. J.: Human blood platelets showing no response to collagen fail to express surface glycoprotein Ia. *Nature* 318: 470–472, 1985.

[41683] 15235. Santos, S.; Amrhein, J.; Hofmann, H. A.; Sachs, U. J. H.; Walka, M. M.; Kroll, H.; Kiefel, V.: A point mutation thr799met on the alpha-2 integrin leads to the formation of new human platelet alloantigen Sit(a) and affects collagen-induced aggregation. *Blood* 94: 4103–4111, 1999.

[41684] 15236. Santos, S.; Kalb, R.; Walka, M.; Kiefel, V.; Mueller-

Eckhardt,C.; Newman, P. J.: The human platelet alloantigens Br(a) and Br(b)are associated with a single amino acid polymorphism on glycoproteinIa (integrin subunit alpha-2). J. Clin. Invest. 92: 2427–2432, 1993.

[41685] 15237.Santoso, S.; Kunicki, T. J.; Kroll, H.; Haberbosch, W.; Gardemann,A.: Association of the platelet glycoprotein Ia C807T gene polymorphismwith nonfatal myocardial infarction in younger patients. Blood 93:2449–2453, 1999.

[41686] 15238.Takada, Y.; Hemler, M. E.: The primary structure of the VLA-2/collagenreceptor alpha-2 subunit (platelet GPIa): homology to other integrinsand the presence of a possible collagen-binding domain. J. Cell Biol. 109:397–407, 1989.

[41687] 15239.von Beckerath, N.; Koch, W.; Mehilli, J.; Bottiger, C.; Schomig,A.; Kastrati, A.: Glycoprotein Ia gene C807T polymorphism and riskfor major adverse cardiac events within the first 30 days after coronaryartery stenting. Blood 95: 3297–3301, 2000.

[41688] 15240.Fernandez-Ruiz, E.; Pardo-Manuel de Villena, F.; Rubio, M. A.;Corbi, A. L.; Rodriguez de Cordoba, S.; Sanchez-Madrid, F.: Mappingof the human VLA-alpha-4 gene to chromosome 2q31-q32. Europ. J. Immun. 22:587–590, 1992.

- [41689] 15241. Roseblatt, M.; Vuillet-Gaugler, M. H.; Leroy, C.; Coulombel, L.: Coexpression of two fibronectin receptors, VLA-4 and VLA-5, by immature human erythroblastic precursor cells. *J. Clin. Invest.* 87: 6-11, 1991.
- [41690] 15242. Zhang, Z.; Vekemans, S.; Aly, M. S.; Jaspers, M.; Marynen, P.; Cassiman, J.-J.: The gene for the alpha-4 subunit of the VLA-4 integrin maps to chromosome 2q31-32. *Blood* 78: 2396-2399, 1991.
- [41691] 15243. Hiesberger, T.; Trommsdorff, M.; Howell, B. W.; Goffinet, A.; Mumby, M. C.; Cooper, J. A.; Herz, J.: Direct binding of reelin to VLDL receptor and apoE receptor 2 induces tyrosine phosphorylation of disabled-1 and modulates tau phosphorylation. *Neuron* 24: 481-489, 1999.
- [41692] 15244. Oka, K.; Tzung, K.-W.; Sullivan, M.; Lindsay, E.; Baldini, A.; Chan, L.: Human very-low-density lipoprotein receptor complementary DNA and deduced amino acid sequence and localization of its gene (VLDLR) to chromosome band 9q24 by fluorescence in situ hybridization. *Genomics* 20: 298-300, 1994.
- [41693] 15245. Okuizumi, K.; Onodera, O.; Namba, Y.; Ikeda, K.; Yamamoto, T.; Seki, K.; Ueki, A.; Nanko, S.; Tanaka, H.; Takahashi, H.; Oyanagi, K.; Mizusawa, H.; Kanazawa, I.; Tsuji, S.: Genetic association of the very low density

lipoprotein (VLDL) receptor gene with sporadic Alzheimer's disease. *Nature Genet.* 11: 207–209, 1995.

[41694] 15246. Gafvels, M. E.; Paavola, L. G.; Boyd, C. O.; Nolan, P. M.; Wittmaack, F.; Chawla, A.; Lazar, M. A.; Bucan, M.; Angelin, B.; Strauss, J. F., III: Cloning of a complementary deoxyribonucleic acid encoding the murine homolog of the very low density lipoprotein/apolipoprotein-E receptor: expression pattern and assignment of the gene to mouse chromosome 19. *Endocrinology* 135: 387–394, 1994.

[41695] 15247. Sakai, J.; Hoshino, A.; Takahashi, S.; Miura, Y.; Ishii, H.; Suzuki, H.; Kawarabayashi, Y.; Yamamoto, T.: Structure, chromosome location, and expression of the human very low density lipoprotein receptor gene. *J. Biol. Chem.* 269: 2173–2182, 1994.

[41696] 15248. Trommsdorff, M.; Gotthardt, M.; Hiesberger, T.; Shelton, J.; Stockinger, W.; Nimpf, J.; Hammer, R. E.; Richardson, J. A.; Herz, J.: Reeler/Disabled-like disruption of neuronal migration in knockout mice lacking the VLDL-receptor and ApoE receptor 2. *Cell* 97: 689–701, 1999.

[41697] 15249. Huebner, K.; Druck, T.; Croce, C. M.; Thiesen, H. J.: Twenty-seven nonoverlapping zinc finger cDNAs from human T cells map to nine different chromosomes with apparent clustering. *Am. J. Hum. Genet.* 48: 726–740, 1991.

- [41698] 15250. Rousseau-Merck, M.-F.; Hillion, J.; Jonveaux, P.; Couillin, P.; Seite, P.; Thiesen, H.-J.; Berger, R.: Chromosomal localization of 9 KOX zinc finger genes: physical linkages suggest clustering of KOX genes on chromosomes 12, 16, and 19. *Hum. Genet.* 92: 583-587, 1993.
- [41699] 15251. Bray, P.; Lichter, P.; Thiesen, H.-J.; Ward, D. C.; Dawid, I. B.: Characterization and mapping of human genes encoding zinc finger proteins. *Proc. Nat. Acad. Sci.* 88: 9563-9567, 1991.
- [41700] 15252. Lehmann, O. J.; El-ashry, M. F.; Ebenezer, N. D.; Ocaka, L.; Francis, P. J.; Wilkie, S. E.; Patel, R. J.; Ficker, L.; Jordan, T.; Khaw, P. T.; Bhattacharya, S. S.: A novel keratocan mutation causing autosomal recessive cornea plana. *Invest. Ophthalm. Vis. Sci.* 42: 3118-3122, 2001.
- [41701] 15253. Bermingham, N.; Hernandez, D.; Balfour, A.; Gilmour, F.; Martin, J. E.; Fisher, E. M. C.: Mapping TNNC1, the gene that encodes cardiac troponin I in the human and mouse. *Genomics* 30: 620-622, 1995.
- [41702] 15254. Roher, A.; Lieska, N.; Spitz, W.: The amino acid sequence of human cardiac troponin-C. *Muscle Nerve* 9: 73-77, 1986.
- [41703] 15255. Romero-Herrera, A. E.; Castillo, O.; Lehmann, H.: Human skeletal muscle proteins: the primary structure of

troponin C. J. Molec. Evol. 8:251–270, 1976.

- [41704] 15256. Schreier, T.; Kedes, L.; Gahlmann, R.: Cloning, structural analysis, and expression of the human slow twitch skeletal muscle/cardiac troponin C gene. J. Biol. Chem. 265: 21247–21253, 1990.
- [41705] 15257. Song, W.-J.; Van Keuren, M. L.; Drabkin, H. A.; Cypser, J. R.; Gemmill, R. M.; Kurnit, D. M.: Assignment of the human slow twitch skeletal muscle/cardiac troponin C gene (TNNC1) to human chromosome 3p21.3–3p14.3 using somatic cell hybrids. Cytogenet. Cell Genet. 75:36–37, 1996.
- [41706] 15258. Song, W. J.: Personal Communication. Ann Arbor, Mich. 1/17/1997.
- [41707] 15259. Townsend, P. J.; Yacoub, M. H.; Barton, P. J. R.: Assignment of the human cardiac/slow skeletal muscle troponin C gene (TNNC1) between D3S3118 and GCT4B10 on the short arm of chromosome 3 by somatic cell hybrid analysis. Ann. Hum. Genet. 61: 375–377, 1997.
- [41708] 15260. Haldane, J. B. S.; Sprunt, A. D.; Haldane, N. M.: Reduplication in mice. J. Genet. 5: 133–135, 1915.
- [41709] 15261. Lyon, M. F.; King, T. R.; Gondo, Y.; Gardner, J. M.; Nakatsu, Y.; Eicher, E. M.; Brilliant, M. H.: Genetic and molecular analysis of recessive alleles at the pink-eyed di-

lution (p) locus of the mouse. *Proc.Nat. Acad. Sci.* 89: 6968–6972, 1992.

- [41710] 15262.Stohl, W.; Kunkel, H. G.: Heterogeneity in expression of theT4 epitope in black individuals. *Scand. J. Immun.* 20: 273–278, 1984.
- [41711] 15263.Tishkoff, S. A.; Dietzsch, E.; Speed, W.; Pakstis, A. J.; Kidd,J. R.; Cheung, K.; Bonne–Tamir, B.; Santachiara–Benerecetti, A. S.;Moral, P.; Krings, M.; Paabo, S.; Watson, E.; Risch, N.; Jenkins,T.; Kidd, K. K.: Global patterns of linkage disequilibrium at theCD4 locus and modern human origins. *Science* 271: 1380–1387, 1996.
- [41712] 15264.van Dongen, J. J. M.; Wolvers–Tettero, I. L. M.; Versnel, M. A.;Westerveld, A.; Geurts van Kessel, A. H. M.: Assignment of the genes coding for the T–cell antigens CD7 (Tp41), CD5 (T1) and CD4 (T4) to human chromosome 17, 11, 12 respectively. (Abstract) *Cytogenet. CellGenet.* 40: 767, 1985.
- [41713] 15265.Zou, Y.–R.; Sunshine, M.–J.; Taniuchi, I.; Hatam, F.; Killeen,N.; Littman, D. R.: Epigenetic silencing of CD4 in T cells committed to the cytotoxic lineage. *Nature Genet.* 332–336, 2001.
- [41714] 15266.DiLella, A. G.: Chromosomal assignment of the human immunophilinFKBP–12 gene. *Biochem. Biophys. Res.*

Commun. 179: 1427–1433, 1991.

- [41715] 15267. DiLella, A. G.; Hawkins, A.; Craig, R. J.; Schreiber, S. L.; Griffin, C. A.: Chromosomal band assignments of the genes encoding human FKBP12 and FKBP13. Biochem. Biophys. Res. Commun. 189: 819–823, 1992.
- [41716] 15268. Goebel, M. G.: The peptidyl–prolyl isomerase, FK506–binding protein, is most likely the 12 kd endogenous inhibitor 2 of protein kinase C. (Letter) Cell 64: 1051–1052, 1991.
- [41717] 15269. Jin, Y.–J.; Albers, M. W.; Lane, W. S.; Bierer, B. E.; Schreiber, S. L.; Burakoff, S. J.: Molecular cloning of a membrane–associated human FK506– and rapamycin–binding protein, FKBP–13. Proc. Nat. Acad. Sci. 88: 6677–6681, 1991.
- [41718] 15270. Maki, N.; Sekiguchi, F.; Nishimaki, J.; Miwa, K.; Hayano, T.; Takahashi, N.; Suzuki, M.: Complementary DNA encoding the human T–cell FK506–binding protein, a peptidylprolyl cis–trans isomerase distinct from cyclophilin. Proc. Nat. Acad. Sci. 87: 5440–5443, 1990.
- [41719] 15271. Peattie, D. A.; Hsiao, K.; Benasutti, M.; Lippke, J. A.: Three distinct messenger RNAs can encode the human immunosuppressant–binding protein FKBP12. Gene 150: 251–257, 1994.

- [41720] 15272.Shou, W.; Aghdasi, B.; Armstrong, D. L.; Guo, Q.; Bao, S.; Charng,M.-J.; Mathews, L. M.; Schneider, M. D.; Hamilton, S. L.; Matzuk,M. M.: Cardiac defects and altered ryanodine receptor function in mice lacking FKBP12. Nature 391: 489–492, 1998.
- [41721] 15273.Standaert, R. F.; Galat, A.; Verdine, G. L.; Schreiber, S. L.:Molecular cloning and overexpression of the human FK506–binding proteinFKBP. Nature 346: 671–674, 1990.
- [41722] 15274.Wang, T.; Donahoe, P. K.; Zervos, A. S.: Specific interaction of type I receptors of the TGF–beta family with the immunophilin FKBP–12. Science 265:674–676, 1994.
- [41723] 15275.Jin, Y. J.; Albers, M. W.; Lane, W. S.; Bierer, B. E.; Schreiber,S. L.; Burakoff, S. J.: Molecular cloning of a membrane–associated human FK506– and rapamycin–binding protein, FKBP–13. Proc. Nat. Acad.Sci. 88: 6677–6681, 1991.
- [41724] 15276.Howard, H. C.; Mount, D. B.; Rochefort, D.; Byun, N.; Dupre, N.;Lu, J.; Fan, X.; Song, L.; Riviere, J.–B.; Prevost, C.; Horst, J.;Simonati, A.; and 12 others: The K–Cl cotransporter KCC3 is mutant in a severe peripheral neuropathy associated with agenesis of the corpus callosum. Nature Genet. 7Oct, 2002. Note: Advance Electronic Publication.

- [41725] 15277.Haire, R. N.; Litman, G. W.: The murine form of TXK, a novel TECkinase expressed in thymus maps to chromosome 5. *Mammalian Genome* 6:476–480, 1995.
- [41726] 15278.Haire, R. N.; Ohta, Y.; Lewis, J. E.; Fu, S. M.; Kroisel, P.; Litman,G. W.: TXK, a novel human tyrosine kinase expressed in T cells sharessequence identity with Tec family kinases and maps to 4p12. *Hum.Molec. Genet.* 3: 897–901, 1994.
- [41727] 15279.Schneider, H.; Schwartzberg, P. L.; Rudd, C. E.: Resting lymphocytekinase (Rlk/Txk) phosphorylates the YVKM motif and regulates PI 3–kinasebinding to T–cell antigen CTLA–4. *Biochem. Biophys. Res. Commun.* 252:14–19, 1998.
- [41728] 15280.Takeba, Y.; Nagafuchi, H.; Takeno, M.; Kashiwakura, J.; Suzuki,N.: Txk, a member of nonreceptor tyrosine kinase of Tec family, actsas a Th1 cell–specific transcription factor and regulates IFN–gammagene transcription. *J. Immun.* 168: 2365–2370, 2002.
- [41729] 15281.Al–Maghtheh, M.; Vithana, E.; Tarttelin, E.; Jay, M.; Evans, K.;Moore, T.; Bhattacharya, S.; Inglehearn, C. F.: Evidence for a majorretinitis pigmentosa locus on 19q13.4 (RP11), and association witha unique bimodal expressivity phenotype. *Am. J. Hum. Genet.* 59:864–871, 1996.

- [41730] 15282. Gasdaska, J. R.; Gasdaska, P. Y.; Gallegos, A.; Powis, G.: Human thioredoxin reductase gene localization to chromosomal position 12q23–q24.1 and mRNA distribution in human tissue. *Genomics* 37: 257–259, 1996.
- [41731] 15283. Gasdaska, P. Y.; Gasdaska, J. R.; Cochran, S.; Powis, G.: Cloning and sequencing of a human thioredoxin reductase. *FEBS Lett.* 373: 5–9, 1995.
- [41732] 15284. Svaren, J.; Apel, E. D.; Simburger, K. S.; Jenkins, N. A.; Gilbert, D. J.; Copeland, N. A.; Milbrandt, J.: The Nab2 and Stat6 genes share a common transcription termination region. *Genomics* 41: 33–39, 1997.
- [41733] 15285. Weichenrieder, O.; Wild, K.; Strub, K.; Cusack, S.: Structure and assembly of the Alu domain of the mammalian signal recognition particle. *Nature* 408: 167–173, 2000.
- [41734] 15286. Nichols, R. C.; Blinder, J.; Pai, S. I.; Ge, Q.; Targoff, I. N.; Plotz, P. H.; Liu, P.: Assignment of two human autoantigen genes: isoleucyl-tRNA synthetase locates to 9q21 and lysyl-tRNA synthetase locates to 16q23–q24. *Genomics* 36: 210–213, 1996.
- [41735] 15287. Nichols, R. C.; Raben, N.; Boerkoel, C. F.; Plotz, P. H.: Human isoleucyl-tRNA synthetase: sequence of the cDNA, alternative mRNA splicing, and the characteristics of

an unusually long C-terminal extension. Gene 155: 299–304, 1995.

- [41736] 15288. Dejgaard, K.; Leffers, H.; Rasmussen, H. H.; Madsen, P.; Kruse, T. A.; Gesser, B.; Nielsen, H.; Celis, J. E.: Identification, molecular cloning, expression and chromosome mapping of a family of transformation upregulated hnRNP-K proteins derived by alternative splicing. J. Molec. Biol. 236: 33–48, 1994.
- [41737] 15289. Tommerup, N.; Leffers, H.: Assignment of human KH-box-containing genes by in situ hybridization: HNRNP-K maps to 9q21.32–q21.33, PCBP1 to 2p12–p13, and PCBP2 to 12q13.12–q13.13, distal to FRA12A. Genomics 32:297–298, 1996.
- [41738] 15290. Agarwal, A. K.; Monder, C.; Eckstein, B.; White, P. C.: Cloning and expression of rat cDNA encoding corticosteroid 11-beta-dehydrogenase. J. Biol. Chem. 264: 18939–18943, 1989.
- [41739] 15291. Agarwal, A. K.; Rogerson, F. M.; Mune, T.; White, P. C.: Gene structure and chromosomal localization of the human HSD11K gene encoding the kidney (type 2) isozyme of 11-beta-hydroxysteroid dehydrogenase. Genomics 29:195–199, 1995.
- [41740] 15292. Lakshmi, V.; Monder, C.: Purification and charac-

terization of the corticosteroid 11- β -dehydrogenase component of the rat liver 11- β -hydroxysteroid dehydrogenase complex. *Endocrinology* 123:2390–2398, 1988.

[41741] 15293. Kotelevtsev, Y.; Holmes, M. C.; Burchell, A.; Houston, P. M.; Schmall, D.; Jamieson, P.; Best, R.; Brown, R.; Edwards, C. R. W.; Seckl, J. R.; Mullins, J. J.: 11- β -hydroxysteroid dehydrogenase type 1 knockout-mice show attenuated glucocorticoid-inducible responses and resist hyperglycemia on obesity or stress. *Proc. Nat. Acad. Sci.* 94: 14924–14929, 1997.

[41742] 15294. Masuzaki, H.; Paterson, J.; Shinyama, H.; Morton, N. M.; Mullins, J. J.; Seckl, J. R.; Flier, J. S.: A transgenic model of visceral obesity and the metabolic syndrome. *Science* 294: 2166–2170, 2001.

[41743] 15295. Morton, N. M.; Holmes, M. C.; Fievet, C.; Staels, B.; Tailleux, A.; Mullins, J. J.; Seckl, J. R.: Improved lipid and lipoprotein profile, hepatic insulin sensitivity, and glucose tolerance in 11- β -hydroxysteroid dehydrogenase type 1 null mice. *J. Biol. Chem.* 276: 41293–41300, 2001.

[41744] 15296. Ricketts, M. L.; Verhaeg, J. M.; Bujalska, I.; Howie, A. J.; Rainey, W. E.; Stewart, P. M.: Immunohistochemical localization of type 11- β -hydroxysteroid dehydroge-

nase in human tissues. J.Clin. Endocr.Metab. 83:
1325–1335, 1998.

- [41745] 15297.Alessi, D. R.; Smythe, C.; Keyse, S. M.: The human CL100 gene encodes a tyr/thr–protein phosphatase which potently and specifically inactivates MAP kinase and suppresses its activation by oncogenic ras in Xenopus oocyte extracts. Oncogene 8: 2015–2020, 1993.
- [41746] 15298.Brondello, J.–M.; Pouyssegur, J.; McKenzie, F. R.: Reduced MAP kinase phosphatase–1 degradation after p42/p44(MAPK)–dependent phosphorylation. Science 286:2514–2517, 1999.
- [41747] 15299.Emslie, E. A.; Jones, T. A.; Sheer, D.; Keyse, S. M.: The CL100 gene, which encodes a dual specificity (tyr/thr) MAP kinase phosphatase, is highly conserved and maps to human chromosome 5q34. Hum. Genet. 93:513–516, 1994.
- [41748] 15300.Keyse, S. M.; Emslie, E. A.: Oxidative stress and heat shock induce a human gene encoding a protein–tyrosine phosphatase. Nature 359:644–647, 1992.
- [41749] 15301.Martell, K. J.; Kwak, S.; Hakes, D. J.; Dixon, J. E.; Trent, J.M.: Chromosomal localization of four human VH1–like protein–tyrosine phosphatases. Genomics 22: 462–464, 1994.

- [41750] 15302.Chrast, R.; Chen, H.; Morris, M. A.; Antonarakis, S. E.: Mapping of the human transcription factor GABPA (E4TF1-60) gene to chromosome 21. *Genomics* 28: 119-122, 1995.
- [41751] 15303.Gugneja, S.; Virbasius, J. V.; Scarpulla, R. C.: Four structurally distinct, non-DNA-binding subunits of human nuclear respiratory factor 2 share a conserved transcriptional activation domain. *Molec. Cell Biol.* 15: 102-111, 1995.
- [41752] 15304.Guo, A.; Nie, F.; Wong-Riley, M.: Human nuclear respiratory factor 2-alpha subunit cDNA: isolation, subcloning, sequencing, and in situ hybridization of transcripts in normal and monocularly deprived macaque visual system. *J. Comp. Neurol.* 417: 221-232, 2000.
- [41753] 15305.Sawada, J.; Goto, M.; Watanabe, H.; Handa, H.; Yoshida, M. C.: Regional mapping of two subunits of transcription factor E4TF1 to human chromosome. *Jpn. J. Cancer Res.* 86: 10-12, 1995.
- [41754] 15306.Virbasius, J. V.; Scarpulla, R. C.: Activation of the human mitochondrial transcription factor A gene by nuclear respiratory factors: a potential regulatory link between nuclear and mitochondrial gene expression in organelle biogenesis. *Proc. Nat. Acad. Sci.* 91: 1309-1313,

1994.

- [41755] 15307.Watanabe, H.; Sawada, J.; Yano, K.-I.; Yamaguchi, K.; Goto, M.;Handa, H.: cDNA cloning of transcription factor E4TF1 subunits withEts and notch motifs. *Molec. Cell. Biol.* 13: 1385–1391, 1993.
- [41756] 15308.Becker, T.; Gerke, V.; Kube, E.; Weber, K.: S100P, a novel $\text{Ca}(2+)$ –bindingprotein from human placenta: cDNA cloning, recombinant protein expressionand $\text{Ca}(2+)$ binding properties. *Europ. J. Biochem.* 207: 541–547,1992.
- [41757] 15309.Dixon, J.; Loftus, S. K.; Gladwin, A. J.; Scambler, P. J.; Wasmuth,J. J.; Dixon, M. J.: Cloning of the human heparan sulfate–N–deacetylase/N–sulfotransferasegene from the Treacher Collins syndrome candidate region at 5q32–q33.1. *Genomics* 26:239–244, 1995.
- [41758] 15310.Gladwin, A. J.; Dixon, J.; Loftus, S. K.; Wasmuth, J. J.; Dixon,M. J.: Genomic organization of the human heparan sulfate–N–deacetylase/N–sulfotransferasegene: exclusion from a causative role in the pathogenesis of TreacherCollins syndrome. *Genomics* 32: 471–473, 1996.
- [41759] 15311.Hashimoto, Y.; Orellana, A.; Gil, G.; Hirschberg, C. B.: Molecularcloning and expression of rat liver N–heparan sulfate sulfotransferase. *J.Biol. Chem.* 267: 15744–15750, 1995.

- [41760] 15312.Chen, H.; Antonarakis, S. E.: Localisation of a human homologue of the *Drosophila* *mnb* and rat *Dyrk* genes to chromosome 21q22.2. *Hum.Genet.* 99: 262–265, 1997.
- [41761] 15313.Patil, N.; Cox, D. R.; Bhat, D.; Faham, M.; Myers, R. M.; Peterson, A. S.: A potassium channel mutation in weaver mice implicates membrane excitability in granule cell differentiation. *Nature Genet.* 11:126–129, 1995.
- [41762] 15314.Shindoh, N.; Kudoh, J.; Maeda, H.; Yamaki, A.; Minoshima, S.; Shimizu, Y.; Shimizu, N.: Cloning of a human homolog of the *Drosophila* *minibrain*/rat *Dyrk* gene from 'the Down syndrome critical region' of chromosome 21. *Biochem.Biophys. Res. Commun.* 225: 92–99, 1996.
- [41763] 15315.Smith, D. J.; Stevens, M. E.; Sudanagunta, S. P.; Bronson, R. T.; Makhinson, M.; Watabe, A. M.; O'Dell, T. J.; Fung, J.; Weier, H.-U.G.; Cheng, J.-F.; Rubin, E. M.: Functional screening of 2 Mb of human chromosome 21q22.2 in transgenic mice implicates *minibrain* in learning defects associated with Down syndrome. *Nature Genet.* 16: 28–36, 1997.
- [41764] 15316.Song, W.-J.; Chung, S.-H.; Kurnit, D. M.: The murine *Dyrk* protein maps to chromosome 16, localizes to the nucleus, and can form multimers. *Biochem.Biophys. Res. Commun.* 231: 640–644, 1997.

- [41765] 15317.Song, W.-J.; Sternberg, L. R.; Kasten-Sportes, C.; Van Keuren,M. L.; Chung, S.-H.; Slack, A. C.; Miller, D. E.; Glover, T. W.; Chiang,P.-W.; Lou, L.; Kurnit, D. M.: Isolation of human and murine homologues of the *Drosophila* minibrain gene: human homologue maps to 21q22.2 in the Down syndrome 'critical region.' *Genomics* 38: 331–339, 1996.
- [41766] 15318.Tejedor, F.; Zhu, X. R.; Kaltenbach, E.; Ackermann, A.; Baumann,A.; Canal, I.; Heisenberg, M.; Fischbach, K. F.; Pongs, O.: Minibrain:a new protein kinase family involved in postembryonic neurogenesis in *Drosophila*. *Neuron* 14: 287–301, 1995.
- [41767] 15319.Niethammer, D.; Dieterle, U.; Kleihauer, E.; Wildfeuer, A.; Haferkamp,O.; Hitzig, W. H.: An inherited defect in granulocyte function: impaired chemotaxis, phagocytosis and intracellular killing of microorganisms. *Helv.Paediat. Acta* 30: 537–541, 1976.
- [41768] 15320.Ostermann, G.; Weber, K. S. C.; Zerneck, A.; Schroder, A.; Weber,C.: JAM-1 is a ligand of the beta-2 integrin LFA-1 involved in transendothelial migration of leukocytes. *Nature Immun.* 3: 151–158, 2002.
- [41769] 15321.Rivera-Matos, I. R.; Rakita, R. M.; Mariscalco, M. M.; Elder,F. F. B.; Dreyer, S. A.; Cleary, T. G.: Leukocyte adhe-

sion deficiency mimicking Hirschsprung disease. J. Pediat. 127: 755–757, 1995.

[41770] 15322. Rosmarin, A. G.; Caprio, D.; Levy, R.; Simkevich, C.: CD18 (beta-2 leukocyte integrin) promoter requires PU.1 transcription factor for myeloid activity. Proc. Nat. Acad. Sci. 92: 801–805, 1995.

[41771] 15323. Ross, G. D.: Clinical and laboratory features of patients with an inherited deficiency of neutrophil membrane complement receptor type 3 (CR3) and the related membrane antigens LFA-1 and p150,95. J. Clin. Immun. 6: 107–113, 1986.

[41772] 15324. Shuster, D. E.; Kehrli, M. E., Jr.; Ackermann, M. R.; Gilbert, R. O.: Identification and prevalence of a genetic defect that causes leukocyte adhesion deficiency in Holstein cattle. Proc. Nat. Acad. Sci. 89: 9225–9229, 1992.

[41773] 15325. Sligh, J. E., Jr.; Anderson, D. C.; Beaudet, A. L.: A mutation in the initiation codon of the CD18 gene in a patient with the moderate phenotype of leukocyte adhesion deficiency. (Abstract) Am. J. Hum. Genet. 45 (suppl.): A219, 1989.

[41774] 15326. Solomon, E.; Palmer, R. W.; Hing, S.; Law, S. K. A.: Regional localization of CD18, the beta-subunit of the cell surface adhesion molecule LFA-1, on human chromosome

21 by in situ hybridization. *Ann.Hum. Genet.* 52:
123–128, 1988.

[41775] 15327.Springer, T. A.; Miller, L. J.; Anderson, D. C.:
p150,95, the third member of the Mac-1, LFA-1 human
leukocyte adhesion glycoprotein family. *J. Immun.* 136:
240–245, 1986.

[41776] 15328.Springer, T. A.; Thompson, W. S.; Miller, L. J.;
Schmalstieg, F. C.; Anderson, D. C.: Inherited deficiency of
the Mac-1, LFA-1, p150,95 glycoprotein family and its
molecular basis. *J. Exp. Med.* 160:1901–1918, 1984.

[41777] 15329.Ross, G. D.; Thompson, R. A.; Walport, M. J.;
Springer, T. A.; Watson, J. V.; Ward, R. H. R.; Lida, J.; New-
man, S. L.; Harrison, R. A.; Lachmann, P. J.: Characteriza-
tion of patients with an increased susceptibility to bacterial
infections and a genetic deficiency of leukocyte membrane
complement receptor type three (CR3) and the related-
membrane antigen LFA-1. *Blood* 66: 882–890, 1985.

[41778] 15330.Suomalainen, H. A.; Gahmberg, C. G.; Patarroyo,
M.; Beatty, P.G.; Schroder, J.: Genetic assignment of GP90,
leukocyte adhesion glycoprotein to human chromosome
21. *Somat. Cell Molec. Genet.* 12:297–302, 1986.

[41779] 15331.Suomalainen, H. A.; Gahmberg, C. G.; Patarroyo,
M.; Schroder, J.: GP90 (Leu-CAM antigen) is coded for by

genes on chromosome 21.(Abstract) Cytogenet. Cell Genet. 40: 755, 1985.

- [41780] 15332.Taylor, G. M.; Williams, A.; D'Souza, S. W.; Ferguson, W. D.;Donnai, D.; Fennell, J.; Harris, R.: The expression of CD18 is increasedon trisomy 21 (Down syndrome) lymphoblastoid cells. Clin. Exp. Immun. 71:324–328, 1988.
- [41781] 15333.Todd, R. F., III; Freyer, D. R.: The CD11/CD18 leukocyte glycoproteindeficiency. Hemat. Oncol. Clin. North Am. 2: 13–31, 1988.
- [41782] 15334.van der Meer, J. W. M.; van Zwet, T. L.; van Furth, R.: New familialdefect in microbicidal function of polymorphonuclear leucocytes. Lancet II:630–632, 1975.
- [41783] 15335.Vazquez–Torres, A.; Jones–Carson, J.; Baumler, A. J.; Falkow,S.; Valdivia, R.; Brown, W.; Le, M.; Berggren, R.; Parks, W. T.; Fang,F. C.: Extraintestinal dissemination of Salmonella by CD18–expressingphagocytes. Nature 401: 804–808, 1999.
- [41784] 15336.Vedder, N. B.; Winn, R. K.; Rice, C. L.; Chi, E. Y.; Arfors, K.–E.;Harlan, J. M.: A monoclonal antibody to the adherence–promoting leukocyteglycoprotein, CD18, reduces organ injury and improves survival fromhemorrhagic shock and resuscitation in rabbits. J. Clin. Invest.

81:939–944, 1988.

- [41785] 15337.Wardlaw, A. J.; Hibbs, M. L.; Stacker, S. A.; Springer, T. A.: Distinct mutations in two patients with leukocyte adhesion deficiency and their functional correlates. *J. Exp. Med.* 172: 335–345, 1990.
- [41786] 15338.Weening, R. S.; Roos, D.; Weemaes, C. M. R.; Homan–Muller, J.W. T.; van Schaik, M. L. J.: Defective initiation of the metabolic stimulation in phagocytizing granulocytes: a new congenital defect. *J.Lab. Clin. Med.* 88: 757–768, 1976.
- [41787] 15339.Weitzman, J. B.; Wells, C. E.; Wright, A. H.; Clark, P. A.; Law, S. K. A.: The gene organisation of the human beta–2 integrin subunit(CD18). *FEBS Lett.* 294: 97–103, 1991.
- [41788] 15340.Wilson, J. M.; Ping, A. J.; Krauss, J. C.; Mayo–Bond, L.; Rogers, C. E.; Anderson, D. C.; Todd, R. F., III: Correction of CD18–deficient lymphocytes by retrovirus–mediated gene transfer. *Science* 248: 1413–1416, 1990.
- [41789] 15341.Wilson, R. W.; Ballantyne, C. M.; Smith, C. W.; Montgomery, C.; Bradley, A.; O'Brien, W. E.; Beaudet, A. L.: Gene targeting yields a CD18–mutant mouse for study of inflammation. *J. Immun.* 151: 1571–1578, 1993.
- [41790] 15342.Eichwald, E. J.; Silmsker, C. R.: *Skin. Transplant. Bull.*

2:148–149, 1955.

- [41791] 15343.Gasser, D. L.; Silvers, W. K.: Genetics and immunology of sex-linked antigens. *Adv. Immun.* 15: 215–247, 1972.
- [41792] 15344.Kent-First, M. G.; Maffitt, M.; Muallem, A.; Brisco, P.; Shultz, J.; Ekenberg, S.; Agulnik, A. I.; Agulnik, I.; Shramm, D.; Bavister, B.; Abdul-Mawgood, A.; VandeBerg, J.: Gene sequence and evolutionary conservation of human SMCY. (Letter) *Nature Genet.* 14: 128–129, 1996.
- [41793] 15345.Scott, D. M.; Ehrmann, I. E.; Ellis, P. S.; Bishop, C. E.; Agulnik, A. I.; Simpson, E.; Mitchell, M. J.: Identification of a mouse male-specific transplantation antigen, H-Y. *Nature* 376: 695–698, 1995.
- [41794] 15346.Wachtel, S. S.; Koo, G. C.; Breg, W. R.; Elias, S.; Boyse, E.A.; Miller, O. J.: Expression of H-Y antigen in human males with two Y chromosomes. *New Eng. J. Med.* 293: 1070–1072, 1975.
- [41795] 15347.Wachtel, S. S.; Koo, G. C.; Zuckerman, E. E.; Hammerling, U.; Scheid, M. P.; Boyse, E. A.: Serological cross-reactivity between H-Y (male) antigens of mouse and man. *Proc. Nat. Acad. Sci.* 71:1215–1218, 1974.
- [41796] 15348.Wang, W.; Meadows, L. R.; den Haan, J. M. M.; Sherman, N. E.; Chen, Y.; Blokland, E.; Shabanowitz, J.; Ag-

ulnik, A. I.; Hendrickson, R. C.; Bishop, C. E.; Hunt, D. F.; Goulmy, E.; Engelhard, V. H.: Human H-Y: a male-specific histocompatibility antigen derived from the SMCY protein. Science 269: 1588–1590, 1995.

[41797] 15349. Redmond, T. M.; Yu, S.; Lee, E.; Bok, D.; Hamasaki, D.; Chen, N.; Goletz, P.; Ma, J.-X.; Crouch, R. K.; Pfeifer, K.: Rpe65 is necessary for production of 11-cis-vitamin A in the retinal visual cycle. Nature Genet. 20: 344–351, 1998.

[41798] 15350. Seeliger, M. W.; Grimm, C.; Stahlberg, F.; Friedburg, C.; Jaissle, G.; Zrenner, E.; Guo, H.; Reme, C. E.; Humphries, P.; Hofmann, F.; Biel, M.; Fariss, R. N.; Redmond, T. M.; Wenzel, A.: New views on RPE65 deficiency: the rod system is the source of vision in a mouse model of Leber congenital amaurosis. Nature Genet. 29: 70–74, 2001.

[41799] 15351. Thompson, D. A.; Gyurus, P.; Fleischer, L.; Bingham, E. L.; McHenry, C. L.; Apfelstedt-Sylla, E.; Zrenner, E.; Lorenz, B.; Richards, J. E.; Jacobson, S. G.; Sieving, P. A.; Gal, A.: Genetics and phenotypes of RPE65 mutations in inherited retinal degeneration. Invest. Ophthalmol. Vis. Sci. 41: 4293–4299, 2000.

[41800] 15352. Thompson, D. A.; McHenry, C. L.; Li, Y.; Richards, J. E.; Othman, M. I.; Schwinger, E.; Vollrath, D.; Jacobson, S.

G.; Gal, A.: Retinal dystrophy due to paternal isodisomy for chromosome 1 or chromosome 2, with homoallelism for mutations in RPE65 or MERTK, respectively. *Am.J. Hum. Genet.* 70: 224–229, 2002.

- [41801] 15353. Wrigstad, A.; Narfstrom, K.; Nilsson, S. E.: Slowly progressive changes of the retina and retinal pigment epithelium in Briard dogs with hereditary retinal dystrophy: a morphological study. *Doc. Ophthalmol.* 87:337–354, 1994.
- [41802] 15354. Bogenmann, E.; Lochrie, M. A.; Simon, M. I.: Cone cell-specific genes expressed in retinoblastoma. *Science* 240: 76–78, 1988.
- [41803] 15355. Bonaiti-Pellie, C.; Briard-Guillemot, M. L.: Segregation analysis in hereditary retinoblastoma. *Hum. Genet.* 57: 411–419, 1981.
- [41804] 15356. Harbitz, I.; Chowdhary, B.; Thomsen, P. D.; Davies, W.; Kaufmann, U.; Kran, S.; Gustavsson, I.; Christensen, K.; Hauge, J. G.: Assignment of the porcine calcium release channel gene, a candidate for the malignant hyperthermia locus, to the 6p11–q21 segment of chromosome 6. *Genomics* 8:243–248, 1990.
- [41805] 15357. Hunter, A. G. W.; Thompson, M. W.: Acromesomelic dwarfism: description of a patient and comparison with previously reported cases. *Hum. Genet.* 34: 107–113,

1976.

- [41806] 15358. Harbitz, I.; Kristensen, T.; Bosnes, M.; Kran, S.; Davies, W.: DNA sequence of the skeletal muscle calcium release channel cDNA and verification of the arg615-to-cys615 mutation, associated with porcine malignant hyperthermia, in Norwegian Landrace pigs. *Animal Genet.* 23: 395–402, 1992.
- [41807] 15359. Keating, K. E.; Quane, K. A.; Manning, B. M.; Lehane, M.; Hartung, E.; Censier, K.; Urwyler, A.; Klausnitzer, M.; Muller, C. R.; Heffron, J. J. A.; McCarthy, T. V.: Detection of a novel RYR1 mutation in four malignant hyperthermia pedigrees. *Hum. Molec. Genet.* 3: 1855–1858, 1994.
- [41808] 15360. Lynch, P. J.; Tong, J.; Lehane, M.; Mallet, A.; Giblin, L.; Heffron, J. J. A.; Vaughan, P.; Zafra, G.; MacLennan, D. H.; McCarthy, T. V.: A mutation in the transmembrane/luminal domain of the ryanodine receptor is associated with abnormal Ca^{2+} release channel function and severe central core disease. *Proc. Nat. Acad. Sci.* 96: 4164–4169, 1999.
- [41809] 15361. MacKenzie, A. E.; Korneluk, R. G.; Duff, C.; Worton, R. G.; MacLennan, D. H.: Location of the human ryanodine receptor gene with respect to an established human chromosome 19 linkage group. (Abstract) *Am. J. Hum. Genet.*

45 (suppl.): A149, 1989.

- [41810] 15362. MacKenzie, A. E.; Korneluk, R. G.; Zorzato, F.; Fujii, J.; Phillips, M.; Iles, D.; Wieringa, B.; Leblond, S.; Bailly, J.; Willard, H. F.; Duff, C.; Worton, R. G.; MacLennan, D. H.: The human ryanodine receptor gene: its mapping to 19q13.1, placement in a chromosome 19 linkage group, and exclusion as the gene causing myotonic dystrophy. *Am. J. Hum. Genet.* 46: 1082–1089, 1990.
- [41811] 15363. MacLennan, D. H.: Personal Communication. Toronto, Ontario, Canada 7/5/1991.
- [41812] 15364. Manning, B. M.; Quane, K. A.; Lynch, P. J.; Urwyler, A.; Tegazzin, V.; Krivosic-Horber, R.; Censier, K.; Comi, G.; Adnet, P.; Wolz, W.; Lunardi, J.; Muller, C. R.; McCarthy, T. V.: Novel mutations at a CpG dinucleotide in the ryanodine receptor in malignant hyperthermia. *Hum. Mutat.* 11: 45–50, 1998.
- [41813] 15365. Mattei, M. G.; Giannini, G.; Moscatelli, F.; Sorrentino, V.: Chromosomal localization of murine ryanodine receptor genes RYR1, RYR2, and RYR3 by in situ hybridization. *Genomics* 22: 202–204, 1994.
- [41814] 15366. McCarthy, T. V.; Quane, K. A.; Lynch, P. J.: Ryanodine receptor mutations in malignant hyperthermia and central core disease. *Hum. Mutat.* 15: 410–417, 2000.

- [41815] 15367. Monnier, N.; Romero, N. B.; Lerale, J.; Landrieu, P.; Nivoche, Y.; Fardeau, M.; Lunardi, J.: Familial and sporadic forms of centralcore disease are associated with mutations in the C-terminal domain of the skeletal muscle ryanodine receptor. *Hum. Molec. Genet.* 10:2581–2592, 2001.
- [41816] 15368. MacLennan, D. H.; Zorzato, F.; Fujii, J.; Otsu, K.; Phillips, M.; Lai, F. A.; Meissner, G.; Green, N. M.; Willard, H. F.; Britt, B. A.; Worton, R. G.; Korneluk, R. G.: Cloning and localization of the human calcium release channel (ryanodine receptor) gene to the proximal long arm (cen-q13.2) of human chromosome 19. (Abstract) *Am. J. Hum. Genet.* 45 (suppl.): A205, 1989.
- [41817] 15369. Manning, B. M.; Quane, K. A.; Ording, H.; Urwyler, A.; Tegazzin, V.; Lehane, M.; O'Halloran, J.; Hartung, E.; Giblin, L. M.; Lynch, P. J.; Vaughan, P.; Censier, K.; Bendixen, D.; Comi, G.; Heytens, L.; Monsieurs, K.; Fagerlund, T.; Wolz, W.; Heffron, J. J. A.; Muller, C. R.; McCarthy, T. V.: Identification of novel mutations in the ryanodine-receptor gene (RYR1) in malignant hyperthermia: genotype-phenotype correlation. *Am. J. Hum. Genet.* 62: 599–609, 1998.
- [41818] 15370. Monnier, N.; Romero, N. B.; Lerale, J.; Nivoche, Y.;

Qi, D.; MacLennan, D. H.; Fardeau, M.; Lunardi, J.: An autosomal dominant congenital myopathy with cores and rods is associated with a neomutation in the RYR1 gene encoding the skeletal muscle ryanodine receptor. *Hum. Molec. Genet.* 9: 2599–2608, 2000.

[41819] 15371. Monsieus, K. G.; Van Broekhoven, C.; Martin, J.-J.; Van Hoof, V. O.; Heytens, L.: Gly341arg mutation indicating malignant hyperthermia susceptibility: specific cause of chronically elevated serum creatine kinase activity. *J. Neurol. Sci.* 154: 62–65, 1998.

[41820] 15372. O'Brien, R. O.; Taske, N. L.; Hansbro, P. M.; Matthaei, K. I.; Hogan, S. P.; Denborough, M. A.; Foster, P. S.: Exclusion of defects in the skeletal muscle specific regions of the DHPR alpha-1 subunit as frequent causes of malignant hyperthermia. *J. Med. Genet.* 32: 913–914, 1995.

[41821] 15373. Otsu, K.; Khanna, V. K.; Archibald, A. L.; MacLennan, D. H.: Cosegregation of porcine malignant hyperthermia and a probable causal mutation in the skeletal muscle ryanodine receptor gene in backcross families. *Genomics* 11: 744–750, 1991.

[41822] 15374. Otsu, K.; Nishida, K.; Kimura, Y.; Kuzuya, T.; Hori, M.; Kamada, T.; Tada, M.: The point mutation

arg615-to-cys in the Ca(2+) release channel of skeletal sarcoplasmic reticulum is responsible for hypersensitivity to caffeine and halothane in malignant hyperthermia. J. Biol. Chem. 269:9413-9415, 1994.

[41823] 15375.Otsu, K.; Phillips, M. S.; Khanna, V. K.; de Leon, S.; MacLennan, D. H.: Refinement of diagnostic assays for a probable causal mutation for porcine and human malignant hyperthermia. Genomics 13: 835-837, 1992.

[41824] 15376.Phillips, M. S.; Fujii, J.; Khanna, V. K.; DeLeon, S.; Yokobata, K.; De Jong, P. J.; MacLennan, D. H.: The structural organization of the human skeletal muscle ryanodine receptor (RYR1) gene. Genomics 34:24-41, 1996.

[41825] 15377.Quane, K. A.; Healy, J. M. S.; Keating, K. E.; Manning, B. M.; Couch, F. J.; Palmucci, L. M.; Doriguzzi, C.; Fagerlund, T. H.; Berg, K.; Ording, H.; Bendixen, D.; Mortier, W.; Linz, U.; Muller, C. R.; McCarthy, T. V.: Mutations in the ryanodine receptor gene in central core disease and malignant hyperthermia. Nature Genet. 5: 51-55, 1993.

[41826] 15378.Richter, M.; Schleithoff, L.; Deufel, T.; Lehmann-Horn, F.; Herrmann-Frank, A.: Functional characterization of a distinct ryanodine receptor mutation in human malignant hyperthermia-susceptible muscle. J. Biol. Chem. 272:

5256–5260, 1997.

- [41827] 15379. Robinson, R. L.; Brooks, C.; Brown, S. L.; Ellis, F. R.; Halsall, P. J.; Quinnell, R. J.; Shaw, M.-A.; Hopkins, P. M.: RYR1 mutations causing central core disease are associated with more severe malignant hyperthermia in vitro contracture test phenotypes. *Hum. Mutat.* 20:88–97, 2002.
- [41828] 15380. Sambuughin, N.; McWilliams, S.; de Bantel, A.; Sivakumar, K.; Nelson, T. E.: Single-amino-acid deletion in the RYR1 gene, associated with malignant hyperthermia susceptibility and unusual contraction phenotype. *Am. J. Hum. Genet.* 69: 204–208, 2001.
- [41829] 15381. Oesterling, J. E.; Chan, D. W.; Epstein, J. I.; Kimball, A. W., Jr.; Bruzek, D. J.; Rock, R. C.; Brendler, C. B.; Walsh, P. C.: Prostate specific antigen in the preoperative and postoperative evaluation of localized prostatic cancer treated with radical prostatectomy. *J. Urol.* 139: 766–772, 1988.
- [41830] 15382. Riegman, P. H. J.; Vlietstra, R. J.; van der Korput, J. A. G. M.; Brinkmann, A. O.; Trapman, J.: The promoter of the prostate-specific antigen gene contains a functional androgen responsive element. *Molec. Endocr.* 5: 1921–1930, 1991.

- [41831] 15383.van den Elsen, P.; Bruns, G.; Gerhard, D. S.; Pravtcheva, D.; Jones, C.; Housman, D.; Ruddle, F. A.; Orkin, S.; Terhorst, C.: Assignment of the gene coding for the T3–delta subunit of the T3–T-cell receptor complex to the long arm of human chromosome 11 and to mouse chromosome 9. *Proc. Nat. Acad. Sci.* 82: 2920–2924, 1985.
- [41832] 15384.van Rijs, J.; Giguere, V.; Hurst, J.; van Agthoven, T.; Geurts van Kessel, A.; Goyert, S.; Grosveld, F.: Chromosomal localization of the human Thy–1 gene. *Proc. Nat. Acad. Sci.* 82: 5832–5835, 1985.
- [41833] 15385.Williams, A. F.; Gagnon, J.: Neuronal cell Thy–1 glycoprotein:homology with immunoglobulin. *Science* 216: 696–703, 1982.
- [41834] 15386.Johansson, M.; Karlsson, A.: Cloning of the cDNA and chromosomal localization of the gene for human thymidine kinase 2. *J. Biol. Chem.* 272:8454–8458, 1997.
- [41835] 15387.Mandel, H.; Szargel, R.; Labay, V.; Elpeleg, O.; Saada, A.; Shalata, A.; Anbinder, Y.; Berkowitz, D.; Hartman, C.; Barak, M.; Eriksson, S.; Cohen, N.: The deoxyguanosine kinase gene is mutated in individuals with depleted hepatocerebral mitochondrial DNA. *Nature Genet.* 29:337–341, 2001. Note: Erratum: *Nature Genet.*

29: 491 only, 2001.

- [41836] 15388.Saada, A.; Shaag, A.; Mandel, H.; Nevo, Y.; Eriksson, S.; Elpeleg,O.: Mutant mitochondrial thymidine kinase in mitochondrial DNA depletionmyopathy. *Nature Genet.* 29: 342–344, 2001.
- [41837] 15389.Wang, L.; Munch–Petersen, B.; Herrstrom Sjoberg, A.; Hellman, U.;Bergman, T.; Jornvall, H.; Eriksson, S.: Human thymidine kinase 2:molecular cloning and characterisation of the enzyme activity withantiviral and cytostatic nucleoside substrates. *FEBS Lett.* 443:170–174, 1999.
- [41838] 15390.Willecke, K.; Reuber, T.; Kucherlapati, R. S.; Ruddle, F. H.:Human mitochondrial thymidine kinase is coded for by a gene on chromosome16 of the nucleus. *Somat. Cell Genet.* 3: 237–245, 1977.
- [41839] 15391.Chang, J. G.; Scarpa, A.; Eddy, R. L.; Byers, M. G.; Harris, A.S.; Morrow, J. S.; Watkins, P.; Shows, T. B.; Forget, B. G.: Cloningof a portion of the chromosomal gene and cDNA for human beta–fodrin,the nonerythroid form of beta–spectrin. *Genomics* 17: 287–293, 1993.
- [41840] 15392.Watkins, P. C.; Eddy, R.; Forget, B. G.; Chang, J. G.; Rochelle,R.; Shows, T. B.: Assignment of a non–erythroid spectrin gene tohuman chromosome 2. (Abstract) *Am. J. Hum. Genet.* 43: A161, 1988.

- [41841] 15393.Miceli-Richard, C.; Lesage, S.; Rybojad, M.; Prieur, A.-M.; Manouvrier-Hanu,S.; Hafner, R.; Chamaillard, M.; Zouali, H.; Thomas, G.; Hugot, J.-P.: CARD15 mutations in Blau syndrome. *Nature Genet.* 29: 19-20, 2001.
- [41842] 15394.Cong, F.; Spencer, S.; Cote, J.-F.; Wu, Y.; Tremblay, M. L.; Lasky,L. A.; Goff, S. P.: Cytoskeletal protein PSTPIP1 directs the PEST-typeprotein tyrosine phosphatase to the c-Abl kinase to mediate Abl dephosphorylation. *Molec.Cell* 6: 1413-1423, 2000.
- [41843] 15395.Han, L. Wong, D.; Dhaka, A.; Afar, D.; White, M.; Xie, W.; Herschman,H.; Witte, O.: Colicelli, J.: Protein binding and signaling propertiesof RIN1 suggest a unique effector function. *Proc. Nat. Acad. Sci.* 94:4954-4959, 1997.
- [41844] 15396.Jhanwar, S. C.; Neel, B. G.; Hayward, W. S.; Chaganti, R. S. K.: Localization of the cellular oncogenes ABL, SIS, and FES on humangerm-line chromosomes. *Cytogenet. Cell Genet.* 38: 73-75, 1984.
- [41845] 15397.Blank, V.; Andrews, N. C.: The Maf transcription factors: regulatorsof differentiation. *Trends Biochem. Sci.* 22: 437-441, 1997.
- [41846] 15398.Jamieson, R. V.; Perveen, R.; Kerr, B.; Carette, M.; Yardley, J.;Heon, E.; Wirth, M. G.; van Heyningen, V.; Donnai, D.; Munier, F.;Black, G. C. M.: Domain disruption and

mutation of the bZIP transcriptionfactor, MAF, associated with cataract, ocular anterior segment dysgenesisand coloboma. Hum. Molec. Genet. 11: 33–42, 2002.

[41847] 15399.Kim, J. I.; Li, T.; Ho, I. C.; Grusby, M. J.; Glimcher, L. H.:Requirement for the c-Maf transcription factor in crystallin generegulation and lens development. Proc. Nat. Acad. Sci. 96: 3781–3785,1999.

[41848] 15400.Nishizawa, M.; Kataoka, K.; Goto, N.; Fujiwara, K. T.; Kawai, S.: v-maf, a viral oncogene that encodes a 'leucine zipper' motif. Proc.Nat. Acad. Sci. 86: 7711–7715, 1989.

[41849] 15401.Yoshida, M. C.; Nishizawa, M.; Kataoka, K.; Goto, N.; Fujiwara,K. T.; Kawai, S.: Localization of the human MAF protooncogene onchromosome 16 to bands q22–q23. (Abstract) Cytogenet. Cell Genet. 58:2003 only, 1991.

[41850] 15402.Bao, S.; Tibbetts, R. S.; Brumbaugh, K. M.; Fang, Y.; Richardson,D. A.; Ali, A.; Chen, S. M.; Abraham, R. T.; Wang, X.–F.: ATR/ATM–mediatedphosphorylation of human Rad17 is required for genotoxic stress responses. Nature 411:969–974, 2001.

[41851] 15403.Barbosa, M. D. F. S.; Barrat, F. J.; Tchernev, V. T.; Nguyen, Q.A.; Mishra, V. S.; Colman, S. D.; Pastural, E.;

Dufourcq-Lagelouse, R.; Fischer, A.; Holcombe, R. F.; Wallace, M. R.; Brandt, S. J.; deSaint Basile, G.; Kingsmore S. F.: Identification of mutations in two major mRNA isoforms of the Chediak-Higashi syndrome gene in human and mouse. *Hum. Molec. Genet.* 6: 1091-1098, 1997.

- [41852] 15404. Barrat, F. J.; Auloge, L.; Pastural, E.; Dufourcq Lagelouse, R.; Vilmer, E.; Cant, A. J.; Weissenbach, J.; Le Paslier, D.; Fischer, A.; de Saint Basile, G.: Genetic and physical mapping of the Chediak-Higashi syndrome on chromosome 1q42-43. *Am. J. Hum. Genet.* 59: 625-632, 1996.
- [41853] 15405. Dufourcq-Lagelouse, R.; Lambert, N.; Duval, M.; Viot, G.; Vilmer, E.; Fischer, A.; Prieur, M.; de Saint Basile, G.: Chediak-Higashi syndrome associated with maternal uniparental isodisomy of chromosome 1. *Europ. J. Hum. Genet.* 7: 633-637, 1999.
- [41854] 15406. Faigle, W.; Raposo, G.; Tenza, D.; Pinet, V.; Vogt, A. B.; Kropshofer, H.; Fischer, A.; de Saint-Basile, G.; Amigorena, S.: Deficient peptide loading and MHC class II endosomal sorting in a human genetic immunodeficiency-disease: the Chediak-Higashi syndrome. *J. Cell Biol.* 141: 1121-1134, 1998.
- [41855] 15407. Karim, M. A.; Suzuki, K.; Fukai, K.; Oh, J.; Nagle, D.

L.; Moore, K. J.; Barbosa, E.; Falik-Borenstein, T.; Filipovich, A.; Ishida, Y. Kivrikko, S.; Klein, C.; and 8 others: Apparent genotype-phenotype correlation in childhood, adolescent, and adult Chediak-Higashi syndrome. *Am. J. Med. Genet.* 108: 16-22, 2002.

[41856] 15408. Kunieda, T.; Ide, H.; Nakagiri, M.; Yoneda, K.; Konfortov, B.; Ogawa, H.: Localization of the locus responsible for Chediak-Higashi syndrome in cattle to bovine chromosome 28. *Anim. Genet.* 31: 87-90, 2000.

[41857] 15409. Akama, T. O.; Nishida, K.; Nakayama, J.; Watanabe, H.; Ozaki, K.; Nakamura, T.; Dota, A.; Kawasaki, S.; Inoue, Y.; Maeda, N.; Yamamoto, S.; Fujiwara, T.; Thonar, E. J.-M. A.; Shimomura, Y.; Kinoshita, S.; Tanigami, A.; Fukuda, M. N.: Macular corneal dystrophy type I and type II are caused by distinct mutations in a new sulphotransferase gene. *Nature Genet.* 26: 237-241, 2000.

[41858] 15410. Enomoto, A.; Kimura, H.; Chairoungdua, A.; Shigeta, Y.; Jutabha, P.; Cha, S. H.; Hosoyamada, M.; Takeda, M.; Sekine, T.; Igarashi, T.; Matsuo, H.; Kikuchi, Y.; Oda, T.; Ichida, K.; Hosoya, T.; Shimokata, K.; Niwa, T.; Kanai, Y.; Endou, H.: Molecular identification of a renal urate-anion exchanger that regulates blood urate levels. *Nature* 417: 447-452, 2002.

- [41859] 15411.Goedert, M.; Hasegawa, J.; Craxton, M.; Leversha, M. A.; Clegg,S.: Assignment of the human stress-activated protein kinase-3 gene(SAPK3) to chromosome 22q13.3 by fluorescence in situ hybridization. *Genomics* 41:501-502, 1997.
- [41860] 15412.Yang, H.; Jeffrey, P. D.; Miller, J.; Kinnucan, E.; Sun, Y.; Thoma,N. H.; Zheng, N.; Chen, P.-L.; Lee, W.-H.; Pavletich, N. P.: BRCA2function in DNA binding and recombination from a BRCA2-DSS1-ssDNAstructure. *Science* 297: 1837-1848, 2002.
- [41861] 15413.Koettnitz, K.; Kappel, B.; Baumruker, T.; Hauber, J.; Bevec, D.: The genomic structure encoding human initiation factor eIF-5A. *Gene* 144:249-252, 1994.
- [41862] 15414.Koettnitz, K.; Wohl, T.; Kappel, B.; Lottspeich, F.; Hauber, J.;Bevec, D.: Identification of a new member of the human eIF-5A genefamily. *Gene* 159: 283-284, 1995.
- [41863] 15415.Steinkasserer, A.; Jones, T.; Sheer, D.; Koettnitz, K.; Hauber,J.; Bevec, D.: The eukaryotic cofactor for the human immunodeficiencyvirus type 1 (HIV-1) rev protein, eIF-5A, maps to chromosome 17p12-p13:three eIF-5A pseudogenes map to 10q23.3, 17q25, and 19q13.2. *Genomics* 25:749-752, 1995.
- [41864] 15416.Hartley, D. A.; Preiss, A.; Artavanis-Tsakonas, S.: A

deduced gene product from the *Drosophila* neurogenic locus, enhancer of split, shows homology to mammalian G-protein beta subunit. *Cell* 55: 785–795, 1988.

[41865] 15417. Hou, E. W.; Li, S.-L.: Genomic organization and chromosome localization to band 19p13.3 of the human AES gene: gene product exhibits strong similarity to the N-terminal domain of *Drosophila* enhancer of split Groucho protein. *DNA Cell Biol.* 17: 911–913, 1998.

[41866] 15418. Miyasaka, H.; Choudhury, B. K.; Hou, E. W.; Li, S. S.-L.: Molecular cloning and expression of mouse and human cDNA encoding AES and ESG proteins with strong similarity to *Drosophila* enhancer of split groucho protein. *Europ. J. Biochem.* 216: 343–352, 1993.

[41867] 15419. Imai, Y.; Kurokawa, M.; Tanaka, K.; Friedman, A. D.; Ogawa, S.; Mitani, K.; Yazaki, Y.; Hirai, H.: TLE, the human homolog of groucho, interacts with AML1 and acts as a repressor of AML1-induced transactivation. *Biochem. Biophys. Res. Commun.* 252: 582–589, 1998.

[41868] 15420. Liu, Y.; Dehni, G.; Purcell, K. J.; Sokolow, J.; Canciani, M. L.; Artavanis-Tsakonas, S.; Stifani, S.: Epithelial expression and chromosomal location of human TLE genes: implications for Notch signaling and neoplasia. *Genomics* 31: 58–64, 1996.

- [41869] 15421.Stifani, S.; Blaumueller, C. M.; Redhead, N. J.; Hill, R. E.; Artavanis-Tsakonas,S.: Human homologs of a *Drosophila* enhancer of split gene productdefine a novel family of nuclear proteins. *Nature Genet.* 2: 119–127,1992.
- [41870] 15422.de Bruijn, D. R. H.; Baats, E.; Zechner, U.; de Leeuw, B.; Balemans,M.; Olde Weghuis, D.; Hirning-Folz, U.; Geurts van Kessel, A.: Isolationand characterization of the mouse homolog of SYT, a gene implicatedin the development of human synovial sarcomas. *Oncogene* 13: 643–648,1996.
- [41871] 15423.de Bruijn, D. R. H.; Kater-Baats, E.; Eleveld, M.; Merkx, G.; vanKessel, A. G.: Mapping and characterization of the mouse and humanSS18 genes, two human SS18-like genes and a mouse Ss18 pseudogene. *Cytogenet.Cell Genet.* 92: 310–319, 2001.
- [41872] 15424.Thaete, C.; Brett, D.; Monaghan, P.; Whitehouse, S.; Rennie, G.;Rayner, E.; Cooper, C. S.; Goodwin, G.: Functional domains of theSYT and SYT-SSX synovial sarcoma translocation proteins and co-localizationwith the SNF protein BRM in the nucleus. *Hum. Molec. Genet.* 8: 585–591,1999.
- [41873] 15425.Ge, Q.; Trieu, E. P.; Targoff, I. N.: Primary structure

and functional expression of human glycyl-tRNA synthetase, an autoantigen in myositis. *J. Biol. Chem.* 269: 28790–28797, 1994.

[41874] 15426. Nichols, R. C.; Pai, S. I.; Ge, Q.; Targoff, I. N.; Plotz, P. H.; Liu, P.: Localization of two human autoantigen genes by PCR screening and in situ hybridization--Glycyl-tRNA synthetase locates to 7p15 and alanyl-tRNA synthetase locates to 16q22. *Genomics* 30: 131–132, 1995.

[41875] 15427. Shiba, K.; Schimmel, P.; Motegi, H.; Noda, T.: Human glycyl-tRNA synthetase: wide divergence of primary structure from bacterial counterpart and species-specific aminoacylation. *J. Biol. Chem.* 269: 30049–30055, 1994.

[41876] 15428. Williams, J.; Osvath, S.; Khong, T. F.; Pearse, M.; Power, D.: Cloning, sequencing and bacterial expression of human glycine tRNA synthetase. *Nucleic Acids Res.* 23: 1307–1310, 1995.

[41877] 15429. Avraham, K. B.; Prezioso, V. R.; Chen, W. S.; Lai, E.; Sladek, F. M.; Zhong, W.; Darnell, J. E., Jr.; Jenkins, N. A.; Copeland, N. G.: Murine chromosomal location of four hepatocyte-enriched transcription factors: HNF-3-alpha, HNF-3-beta, HNF-3-gamma, and HNF-4. *Genomics* 13: 264–268, 1992.

[41878] 15430. Deleuze, J. F.; Dhorne, S.; Hazan, J.; Borghi, E.;

Raynaud, N.; Pollet, N.; Meunier-Rotival, M.; Deschatrette, J.; Alagille, D.; Hadchouel, M.: Deleted chromosome 20 from a patient with Alagille syndrome isolated in a cell hybrid through leucine transport selection: study of three-candidate genes. *Mammalian Genome* 5: 663–669, 1994.

[41879] 15431. Kaestner, K. H.; Hiemisch, H.; Luckow, B.; Schutz, G.: The HNF-3 gene family of transcription factors in mice: gene structure, cDNA sequence, and mRNA distribution. *Genomics* 20: 377–385, 1994.

[41880] 15432. Mincheva, A.; Lichter, P.; Schutz, G.; Kaestner, K. H.: Assignment of the human genes for hepatocyte nuclear factor 3- α , - β , and - γ (HNF3A, HNF3B, HNF3G) to 14q12–q13, 20p11, and 19q13.2–q13.4. *Genomics* 39:417–419, 1997.

[41881] 15433. Adams, R. H.; Porras, A.; Alonso, G.; Jones, M.; Vintersten, K.; Panelli, S.; Valladares, A.; Perez, L.; Klein, R.; Nebreda, A. R.: Essential role of p38- α MAP kinase in placental but not embryonic cardiovascular development. *Molec. Cell* 6: 109–116, 2000.

[41882] 15434. Ge, B.; Gram, H.; Di Padova, F.; Huang, B.; New, L.; Ulevitch, R. J.; Luo, Y.; Han, J.: MAPKK-independent activation of p38- α mediated by TAB1-dependent autophosphorylation of p38- α . *Science* 295:1291–1294,

2002.

- [41883] 15435.Han, J.; Lee, J.-D.; Bibbs, L.; Ulevitch, R. J.: A MAP kinase targeted by endotoxin and hyperosmolarity in mammalian cells. *Science* 265:808–811, 1994.
- [41884] 15436.Kim, D. H.; Feinbaum, R.; Alloing, G.; Emerson, F. E.; Garsin, D. A.; Inoue, H.; Tanaka-Hino, M.; Hisamoto, N.; Matsumoto, K.; Tan, M.-W.; Ausubel, F. M.: A conserved p38 MAP kinase pathway in *Caenorhabditis elegans* innate immunity. *Science* 297: 623–626, 2002.
- [41885] 15437.Kumar, S.; McLaughlin, M. M.; McDonnell, P. C.; Lee, J. C.; Livi, G. P.; Young, P. R.: Human mitogen-activated protein kinase CSBP1, but not CSBP2, complements a *hog1* deletion in yeast. *J. Biol. Chem.* 270:29043–29046, 1995.
- [41886] 15438.Lee, J. C.; Laydon, J. T.; McDonnell, P. C.; Gallagher, T. F.; Kumar, S.; Green, D.; McNulty, D.; Blumenthal, M. J.; Heys, J. R.; Landvatter, S. W.; Stickler, J. E.; McLaughlin, M. M.; Siemens, I. R.; Fisher, S. M.; Livi, G. P.; White, J. R.; Adams, J. L.; Young, P. R.: A protein kinase involved in the regulation of inflammatory cytokine biosynthesis. *Nature* 372: 739–746, 1994.
- [41887] 15439.Liao, P.; Georgakopoulos, D.; Kovacs, A.; Zheng, M.; Lerner, D.; Pu, H.; Saffitz, J.; Chien, K.; Xiao, R.-P.;

Kass, D. A.; Wang, Y.: The in vivo role of p38 MAP kinases in cardiac remodeling and restrictive cardiomyopathy.

Proc. Nat. Acad. Sci. 98: 12283–12288, 2001.

[41888] 15440. Maizels, E. T.; Mukherjee, A.; Sithanandam, G.; Peters, C. A.; Cottom, J.; Mayo, K. E.; Hunzicker-Dunn, M.: Developmental regulation of mitogen-activated protein kinase-activated kinases-2 and -3 (MAPKAPK-2/-3) in vivo during corpus luteum formation in the rat. Molec. Endocr. 15:716–733, 2001.

[41889] 15441. McDonnell, P. C.; DiLella, A. G.; Lee, J. C.; Young, P. R.: Localization of the human stress responsive MAP kinase-like CSAIDs binding protein (CSBP) gene to chromosome 6p21.3/21.2. Genomics 29: 301–302, 1995.

[41890] 15442. New, L.; Jiang, Y.; Zhao, M.; Liu, K.; Zhu, W.; Flood, L. J.; Kato, Y.; Parry, G. C. N.; Han, J.: PRAK, a novel protein kinase regulated by the p38 MAP kinase. EMBO J. 17: 3372–3384, 1998.

[41891] 15443. Ni, H.; Wang, X. S.; Diener, K.; Yao, Z.: MAPKAPK5, a novel mitogen-activated protein kinase (MAPK)-activated protein kinase, is a substrate of the extracellular-regulated kinase (ERK) and p38 kinase. Biochem. Biophys. Res. Commun. 243: 492–496, 1998.

[41892] 15444. Takekawa, M.; Maeda, T.; Saito, H.: Protein phos-

phatase 2C- α inhibits the human stress-responsive p38 and JNK MAPK pathways. EMBOJ. 17: 4744–4752, 1998.

- [41893] 15445. Tamura, K.; Sudo, T.; Senftleben, U.; Dadgar, A. M.; Johnson, R.; Karin, M.: Requirement for p38- α in erythropoietin expression: a role for stress kinases in erythropoiesis. Cell 102: 221–231, 2000.
- [41894] 15446. Haber, N.; Stengle, D.; Defer, N.; Roedel, N.; Mattei, M.-G.; Hanoune, J.: Chromosomal mapping of human adenylyl cyclase gene type III, type V and type VI. Hum. Genet. 94: 69–73, 1994.
- [41895] 15447. Sinnarajah, S.; Dessauer, C. W.; Srikumar, D.; Chen, J.; Yuen, J.; Yilma, S.; Dennis, J. C.; Morrison, E. E.; Vodyanoy, V.; Kehrl, J. H.: RGS2 regulates signal transduction in olfactory neurons by attenuating activation of adenylyl cyclase III. Nature 409: 1051–1055, 2001.
- [41896] 15448. Pellizzoni, L.; Kataoka, N.; Charroux, B.; Dreyfuss, G.: A novel function for SMN, the spinal muscular atrophy gene product, in pre-mRNA splicing. Cell 95: 615–624, 1998.
- [41897] 15449. Brown, K. A.; Leek, J. P.; Lench, N. J.; Moynihan, L. M.; Markham, A. F.; Mueller, R. F.: Human sequences homologous to the gene for the cochlear protein Ocp-II do

not map to currently known non-syndromic hearing loss loci. *Ann. Hum. Genet.* 60: 385–389, 1996.

- [41898] 15450. Chen, H.; Thalmann, I.; Adams, J. C.; Avraham, K. B.; Copeland, N. G.; Jenkins, N. A.; Beier, D. R.; Corey, D. P.; Thalmann, R.; Duyk, G. M.: cDNA cloning, tissue distribution, and chromosomal localization of Ocp2, a gene encoding a putative transcription-associated factor predominantly expressed in the auditory organs. *Genomics* 27: 389–398, 1995.
- [41899] 15451. Demetrick, D. J.; Zhang, H.; Beach, D. H.: Chromosomal mapping of the genes for the human CDK2/cyclin A-associated proteins p19 (SKP1A and SKP1B) and p45 (SKP2). *Cytogenet. Cell Genet.* 73: 104–107, 1996.
- [41900] 15452. Liang, Y.; Chen, H.; Asher, J. H., Jr.; Chang, C.-C.; Friedman, T. B.: Human inner ear OCP2 cDNA maps to 5q22–5q35.2 with related sequences on chromosomes 4p16.2–4p14, 5p13–5q22, 7pter–q22, 10 and 12p13–12qter. *Gene* 184: 163–167, 1997.
- [41901] 15453. Sowden, J.; Morrison, K.; Schofield, J.; Putt, W.; Edwards, Y.: A novel cDNA with homology to an RNA polymerase II elongation factor maps to human chromosome 5q31 (TCEB1L) and to mouse chromosome 11 (Tceb1l). *Genomics* 29: 145–151, 1995.

- [41902] 15454. Thalmann, I.; Rosenthal, H. L.; Moore, B. W.; Thalmann, R.: Organ of Corti-specific polypeptides: OCP-I and OCP-II. *Arch. Oto-Rhino-Laryngol.* 226:123–128, 1980.
- [41903] 15455. Thalmann, I.; Takahashi, K.; Varghese, J.; Comegys, T. H.; Thalmann, R.: Biochemical features of major organ of Corti proteins (OCP-I and OCP-II) including partial amino acid sequence. *Laryngoscope* 100:99–105, 1990.
- [41904] 15456. Zhang, H.; Kobayashi, R.; Galaktionov, K.; Beach, D.: p19Skp1 and p45Skp2 are essential elements of the cyclin A-CDK2 S phase kinase. *Cell* 82:915–925, 1995.
- [41905] 15457. Anand, A.; Chada, K.: In vivo modulation of Hmgic reduces obesity. *Nature Genet.* 24: 377–380, 2000.
- [41906] 15458. Arlotta, P.; Tai, A. K.-F.; Manfioletti, G.; Clifford, C.; Jay, G.; Ono, S. J.: Transgenic mice expressing a truncated form of the high mobility group I-C protein develop adiposity and an abnormally high prevalence of lipomas. *J. Biol. Chem.* 275: 14394–14400, 2000.
- [41907] 15459. Ashar, H. R.; Cherath, L.; Przybysz, K. M.; Chada, K.: Genomic characterization of human HMGIC, a member of the accessory transcription factor family found at translocation breakpoints in lipomas. *Genomics* 31:207–214, 1996.
- [41908] 15460. Ashar, H. R.; Schoenberg Fejzo, M.; Tkachenko, A.;

Zhou, X.; Fletcher, J. A.; Weremowicz, S.; Morton, C. C.; Chada, K.: Disruption of the architectural factor HMGI-C: DNA-binding AT hook motifs fused in lipoma to distinct transcriptional regulatory domains. *Cell* 82: 57–65, 1995.

[41909] 15461. Chau, K.-Y.; Patel, U. A.; Lee, K.-L. D.; Lam, H.-Y. P.; Crane-Robinson, C.: The gene for the human architectural transcription factor HMGI-C consists of five exons each coding for a distinct functional element. *Nucleic Acids Res.* 23: 4262–4266, 1995.

[41910] 15462. Danforth, E., Jr.: Failure of adipocyte differentiation causes type II diabetes mellitus. *Nature Genet.* 26: 13 only, 2000.

[41911] 15463. Friedmann, M.; Holth, L. T.; Zoghbi, H. Y.; Reeves, R.: Organization, inducible-expression and chromosome localization of the human HMG-I(Y) nonhistone protein gene. *Nucleic Acids Res.* 21: 4259–4267, 1993.

[41912] 15464. Ishwad, C. S.; Shriver, M. D.; Lassig, D. M.; Ferrell, R. E.: The high mobility group I-C gene (HMGI-C): polymorphism and genetic localization. *Hum. Genet.* 99: 103–105, 1997.

[41913] 15465. Kazmierczak, B.; Dal Cin, P.; Wanschura, S.; Bartnitzke, S.; Vanden Berghe, H.; Bullerdiek, J.: Cloning and molecular characterization of part of a new gene fused to

HMGIC in mesenchymal tumors. Am. J.Path. 152:
431–435, 1998.

- [41914] 15466.Kazmierczak, B.; Pohnke, Y.; Bullerdiek, J.: Fusion transcripts between the HMGIC gene and RTVL–H–related sequences in mesenchymal tumors without cytogenetic aberrations. Genomics 38: 223–226, 1996.
- [41915] 15467.Manfioletti, G.; Rustighi, A.; Mantovani, F.; Goodwin, G. H.;Giancotti, V.: Isolation and characterization of the gene coding for murine high–mobility–group protein HMGI–C. Gene 167: 249–253,1995.
- [41916] 15468.Mine, N.; Kurose, K.; Nagai, H.; Doi, D.; Ota, Y.; Yoneyama, K.;Konishi, H.; Araki, T.; Emi, M.: Gene fusion involving HMGIC is a frequent aberration in uterine leiomyomas. J. Hum. Genet. 46: 408–412,2001.
- [41917] 15469.Nucci, M. R.; Weremowicz, S.; Neskey, D. M.; Sornberger, K.; Tallini,G.; Morton, C. C.; Quade, B. J.: Chromosomal translocation t(8;12)induces aberrant HMGIC expression in aggressive angiomyxoma of the vulva. Genes Chromosomes Cancer 32: 172–176, 2001.
- [41918] 15470.Rodrigues, N. R.; Owen, N.; Talbot, K.; Ignatius, J.; Dubowitz,V.; Davies, K. E.: Deletions in the survival motor neuron gene on5q13 in autosomal recessive spinal muscular atrophy. Hum. Molec.Genet. 4: 631–634, 1995.

- [41919] 15471. Rossoll, W.; Kroning, A.-K.; Ohndorf, U.-M.; Steegborn, C.; Jablonka, S.; Sendtner, M.: Specific interaction of Smn, the spinal muscular atrophy determining gene product, with hnRNP-R and gry-rbp/hnRNP-Q: a role for Smn in RNA processing in motor axons? *Hum. Molec. Genet.* 11:93–105, 2002.
- [41920] 15472. Simard, L. R.; Rochette, C.; Semionov, A.; Morgan, K.; Vanasse, M.: SMN(T) and NAIP mutations in Canadian families with spinal muscular atrophy (SMA): genotype/phenotype correlations with disease severity. *Am. J. Med. Genet.* 72: 51–58, 1997.
- [41921] 15473. Sossi, V.; Giuli, A.; Vitali, T.; Tiziano, F.; Mirabella, M.; Antonelli, A.; Neri, G.; Brahe, C.: Premature termination mutations in exon 3 of the SMN1 gene are associated with exon skipping and a relatively mild SMA phenotype. *Europ. J. Hum. Genet.* 9: 113–120, 2001.
- [41922] 15474. Stewart, H.; Wallace, A.; McGaughan, J.; Mountford, R.; Kingston, H.: Molecular diagnosis of spinal muscular atrophy. *Arch. Dis. Child.* 78:531–535, 1998.
- [41923] 15475. Talbot, K.; Ponting, C. P.; Theodosiou, A. M.; Rodrigues, N. R.; Surtees, R.; Mountford, R.; Davies, K. E.: Missense mutation clustering in the survival motor neuron gene: a role for a conserved tyrosine and glycine rich re-

gion of the protein in RNA metabolism? Hum.

Molec.Genet. 6: 497–500, 1997.

[41924] 15476.van der Steege, G.; Grootschloten, P. M.; Cobben, J. M.; Zappata,S.; Scheffer, H.; den Dunnen, J. T.; van Ommen, G.–J. B.; Brahe, C.;Buys, C. H. C. M.: Apparent gene conversions involving the SMN genein the region of the spinal muscular atrophy locus on chromosome 5. Am.J. Hum. Genet. 59: 834–838, 1996.

[41925] 15477.Viollet, L.; Bertrand, S.; Bueno Brunialti, A. L.; Lefebvre,S.; Burlet, P.; Clermont, O.; Cruaud, C.; Guenet, J.–L.; Munnich,A.; Melki, J.: cDNA isolation, expression, and chromosomal localizationof the mouse survival motor neuron gene (Smn). Genomics 40: 185–188,1997.

[41926] 15478.Wang, C. H.; Xu, J.; Carter, T. A.; Ross, B. M.; Dominski, M.K.; Bellcross, C. A.; Penchaszadeh, G. K.; Munsat, T. L.; Gilliam,T. C.: Characterization of survival motor neuron (SMNT) gene deletionsin asymptomatic carriers of spinal muscular atrophy. Hum. Molec.Genet. 5: 359–365, 1996.

[41927] 15479.Wang, J.; Dreyfuss, G.: A cell system with targeted disruptionof the SMN gene: functional conservation of the SMN protein and dependenceof Gemin2 on SMN. J. Biol. Chem. 276: 9599–9605, 2001.

- [41928] 15480. Wirth, B.: An update of the mutation spectrum of the survival motor neuron gene (SMN1) in autosomal recessive spinal muscular atrophy (SMA). *Hum. Mutat.* 15: 228–237, 2000.
- [41929] 15481. Wirth, B.; Hahnen, E.; Morgan, K.; DiDonato, C. J.; Dadze, A.; Rudnik-Schoneborn, S.; Simard, L. R.; Zerres, K.; Burghes, A. H. M.: Allelic association and deletions in autosomal recessive proximal spinal muscular atrophy: association of marker genotype with disease severity and candidate cDNAs. *Hum. Molec. Genet.* 4: 1273–1284, 1995.
- [41930] 15482. Wirth, B.; Herz, M.; Wetter, A.; Moskau, S.; Hahnen, E.; Rudnik-Schoneborn, S.; Wienker, T.; Zerres, K.: Quantitative analysis of survival motor neuron copies: identification of subtle SMN1 mutations in patients with spinal muscular atrophy, genotype–phenotype correlation, and implications for genetic counseling. *Am. J. Hum. Genet.* 64: 1340–1356, 1999.
- [41931] 15483. Young, P. J.; Man, N.; Lorson, C. L.; Le, T. T.; Androphy, E. J.; Burghes, A. H. M.; Morris, G. E.: The exon 2b region of the spinal muscular atrophy protein, SMN, is involved in self-association and SIP1 binding. *Hum. Molec. Genet.* 9: 2869–2877, 2000.
- [41932] 15484. David, C.; Solimena, M.; De Camilli, P.: Autoimmu–

nity in stiff-mansyndrome with breast cancer is targeted to the C-terminal region of human amphiphysin, a protein similar to the yeast proteins, Rvs167 and Rvs161. FEBS Lett. 351: 73–79, 1994.

- [41933] 15485. De Camilli, P.; Thomas, A.; Cofield, R.; Folli, F.; Lichte, B.; Piccolo, G.; Meinck, H.-M.; Austoni, M.; Fassetta, G.; Bottazzo, G.; Bates, D.; Cartledge, N.; Solimena, M.; Kilimann, M. W.: The synaptic vesicle-associated protein amphiphysin is the 128-kD autoantigen of stiff-man syndrome with breast cancer. J. Exp. Med. 178: 2219–2223, 1993.
- [41934] 15486. Jenkins, N. A.; Gilbert, D. J.; Yamamoto, R.; Kilimann, M. W.; Copeland, N. G.: Amphiphysin (Amph) maps to the proximal region of mouse chromosome 13. Genomics 28: 363–365, 1995.
- [41935] 15487. Lichte, B.; Veh, R. W.; Meyer, H. E.; Kilimann, M. W.: Amphiphysin, a novel protein associated with synaptic vesicles. EMBO J. 11: 2521–2530, 1992.
- [41936] 15488. Yamamoto, R.; Li, X.; Francke, U.; Kilimann, M. W.: Genetic mapping of the human amphiphysin gene (AMPH) at 7p14–p13 excludes its involvement in retinitis pigmentosa 9 or dominant cystoid macular dystrophy. (Letter) Am. J. Hum. Genet. 57: 970–972, 1995.

- [41937] 15489.Yamamoto, R.; Li, X.; Winter, S.; Francke, U.; Kili-
mann, M. W.: Primary structure of human amphiphysin,
the dominant autoantigen of paraneoplastic Stiff-Man syn-
drome, and mapping of its gene (AMPH) to chromosome
7p13-p14. *Hum. Molec. Genet.* 4: 265-268, 1995.
- [41938] 15490.Kingsmore, S. F.; Suh, D.; Seldin, M. F.: Genetic
mapping of the glycine receptor alpha-3 subunit on mouse
chromosome 8. *Mammalian Genome* 5: 831-832, 1994.
- [41939] 15491.Kuhse, J.; Schmieden, V.; Betz, H.: Identification
and functional expression of a novel ligand binding sub-
unit of the inhibitory glycine receptor. *J. Biol. Chem.* 265:
22317-22320, 1990.
- [41940] 15492.Nikolic, Z.; Laube, B.; Weber, R. G.; Lichter, P.;
Kioschis, P.; Poustka, A.; Mulhardt, C.; Becker, C.-M.: The
human glycine receptor subunit alpha-3: GLRA3 gene
structure, chromosomal localization, and functional char-
acterization of alternative transcripts. *J. Biol. Chem.* 273:
19708-19714, 1998.
- [41941] 15493.Birkenmeier, E. H.; Rowe, L. B.; Crossman, M. W.;
Gordon, J. I.: Ileal lipid-binding protein (Illbp) gene maps
to mouse chromosome 11. *Mammalian Genome* 5:
805-806, 1994.
- [41942] 15494.Oelkers, P.; Dawson, P. A.: Cloning and chromoso-

mal localization of the human ileal lipid-binding protein.

Biochim. Biophys. Acta 1257:199–202, 1995.

[41943] 15495. Johnson, D. W.; Berg, J. N.; Baldwin, M. A.; Gallione, C. J.; Marondel, I.; Yoon, S.-J.; Stenzel, T. T.; Speer, M.; Pericak-Vance, M. A.; Diamond, A.; Guttmacher, A. E.; Jackson, C. E.; Attisano, L.; Kucherlapati, R.; Porteous, M. E. M.; Marchuk, D. A.: Mutations in the activin receptor-like kinase 1 gene in hereditary haemorrhagic telangiectasia-type 2. Nature Genet. 13: 189–195, 1996.

[41944] 15496. Seimiya, H.; Sawabe, T.; Inazawa, J.; Tsuruo, T.: Cloning, expression and chromosomal localization of a novel gene for protein tyrosine phosphatase (PTP-U2) induced by various differentiation-inducing agents. Oncogene 10:1731–1738, 1995.

[41945] 15497. Wiggins, R. C.; Wiggins, J. E.; Goyal, M.; Wharram, B. L.; Thomas, P. E.: Molecular cloning of cDNAs encoding human GLEPP1, a membrane protein tyrosine phosphatase: characterization of the GLEPP1 protein distribution in human kidney and assignment of the GLEPP1 gene to human chromosome 12p12–p13. Genomics 27: 174–181, 1995.

[41946] 15498. Borsani, G.; Rugarli, E. I.; Tagliatela, M.; Wong, C.; Ballabio, A.: Characterization of a human and murine gene (CLCN3) sharing similarity to voltage-gated chloride

channels and to a yeast integral membraneprotein. *Genomics* 27: 131–141, 1995.

[41947] 15499.Mills, K. A.; Mathews, K. D.; Scherpbier–Heddema, T.; Buetow, K.H.; Baldini, A.; Ballabio, A.; Borsani, G.: Genetic and physicalmapping of a voltage–dependent chloride channel gene to human 4q32and to mouse 8. *Genomics* 36: 374–376, 1996.

[41948] 15500.Stobrawa, S. M.; Breiderhoff, T.; Takamori, S.; Engel, D.; Schweizer,M.; Zdebik, A. A.; Bosl, M. R.; Ruether, K.; Jahn, H.; Draguhn, A.;Jahn, R.; Jentsch, T. J.: Disruption of CLC–3, a chloride channelexpressed on synaptic vesicles, leads to a loss of the hippocampus. *Neuron* 29:185–196, 2001.

[41949] 15501.Taine, L.; Coupry, I.; Boisseau, P.; Saura, R.; Lacombe, D.; Arveiler,B.: Refined localisation of the voltage–gated chloride channel, CLCN3,to 4q33. *Hum. Genet.* 102: 178–181, 1998.

[41950] 15502.Yoshikawa, M.; Uchida, S.; Ezaki, J.; Rai, T.; Hayama, A.; Kobayashi,K.; Kida, Y.; Noda, M.; Koike, M.; Uchiyama, Y.; Marumo, F.; Kominami,E.; Sasaki, S.: CLC–3 deficiency leads to phenotypes similar to humanneuronal ceroid lipofuscinosis. *Genes Cells* 7: 597–605, 2002.

[41951] 15503.Pagliuca, A.; Bartoli, P. C.; Saccone, S.; Valle, G. D.;

Lania,L.: Molecular cloning of ID4, a novel dominant negative helix-loop-helix human gene on chromosome 6p21.3-p22. Genomics 27: 200-203, 1995.

[41952] 15504.Rigolet, M.; Rich, T.; Gross-Morand, M.-S.; Molina-Gomes, D.; Viegas-Pequignot,E.; Junien, C.: cDNA cloning, tissue distribution and chromosomal localization of the human ID4 gene. DNA Res. 5: 309-313, 1998.

[41953] 15505.Jia, S.; VanDusen, W. J.; Diehl, R. E.; Kohl, N. E.; Dixon, R.A. F.; Elliston, K. O.; Stern, A. M.; Friedman, P. A.: cDNA cloning and expression of bovine aspartyl (asparaginyl) beta-hydroxylase. J.Biol. Chem. 267: 14322-14327, 1992.

[41954] 15506.Korioth, F.; Gieffers, C.; Frey, J.: Cloning and characterization of the human gene encoding aspartyl beta-hydroxylase. Gene 150:395-399, 1994.

[41955] 15507.Lavaissiere, L.; Jia, S.; Nishiyama, M.; de la Monte, S.; Stern,A. M.; Wands, J. R.; Friedman, P. A.: Overexpression of human aspartyl(asparaginyl)-beta-hydroxylase in hepatocellular carcinoma and cholangiocarcinoma. J. Clin. Invest. 98:1313-1323, 1996.

[41956] 15508.Lim, K. Y.; Hong, C.-S.; Kim, D. H.: cDNA cloning and characterization of human cardiac junctin. Gene 255: 35-42, 2000.

- [41957] 15509.Scott, A. F.: Personal Communication. Baltimore, Md. 2/19/2001.
- [41958] 15510.Treves, S.; Feriotto, G.; Moccagatta, L.; Gambari, R.; Zorzato,F.: Molecular cloning, expression, functional characterization, chromosomallocalization, and gene structure of junctate, a novel integral calciumbinding protein of sarco(endo)plasmic reticulum membrane. J. Biol.Chem. 275: 39555–39568, 2000.
- [41959] 15511.Wetzel, G. T.; Ding, S.; Chen, F.: Molecular cloning of junctinfrom human and developing rabbit heart. Molec. Genet. Metab. 69:252–258, 2000.
- [41960] 15512.Zhou, P.; Sun, L. J.; Dotsch, V.; Wagner, G.; Verdine, G. L.:Solution structure of the core NFATC1/DNA complex. Cell 92: 687–696,1998.
- [41961] 15513.McCaffrey, P. G.; Luo, C.; Kerppola, T. K.; Jain, J.; Badalian,T. M.; Ho, A. M.; Burgeon, E.; Lane, W. S.; Lambert, J. N.; Curran,T.; Verdine, G. L.; Rao, A.; Hogan, P. G.: Isolation of the cyclosporin–sensitiveT cell transcription factor NFATp. Science 262: 750–754, 1993.
- [41962] 15514.Rengarajan, J.; Tang, B.; Glimcher, L. H.: NFATc2 and NFATc3 regulateTH2 differentiation and modulate TCR–responsiveness of naive TH cells. NatureImmun. 3: 48–54, 2002.

- [41963] 15515.Chan, K.; Han, X.-D.; Kan, Y. W.: An important function of Nrf2 in combating oxidative stress: detoxification of acetaminophen. *Proc.Nat. Acad. Sci.* 98: 4611–4616, 2001.
- [41964] 15516.Baud, V.; Chissoe, S. L.; Viegas-Pequignot, E.; Diriong, S.; N'Guyen,V. C.; Roe, B. A.; Lipinski, M.: EMR1, an unusual member in the family of hormone receptors with seven transmembrane segments. *Genomics* 26:334–344, 1995.
- [41965] 15517.McKnight, A. J.; Macfarlane, A. J.; Seldin, M. F.; Gordon, S.:Chromosome mapping of the Emr1 gene. *Mammalian Genome* 8: 946–947,1997.
- [41966] 15518.Atanasoski, S.; Toldo, S. S.; Malipiero, U.; Schreiber, E.; Fries,R.; Fontana, A.: Isolation of the human genomic brain-2/N-Oct 3 gene(POUF3) and assignment to chromosome 6q16. *Genomics* 26: 272–280,1995.
- [41967] 15519.Schreiber, E.; Tobler, A.; Malipiero, U.; Schaffner, W.; Fontana,A.: cDNA cloning of human N-Oct 3, a nervous-system specific POUdomain transcription factor binding to the octamer DNA motif. *NucleicAcids Res.* 21: 253–258, 1993.
- [41968] 15520.Xia, Y.-R.; Andersen, B.; Mehrabian, M.; Diep, A. T.; Warden, C.H.; Mohandas, T.; McEvilly, R. J.; Rosenfeld, M.

G.; Lusis, A. J.: Chromosomal organization of mammalian POU domain factors. *Genomics* 18:126–130, 1993.

[41969] 15521.Brass, N.; Heckel, D.; Sahin, U.; Pfreundschuh, M.; Sybrecht, G.W.; Meese, E.: Translation initiation factor eIF-4gamma is encoded by an amplified gene and induces an immune response in squamous cell carcinoma. *Hum. Molec. Genet.* 6: 33–39, 1997.

[41970] 15522.Gradi, A.; Imataka, H.; Svitkin, Y. V.; Rom, E.; Raught, B.; Morino, S.; Sonenberg, N.: A novel functional human eukaryotic translation initiation factor 4G. *Molec. Cell. Biol.* 18: 334–342, 1998.

[41971] 15523.Imataka, H.; Gradi, A.; Sonenberg, N.: A newly identified N-terminal amino acid sequence of human eIF4G binds poly(A)-binding protein and functions in poly(A)-dependent translation. *EMBO J.* 17: 7480–7489, 1998.

[41972] 15524.Imataka, H.; Sonenberg, N.: Human eukaryotic translation initiation factor 4G (eIF4G) possesses two separate and independent binding sites for eIF4A. *Molec. Cell. Biol.* 17: 6940–6947, 1997.

[41973] 15525.Yan, R.; Rhoads, R. E.: Human protein synthesis initiation factor eIF-4-gamma is encoded by a single gene (EIF4G) that maps to chromosome 3q27-qter. *Genomics*

26: 394–398, 1995.

- [41974] 15526. Yan, R.; Rychlik, W.; Etchison, D.; Rhoads, R. E.: Amino acid sequence of the human protein synthesis initiation factor eIF-4-gamma. *J. Biol. Chem.* 267: 23226–23231, 1992.
- [41975] 15527. Corrigall, A. V.; Hift, R. J.; Hancock, V.; Meissner, D.; Davids, L.; Kirsch, R. E.; Meissner, P. N.: Identification and characterisation of a deletion (537delAT) in the proto-porphyrinogen oxidase gene in a South African variegate porphyria family. *Hum. Mutat.* 12: 403–407, 1998.
- [41976] 15528. Aso, T.; Haque, D.; Fukudome, K.; Brower, C. S.; Conaway, J. W.; Conaway, R. C.: A human cDNA encoding the 110-kDa A subunit of RNA polymerase II transcription factor elongin. *Gene* 168: 277–278, 1996.
- [41977] 15529. Aso, T.; Lane, W. S.; Conaway, J. W.; Conaway, R. C.: Elongin(SIII): a multisubunit regulator of elongation by RNA polymerase II. *Science* 269: 1439–1443, 1995.
- [41978] 15530. Aso, T.; Mokady, N.; Haque, D.; Conaway, R. C.; Conaway, J. W.: Assignment of a human gene encoding the 110-kDa subunit of general transcription factor elongin (SIII) to chromosome 1p36.1. *Genomics* 30: 393–394, 1995.
- [41979] 15531. Vikkula, M.; Boon, L. M.; Carraway, K. L., III;

Calvert, J. T.; Diamonti, A. J.; Goumnerov, B.; Pasyk, K. A.; Marchuk, D. A.; Warman, M. L.; Cantley, L. C.; Mulliken, J. B.; Olsen, B. R.: Vascular dysmorphogenesis caused by an activating mutation in the receptor tyrosine kinase TIE2. *Cell* 87:1181–1190, 1996.

[41980] 15532. Iwata, T.; Kogame, K.; Toki, T.; Yokoyama, M.; Yamamoto, M.; Ito, E.: Structure and chromosome mapping of the human small maf genes MAFG and MAFK. *Cytogenet. Cell Genet.* 82: 88–90, 1998.

[41981] 15533. Motohashi, H.; Katsuoka, F.; Shavit, J. A.; Engel, J. D.; Yamamoto, M.: Positive or negative MARE-dependent transcriptional regulation is determined by the abundance of small Maf proteins. *Cell* 103:865–875, 2000.

[41982] 15534. Peters, L. L.; Eicher, E. M.: The ubiquitous subunit of the globin enhancer-binding protein NF-E2 (Nfe2u) maps to mouse chromosome 5. *Genomics* 22:490–491, 1994.

[41983] 15535. Shavit, J. A.; Motohashi, H.; Onodera, K.; Akasaka, J.; Yamamoto, M.; Engel, J. D.: Impaired megakaryopoiesis and behavioral defects in mafG-null mutant mice. *Genes Dev.* 12: 2164–2174, 1998.

[41984] 15536. Toki, T.; Itoh, J.; Kitazawa, J.; Arai, K.; Hatakeyama, K.; Akasaka, J.; Igarashi, K.; Nomura, N.; Yokoyama, M.;

Yamamoto, M.; Ito, E.: Human small Maf proteins form heterodimers with CNC family transcription factors and recognize the NF-E2 motif. *Oncogene* 14: 1901–1910, 1997.

- [41985] 15537. Yang, B.; He, B.; Abdel-Halim, S. M.; Tibell, A.; Brendel, M. D.; Bretzel, R. G.; Efendic, S.; Hillert, J.: Molecular cloning of a full-length cDNA for human type 3 adenylyl cyclase and its expression in human islets. *Biochem. Biophys. Res. Commun.* 254: 548–551, 1999.
- [41986] 15538. Arden, K. C.; Viars, C. S.; Weiss, S.; Argentin, S.; Nemer, M.: Localization of the human B-type natriuretic peptide precursor (NPPB) gene to chromosome 1p36. *Genomics* 26: 385–389, 1995.
- [41987] 15539. Jordan, C. E.; White, R. I., Jr.; Fischer, K. C.; Neill, C.; Dorst, J. P.: The scoliosis of congenital heart disease. *Am. Heart J.* 84:463–469, 1972.
- [41988] 15540. Ogawa, Y.; Itoh, H.; Yoshitake, Y.; Inoue, M.; Yoshimasa, T.; Serikawa, T.; Nakao, K.: Molecular cloning and chromosomal assignment of the mouse C-type natriuretic peptide (CNP) gene (Nppc): comparison with the human CNP gene (NPPC). *Genomics* 24: 383–387, 1994.
- [41989] 15541. Reckles, L. N.; Peterson, H. A.; Weidman, W. H.; Bianco, A. J., Jr. The association of scoliosis and congenital

heart disease. *J. Bone Joint Surg.* 57A: 449–455, 1975.

- [41990] 15542. Steinhilber, M. E.: Structure, expression, and genomic mapping of the mouse natriuretic peptide type-B gene. *Circ. Res.* 72: 984–992, 1993.
- [41991] 15543. Tamura, N.; Ogawa, Y.; Chusho, H.; Nakamura, K.; Nakao, K.; Suda, M.; Kasahara, M.; Hashimoto, R.; Katsura, G.; Mukoyama, M.; Itoh, H.; Saito, Y.; Tanaka, I.; Otani, H.; Katsuki, M.; Nakao, K.: Cardiac fibrosis in mice lacking brain natriuretic peptide. *Proc. Nat. Acad. Sci.* 97: 4239–4244, 2000.
- [41992] 15544. Chawengsaksophak, K.; James, R.; Hammond, V. E.; Kontgen, F.; Beck, F.: Homeosis and intestinal tumours in *Cdx2* mutant mice. *Nature* 386:84–87, 1997.
- [41993] 15545. Drummond, F.; Putt, W.; Fox, M.; Edwards, Y. H.: Cloning and chromosome assignment of the human *CDX2* gene. *Ann. Hum. Genet.* 61: 393–400, 1997.
- [41994] 15546. German, M. S.; Wang, J.; Fernald, A. A.; Espinosa, R., III; LeBeau, M. M.; Bell, G. I.: Localization of the genes encoding two transcription factors, *LMX1* and *CDX3*, regulating insulin gene expression to human chromosomes 1 and 13. *Genomics* 24: 403–404, 1994.
- [41995] 15547. Woodford-Richens, K. L.; Halford, S.; Rowan, A.; Bevan, S.; Aaltonen, L. A.; Wasan, H.; Bicknell, D.; Bodmer,

W. F.; Houlston, R. S.; Tomlinson, I. P. M.: CDX2 mutations do not account for juvenile polyposis or Peutz-Jeghers syndrome and occur infrequently in sporadic colorectal cancers. *Brit. J. Cancer* 84: 1314–1316, 2001.

[41996] 15548. Dubbink, H. J.; Verkaik, N. S.; Faber, P. W.; Trapman, J.; Schroder, F. H.; Romijn, J. C.: Tissue-specific and androgen-regulated expression of human prostate-specific transglutaminase. *Biochem. J.* 315: 901–908, 1996.

[41997] 15549. Gentile, V.; Grant, F. J.; Porta, R.; Baldini, A.: Localization of the human prostate transglutaminase (type IV) gene (TGM4) to chromosome 3p21.33–p22 by fluorescence in situ hybridization. *Genomics* 27: 219–220, 1995.

[41998] 15550. Grant, F. J.; Taylor, D. A.; Sheppard, P. O.; Mathewes, S. L.; Lint, W.; Vanaja, E.; Bishop, P. D.; O'Hara, P. J.: Molecular cloning and characterization of a novel transglutaminase cDNA from a human prostate cDNA library. *Biochem. Biophys. Res. Commun.* 203: 1117–1123, 1994.

[41999] 15551. Barker, H. M.; Brewis, N. D.; Street, A. J.; Spurr, N. K.; Cohen, P. T. W.: Three genes for protein phosphatase 1 map to different human chromosomes: sequence, expression and gene localisation of protein serine/threonine phosphatase 1 beta (PPP1CB). *Biochim. Biophys. Acta* 1220: 212–218, 1994.

- [42000] 15552.Saadat, M.; Kakinoki, Y.; Mizuno, Y.; Kikuchi, K.; Yoshida, M.C.: Chromosomal localization of human, rat, and mouse protein phosphatase type 1 beta catalytic subunit genes (PPP1CB) by fluorescence in situ hybridization. *Jpn. J. Genet.* 69: 697–700, 1994.
- [42001] 15553.King, L. S.; Nielsen, S.; Agre, P.: Aquaporins and the respiratory system: advice for a lung investigator. *J. Clin. Invest.* 105: 15–16, 2000.
- [42002] 15554.Lee, M. D.; Bhakta, K. Y.; Raina, S.; Yonescu, R.; Griffin, C.A.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Preston, G. M.; Agre, P.: The human aquaporin–5 gene: molecular characterization and chromosomal localization. *J. Biol. Chem.* 271: 8599–8604, 1996.
- [42003] 15555.Ma, T.; Fukuda, N.; Song, Y.; Matthay, M. A.; Verkman, A. S.: Lung fluid transport in aquaporin–5 knockout mice. *J. Clin. Invest.* 105:93–100, 2000.
- [42004] 15556.Raina, S.; Preston, G. M.; Guggino, W. B.; Agre, P.: Molecular cloning and characterization of an aquaporin cDNA from salivary, lacrimal, and respiratory tissues. *J. Biol. Chem.* 270: 1908–1912, 1995.
- [42005] 15557.Padilla, C. A.; Bajalica, S.; Lagercrantz, J.; Holmgren, A.: The gene for human glutaredoxin (GLRX) is localized to human chromosome 5q14. *Genomics* 32: 455–457,

1996.

- [42006] 15558. Padilla, C. A.; Martinez-Galisteo, E.; Barcena, J. A.; Spyrou, G.; Holmgren, A.: Purification from placenta, amino acid sequence, structure comparisons and cDNA cloning of human glutaredoxin. *Europ. J. Biochem.* 227: 27–34, 1995.
- [42007] 15559. Raghavachari, N.; Krysan, K.; Xing K.; Lou, M. F.: Regulation of thioltransferase expression in human lens epithelial cells. *Invest. Ophthalm. Vis. Sci.* 42: 1002–1008, 2001.
- [42008] 15560. Corset, V.; Nguyen-Ba-Charvet, K. T.; Forcet, C.; Moyse, E.; Chedotal, A.; Mehlen, P.: Netrin-1-mediated axon outgrowth and cAMP production requires interaction with adenosine A2b receptor. *Nature* 407: 747–750, 2000.
- [42009] 15561. Mino, R. P.; Spoerri, P. E.; Caballero, S.; Player, D.; Belardinelli, L.; Biaggioni, I.; Grant, M. B.: Adenosine receptor antagonists and retinal neovascularization in vivo. *Invest. Ophthalm. Vis. Sci.* 42: 3320–3324, 2001.
- [42010] 15562. Stein, E.; Zou, Y.; Poo, M.; Tessier-Lavigne, M.: Binding of DCC by netrin-1 to mediate axon guidance independent of adenosine A2B receptor activation. *Science* 291: 1976–1982, 2001.
- [42011] 15563. Townsend-Nicholson, A.; Baker, E.; Sutherland, G.

R.; Schofield, P. R.: Localization of the adenosine A2b receptor subtype gene (ADORA2B) to chromosome 17p11.2–p12 by FISH and PCR screening of somatic cell-hybrids. *Genomics* 25: 605–607, 1995.

[42012] 15564. von Gall, C.; Garabette, M. L.; Kell, C. A.; Frenzel, S.; Dehghani, F.; Schumm–Draeger, P.–M.; Weaver, D. R.; Korf, H.–W.; Hastings, M. H.; Stehle, J. H.: Rhythmic gene expression in pituitary depends on heterologous sensitization by the neurohormone melatonin. *Nature Neurosci.* 5: 234–238, 2002.

[42013] 15565. Becq, F.; Hamon, Y.; Bajetto, A.; Gola, M.; Verrier, B.; Chimini, G.: ABC1, an ATP binding cassette transporter required for phagocytosis of apoptotic cells, generates a regulated anion flux after expression in *Xenopus laevis* oocytes. *J. Biol. Chem.* 272: 2695–2699, 1997.

[42014] 15566. Decottignies, A.; Goffeau, A.: Complete inventory of the yeast ABC proteins. *Nature Genet.* 15: 137–145, 1997.

[42015] 15567. Guo, Z.; Inazu, A.; Yu, W.; Suzumura, T.; Okamoto, M.; Nohara, A.; Higashikata, T.; Sano, R.; Wakasugi, K.; Hayakawa, T.; Yoshida, K.; Suehiro, T.; Schmitz, G.; Mabuchi, H.: Double deletions and missense mutations in the first nucleotide-binding fold of the ATP–

binding cassette transporter A1 (ABCA1) gene in Japanese patients with Tangier disease. *J. Hum. Genet.* 47: 325–329, 2002.

- [42016] 15568. Hong, S. H.; Rhyne, J.; Zeller, K.; Miller, M.: Novel ABCA1 compound variant associated with HDL cholesterol deficiency. *Biochim. Biophys. Acta* 1587: 60–64, 2002.
- [42017] 15569. Huang, W.; Moriyama, K.; Koga, T.; Hua, H.; Ageta, M.; Kawabata, S.; Mawatari, K.; Imamura, T.; Eto, T.; Kawamura, M.; Teramoto, T.; Sasaki, J.: Novel mutations in ABCA1 gene in Japanese patients with Tangier disease and familial high density lipoprotein deficiency with coronary heart disease. *Biochim. Biophys. Acta* 1537: 71–78, 2001.
- [42018] 15570. Ishii, J.; Nagano, M.; Kujiraoka, T.; Ishihara, M.; Egashira, T.; Takada, D.; Tsuji, M.; Hattori, H.; Emi, M.: Clinical variant of Tangier disease in Japan: mutation of the ABCA1 gene in hypoalphalipoproteinemia with corneal lipidoses. *J. Hum. Genet.* 47: 366–369, 2002.
- [42019] 15571. Jennings, M. W.; Jones, R. W.; Wood, W. G.; Weatherall, D. J.: Analysis of an inversion within the human beta globin gene cluster. *Nucleic Acids Res.* 13: 2897–2906, 1985.
- [42020] 15572. Kulozik, A. E.; Bellan-Koch, A.; Kohne, E.; Kleihauer, E.: Deletion/inversion rearrangement of the beta-

globin gene cluster in a Turkish family with delta-beta(0)-thalassemia intermedia. *Blood* 79:2455–2459, 1992.

- [42021] 15573. Langmann, T.; Klucken, J.; Reil, M.; Liebisch, G.; Luciani, M.-F.; Chimini, G.; Kaminski, W. E.; Schmitz, G.: Molecular cloning of the human ATP-binding cassette transporter 1 (hABC1): evidence for sterol-dependent regulation in macrophages. *Biochem. Biophys. Res. Commun.* 257: 29–33, 1999.
- [42022] 15574. Lawn, R. M.; Wade, D. P.; Garvin, M. R.; Wang, X.; Schwartz, K.; Porter, J. G.; Seilhamer, J. J.; Vaughan, A. M.; Oram, J. F.: The Tangier disease gene product ABC1 controls the cellular apolipoprotein-mediated lipid removal pathway. *J. Clin. Invest.* 104: R25–R31, 1999.
- [42023] 15575. Luciani, M. F.; Denizot, F.; Savary, S.; Mattei, M. G.; Chimini, G.: Cloning of two novel ABC transporters mapping on human chromosome 9. *Genomics* 21: 150–159, 1994.
- [42024] 15576. Lapicka-Bodzioch, K.; Bodzioch, M.; Krull, M.; Kielar, D.; Probst, M.; Kiec, B.; Andrikovics, H.; Bottcher, A.; Hubacek, J.; Aslanidis, C.; Suttorp, N.; Schmitz, G.: Homogeneous assay based on 52 primer sets to scan for mutations of the ABCA1 gene and its application in genetic

analysis of a new patient with familial high-density lipoprotein deficiency syndrome. *Biochim. Biophys. Acta* 1537: 42–48, 2001.

- [42025] 15577. Marcil, M.; Boucher, B.; Krimbou, L.; Solymoss, B. C.; Davignon, J.; Frohlich, J.; Genest, J., Jr.: Severe familial HDL deficiency in French-Canadian kindreds: clinical, biochemical, and molecular characterization. *Arterioscler. Thromb. Vasc. Biol.* 15: 1015–1024, 1995.
- [42026] 15578. Marcil, M.; Yu, L.; Krimbou, L.; Boucher, B.; Oram, J. F.; Cohn, J. S.; Genest, J., Jr.: Cellular cholesterol transport and efflux in fibroblasts are abnormal in subjects with familial HDL deficiency. *Arterioscler. Thromb. Vasc. Biol.* 19: 159–169, 1999.
- [42027] 15579. McNeish, J.; Aiello, R. J.; Guyot, D.; Turi, T.; Gabel, C.; Aldinger, C.; Hoppe, K. L.; Roach, M. L.; Royer, L. J.; de Wet, J.; Broccardo, C.; Chimini, G.; Francone, O. L.: High density lipoprotein deficiency and foam cell accumulation in mice with targeted disruption of ATP-binding cassette transporter-1. *Proc. Nat. Acad. Sci.* 97: 4245–4250, 2000.
- [42028] 15580. Pullinger, C. R.; Hakamata, H.; Duchateau, P. N.; Eng, C.; Aouizerat, B. E.; Cho, M. H.; Fielding, C. J.; Kane, J. P.: Analysis of hABC1 gene 5-prime end: additional peptide sequence, promoter region, and four polymorphisms.

Biochem. Biophys. Res. Commun. 271: 451–455,2000.

- [42029] 15581. Petit, M. M. R.; Schoenmakers, E. F. P. M.; Huysmans, C.; Geurts, J. M. W.; Mandahl, N.; Van de Ven, W. J. M.: LHFP, a novel translocation partner gene of HMGIC in a lipoma, is a member of a new family of LHFP-like genes. Genomics 57: 438–441, 1999.
- [42030] 15582. Nishita, Y.; Yoshida, I.; Sado, T.; Takagi, N.: Genomic imprinting and chromosomal localization of the human MEST gene. Genomics 36: 539–542, 1996.
- [42031] 15583. Kohen, R.; Metcalf, M. A.; Khan, N.; Druck, T.; Huebner, K.; Lachowicz, J. E.; Meltzer, H. Y.; Sibley, D. R.; Roth, B. L.; Hamblin, M. W.: Cloning, characterization, and chromosomal localization of a human 5-HT-6 serotonin receptor. J. Neurochem. 66: 47–56, 1996.
- [42032] 15584. Frank, J.; Aita, V. M.; Ahmad, W.; Lam, H.; Wolff, C.; Christiano, A. M.: Identification of a founder mutation in the protoporphyrinogen oxidase gene in variegate porphyria patients from Chile. Hum. Hered. 51: 160–168, 2001.
- [42033] 15585. Frank, J.; Jugert, F. K.; Breitkopf, C.; Goerz, G.; Merk, H. F.; Christiano, A. M.: Recurrent missense mutation in the protoporphyrinogen oxidase gene underlies variegate porphyria. Am. J. Med. Genet. 79: 22–26, 1998.

- [42034] 15586.Orso, E.; Broccardo, C.; Kaminski, W. E.; Bottcher, A.; Liebisch,G.; Drobnik, W.; Gotz, A.; Chambenoit, O.; Diederich, W.; Langmann,T.; Spruss, T.; Luciani, M.-F.; Rothe, G.; Lackner, K. J.; Chimini,G.; Schmitz, G.: Transport of lipids from Golgi to plasma membrane is defective in Tangier disease patients and Abc1-deficient mice. *NatureGenet.* 24: 192–196, 2000.
- [42035] 15587.Hift, R. J.; Meissner, P. N.; Corrigall, A. V.; Ziman, M. R.; Petersen,L. A.; Meissner, D. M.; Davidson, B. P.; Sutherland, J.; Dailey, H.A.; Kirsch, R. E.: Variegate porphyria in South Africa, 1688–1996:new developments in an old disease. *S. Afr. Med. J.* 87: 722–731,1997.
- [42036] 15588.Hift, R. J.; Meissner, P. N.; Todd, G.; Kirby, P.; Bilsland, D.;Collins, P.; Ferguson, J.; Moore, M. R.: Homozygous variegate porphyria:an evolving clinical syndrome. *Postgrad. Med. J.* 69: 781–786, 1993.
- [42037] 15589.Lam, H.; Dragan, L.; Tsou, H. C.; Merk, H.; Peacocke, M.; Goerz,G.; Sassa, S.; Poh-Fitzpatrick, M.; Bickers, D. R.; Christiano, A.M.: Molecular basis of variegate porphyria: a de novo insertion mutation in the protoporphyrinogen oxidase gene. *Hum. Genet.* 99: 126–129,1997.
- [42038] 15590.Puy, H.; Robreau, A.-M.; Rosipal, R.; Nordmann, Y.;

Deybach, J.-C.: Protoporphyrinogen oxidase: complete genomic sequence and polymorphisms in the human gene. *Biochem. Biophys. Res. Comm.* 226: 226–230, 1996.

[42039] 15591. Roberts, A. G.; Puy, H.; Dailey, T. A.; Morgan, R. R.; Whatley, S. D.; Dailey, H. A.; Martasek, P.; Nordmann, Y.; Deybach, J.-C.; Elder, G. H.: Molecular characterization of homozygous variegate porphyria. *Hum. Molec. Genet.* 7: 1921–1925, 1998.

[42040] 15592. Honda, H.; Inazawa, J.; Nishida, J.; Yazaki, Y.; Hirai, H.: Molecular cloning, characterization, and chromosomal localization of a novel protein-tyrosine phosphatase, HPTP. *Blood* 84: 4186–4194, 1994.

[42041] 15593. Ostman, A.; Yang, Q.; Tonks, N. K.: Expression of DEP-1, a receptor-like protein-tyrosine-phosphatase, is enhanced with increasing cell density. *Proc. Nat. Acad. Sci.* 91: 9680–9684, 1994.

[42042] 15594. Katoh, M.; Hirai, M.; Sugimura, T.; Terada, M.: Cloning and characterization of MST, a novel (putative) serine/threonine kinase with SH3 domain. *Oncogene* 10: 1447–1451, 1995.

[42043] 15595. Morgan, H. D.; Sutherland, H. G. E.; Martin, D. I. K.; Whitelaw, E.: Epigenetic inheritance at the agouti locus in the mouse. *Nature Genet.* 23: 314–318, 1999.

- [42044] 15596. Jacoby, A. S.; Webb, G. C.; Liu, M. L.; Kofler, B.; Hort, Y. J.; Fathi, Z.; Bottema, C. D. K.; Shine, J.; Iismaa, T. P.: Structural organization of the mouse and human GALR1 galanin receptor genes (Galnr and GALNR) and chromosomal localization of the mouse gene. *Genomics* 45:496–508, 1997.
- [42045] 15597. Tomoda, T.; Kurashige, T.; Moriki, T.; Yamamoto, H.; Fujimoto, S.; Taniguchi, T.: Enhanced expression of poly(ADP-ribose) synthetase gene in malignant lymphoma. *Am. J. Hemat.* 37: 223–227, 1991.
- [42046] 15598. Vasquez, K. M.; Marburger, K.; Intody, Z.; Wilson, J. H.: Manipulating the mammalian genome by homologous recombination. *Proc. Nat. Acad. Sci.* 98: 8403–8410, 2001.
- [42047] 15599. Yu, S.-W.; Wang, H.; Poitras, M. F.; Coombs, C.; Bowers, W. J.; Federoff, H. J.; Poirier, G. G.; Dawson, T. M.; Dawson, V. L.: Mediation of poly(ADP-ribose) polymerase-1-dependent cell death by apoptosis-inducing factor. *Science* 297: 259–263, 2002.
- [42048] 15600. Zabel, B. U.; Herzog, H.; Schneider, R.; Auer, B.; Hirsch-Kauffmann, M.; Schweiger, M.: Chromosomal sublocalization of the gene for human poly (ADP-ribose) polymerase (NAD⁺ ADP-ribosyltransferase) at 1q41–42. (Abstract) *Cytogenet. Cell Genet.* 51: 1115,

1989.

- [42049] 15601.Hood, L.; Kronenberg, M.; Hunkapiller, T.: T cell antigen receptors and the immunoglobulin supergene family. *Cell* 40: 225–229, 1985.
- [42050] 15602.de Villiers, J. N. P.; Hillermann, R.; Loubser, L.; Kotze, M. J.: Spectrum of mutations in the HFE gene implicated in haemochromatosis and porphyria. *Hum. Molec. Genet.* 8: 1517–1522, 1999.
- [42051] 15603.Deybach, J.-C.; Puy, H.; Robreau, A.-M.; Lamoril, J.; Da Silva, V.; Grandchamp, B.; Nordmann, Y.: Mutations in the protoporphyrinogen oxidase gene in patients with variegate porphyria. *Hum. Molec. Genet.* 5:407–410, 1996.
- [42052] 15604.Meissner, P. N.; Dailey, T. A.; Hift, R. J.; Ziman, M.; Corrigan, A. V.; Roberts, A. G.; Meissner, D. M.; Kirsch, R. E.; Dailey, H.A.: A R59W mutation in human protoporphyrinogen oxidase results in decreased enzyme activity and is prevalent in South Africans with variegate porphyria. *Nature Genet.* 13: 95–97, 1996.
- [42053] 15605.Kalman, K.; Nguyen, A.; Tseng-Crank, J.; Dukes, I. D.; Chandy, G.; Hustad, C. M.; Copeland, N. G.; Jenkins, N. A.; Mohrenweiser, H.; Brandriff, B.; Cahalan, M.; Gutman, G. A.; Chandy, K. G.: Genomic organization, chromosomal localization, tissue distribution, and biophysical character-

ization of a novel mammalian Shaker-related voltage-gated potassium channel, Kv1.7. *J. Biol. Chem.* 273: 5851–5857, 1998.

- [42054] 15606. Stewart, S. L.; Rosenberg, H.; Fletcher, J. E.: Failure to identify the ryanodine receptor G1021A mutation in a large North American population with malignant hyperthermia. *Clin. Genet.* 54: 358–361, 1998.
- [42055] 15607. Orkiszewski, K. G.; Tedesco, T. A.; Mellman, W. J.; Croce, C. M.: Linkage relationship between the genes for thymidine kinase and galactokinase in different primates. *Somat. Cell Genet.* 2: 21–26, 1976.
- [42056] 15608. Dunham, I.; Collins, J.; Wadey, R.; Scambler, P.: Possible role for COMT in psychosis associated with velocardio-facial syndrome. (Letter) *Lancet* 340: 1361–1362, 1992.
- [42057] 15609. Pulkka, A.; Ihalainen, R.; Suorsa, A.; Riviere, M.; Szpirer, J.; Pajunen, A.: Structures and chromosomal localizations of two rat genes encoding S-adenosylmethionine decarboxylase. *Genomics* 16: 342–349, 1993.
- [42058] 15610. Radford, D. M.; Eddy, R.; Haley, L.; Henry, W. M.; Pegg, A. E.; Pajunen, A.; Shows, T. B.: Gene sequences coding for S-adenosylmethionine decarboxylase are present on human chromosome 6 and the X and are not

amplified in colon neoplasia. *Cytogenet. Cell Genet.* 49: 285–288,1989.

- [42059] 15611.Grignani, F.; De Matteis, S.; Nervi, C.; Tomassoni, L.; Gelmetti,V.; Cioce, M.; Fanelli, M.; Ruthardt, M.; Ferrara, F. F.; Zamir, I.;Seiser, C.; Grignani, F.; Lazar, M. A.; Minucci, S.; Pelicci, P. G.: Fusion proteins of the retinoic acid receptor- α recruit histone deacetylase in promyelocytic leukaemia. *Nature* 391: 815–818, 1998.
- [42060] 15612.Lin, R. J.; Nagy, L.; Inoue, S.; Shao, W.; Miller, W. H., Jr.;Evans, R. M.: Role of the histone deacetylase complex in acute promyelocytic leukaemia. *Nature* 391: 811–814, 1998.
- [42061] 15613.Mendez, R.; Hake, L. E.; Andresson, T.; Littlepage, L. E.; Ruderman,J. V.; Richter, J. D.: Phosphorylation of CPE binding factor by Eg2 regulates translation of c-mos mRNA. *Nature* 404: 302–307, 2000.
- [42062] 15614.Ahrendt, S. A.; Decker, P. A.; Alawi, E. A.; Zhu, Y.; Sanchez-Cespedes,M.; Yang, S. C.; Haasler, G. B.; Kajdacsy-Balla, A.; Demeure, M. J.;Sidransky, D.: Cigarette smoking is strongly associated with mutation of the K-ras gene in patients with primary adenocarcinoma of the lung. *Cancer* 92:1525–1530, 2001.
- [42063] 15615.Almoguera, C.; Shibata, D.; Forrester, K.; Martin, J.;

Arnheim,N.; Perucho, M.: Most human carcinomas of the exocrine pancreas containmutant c-K-ras genes. Cell 53: 549–554, 1988.

[42064] 15616.Andreyev, H. J. N.; Tilsed, J. V. T.; Cunningham, D.; Sampson,S. A.; Norman, A. R.; Schneider, H. J.; Clarke, P. A.: K-ras mutationsin patients with early colorectal cancers. Gut 41: 323–329, 1997.

[42065] 15617.Bollag, G.; Adler, F.; elMasry, N.; McCabe, P. C.; Connor, E.,Jr.; Thompson, P.; McCormick, F.; Shannon, K.: Biochemical characterizationof a novel KRAS insertion mutation from a human leukemia. J. Biol.Chem. 271: 32491–32494, 1996.

[42066] 15618.Burmer, G. C.; Loeb, L. A.: Mutations in the KRAS2 oncogene duringprogressive stages of human colon carcinoma. Proc. Nat. Acad. Sci. 86:2403–2407, 1989.

[42067] 15619.Capon, D. J.; Seeburg, P. H.; McGrath, J. P.; Hayflick, J. S.;Edman, U.; Levinson, A. D.; Goeddel, D. V.: Activation of Ki-ras2gene in human colon and lung carcinomas by two different point mutations. Nature 304:507–513, 1983.

[42068] 15620.Cubilla, A. L.; Fitzgerald, P. J. :Cancer Res. 36: 2690–2698,1976.

[42069] 15621.Der, C. J.; Cooper, G. M.: Altered gene products are

associated with activation of cellular ras-k genes in human lung and colon carcinomas. Cell 32:201–208, 1983.

[42070] 15622. Feig, L. A.; Bast, R. C., Jr.; Knapp, R. C.; Cooper, G. M.: Somatic activation of ras-K gene in a human ovarian carcinoma. Science 223:698–701, 1984.

[42071] 15623. Grimmond, S. M.; Raghavan, D.; Russell, P. J.: Detection of a rare point mutation in Ki-ras of a human bladder cancer xenograft by polymerase chain reaction and direct sequencing. Urol. Res. 20:121–126, 1992.

[42072] 15624. Hayashi, N.; Sugai, S.; Ito, I.; Nakamori, S.; Ogawa, M.; Nakamura, Y.: Ethnic difference in the pattern of K-ras oncogene mutations in human colorectal cancers. Hum. Mutat. 8: 258–261, 1996.

[42073] 15625. Tegazzin, V.; Accorsi, A.; Gritti, G.; Arcelli, L.; Di Giovanni, A.: MH fulminant reaction after eight anaesthetics: a case report. Minerva Anesthesiol. 60: 217–219, 1994.

[42074] 15626. Tilgen, N.; Zorzato, F.; Halliger-Keller, B.; Muntoni, F.; Sewry, C.; Palumucci, L. M.; Schneider, C.; Hauser, E.; Lehmann-Horn, F.; Muller, C. R.; Treves, S.: Identification of four novel mutations in the C-terminal membrane spanning domain of the ryanodine receptor1: association with central core disease and alteration of calcium homeostasis.

Hum. Molec. Genet. 10: 2879–2887, 2001.

[42075] 15627.Tobin, J. R.; Jason, D. R.; Challa, V. R.; Nelson, T. E.; Sambuughin,N.: Malignant hyperthermia and apparent heat stroke. (Letter) JAMA 286:168–169, 2001.

[42076] 15628.Wang, S.–Q.; Song, L.–S.; Lakatta, E. G.; Cheng, H.: Ca(2+) signallingbetween single L–type Ca(2+) channels and ryanodine receptors in heartcells. Nature 410: 592–596, 2001.

[42077] 15629.Zhang, Y.; Chen, H. S.; Khanna, V. K.; De Leon, S.; Phillips,M. S.; Schappert, K.; Britt, B. A.; Brownell, A. K. W.; MacLennan,D. H.: A mutation in the human ryanodine receptor gene associatedwith central core disease. Nature Genet. 5: 46–50, 1993.

[42078] 15630.Zorzato, F.; Fujii, J.; Otsu, K.; Phillips, M.; Green, N. M.;Lai, F. A.; Meissner, G.; MacLennan, D. H.: Molecular cloning ofcDNA encoding human and rabbit forms of the Ca(2+) release channel(ryanodine receptor) of skeletal muscle sarcoplasmic reticulum. J.Biol. Chem. 265: 2244–2256, 1990.

[42079] 15631.Marx, S. O.; Reiken, S.; Hisamatsu, Y.; Jayaraman, T.; Burkhoff,D.; Rosemlit, N.; Marks, A. R.: PKA phosphorylation dissociatesFKBP12.6 from the calcium release channel (ryanodine receptor): defectiveregulation in failing

hearts. Cell 101: 365–376, 2000.

- [42080] 15632. Mattei, M. G.; Giannini, G.; Moscatelli, F.; Sorrentino, V.: Chromosomal localization of murine ryanodine receptor genes RYR1, RYR2, and RYR3 by in situ hybridization. Genomics 22: 202–204, 1994.
- [42081] 15633. Bertocchini, F.; Ovitt, C. E.; Conti, A.; Barone, V.; Scholer, H. R.; Bottinelli, R.; Reggiani, C.; Sorrentino, V.: Requirement for the ryanodine receptor type 3 for efficient contraction in neonatal skeletal muscles. EMBO J. 16: 6956–6963, 1997.
- [42082] 15634. Futatsugi, A.; Kato, K.; Ogura, H.; Li, S.-T.; Nagata, E.; Kuwajima, G.; Tanaka, K.; Itohara, S.; Mikoshiba, K.: Facilitation of NMDAR-independent LTP and spatial learning in mutant mice lacking ryanodine receptor type 3. Neuron 24: 701–713, 1999.
- [42083] 15635. Hakamata, Y.; Nakai, J.; Takeshima, H.; Imoto, K.: Primary structure and distribution of a novel ryanodine receptor/calcium release channel from rabbit brain. FEBS Lett. 312: 229–235, 1992.
- [42084] 15636. Leeb, T.; Brenig, B.: cDNA cloning and sequencing of the human ryanodine receptor type 3 (RYR3) reveals a novel alternative splice site in the RYR3 gene. FEBS Lett. 423: 367–370, 1998.

- [42085] 15637.Sorrentino, V.; Giannini, G.; Malzac, P.; Mattei, M. G.: Localization of a novel ryanodine receptor gene (RYR3) to human chromosome 15q14–q15 by in situ hybridization. *Genomics* 18: 163–165, 1993.
- [42086] 15638.Takeshima, H.; Ikemoto, T.; Nishi, M.; Nishiyama, N.; Shimuta, M.; Sugitani, Y.; Kuno, J.; Saito, I.; Saito, H.; Endo, M.; Iino, M.; Noda, T.: Generation and characterization of mutant mice lacking ryanodine receptor type 3. *J. Biol. Chem.* 271: 19649–19652, 1996.
- [42087] 15639.Bigler, R. D.; Bushkin, Y.; Chiorazzi, N.: S152 (CD27): a modulating disulfide-linked T cell activation antigen. *J. Immun.* 141: 21–28, 1988.
- [42088] 15640.Hendriks, J.; Gravestien, L. A.; Tesselaar, K.; van Lier, R. A.W.; Schumacher, T. N. M.; Borst, J.: CD27 is required for generation and long-term maintenance of T cell immunity. *Nature Immun.* 1: 433–440, 2000.
- [42089] 15641.Prasad, K. V. S.; Ao, Z.; Yoon, Y.; Wu, M. X.; Rizk, M.; Jacquot, S.; Schlossman, S. F.: CD27, a member of the tumor necrosis factor receptor family, induces apoptosis and binds to Siva, a proapoptotic protein. *Proc. Nat. Acad. Sci.* 94: 6346–6351, 1997.
- [42090] 15642.Arnaiz-Villena, A.; Perez-Aciego, P.; Ballestin, C.; Sotelo, T.; Perez-Seoane, C.; Martin-Villa, J. M.; Regueiro, J.

R.: Biochemical basis of a novel T lymphocyte receptor immunodeficiency by immunohistochemistry: a possible CD3-gamma abnormality. *Lab. Invest.* 64: 675-681, 1991.

[42091] 15643. Arnaiz-Villena, A.; Timon, M.; Corell, A.; Perez-Aciego, P.; Martin-Villa, J. M.; Regueiro, J. R.: Primary immunodeficiency caused by mutations in the gene encoding the CD3-gamma subunit of the T-lymphocyte receptor. *New Eng. J. Med.* 327: 529-533, 1992.

[42092] 15644. Das, S.; Cotter, F. E.; Gibbons, B.; Dhut, S.; Young, B. D.: CD3G is within 200 kb of the leukemic t(4;11) translocation breakpoint. *Genes Chromosomes Cancer* 3: 44-47, 1991.

[42093] 15645. Evans, G. A.; Lewis, K. A.; Lawless, G. M.: Molecular organization of the human CD3 gene family on chromosome 11q23. *Immunogenetics* 28: 365-373, 1988.

[42094] 15646. Krissansen, G. W.; Gorman, P. A.; Kozak, C. A.; Spurr, N. K.; Sheer, D.; Goodfellow, P. N.; Crumpton, M. J.: Chromosomal locations of the gene coding for the CD3 (T3) gamma-subunit of the human and mouse CD3/T-cell antigen receptor complexes. *Immunogenetics* 26: 258-266, 1987.

[42095] 15647. Krissansen, G. W.; Owen, M. J.; Verbi, W.; Crump-

ton, M. J.: Primary structure of the T3 gamma subunit of the T3/T cell antigen receptor complex deduced from cDNA sequences: evolution of the T3 gamma and delta subunits. EMBO J. 5: 1799–1808, 1986.

- [42096] 15648. Saito, H.; Koyama, T.; Georgopoulos, K.; Clevers, H.; Haser, W.G.; LeBien, T.; Tonegawa, S.; Terhorst, C.: Close linkage of the mouse and human CD3 gamma- and delta-chain genes suggests that their transcription is controlled by common regulatory elements. Proc. Nat. Acad. Sci. 84: 9131–9134, 1987.
- [42097] 15649. Tunnacliffe, A.; Buluwela, L.; Rabbitts, T. H.: Physical linkage of three CD3 genes on human chromosome 11. EMBO J. 6: 2953–2957, 1987.
- [42098] 15650. Tunnacliffe, A.; McGuire, R. S.: A physical linkage group in human chromosome band 11q23 covering a region implicated in leukocyte neoplasia. Genomics 8: 447–453, 1990.
- [42099] 15651. Tunnacliffe, A.; Olsson, C.; Buluwela, L.; Rabbitts, T. H.: Organization of the human CD3 locus on chromosome 11. Europ. J. Immun. 18: 1639–1642, 1988.
- [42100] 15652. Yunis, J. J.; Jones, C.; Madden, M. T.; Lu, D.; Mayer, M. G.: Gene order, amplification, and rearrangement of chromosome band 11q23 in hematologic malignancies.

Genomics 5: 84–90, 1989.

- [42101] 15653. Park, S.-H.; Bendelac, A.: CD1-restricted T-cell responses and microbial infection. *Nature* 406: 788–792, 2000.
- [42102] 15654. Beckman, E. M.; Porcelli, S. A.; Morita, C. T.; Behar, S. M.; Furlong, S. T.; Brenner, M. B.: Recognition of a lipid antigen by CD1-restricted α - β (+) T cells. *Nature* 372: 691–694, 1994.
- [42103] 15655. Sieling, P. A.; Chatterjee, D.; Porcelli, S. A.; Prigozy, T. I.; Mazzaccaro, R. J.; Soriano, T.; Bloom, B. R.; Brenner, M. B.; Kronenberg, M.; Brennan, P. J.; Modlin, R. L.: CD1-restricted T cell recognition of microbial lipoglycan antigens. *Science* 269: 227–230, 1995.
- [42104] 15656. Sieling, P. A.; Jullien, D.; Dahlem, M.; Tedder, T. F.; Rea, T. H.; Modlin, R. L.; Porcelli, S. A.: CD1 expression by dendritic cells in human leprosy lesions: correlation with effective host immunity. *J. Immunol.* 162: 1851–1858, 1999.
- [42105] 15657. Nishimura, K.; Taketani, S.; Inokuchi, H.: Cloning of a human cDNA for protoporphyrinogen oxidase by complementation in vivo of a hemG mutant of *Escherichia coli*. *J. Biol. Chem.* 270: 8076–8080, 1995.
- [42106] 15658. Roberts, A. G.; Whatley, S. D.; Daniels, J.; Holmans, P.; Fenton, I.; Owen, M. J.; Thompson, P.; Long, C.; Elder,

G. H.: Partial characterization and assignment of the gene for protoporphyrinogen oxidase and variegate porphyria to human chromosome 1q23. *Hum. Molec. Genet.* 4: 2387–2390, 1995.

[42107] 15659. Taketani, S.; Inazawa, J.; Abe, T.; Furukawa, T.; Kohno, H.; Tokunaga, R.; Nishimura, K.; Inokuchi, H.: The human protoporphyrinogen oxidase gene (PPOX): organization and location to chromosome 1. *Genomics* 29:698–703, 1995.

[42108] 15660. Warnich, L.; Kotze, M. J.; Groenewald, I. M.; Groenewald, J. Z.; van Brakel, M. G.; van Heerden, C. J.; de Villiers, J. N. P.; vande Ven, W. J. M.; Schoenmakers, E. F. P. M.; Taketani, S.; Retief, A. E.: Identification of three mutations and associated haplotypes in the protoporphyrinogen oxidase gene in South African families with variegate porphyria. *Hum. Molec. Genet.* 5: 981–984, 1996.

[42109] 15661. Haas, M.; Ward, D. C.; Lee, J.; Roses, A. D.; Clarke, V.; D'Eustachio, P.; Lau, D.; Vega-Saenz de Miera, E.; Rudy, B.: Localization of Shaw-related K(+) channel genes on mouse and human chromosomes. *Mammalian Genome* 4:711–715, 1993.

[42110] 15662. Albrecht, B.; Weber, K.; Pongs, O.: Characterization of a voltage-activated K-channel gene cluster on human

chromosome 12p13. Receptors Channels 3:213–220, 1995.

[42111] 15663.Grupe, A.; Schroter, K. H.; Ruppertsberg, J. P.; Stocker, M.; Drewes, T.; Beckh, S.; Pongs, O.: Cloning and expression of a human voltage-gated potassium channel: a novel member of the RCK potassium channel family. EMBOJ. 9: 1749–1756, 1990.

[42112] 15664.Klocke, R.; Roberds, S. L.; Tamkun, M. M.; Grone-meier, M.; Augustin, A.; Albrecht, B.; Pongs, O.; Jockusch, H.: Chromosomal mapping in the mouse of eight K(+)-channel genes representing the four Shaker-like subfamilies Shaker, Shab, Shaw, and Shal. Genomics 18: 568–574, 1993.

[42113] 15665.Grissmer, S.; Ghanshani, S.; Dethlefs, B.; McPherson, J. D.; Wasmuth, J. J.; Gutman, G. A.; Cahalan, M. D.; Chandy, K. G.: The Shaw-related potassium channel gene, Kv3.1, on human chromosome 11, encodes the type 1 K⁺ channel in T cells. J. Biol. Chem. 267: 20971–20979, 1992.

[42114] 15666.Ried, T.; Rudy, B.; Vega-Saenz de Miera, E.; Lau, D.; Ward, D.C.; Sen, K.: Localization of a highly conserved human potassium channel gene (NGK2-KV4; KCNC1) to chromosome 11p15. Genomics 15: 405–411, 1993.

- [42115] 15667.Stubbs, L.; Rinchik, E. M.; Goldberg, E.; Rudy, B.; Handel, M.A.; Johnson, D.: Clustering of six human 11p15 gene homologs within a 500-kb interval of proximal mouse chromosome 7. *Genomics* 24: 324–332, 1994.
- [42116] 15668.Curran, M. E.; Landes, G. M.; Keating, M. T.: Molecular cloning, characterization, and genomic localization of a human potassium channel gene. *Genomics* 12: 729–737, 1992.
- [42117] 15669.Al-Chalabi, A.; Andersen, P. M.; Nilsson, P.; Chioza, B.; Andersson, J. L.; Russ, C.; Shaw, C. E.; Powell, J. F.; Leigh, P. N.: Deletion of the heavy neurofilament subunit tail in amyotrophic lateral sclerosis. *Hum. Molec. Genet.* 8: 157–164, 1999.
- [42118] 15670.Collard, J.-F.; Cote, F.; Julien, J.-P.: Defective axonal transport in a transgenic mouse model of amyotrophic lateral sclerosis. *Nature* 375:61–64, 1995.
- [42119] 15671.Figlewicz, D. A.; Krizus, A.; Martinoli, M. G.; Meininger, V.; Dib, M.; Rouleau, G. A.; Julien, J.-P.: Variants of the heavy neurofilament subunit are associated with the development of amyotrophic lateral sclerosis. *Hum. Molec. Genet.* 3: 1757–1761, 1994.
- [42120] 15672.Lees, J. F.; Shneiderman, P. S.; Skuntz, S. F.; Carden, M. J.; Lazzarini, R. A.: The structure and organization of

the human heavy neurofilamentsubunit (NF-H) and the gene encoding it. EMBO J. 7: 1947–1955, 1988.

[42121] 15673. Mattei, M.-G.; Dautigny, A.; Pham-Dinh, D.; Passage, E.; Mattei, J.-F.; Jolles, P.: The gene encoding the large human neurofilamentsubunit (NF-H) maps to the q121–q131 region on human chromosome 22.

Hum.Genet. 80: 293–295, 1988.

[42122] 15674. Rooke, K.; Figlewicz, D. A.; Han, F.; Rouleau, G. A.: Analysis of the KSP repeat of the neurofilament heavy subunit in familial amyotrophic lateral sclerosis. Neurology 46: 789–790, 1996.

[42123] 15675. Rouleau, G. A.; Merel, P.; Lutchman, M.; Sanson, M.; Zucman, J.; Marineau, C.; Hoang-Xuan, K.; Demczuk, S.; Desmaze, C.; Plougastel, B.; Pulst, S. M.; Lenoir, G.; Bijlsma, E.; Fashold, R.; Dumanski, J.; de Jong, P.; Parry, D.; Eldridge, R.; Aurias, A.; Delattre, O.; Thomas, G.: Alteration in a new gene encoding a putative membrane-organizing protein causes neuro-fibromatosis type 2. Nature 363: 515–521, 1993.

[42124] 15676. Tomkins, J.; Usher, P.; Slade, J. Y.; Ince, P. G.; Curtis, A.; Bushby, K.; Shaw, P. J.: Novel insertion in the KSP region of the neurofilament heavy gene in amyotrophic lateral sclerosis. Neuroreport 9: 3967–3970, 1998.

- [42125] 15677.Vechio, J. D.; Bruijn, L. I.; Xu, Z.; Brown, R. H., Jr.; Cleveland,D. W.: Sequence variants in human neurofilament proteins: absenceof linkage to familial amyotrophic lateral sclerosis. *Ann. Neurol.* 40:603–610, 1996.
- [42126] 15678.Watson, C. J.; Gaunt, L.; Evans, G.; Patel, K.; Harris, R.; Strachan,T.: A disease-associated germline deletion maps the type 2 neurofibromatosis(NF2) gene between the Ewing sarcoma region and the leukaemia inhibitory-factor locus. *Hum. Molec. Genet.* 2: 701–704, 1993.
- [42127] 15679.Levy, E.; Liem, R. K. H.; D'Eustachio, P.; Cowan, N. J.: Structureand evolutionary origin of the gene encoding NF-M, the middle-molecular-massneurofilament protein. *Europ. J. Biochem.* 166: 71–77, 1987.
- [42128] 15680.Mersiyanova, I. V.; Perepelov, A. V.; Polyakov, A. V.; Sitnikov,V. F.; Dadali, E. L.; Oparin, R. B.; Petrin, A. N.; Evgrafov, O. V.: A new variant of Charcot-Marie-Tooth disease type 2 is probablythe result of a mutation in the neurofilament-light gene. *Am. J.Hum. Genet.* 67: 37–46, 2000.
- [42129] 15681.Myers, M. W.; Lazzarini, R. A.; Lee, V. M.-Y.; Schlaepfer, W. W.;Nelson, D. L.: The human mid-size neurofilament subunit: a repeatedprotein sequence and the relationship of its gene to the intermediatefilament gene

family. EMBO J. 6: 1617–1626, 1987.

- [42130] 15682. Barker, P. E.; Besmer, P.; Ruddle, F. H.: Human c-kit oncogene on human chromosome 4. (Abstract) Am. J. Hum. Genet. 37: A143, 1985.
- [42131] 15683. Beghini, A.; Larizza, L.; Cairoli, R.; Morra, E.: c-kit activating mutations and mast cell proliferation in human leukemia. (Letter) Blood 92:701–702, 1998.
- [42132] 15684. Beghini, A.; Tibiletti, M.; Roversi, G.; Chiaravalli, A.; Serio, G.; Capella, C.; Larizza, L.: Germline mutation in the juxtamembrane domain of the KIT gene in a family with gastrointestinal stromal tumors and urticaria pigmentosa. Cancer 92: 657–662, 2001.
- [42133] 15685. Blume-Jensen, P.; Jiang, G.; Hyman, R.; Lee, K.-F.; O’Gorman, S.; Hunter, T.: Kit/stem cell factor receptor-induced activation of phosphatidylinositol 3-kinase is essential for male fertility. Nature Genet. 24:157–162, 2000.
- [42134] 15686. Bolognia, J. L.; Pawelek, J. M.: Biology of hypopigmentation. J. Am. Acad. Derm. 19: 217–255, 1988.
- [42135] 15687. Brannan, C. I.; Lyman, S. D.; Williams, D. E.; Eisenman, J.; Anderson, D. M.; Cosman, D.; Bedell, M. A.; Jenkins, N. A.; Copeland, N. G.: Steel-Dickie mutation encodes a c-kit ligand lacking transmembrane and cytoplasmic do-

mains. Proc. Nat. Acad. Sci. 88: 4671–4674, 1991.

[42136] 15688.Chabot, B.; Stephenson, D. A.; Chapman, V. M.; Bersmer, P.; Bernstein,A.: The proto-oncogene c-kit encoding a transmembrane tyrosine kinasereceptor maps to the mouse W locus. Nature 335: 88–89, 1988.

[42137] 15689.d'Auriol, L.; Mattei, M.–G.; Andre, C.; Galibert, F.: Localizationof the human c-kit protooncogene on the q11–q12 region of chromosome4. Hum. Genet. 78: 374–376, 1988.

[42138] 15690.De Miguel, M. P.; Cheng, L.; Holland, E. C.; Feder–spiel, M. J.;Donovan, P. J.: Dissection of the c-Kit signaling pathway in mouseprimordial germ cells by retroviral–mediated gene transfer. Proc.Nat. Acad. Sci. 99: 10458–10463, 2002.

[42139] 15691.Dubreuil, P.; Forrester, L.; Rottapel, R.; Reedijk, M.; Fujita,J.; Bernstein, A.: The c-fms gene complements the mitogenic defectin mast cells derived from mutant W mice but not mi (microphthalmia)mice. Proc. Nat. Acad. Sci. 88: 2341–2345, 1991.

[42140] 15692.el–Omar, M.; Davies, J.; Gupta, S.; Ross, H.; Thompson, R.: Leiomyosarcomain leiomyomatosis of the small intestine. Postgrad. Med. J. 70:661–664, 1994.

[42141] 15693.Fleischman, R. A.: Human piebald trait resulting

from a dominantnegative mutant allele of the c-kit membrane receptor gene. J. Clin. Invest. 89: 1713–1717, 1992.

- [42142] 15694. Fleischman, R. A.; Saltman, D. L.; Stastny, V.; Zneimer, S.: Deletion of the c-kit protooncogene in the human developmental defect piebald trait. Proc. Nat. Acad. Sci. 88: 10885–10889, 1991.
- [42143] 15695. Fritsche-Polanz, R.; Jordan, J.-H.; Feix, A.; Sperr, W. R.; Sunder-Plassmann, G.; Valent, P.; Fodinger, M.: Mutation analysis of C-KIT in patients with myelodysplastic syndromes without mastocytosis and cases of systemic mastocytosis. Brit. J. Haemat. 113: 357–364, 2001.
- [42144] 15696. Geissler, E. N.; Ryan, M. A.; Housman, D. E.: The dominant-whitespotting (W) locus of the mouse encodes the c-kit proto-oncogene. Cell 55: 185–192, 1988.
- [42145] 15697. Giebel, L. B.; Spritz, R. A.: Mutation of the KIT (mast/stemcell growth factor receptor) protooncogene in human piebaldism. Proc. Nat. Acad. Sci. 88: 8696–8699, 1991.
- [42146] 15698. Hirota, S.; Isozaki, K.; Moriyama, Y.; Hashimoto, K.; Nishida, T.; Ishiguro, S.; Kawano, K.; Hanada, M.; Kurata, A.; Takeda, M.; Tunio, G. M.; Matsuzawa, Y.; Kanakura, Y.; Shinomura, Y.; Kitamura, Y.: Gain-of-function mutations of c-kit in human gastrointestinal stromal tumors. Science

279: 577–580, 1998.

[42147] 15699. Huizinga, J. D.; Thuneberg, L.; Kluppel, M.; Malysz, J.; Mikkelsen, H. B.; Bernstein, A.: W/kit gene required for interstitial cells of Cajal and for intestinal pacemaker activity. *Nature* 373: 347–349, 1995.

[42148] 15700. Furitsu, T.; Tsujimura, T.; Tono, T.; Ikeda, H.; Kitayama, H.; Koshimizu, U.; Sugahara, H.; Butterfield, J. H.; Ashman, L. K.; Kanayama, Y.; Matsuzawa, Y.; Kitamura, Y.; Kanakura, Y.: Identification of mutations in the coding sequence of the proto-oncogene c-kit in a human mast cell leukemia cell line causing ligand-independent activation of c-kit product. *J. Clin. Invest.* 92: 1736–1744, 1993.

[42149] 15701. Ingram, D. A.; Yang, F.-C.; Travers, J. B.; Wenning, M. J.; Hiatt, K.; New, S.; Hood, A.; Shannon, K.; Williams, D. A.; Clapp, D. W.: Genetic and biochemical evidence that haploinsufficiency of the Nf1 tumor suppressor gene modulates melanocyte and mast cell fates in vivo. *J. Exp. Med.* 191: 181–187, 2000.

[42150] 15702. Isozaki, K.; Terris, B.; Belghiti, J.; Schiffmann, S.; Hirota, S.; Vanderwinden, J.-M.: Germline-activating mutation in the kinase domain of KIT gene in familial gastrointestinal stromal tumors. *Am. J. Path.* 157: 1581–1585, 2000.

[42151] 15703.Joensuu, H.; Roberts, P. J.; Sarlomo-Rikala, M.; Andersson, L.C.; Tervahartiala, P.; Tuveson, D.; Silberman, S. L.; Capdeville,R.; Dimitrijevic, S.; Druker, B.; Demetri, G. D.: Effect of the tyrosinekinase inhibitor STI571 in a patient with a metastatic gastrointestinalstromal tumor. New Eng. J. Med. 344: 1052–1056, 2001.

[42152] 15704.Johansson Moller, M.; Chaudhary, R.; Hellmen, E.; Hoyheim, B.;Chowdhary, B.; Andersson, L.: Pigs with the dominant white coat colorphenotype carry a duplication of the KIT gene encoding the mast/stemcell growth factor receptor. Mammalian Genome 7: 822–830, 1996.

[42153] 15705.Lasota, J.; Jasinski, M.; Sarlomo-Rikala, M.; Miettinen, M.:Mutations in exon 11 of c-kit occur preferentially in malignant versusbenign gastrointestinal stromal tumors and do not occur in leiomyomasor leiomyosarcomas. Am. J. Path. 154: 53–60, 1999.

[42154] 15706.Longley, B. J.; Tyrrell, L.; Lu, S.-Z.; Ma, Y.-S.; Langley, K.;Ding, T.; Duffy, T.; Jacobs, P.; Tang, L. H.; Modlin, I.: Somaticc-KIT activating mutation in urticaria pigmentosa and aggressive mastocytosis:establishment of clonality in a human mast cell neoplasm. NatureGenet. 12: 312–314, 1996.

[42155] 15707.Longley, B. J., Jr.; Metcalfe, D. D.; Tharp, M.; Wang,

X.; Tyrrell, L.; Lu, S.-Z.; Heitjan, D.; Ma, Y.: Activating and dominant inactivating c-KIT catalytic domain mutations in distinct clinical forms of human mastocytosis. *Proc. Nat. Acad. Sci.* 96: 1609–1614, 1999.

[42156] 15708. Marklund, S.; Kijas, J.; Rodriguez-Martinez, H.; Ronnstrand, L.; Funa, K.; Moller, M.; Lange, D.; Edfors-Lilja, I.; Andersson, L.: Molecular basis for the dominant white phenotype in the domestic pig. *Genome Res.* 8: 826–833, 1998.

[42157] 15709. Kiefer, M. C.; Tucker, J. E.; Joh, R.; Landsberg, K. E.; Saltman, D.; Barr, P. J.: Identification of a second human subtilisin-like protease gene in the fes/fps region of chromosome 15. *DNA Cell Biol.* 10:757–769, 1991.

[42158] 15710. Amiel, J.; Audollent, S.; Joly, D.; Dureau, P.; Salomon, R.; Tellier, A.-L.; Auge, J.; Bouissou, F.; Antignac, C.; Gubler, M.-C.; Eccles, M. R.; Munnich, A.; Vekemans, M.; Lyonnet, S.; Attie-Bitach, T.: PAX2 mutations in renal-coloboma syndrome: mutational hotspot and germline mosaicism. *Europ. J. Hum. Genet.* 8: 820–826, 2000.

[42159] 15711. Dehbi, M.; Ghahremani, M.; Lechner, M.; Dressler, G.; Pelletier, J.: The paired-box transcription factor, PAX2, positively modulates expression of the Wilms' tumor suppressor gene. *Oncogene* 13: 447–453, 1996.

- [42160] 15712.Devriendt, K.; Matthijs, G.; Van Damme, B.; Van Caesbroeck, D.;Eccles, M.; Vanrenterghem, Y.; Fryns, J.-P.; Leys, A.: Missense mutationand hexanucleotide duplication in the PAX2 gene in two unrelated familieswith renal-coloboma syndrome (MIM 120330). Hum. Genet. 103: 149–153,1998.
- [42161] 15713.Dressler, G. R.; Deutsch, U.; Chowdhury, K.; Nornes, H. O.; Gruss,P.: Pax2, a new murine paired-box-containing gene and its expressionin the developing excretory system. Development 109: 787–795, 1990.
- [42162] 15714.Ford, B.; Rupps, R.; Lirenman, D.; Van Allen, M. I.; Farquharson,D.; Lyons, C.; Friedman, J. M.: Renal-coloboma syndrome: prenataldetection and clinical spectrum in a large family. Am. J. Med. Genet. 99:137–141, 2001.
- [42163] 15715.Keller, S. A.; Jones, J. M.; Boyle, A.; Barrow, L. L.; Killen,P. D.; Green, D. G.; Kapousta, N. V.; Hitchcock, P. F.; Swank, R.T.; Meisler, M. H.: Kidney and retinal defects (Krd), a transgene-inducedmutation with a deletion of mouse chromosome 19 that includes thePax2 locus. Genomics 23: 309–320, 1994.
- [42164] 15716.Narahara, K.; Baker, E.; Ito, S.; Yokoyama, Y.; Yu, S.; Hewitt,D.; Sutherland, G. R.; Eccles, M. R.; Richards, R.

I.: Localisation of a 10q breakpoint within the PAX2 gene in a patient with a de novo (10;13) translocation and optic nerve coloboma–renal disease. *J. Med. Genet.* 34: 213–216, 1997.

[42165] 15717. Pilz, A. J.; Povey, S.; Gruss, P.; Abbott, C. M.: Mapping of the human homologs of the murine paired-box-containing genes. *Mammalian Genome* 4: 78–82, 1993.

[42166] 15718. Favor, J.; Sandulache, R.; Neuhauser-Klaus, A.; Pretsch, W.; Chatterjee, B.; Senft, E.; Wurst, W.; Blanquet, V.; Grimes, P.; Sporle, R.; Schughart, K.: The mouse Pax2(1^{Neu}) mutation is identical to a human PAX2 mutation in a family with renal–coloboma syndrome and results in developmental defects of the brain, ear, eye, and kidney. *Proc. Nat. Acad. Sci.* 93:13870–13875, 1996.

[42167] 15719. Liou, G. I.; Ma, D.–P.; Yang, Y.–W.; Geng, L.; Zhu, C.; Baehr, W.: Human interstitial retinoid-binding protein: gene structure and primary sequence. *J. Biol. Chem.* 264: 8200–8206, 1989.

[42168] 15720. Nakamura, Y.; Lathrop, M.; Bragg, T.; Leppert, M.; O'Connell, P.; Jones, C.; Lalouel, J. M.; White, R.: An extended genetic linkage map of markers for human chromosome 10. *Genomics* 3: 389–392, 1988.

- [42169] 15721.Sap, J.; Munoz, A.; Damm, K.; Goldberg, Y.; Ghysdael, J.; Leutz,A.; Beug, H.; Vennstrom, B.: The c-erb-A protein is a high-affinityreceptor for thyroid hormone. *Nature* 324: 635-640, 1986.
- [42170] 15722.Bayliss, W.; Starling, E. H.: The mechanism of pancreatic secretion. *J.Physiol. (London)* 28: 325-353, 1902.
- [42171] 15723.Chow, B. K.-C.: Molecular cloning and functional characterizationof a human secretin receptor. *Biochem. Biophys. Res. Commun.* 212:204-211, 1995.
- [42172] 15724.Ishihara, T.; Nakamura, S.; Kaziro, Y.; Takahashi, T.; Takahashi,K.; Nagata, S.: Molecular cloning and expression of a cDNA encodingthe secretin receptor. *EMBO J.* 10: 1635-1641, 1991.
- [42173] 15725.Mark, H. F. L.; Chow, B. K.-C.: Localization of the gene encodingthe secretin receptor, SCTR, on human chromosome 2q14.1 by fluorescencein situ hybridization and chromosome morphometry. *Genomics* 29: 817-818,1995.
- [42174] 15726.Peel, A. L.; Rao, R. V.; Cottrell, B. A.; Hayden, M. R.; Ellerby,L. M.; Bredesen, D. E.: Double-stranded RNA-dependent protein kinase,PKR, binds preferentially to Huntington's disease (HD) transcriptsand is activated in HD tissue. *Hum. Molec. Genet.* 10: 1531-1538,2001.

- [42175] 15727.Squire, J.; Meurs, E. F.; Chong, K. L.; McMillan, N. A. J.; Hovanessian,A. G.; Williams, B. R. G.: Localization of the human interferon-induced,ds-RNA activated p68 kinase gene (PRKR) to chromosome 2p21-p22. *Genomics* 16:768-770, 1993.
- [42176] 15728.Taylor, D. R.; Shi, S. T.; Romano, P. R.; Barber, G. N.; Lal, M.M. C.: Inhibition of the interferon-inducible protein kinase PKRby HCV E2 protein. *Science* 285: 107-110, 1999.
- [42177] 15729.Brott, B. K.; Alessandrini, A.; Largaespada, D. A.; Copeland, N.G.; Jenkins, N. A.; Crews, C. M.; Erikson, R. L.: MEK2 is a kinase related to MEK1 and is differentially expressed in murine tissues. *CellGrowth Differ.* 4: 921-929, 1993.
- [42178] 15730.Crews, C. M.; Alessandrini, A.; Erikson, R. L.: The primary structure of MEK, a protein kinase that phosphorylates the ERK gene product. *Science* 258:478-480, 1992.
- [42179] 15731.Favata, M. F.; Horiuchi, K. Y.; Manos, E. J.; Daulerio, A. J.;Stradley, D. A.; Feeser, W. S.; Van Dyk, D. E.; Pitts, W. J.; Earl,R. A.; Hobbs, F.; Copeland, R. A.; Magolda, R. L.; Scherle, P. A.;Trzaskos, J. M.: Identification of a novel inhibitor of mitogen-activatedprotein kinase kinase. *J. Biol. Chem.* 273: 18623-18632, 1998.

- [42180] 15732.Meloche, S.; Gopalbhai, K.; Beatty, B. G.; Scherer, S. W.; Pellerin,J.: Chromosome mapping of the human genes encoding the MAP kinasekinase MEK1 (MAP2K1) to 15q21 and MEK2 (MAP2K2) to 7q32. Cytogenet.Cell Genet. 88: 249–252, 2000.
- [42181] 15733.Orth, K.; Palmer, L. E.; Bao, Z. Q.; Stewart, S.; Rudolph, A. E.;Bliska, J. B.; Dixon, J. E.: Inhibition of the mitogen–activatedprotein kinase kinase superfamily by a Yersinia effector. Science 285:1920–1923, 1999.
- [42182] 15734.Perry, R. L. S.; Parker, M. H.; Rudnicki, M. A.: Activated MEK1binds the nuclear MyoD transcriptional complex to repress transactivation. Molec.Cell 8: 291–301, 2001.
- [42183] 15735.Pleschka, S.; Wolff, T.; Ehrhardt, C.; Hobom, G.; Planz, O.; Rapp,U. R.; Ludwig, S.: Influenza virus propagation is impaired by inhibitionof the Raf/MEK/ERK signalling cascade. Nature Cell Biol. 3: 301–305,2001.
- [42184] 15736.Rampoldi, L.; Zimbello, R.; Bortoluzzi, S.; Tiso, N.; Valle, G.;Lanfranchi, G.; Danieli, G. A.: Chromosomal localization of fourMAPK signaling cascade genes: MEK1, MEK3, MEK4 and MEKK5. Cytogenet.Cell Genet. 78: 301–303, 1997.
- [42185] 15737.Ryan, K. M.; Ernst, M. K.; Rice, N. R.; Vousden, K.

H.: Role of NF- κ B in p53-mediated programmed cell death. *Nature* 404: 892–897, 2000.

[42186] 15738. Sebolt-Leopold, J. S.; Dudley, D. T.; Herrera, R.; Van Becelaere, K.; Wiland, A.; Gowan, R. C.; Tecle, H.; Barrett, S. D.; Bridges, A.; Przybranowski, S.; Leopold, W. R.; Saltiel, A. R.: Blockade of the MAP kinase pathway suppresses growth of colon tumors in vivo. *Nature Med.* 5: 810–816, 1999.

[42187] 15739. Seger, R.; Krebs, E. G.: The MAPK signaling cascade. *FASEB J.* 9: 726–735, 1995.

[42188] 15740. Seger, R.; Seger, D.; Lozeman, F. J.; Ahn, N. G.; Graves, L. M.; Campbell, J. S.; Ericsson, L.; Harrylock, M.; Jensen, A. M.; Krebs, E. G.: Human T-cell mitogen-activated protein kinase kinases are related to yeast signal transduction kinases. *J. Biol. Chem.* 267: 25628–25631, 1992.

[42189] 15741. Zheng, C. F.; Guan, K. L.: Cloning and characterization of two distinct human extracellular signal-regulated kinase activator kinases, MEK1 and MEK2. *J. Biol. Chem.* 268: 11435–11439, 1993.

[42190] 15742. Bunnell, B.; Heath, L. S.; Adams, D. E.; Lahti, J. M.; Kidd, V. J.: Elevated expression of a p58 protein kinase leads to changes in the CHO cell cycle. *Proc. Nat. Acad. Sci.*

87: 7467–7471, 1990.

- [42191] 15743.Cornelis, S.; Bruynooghe, Y.; Denecker, G.; Van Huffel, S.; Tinton,S.; Beyaert, R.: Identification and characterization of a novel cellcycle–regulated internal ribosome entry site. *Molec. Cell* 5: 597–605,2000.
- [42192] 15744.Eipers, P. G.; Barnoski, B. L.; Han, J.; Carroll, A. J.; Kidd,V. J.: Localization of the expressed human p58 protein kinase chromosomalgene to chromosome 1p36 and a highly related sequence to chromosome15. *Genomics* 11: 621–629, 1991.
- [42193] 15745.Eipers, P. G.; Lahti, J. M.; Kidd, V. J.: Structure and expressionof the human p58(clk–1) protein kinase chromosomal gene. *Genomics* 13:613–621, 1992.
- [42194] 15746.Gururajan, R.; Lahti, J. M.; Grenet, J.; Easton, J.; Gruber, I.;Ambros, P. F.; Kidd, V. J.: Duplication of a genomic region containingthe Cdc2L1–2 and MMP21–22 genes on human chromosome 1p36.3 and theirlinkage to D1Z2. *Genome Res.* 8: 929–939, 1998.
- [42195] 15747.Lahti, J. M.; Valentine, M.; Xiang, J.; Jones, B.; Amann, J.; Grenet,J.; Richmond, G.; Look, A. T.; Kidd, V. J.: Alterations in the PITSLREprotein kinase gene complex on chromosome 1p36 in childhood neuroblastoma. *Nature–Genet.* 7: 370–375, 1994.

- [42196] 15748.White, P. S.; Maris, J. M.; Beltinger, C.; Sulman, E.; Marshall,H. N.; Fujimori, M.; Kaufman, B. A.; Biegel, J. A.; Allen, C.; Hilliard,C.; Valentine, M. B.; Look, A. T.; Enomoto, H.; Sakiyama, S.; Brodeur,G. M.: A region of consistent deletion in neuroblastoma maps withinhuman chromosome 1p36.2–36.3. *Proc. Nat. Acad. Sci.* 92: 5520–5524,1995.
- [42197] 15749.Xiang, J.; Lahti, J. M.; Grenet, J.; Easton, J.; Kidd, V. J.:Molecular cloning and expression of alternatively spliced PITSLREprotein kinase isoforms. *J. Biol. Chem.* 269: 15786–15794, 1994.
- [42198] 15750.Collu, R.; Tang, J.; Castagne, J.; Lagace, G.; Masson, N.; Huot,C.; Deal, C.; Delvin, E.; Faccenda, E.; Eidne, K. A.; Van Vliet, G.: A novel mechanism for isolated central hy–pothyroidism: inactivatingmutations in the thyrotropin–releasing hormone receptor gene. *J.Clin. Endocr. Metab.* 82: 1361–1365, 1997.
- [42199] 15751.Amin, R. S.; Wert, S. E.; Baughman, R. P.; Toma–shevski, J. F.,Jr.; Nogee, L. M.; Brody, A. S.; Hull, W. M.; Whitsett, J. A.: Surfactantprotein deficiency in familial in–terstitial lung disease. *J. Pediat.* 139:85–92, 2001.
- [42200] 15752.Glasser, S. W.; Burhans, M. S.; Korfhagen, T. R.; Na, C.–L.; Sly,P. D.; Ross, G. F.; Ikegami, M.; Whitsett, J. A.: Al–

tered stability of pulmonary surfactant in SP-C-deficient mice. *Proc. Nat. Acad.Sci.* 98: 6366–6371, 2001.

[42201] 15753. Keller, D. M.; Zeng, X.; Wang, Y.; Zhang, Q. H.; Kapoor, M.; Shu, H.; Goodman, R.; Lozano, G.; Zhao, Y.; Lu, H.: A DNA damage-induced p53 serine 392 kinase complex contains CK2, hSpt16, and SSRP1. *Molec. Cell* 283–292, 2001.

[42202] 15754. Maki, R. G.; Eddy, R. L.; Byers, M. G.; Shows, T. B.; Srivastava, P. K.: Localization of genes for the stress-inducible tumor rejection antigen gp96. (Abstract) *Cytogenet. Cell Genet.* 58: 1978 only, 1991.

[42203] 15755. Maki, R. G.; Old, L. J.; Srivastava, P. K.: Human homologue of murine tumor rejection antigen gp96: 5-prime-regulatory and coding regions and relationship to stress-induced proteins. *Proc. Nat. Acad.Sci.* 87: 5658–5662, 1990.

[42204] 15756. Schild, H.; Rammensee, H.-G.: gp96: the immune system's Swiss army knife. *Nature Immun.* 1: 100–101, 2000.

[42205] 15757. Lanske, B.; Karaplis, A. C.; Lee, K.; Luz, A.; Vortkamp, A.; Pirro, A.; Karperien, M.; Defize, L. H. K.; Ho, C.; Mulligan, R. C.; Abou-Samra, A.-B.; Juppner, H.; Segre, G. V.; Kronenberg, H. M.: PTH/PTHrP receptor in early de-

velopment and Indian hedgehog-regulated bone growth. Science 273:663–666, 1996.

- [42206] 15758. Albright, F.: Case records of the Massachusetts General Hospital(case 27461). New Eng. J. Med. 225: 789–791, 1941.
- [42207] 15759. Bakre, M. M.; Zhu, Y.; Yin, H.; Burton, D. W.; Terkeltaub, R.; Deftos, L. J.; Varner, J. A.: Parathyroid hormone-related peptide is a naturally occurring, protein kinase A-dependent angiogenesis inhibitor. Nature Med. 8: 995–1003, 2002.
- [42208] 15760. Broadus, A. E.; Mangin, M.; Ikeda, K.; Insogna, K. L.; Weir, E. C.; Burtis, W. J.; Stewart, A. F.: Humoral hypercalcemia of cancer: identification of a novel parathyroid hormone-like peptide. New Eng. J. Med. 319: 556–563, 1988.
- [42209] 15761. Hammonds, R. G., Jr.; McKay, P.; Winslow, G. A.; Diefenbach-Jagger, H.; Grill, V.; Glatz, J.; Rodda, C. P.; Moseley, J. M.; Wood, W. I.; Martin, T. J.: Purification and characterization of recombinant human parathyroid hormone-related protein. J. Biol. Chem. 264: 14806–14811, 1989.
- [42210] 15762. Hendy, G. N.; Sakaguchi, A. Y.; Lalley, P. A.; Martinez, L.; Yasuda, T.; Banville, D.; Goltzman, D.: Gene for

parathyroid hormone-likepeptide is on mouse chromosome 6. *Cytogenet. Cell Genet.* 53: 80–82,1990.

[42211] 15763.Hendy, G. N.; Sakaguchi, A. Y.; Lalley, P. A.; Martinez, L.; Yasuda,T.; Banville, D.; Goltzman, D.: Gene for parathyroid hormone-likepeptide is on mouse chromosome 6. (Abstract) *Cytogenet. Cell Genet.* 51:1003 only, 1989.

[42212] 15764.Kamps, M. P.; Look, A. T.; Baltimore, D.: The human t(1:19) translocationin pre-B ALL produces multiple nuclear E2A-Pbx1 fusion proteins withdiffering transforming potentials. *Genes Dev.* 5: 358–368, 1991.

[42213] 15765.Kim, S. K.; Selleri, L.; Lee, J. S.; Zhang, A. Y.; Gu, X.; Jacobs,Y.; Cleary, M. L.: Pbx1 inactivation disrupts pancreas developmentand in lpf1-deficient mice promotes diabetes mellitus. *Nature Genet.* 30:430–435, 2002.

[42214] 15766.Mercader, N.; Leonardo, E.; Azpiazu, N.; Serrano, A.; Morata, G.;Martinez-A, C.; Torres, M.: Conserved regulation of proximodistallimb axis development by Meis1/Hth. *Nature* 402: 425–429, 1999.

[42215] 15767.Monica, K.; Galili, N.; Nourse, J.; Saltman, D.; Cleary, M. L.: PBX2 and PBX3, new homeobox genes with extensive homology to thehuman proto-oncogene PBX1. *Molec. Cell. Biol.* 11: 6149–6157, 1991.

- [42216] 15768.Aguado, B.; Campbell, R. D.: The novel gene G17, located in the human major histocompatibility complex, encodes PBX2, a homeodomain-containing protein. *Genomics* 25: 650–659, 1995.
- [42217] 15769.Katsanis, N.; Fitzgibbon, J.; Fisher, E. M. C.: Paralogy mapping: identification of a region in the human MHC triplicated onto human chromosomes 1 and 9 allows the prediction and isolation of novel PBX and NOTCH loci. *Genomics* 35: 101–108, 1996.
- [42218] 15770.Hourvitz, A.; Widger, A. E.; Filho, F. L. T.; Chang, R. J.; Adashi, E. Y.; Erickson, G. F.: Pregnancy-associated plasma protein-A gene expression in human ovaries is restricted to healthy follicles and corpora lutea. *J. Clin. Endocr. Metab.* 85: 4916–4919, 2000.
- [42219] 15771.Lawrence, J. B.; Oxvig, C.; Overgaard, M. T.; Sottrup-Jensen, L.; Gleich, G. J.; Hays, L. G.; Yates, J. R., III; Conover, C. A.: The insulin-like growth factor (IGF)-dependent IGF binding protein-4 protease secreted by human fibroblasts is pregnancy-associated plasma protein-A. *Proc. Nat. Acad. Sci.* 96: 3149–3153, 1999.
- [42220] 15772.Silahtaroglu, A. N.; Tumer, Z.; Kristensen, T.; Sottrup-Jensen, L.; Tommerup, N.: Assignment of the human gene for pregnancy-associated plasma protein A (PAPPA)

to 9q33.1 by fluorescence in situ hybridization to mitotic and meiotic chromosomes. *Cytogenet. Cell Genet.* 62: 214–216, 1993.

- [42221] 15773. Smith, G. C. S.; Stenhouse, E. J.; Crossley, J. A.; Aitken, D. A.; Cameron, A. D.; Connor, J. M.: Early pregnancy levels of pregnancy-associated plasma protein A and the risk of intrauterine growth restriction, premature birth, preeclampsia, and stillbirth. *J. Clin. Endocr. Metab.* 87:1762–1767, 2002.
- [42222] 15774. Smith, G. C. S.; Stenhouse, E. J.; Crossley, J. A.; Aitken, D. A.; Cameron, A. D.; Connor, J. M.: Early-pregnancy origins of low birth weight. *Nature* 417: 916 only, 2002.
- [42223] 15775. Eisenhofer, G.; Walther, M. M.; Huynh, T.-T.; Li, S.-T.; Bornstein, S. R.; Vortmeyer, A.; Mannelli, M.; Goldstein, D. S.; Linehan, W. M.; Lenders, J. W. M.; Pacak, K.: Pheochromocytomas in von Hippel-Lindau syndrome and multiple endocrine neoplasia type 2 display distinct biochemical and clinical phenotypes. *J. Clin. Endocr. Metab.* 86:1999–2008, 2001.
- [42224] 15776. Glasser, S. W.; Korfhagen, T. R.; Weaver, T. E.; Clark, J. C.; Pilot-Matias, T.; Meuth, J.; Fox, J. L.; Whitsett, J. A.: cDNA, deduced polypeptide structure and chromoso-

mal assignment of human pulmonary surfactant protein-lipid, SPL(pVal). J. Biol. Chem. 263: 9–12, 1988.

- [42225] 15777. Zhang, S.-H.; Liu, J.; Kobayashi, R.; Tonks, N. K.: Identification of the cell cycle regulator VCP (p97/CDC48) as a substrate of the band 4.1-related protein-tyrosine phosphatase PTPH1. J. Biol. Chem. 274:17806–17812, 1999.
- [42226] 15778. Lorimer, D. D.; Benya, R. V.: Cloning and quantification of galanin-1 receptor expression by mucosal cells lining the human gastrointestinal tract. Biochem. Biophys. Res. Commun. 222: 379–385, 1996.
- [42227] 15779. Nicholl, J.; Kofler, B.; Sutherland, G. R.; Shine, J.; Lismaa, T. P.: Assignment of the gene encoding human galanin receptor (GALNR) to 18q23 by in situ hybridization. Genomics 30: 629–630, 1995.
- [42228] 15780. Parker, E. M.; Izzarelli, D. G.; Nowak, H. P.; Mahle, C. D.; Iben, L. G.; Wang, J.; Goldstein, M. E.: Cloning and characterization of the rat GALR1 galanin receptor from Rin14B insulinoma cells. Molec. Brain Res. 34: 179–189, 1995.
- [42229] 15781. Simoneaux, D. K.; Leach, R. J.; O'Connell, P.: Galanin receptor1 gene (Galnr1) is tightly linked to the myelin basic protein gene on chromosome 18 in mouse.

Mammalian Genome 8: 875 only, 1997.

- [42230] 15782.Walli, R.; Schafer, H.; Morys-Wortmann, C.; Paetzold, G.; Nustede,R.; Schmidt, W. E.: Identification and biochemical characterizationof the human brain galanin receptor. J. Molec. Endocr. 13: 347–356,1994.
- [42231] 15783.Janowski, B. A.; Grogan, M. J.; Jones, S. A.; Wisely, G. B.; Kliewer,S. A.; Corey, E. J.; Mangelsdorf, D. J.: Structural requirementsof ligands for the oxysterol liver X receptors LXR-alpha and LXR-beta. Proc.Nat. Acad. Sci. 96: 266–271, 1999.
- [42232] 15784.Le Beau, M. M.; Song, C.; Davis, E. M.; Hiipakka, R. A.; Kokontis,J. M.; Liao, S.: Assignment of the human ubiquitous receptor gene(UNR) to 19q13.3 using fluorescence in situ hybridization. Genomics 26:166–168, 1995.
- [42233] 15785.Repa, J. J.; Berge, K. E.; Pomajzl, C.; Richardson, J. A.; Hobbs,H.; Mangelsdorf, D. J.: Regulation of ATP-binding cassette steroltransporters ABCG5 and ABCG8 by the liver X receptors alpha and beta. J.Biol. Chem. 277: 18793–18800, 2002.
- [42234] 15786.Shinar, D. M.; Endo, N.; Rutledge, S. J.; Vogel, R.; Rodan, G.A.; Schmidt, A.: NER, a new member of the gene family encoding thehuman steroid hormone nuclear receptor. Gene 147: 273–276, 1994.

- [42235] 15787.Song, C.; Hiipakka, R. A.; Kokontis, J. M.; Liao, S.: Ubiquitousreceptor: structures, immunocytochemical localization, and modulationof gene activation by receptors for retinoic acids and thyroid hormones. Ann.N.Y. Acad. Sci. 761: 38–49, 1995.
- [42236] 15788.Tangirala, R. K.; Bischoff, E. D.; Joseph, S. B.; Wagner, B. L.;Walczak, R.; Laffitte, B. A.; Daige, C. L.; Thomas, D.; Heyman, R.A.; Mangelsdorf, D. J.; Wang, X.; Lusis, A. J.; Tontonoz, P.; Schulman,I. G.: Identification of macrophage liver X receptors as inhibitorsof atherosclerosis. Proc. Nat. Acad. Sci. 99: 11896–11901, 2002.
- [42237] 15789.Futterer, A.; Kruppa, G.; Kramer, B.; Lemke, H.; Kronke, M.: Molecularcloning and characterization of human kinectin. Molec. Biol. Cell 6:161–170, 1995.
- [42238] 15790.Plitz, T.; Pfeffer, K.: Intact lysosome transport and phagosomefunction despite kinectin deficiency. Molec. Cell. Biol. 21: 6044–6055,2001.
- [42239] 15791.Print, C. G.; Leung, E.; Harrison, J. E. B.; Watson, J. D.; Krissansen,G. W.: Cloning of a gene encoding a human leukocyte protein characterisedby extensive heptad repeats. Gene 144: 221–228, 1994.
- [42240] 15792.Print, C. G.; Morris, C. M.; Spurr, N. K.; Rooke, L.; Krissansen,G. W.: The CG–1 gene, a member of the

kinectin and ES/130 family, maps to human chromosome band 14q22. Immunogenetics 43: 227–229, 1996.

- [42241] 15793. Rao, P. N.; Yu, H.; Hodge, R.; Pettenati, M. J.; Sheetz, M. P.: Assignment of the human kinectin gene (KTN1), encoding a kinesin-binding protein, to chromosome 14 band q22.1 by in situ hybridization. Cytogenet. Cell Genet. 79: 196–197, 1997.
- [42242] 15794. Yu, H.; Nicchitta, C. V.; Kumar, J.; Becker, M.; Toyoshima, I.; Sheetz, M. P.: Characterization of kinectin, a kinesin-binding protein: primary sequence and N-terminal topogenic signal analysis. Molec. Biol. Cell 6: 171–183, 1995.
- [42243] 15795. Gordenin, D. A.; Kunkel, T. A.; Resnick, M. A.: Repeat expansion— all in a flap? Nature Genet. 16: 116–118, 1997.
- [42244] 15796. Greene, A. L.; Snipe, J. R.; Gordenin, D. A.; Resnick, M. A.: Functional analysis of human FEN1 in *Saccharomyces cerevisiae* and its role in genome stability. Hum. Molec. Genet. 8: 2263–2273, 1999.
- [42245] 15797. Augusseau, S.; Jouk, S.; Jalbert, P.; Prieur, M.: DiGeorge syndrome and 22q11 rearrangements. (Letter) Hum. Genet. 74: 206 only, 1986.
- [42246] 15798. Van Esch, H.; Groenen, P.; Daw, S.; Poffyn, A.;

Holvoet, M.; Scambler, P.; Fryns, J.-P.; Van de Ven, W.; Devriendt, K.: Partial DiGeorges syndrome in two patients with a 10p rearrangement. *Clin. Genet.* 55:269–276, 1999.

[42247] 15799. Wadey, R.; Daw, S.; Taylor, C.; Atif, U.; Kamath, S.; Halford, S.; O'Donnell, H.; Wilson, D.; Goodship, J.; Burn, J.; Scambler, P.: Isolation of a gene encoding an integral membrane protein from the vicinity of a balanced translocation breakpoint associated with DiGeorges syndrome. *Hum. Molec. Genet.* 4: 1027–1033, 1995.

[42248] 15800. Contractor, A.; Rogers, C.; Maron, C.; Henkemeyer, M.; Swanson, G. T.; Heinemann, S. F.: Trans-synaptic Eph receptor–ephrin signaling in hippocampal mossy fiber LTP. *Science* 296: 1864–1869, 2002.

[42249] 15801. Tang, X. X.; Biegel, J. A.; Nycum, L. M.; Yoshioka, A.; Brodeur, G. M.; Pleasure, D. E.; Ikegaki, N.: cDNA cloning, molecular characterization, and chromosomal localization of NET (EPHT2), a human EPH-related receptor–protein–tyrosine kinase gene preferentially expressed in brain. *Genomics* 29:426–437, 1995.

[42250] 15802. Kamp, C.; Hirschmann, P.; Voss, H.; Huellen, K.; Vogt, P. H.: Two long homologous retroviral sequence blocks in proximal Yq11 cause AZFa microdeletions as a result of intrachromosomal recombination events. *Hum.*

Molec. Genet. 9: 2563–2572, 2000.

- [42251] 15803.Krausz, C.; Meyts, E. R.–D.; Frydelund–Larson, L.; Quintana–Murci,L.; McElreavey, K.; Skakkebaek, N. E.: Double–blind Y chromosomemicrodeletion analysis in men with known sperm parameters and reproductive hormone profiles: microdeletions are specific for spermatogenic failure. J.Clin. Endocr. Metab. 86: 2638–2642, 2001.
- [42252] 15804.Simpson, E.; Chandler, P.; Goulmy, E.; Ma, K.; Hargreave, T. B.;Chandley, A. C.: Loss of the 'azoospermia factor' (AZF) on Yq inman is not associated with loss of HYA. Hum. Molec. Genet. 2: 469–471,1993.
- [42253] 15805.Sun, C.; Skaletsky, H.; Rozen, S.; Gromoll, J.; Nieschlag, E.;Oates, R.; Page, D. C.: Deletion of azoospermia factor a (AZFa) regionof human Y chromosome caused by recombination between HERV15 proviruses. Hum.Molec. Genet. 9: 2291–2296, 2000.
- [42254] 15806.Krausz, C.; Quintana–Murci, L.; Barboux, S.; Siffroi, J.–P.; Rouba,H.; Delafontaine, D.; Souleyreau–Therville, N.; Arvis, G.; Antoine,J. M.; Erdei, E.; Taar, J. P.; Tar, A.; Jean–didier, E.; Plessis, G.;Bourgeron, T.; Dadoune, J.–P.; Fellous, M.; McElreavey, K.: A highfrequency of Y chromosome deletions in males with nonidiopathic infertility. J.Clin. Endocr. Metab. 84: 3606–3612, 1999.

- [42255] 15807.Ma, K.; Inglis, J. D.; Sharkey, A.; Bickmore, W. A.; Hill, R.E.; Prosser, E. J.; Speed, R. M.; Thomson, E. J.; Jobling, M.; Taylor,K.; Wolfe, J.; Cooke, H. J.; Hargreave, T. B.; Chandley, A. C.: AY chromosome gene family with RNA-binding protein homology: candidatesfor the azoospermia factor AZF controlling human spermatogenesis. *Cell* 75:1287–1295, 1993.
- [42256] 15808.Vergnaud, G.; Page, D. C.; Simmler, M. C.; Brown, L.; Rouyer,F.; Noel, B.; Botstein, D.; de la Chapelle, A.; Weissenbach, J.:A deletion map of the human Y chromosome based on DNA hybridization. *Am.J. Hum. Genet.* 38: 109–124, 1986.
- [42257] 15809.Vogt, P.; Chandley, A. C.; Hargreave, T. B.; Keil, R.; Ma, K.;Sharkey, A.: Microdeletions in interval 6 of the Y chromosome ofmales with idiopathic sterility point to disruption of AZF, a humanspermatogenesis gene. *Hum. Genet.* 89: 491–496, 1992.
- [42258] 15810.Vogt, P. H.; Edelman, A.; Kirsch, S.; Henegariu, O.; Hirschmann,P.; Kiesewetter, F.; Kohn, F. M.; Schill, W. B.; Farah, S.; Ramos,C.; Hartmann, M.; Hartschuh, W.; Meschede, D.; Behre, H. M.; Castel,A.; Nieschlag, E.; Weidner, W.; Grone, H.–J.; Jung, A.; Engel, W.;Haidl, G.: Human Y chromosome azoospermia factors (AZF) mapped todif–

ferent subregions in Yq11. Hum. Molec. Genet. 5: 933–943, 1996.

[42259] 15811. Agulnik, A. I.; Bishop, C. E.; Lerner, J. L.; Agulnik, S. I.; Solovyev, V. V.: Analysis of mutation rates in the SMCY/SMCX genes shows that mammalian evolution is male driven. Mammalian Genome 8: 134–138, 1997.

[42260] 15812. Hawkins, J. R.: Mutational analysis of SRY in XY females. Hum. Mutat. 2: 347–350, 1993.

[42261] 15813. Hawkins, J. R.; Taylor, A.; Berta, P.; Levilliers, J.; Van der Auwera, B.; Goodfellow, P. N.: Mutational analysis of SRY: nonsense and missense mutations in XY sex reversal. Hum. Genet. 88: 471–474, 1992.

[42262] 15814. Rohen, C.; Caselitz, J.; Stern, C.; Wanschura, S.; Schoenmakers, E. F.; Van de Ven, W. J.; Barnitzke, S.; Bullerdiek, J.: A hamartoma of the breast with an aberration of 12q mapped to the MAR region by fluorescence in situ hybridization. Genes Chromosomes Cancer 84: 82–84, 1995.

[42263] 15815. Schoenmakers, E. F. P. M.; Huysmans, C.; Van de Ven, W. J. M.: Allelic knockout of novel splice variants of human recombination repair gene RAD51B in t(12;14) uterine leiomyomas. Cancer Res. 59: 19–23, 1999.

[42264] 15816. Zhou, X.; Benson, K. F.; Ashar, H. R.; Chada, K.:

Mutation responsible for the mouse pygmy phenotype in the developmentally regulated factor HMGI-C. *Nature* 377: 771–774, 1995.

- [42265] 15817. Agalioti, T.; Lomvardas, S.; Parekh, B.; Yie, J.; Maniatis, T.; Thanos, D.: Ordered recruitment of chromatin modifying and general transcription factors to the IFN- β promoter. *Cell* 103: 667–678, 2000.
- [42266] 15818. Munshi, N.; Agalioti, T.; Lomvardas, S.; Merika, M.; Chen, G.; Thanos, D.: Coordination of a transcriptional switch by HMGI(Y) acetylation. *Science* 293: 1133–1136, 2001.
- [42267] 15819. Thanos, D.; Maniatis, T.: Virus induction of human IFN- β gene expression requires the assembly of an enhanceosome. *Cell* 83: 1091–1100, 1995.
- [42268] 15820. Wood, L. J.; Mukherjee, M.; Dolde, C. E.; Xu, Y.; Maher, J. F.; Bunton, T. E.; Williams, J. B.; Resar, L. M. S.: HMG-I/Y, a new c-Myb target gene and potential oncogene. *Molec. Cell Biol.* 20: 5490–5502, 2000.
- [42269] 15821. Burgess, D. L.; Kohrman, D. C.; Galt, J.; Plummer, N. W.; Jones, J. M.; Spear, B.; Meisler, M. H.: Mutation of a new sodium channel gene, *Scn8a*, in the mouse mutant 'motor endplate disease'. *Nature Genet.* 10: 461–465, 1995.

- [42270] 15822.DeRepentigny, Y.; Cote, P. D.; Pool, M.; Bernier, G.; Girard, S.;Vidal, S. M.; Kothary, R.: Pathological and genetic analysis of the degenerating muscle (dmu) mouse: a new allele of Scn8a. *Hum. Molec.Genet.* 10: 1819–1827, 2001.
- [42271] 15823.Kohrman, D. C.; Harris, J. B.; Meisler, M. H.: Mutation detection in the med and med(j) alleles of the sodium channel Scn8a: unusual splicing due to a minor class AT–AC intron. *J. Biol. Chem.* 271:17576–17581, 1996.
- [42272] 15824.Kohrman, D. C.; Plummer, N. W.; Schuster, T.; Jones, J. M.; Jang, W.; Burgess, D. L.; Galt, J.; Spear, B. T.; Meisler, M. H.: Insertional mutation of the motor endplate disease (med) locus on mouse chromosome 15. *Genomics* 26: 171–177, 1995.
- [42273] 15825.Kohrman, D. C.; Smith, M. R.; Goldin, A. L.; Harris, J.; Meisler, M. H.: A missense mutation in the sodium channel Scn8a is responsible for cerebellar ataxia in the mouse mutant jolting. *J. Neurosci.* 16:5993–5999, 1996.
- [42274] 15826.Meisler, M. H.; Sprunger, L. K.; Plummer, N. W.; Escayg, A.; Jones, J. M.: Ion channel mutations in mouse models of inherited neurological disease. *Ann. Med.* 29: 569–574, 1997.
- [42275] 15827.Plummer, N. W.; Galt, J.; Jones, J. M.; Burgess, D. L.;

Sprunger, L. K.; Kohrman, D. C.; Meisler, M. H.: Exon organization, coding sequence, physical mapping, and polymorphic intragenic markers for the human neuronal sodium channel gene SCN8A. *Genomics* 54: 287–296, 1998.

- [42276] 15828. Plummer, N. W.; McBurney, M. W.; Meisler, M. H.: Alternative splicing of the sodium channel SCN8A predicts a truncated two-domain protein in fetal brain and non-neuronal cells. *J. Biol. Chem.* 272: 24008–24015, 1997.
- [42277] 15829. Reid, E.; Escayg, A.; Dearlove, A. M.; Lee, D. D.; Meisler, M. H.; Rubinstein, D. C.: The spastic paraplegia SPG10 locus: narrowing of critical region and exclusion of sodium channel gene SCN8A as a candidate. *J. Med. Genet.* 38: 65–67, 2001.
- [42278] 15830. Sprunger, L. K.; Escayg, A.; Tallaksen-Greene, S.; Albin, R. L.; Meisler, M. H.: Dystonia associated with mutation of the neuronal sodium channel Scn8a and identification of the modifier locus Scnm1 on mouse chromosome 3. *Hum. Molec. Genet.* 8: 471–479, 1999.
- [42279] 15831. Chang, D. Y.; Nelson, B.; Bilyeu, T.; Hsu, K.; Darlington, G. J.; Maraia, R. J.: A human Alu RNA-binding protein whose expression is associated with accumulation of small cytoplasmic Alu RNA. *Molec. Cell. Biol.* 14:

3949–3959, 1994.

- [42280] 15832.Chen, M.; She, H.; Davis, E. M.; Spicer, C. M.; Kim, L.; Ren, R.;LeBeau, M. M.; Li, W.: Identification of Nck family genes, chromosomallocalization, expression, and signaling specificity. *J. Biol. Chem.* 273:25171–25178, 1998.
- [42281] 15833.Eden, S.; Rohatgi, R.; Podtelejnikov, A. V.; Mann, M.; Kirschner,M. W.: Mechanism of regulation of WAVE1–induced actin nucleationby Rac1 and Nck. *Nature* 418: 790–793, 2002.
- [42282] 15834.Casey, J. L.; Di Jeso, B.; Rao, K.; Klausner, R. D.; Harford, J.B.: Two genetic loci participate in the regulation by iron of thegene for the human transferrin receptor. *Proc. Nat. Acad. Sci.* 85:1787–1791, 1988.
- [42283] 15835.Enns, C. A.; Suomalainen, H. A.; Gebhardt, J. E.; Schroder, J.;Sussman, H. H.: Human transferrin receptor: expression of the receptoris assigned to chromosome 3. *Proc. Nat. Acad. Sci.* 79: 3241–3245,1982.
- [42284] 15836.Gareau, R.; Gagnon, M. G.; Thellend, C.; Chenard, C.; Audran, M.;Chanal, J.–L.; Ayotte, C.; Brisson, G. R.: Transferrin soluble receptor:a possible probe for detection of erythropoietin abuse by athletes. *Horm.Metab. Res.* 26: 311–312, 1994.
- [42285] 15837.Goodfellow, P. N.; Banting, G.; Sutherland, R.;

Greaves, M.; Solomon, E.; Povey, S.: Expression of human transferrin receptor is controlled by a gene on chromosome 3: assignment using species specificity of a monoclonal antibody. *Somat. Cell Genet.* 8: 197–206, 1982.

- [42286] 15838. Larrick, J. W.; Hyman, E. S.: Acquired iron-deficiency anemia caused by an antibody against the transferrin receptor. *New Eng. J. Med.* 311: 214–218, 1984.
- [42287] 15839. Levy, J. E.; Jin, O.; Fujiwara, Y.; Kuo, F.; Andrews, N. C.: Transferrin receptor is necessary for development of erythrocytes and the nervous system. *Nature Genet.* 21: 396–399, 1999.
- [42288] 15840. Miller, Y. E.; Jones, C.; Scoggin, C.; Morse, H.; Seligman, P.: Chromosome 3q (22–ter) encodes the human transferrin receptor. *Am. J. Hum. Genet.* 35: 573–583, 1983.
- [42289] 15841. Newman, R.; Schneider, C.; Sutherland, R.; Vodinelich, L.; Greaves, M.: The transferrin receptor. *Trends Biochem. Sci.* 7: 397–400, 1982.
- [42290] 15842. Nikinmaa, B.; Schroder, J.: Two antigens, the transferrin receptor and p90 assigned to human chromosome 3, are probably the same protein. *Hereditas* 107:55–58, 1987.

- [42291] 15843.Omary, M. B.; Trowbridge, I. S.: Biosynthesis of the human transferrin receptor in cultured cells. *J. Biol. Chem.* 256: 12888–12892, 1981.
- [42292] 15844.Kashuba, V. I.; Gizatullin, R. Z.; Protopopov, A. I.; Allikmets, R.; Korolev, S.; Li, J.; Boldog, F.; Tory, K.; Zabarovska, V.; Marcsek, Z.; Sumegi, J.; Klein, G.; Zabarovsky, E. R.; Kisselev, L.: Not linking/jumping clones of human chromosome 3: mapping of the TFRC, RAB7 and HAUSP genes to regions rearranged in leukemia and deleted in solid tumors. *FEBS Lett.* 419: 181–185, 1997.
- [42293] 15845.Rabin, M.; McClelland, A.; Kuhn, L.; Ruddle, F. H.: Regional localization of the human transferrin receptor gene to 3q26.2–qter. *Am. J. Hum. Genet.* 37: 1112–1116, 1985.
- [42294] 15846.Schneider, C.; Kurkinen, M.; Greaves, M.: Isolation of cDNA clones for the human transferrin receptor. *EMBO J.* 2: 2259–2263, 1983.
- [42295] 15847.Schneider, C.; Owen, M. J.; Banville, D.; Williams, J. G.: Primary structure of human transferrin receptor deduced from the mRNA sequence. *Nature* 311: 675–678, 1984.
- [42296] 15848.Valenzuela, C. Y.; Avendano, A.; Harb, Z.: Association between Rh and plasma iron binding (transferrin).

Hum. Genet. 87: 438–440,1991.

[42297] 15849.Vodinelich, L.; Sutherland, R.; Schneider, C.; Newman, R.; Greaves,M.: Receptor for transferrin may be a 'target' structure for naturalkiller cells. Proc. Nat. Acad. Sci. 80: 835–839, 1983.

[42298] 15850.Carmellet, P.; Ferreira, V.; Breler, G.; Pollefeyt, S.; Kleckens,L.; Gertsenstein, M.; Fahrig, M.; Vandenhoeck, A.; Harpal, K.; Eberhardt,C.; Declercq, C.; Pawling, J.; Moons, L.; Collen, D.; Risau, W.; Nagy,A.: Abnormal blood vessel development and lethality in embryos lacking a single VEGF allele. Nature 380: 435–439, 1996.

[42299] 15851.Crow, R. S.: Peripheral neuritis in myelomatosis. Brit. Med.J. 2: 802–804, 1956.

[42300] 15852.Dantz, D.; Bewersdorf, J.; Fruehwald–Schultes, B.; Kern, W.; Jelkmann,W.; Born, J.; Fehm, H. L.; Peters, A.: Vascular endothelial growthfactor: a novel endocrine defensive response to hypoglycemia. J.Clin. Endocr. Metab. 87: 835–840, 2002.

[42301] 15853.Ferrara, N.; Carver–Moore, K.; Chen, H.; Dowd, M.; Lu, L.; O'Shea,K. S.; Powell–Braxton, L.; Hillan, K. J.; Moore, M. W.: Heterozygousembryonic lethality induced by targeted inactivation of the VEGF gene. Nature 380:439–442, 1996.

- [42302] 15854.Folkman, J.: Angiogenesis in cancer, vascular, rheumatoid and other disease. *Nature Med.* 1: 27–31, 1995.
- [42303] 15855.Fukumura, D.; Xavier, R.; Sugiura, T.; Chen, Y.; Park, E.-C.; Lu, N.; Selig, M.; Nielsen, G.; Taksir, T.; Jain, R. K.; Seed, B.: Tumor induction of VEGF promoter activity in stromal cells. *Cell* 94: 715–725, 1998.
- [42304] 15856.Gerber, H.-P.; Malik, A. K.; Solar, G. P.; Sherman, D.; Liang, X. H.; Meng, G.; Hong, K.; Marsters, J. C.; Ferrara, N.: VEGF regulates haematopoietic stem cell survival by an internal autocrine loop mechanism. *Nature* 417:954–958, 2002.
- [42305] 15857.Gerber, H.-P.; Vu, T. H.; Ryan, A. M.; Kowalski, J.; Werb, Z.; Ferrara, N.: VEGF couples hypertrophic cartilage remodeling, ossification and angiogenesis during endochondral bone formation. *Nature Med.* 5:623–628, 1999.
- [42306] 15858.Giordano, F. J.; Gerber, H.-P.; Williams, S.-P.; Van-Bruggen, N.; Bunting, S.; Ruiz-Lozano, P.; Gu, Y.; Nath, A. K.; Huang, Y.; Hickey, R.; Dalton, N.; Peterson, K. L.; Ross, J., Jr.; Chien, K. R.; Ferrara, N.: A cardiac myocyte vascular endothelial growth factor paracrine pathway is required to maintain cardiac function. *Proc. Nat. Acad. Sci.* 98: 5780–5785, 2001.

- [42307] 15859. Helmlinger, G.; Endo, M.; Ferrara, N.; Hlatky, L.; Jain, R. K.: Formation of endothelial cell networks. *Nature* 405: 139–141, 2000.
- [42308] 15860. Hofman, P.; van Blijswijk, B. C.; Gaillard, P. J.; Vrensen, G.F. J. M.; Schlingemann, R. O.: Endothelial cell hypertrophy induced by vascular endothelial growth factor in the retina: new insights into the pathogenesis of capillary nonperfusion. *Arch. Ophthalmol.* 119: 861–866, 2001.
- [42309] 15861. Holash, J.; Maisonpierre, P. C.; Compton, D.; Boland, P.; Alexander, C. R.; Zagzag, D.; Yancopoulos, G. D.; Wiegand, S. J.: Vessel cooption, regression, and growth in tumors mediated by angiopoietins and VEGF. *Science* 284: 1994–1998, 1999.
- [42310] 15862. Jin, K.; Zhu, Y.; Sun, Y.; Mao, X. O.; Xie, L.; Greenberg, D. A.: Vascular endothelial growth factor (VEGF) stimulates neurogenesis in vitro and in vivo. *Proc. Nat. Acad. Sci.* 99: 11946–11950, 2002.
- [42311] 15863. Mattei, M.-G.; Borg, J.-P.; Rosnet, O.; Marme, D.; Birnbaum, D.: Assignment of vascular endothelial growth factor (VEGF) and placental growth factor (PlGF) genes to human chromosome 6p12–p21 and 14q24–q31 regions, respectively. *Genomics* 32: 168–169, 1996.
- [42312] 15864. Miralles, G. D.; O'Fallon, J. R.; Talley, N. J.: Plasma–

celldyscrasia with polyneuropathy: the spectrum of POEMS syndrome. *NewEng. J. Med.* 327: 1919–1923, 1992.

[42313] 15865. Mueller, M. D.; Vigne, J.-L.; Minchenko, A.; Lebovic, D. I.; Leitman, D. C.; Taylor, R. N.: Regulation of vascular endothelial growth factor(VEGF) gene transcription by estrogen receptors alpha and beta. *Proc.Nat. Acad. Sci.* 97: 10972–10977, 2000.

[42314] 15866. Nakanishi, T.; Sobue, I.; Toyokura, Y.; Nishitani, H.; Kuroiwa, Y.; Satoyoshi, E.; Tsubaki, T.; Igata, A.; Ozaki, Y.: The Crow–Fukasesyndrome: a study of 102 cases in Japan. *Neurology* 34: 712–720, 1984.

[42315] 15867. Niimi, H.; Arimura, K.; Jonosono, M.; Hashiguchi, T.; Kawabata, M.; Osame, M.; Kitajima, I.: VEGF is causative for pulmonary hypertension in a patient with Crow–Fukase (POEMS) syndrome. *Intern. Med.* 39: 1101–1104, 2000.

[42316] 15868. Oosthuyse, B.; Moons, L.; Storkebaum, E.; Beck, H.; Nuyens, D.; Brusselmans, K.; Van Dorpe, J.; Hellings, P.; Gorselink, M.; Heymans, S.; Theilmeier, G.; Dewerchin, M.; and 20 others: Deletion of the hypoxia–response element in the vascular endothelial growth factor promoter causes motor neuron degeneration. *Nature Genet.* 28: 131–138, 2001.

[42317] 15869. Poltorak, Z.; Cohen, T.; Sivan, R.; Kandelis, Y.;

Spira, G.; Vlodavsky, I.; Keshet, E.; Neufeld, G.: VEGF145, a secreted vascular endothelial growth factor isoform that binds to extracellular matrix. *J. Biol. Chem.* 272: 7151–7158, 1997.

[42318] 15870. Shimpo, S.: Solitary myeloma causing polyneuritis and endocrine disorders. *Nippon Rinsho* 26: 2444–2456, 1968.

[42319] 15871. Soker, S.; Takashima, S.; Miao, H. Q.; Neufeld, G.; Klagsbrun, M.: Neuropilin-1 is expressed by endothelial and tumor cells as an isoform-specific receptor for vascular endothelial growth factor. *Cell* 92: 735–745, 1998.

[42320] 15872. Sone, H.; Kawakami, Y.; Sakauchi, M.; Nakamura, Y.; Takahashi, A.; Shimano, H.; Okuda, Y.; Segawa, T.; Suzuki, H.; Yamada, N.: Neutralization of vascular endothelial growth factor prevents collagen-induced arthritis and ameliorates established disease in mice. *Biochem. Biophys. Res. Commun.* 281: 562–568, 2001.

[42321] 15873. Springer, M. L.; Chen, A. S.; Kraft, P. E.; Bednarski, M.; Blau, H. M.: VEGF gene delivery to muscle: potential role for vasculogenesis in adults. *Molec. Cell* 2: 549–558, 1998.

[42322] 15874. Thurston, G.; Suri, C.; Smith, K.; McClain, J.; Sato, T. N.; Yancopoulos, G. D.; McDonald, D. M.: Leakage-re-

sistant blood vessels in mice transgenically overexpressing angiopoietin-1. *Science* 286: 2511–2514, 1999.

- [42323] 15875. Tischer, E.; Mitchell, R.; Hartman, T.; Silva, M.; Gospodarowicz, D.; Fiddes, J. C.; Abraham, J. A.: The human gene for vascular endothelial growth factor: multiple protein forms are encoded through alternative exon splicing. *J. Biol. Chem.* 266: 11947–11954, 1991.
- [42324] 15876. Osborne, L. R.; Martindale, D.; Scherer, S. W.; Shi, X.-M.; Huizenga, J.; Heng, H. H. Q.; Costa, T.; Pober, B.; Lew, L.; Brinkman, J.; Rommens, J.; Koop, B.; Tsui, L.-C.: Identification of genes from a 500-kb region at 7q11.23 that is commonly deleted in Williams syndrome patients. *Genomics* 36:328–336, 1996.
- [42325] 15877. Rampoldi, L.; Dobson-Stone, C.; Rubio, J. P.; Danek, A.; Chalmers, R. M.; Wood, N. W.; Verellen, C.; Ferrer, X.; Malandrini, A.; Fabrizi, G. M.; Brown, R.; Vance, J.; Pericak-Vance, M.; Rudolf, G.; Carre, S.; Alonso, E.; Manfredi, M.; Nemeth, A. H.; Monaco, A. P.: A conserved sorting-associated protein is mutant in chorea-acanthocytosis. *Nature Genet.* 28: 119–120, 2001.
- [42326] 15878. Weese-Mayer, D. E.; Bolk, S.; Silvestri, J. M.; Chakravarti, A.: Idiopathic congenital central hypoventilation syndrome: evaluation of brain-derived neurotrophic

factor genomic DNA sequence variation. *Am.J. Med. Genet.* 107: 306–310, 2002.

- [42327] 15879.Belden, W. J.; Barlowe, C.: Role of Erv29p in collecting solublesecretory proteins into ER–derived transport vesicles. *Science* 294:1528–1531, 2001.
- [42328] 15880.Duhig, T.; Ruhrberg, C.; Mor, O.; Fried, M.: The human surfetlocus. *Genomics* 52: 72–78, 1998.
- [42329] 15881.Reeves, J. E.; Fried, M.: The surf–4 gene encodes a novel 30 kDaintegral membrane protein. *Molec. Membr. Biol.* 12: 201–208, 1995.
- [42330] 15882.Archer, B. T., III; Ozcelik, T.; Jahn, R.; Francke, U.; Sudhof,T. C.: Structures and chromosomal localizations of two human genesencoding synaptobrevins 1 and 2. *J. Biol. Chem.* 265: 17267–17273,1990.
- [42331] 15883.Hu, K.; Carroll, J.; Fedorovich, S.; Rickman, C.; Sukhodub, A.;Davietov, B.: Vesicular restriction of synaptobrevin suggests a rolefor calcium in membrane fusion. *Nature* 415: 646–650, 2002.
- [42332] 15884.Franchi, F.; Biguzzi, E.; Cetin, I.; Facchetti, F.; Radaelli, T.;Bozzo, M.; Pardi, G.; Faioni, E. M.: Mutations in the thrombomodulinand endothelial protein C receptor genes in women with late fetalloss. *Brit. J. Haemat.* 114: 641–646, 2001.

- [42333] 15885.Perez, F.; Diamantopoulos, G. S.; Stalder, R.; Kreis, T. E.: CLIP-170 highlights growing microtubule ends in vivo. *Cell* 96: 517–527, 1999.
- [42334] 15886.Pierre, P.; Pepperkok, R.; Kreis, T. E.: Molecular characterization of two functional domains of CLIP-170 in vivo. *J. Cell Sci.* 107:1909–1920, 1994.
- [42335] 15887.Pierre, P.; Scheel, J.; Rickard, J. E.; Kreis, T. E.: CLIP-170 links endocytic vesicles to microtubules. *Cell* 70: 887–900, 1992.
- [42336] 15888.Acland, G. M.; Aguirre, G. D.; Ray, J.; Zhang, Q.; Aleman, T. S.; Cideciyan, A. V.; Pearce-Kelling, S. E.; Anand, V.; Zeng, Y.; Maguire, A. M.; Jacobson, S. G.; Hauswirth, W. W.; Bennett, J.: Gene therapy restores vision in a canine model of childhood blindness. *Nature* 28:92–95, 2001.
- [42337] 15889.Aguirre, G. D.; Baldwin, V.; Pearce-Kelling, S.; Narfstrom, K.; Ray, K.; Acland, G. M.: Congenital stationary night blindness in the dog: common mutation in the RPE65 gene indicates founder effect. *Molec.Vision* 4: 23, 1998.
Note: Electronic Article.
- [42338] 15890.Bavik, C.-O.; Busch, C.; Eriksson, U.: Characterization of a plasma retinol-binding protein membrane receptor expressed in the retinal pigment epithelium. *J. Biol.*

Chem. 267: 23035–23042, 1992.

- [42339] 15891.Felius, J.; Thompson, D. A.; Khan, N. W.; Bingham, E. L.; Jamison, J. A.; Kemp, J. A.; Sieving P. A.: Clinical course and visual function in a family with mutations in the RPE65 gene. Arch. Ophthalmol. 120:55–61, 2002.
- [42340] 15892.Grimm, C.; Wenzel, A.; Hafezi, F.; Yu, S.; Redmond, T. M.; Reme, C. E.: Protection of Rpe65-deficient mice identifies rhodopsin as a mediator of light-induced retinal degeneration. Nature Genet. 25:63–66, 2000.
- [42341] 15893.Gu, S.; Thompson, D. A.; Srikumari, C. R. S.; Lorenz, B.; Finckh, U.; Nicoletti, A.; Murthy, K. R.; Rathmann, M.; Kumaramanickavel, G.; Denton, M. J.; Gal, A.: Mutations in RPE65 cause autosomal recessive childhood-onset severe retinal dystrophy. Nature Genet. 17: 194–197, 1997.
- [42342] 15894.Hamel, C. P.; Jenkins, N. A.; Gilbert, D. J.; Copeland, N. G.; Redmond, T. M.: The gene for the retinal pigment epithelium-specific protein RPE65 is localized to human 1p31 and mouse 3. Genomics 20:509–512, 1994.
- [42343] 15895.Hamel, C. P.; Tsilou, E.; Pfeiffer, B. A.; Hooks, J. J.; Detrick, B.; Redmond, T. M.: Molecular cloning and expression of RPE65, a novel retinal pigment epithelium-specific microsomal protein that is post-transcriptionally regulated

in vitro. J. Biol. Chem. 268:15751–15757, 1993.

- [42344] 15896. Marlhens, F.; Bareil, C.; Griffoin, J.–M.; Zrenner, E.; Amalric, P.; Eliaou, C.; Liu, S.–Y.; Harris, E.; Redmond, T. M.; Arnaud, B.; Claustres, M.; Hamel, C. P.: Mutations in RPE65 cause Leber's congenital amaurosis. (Letter) Nature Genet. 17: 139–141, 1997.
- [42345] 15897. Morimura, H.; Fishman, G. A.; Grover, S. A.; Fulton, A. B.; Berson, E. L.; Dryja, T. P.: Mutations in the RPE65 gene in patients with autosomal recessive retinitis pigmentosa or Leber congenital amaurosis. Proc. Nat. Acad. Sci. 95: 3088–3093, 1998.
- [42346] 15898. Narfstrom, K.; Wrigstad, A.; Nilsson, S. E.: The Briard dog: a new animal model of congenital stationary night blindness. Brit. J. Ophthalmol. 73: 750–756, 1989.
- [42347] 15899. Nicoletti, A.; Wong, D. J.; Kawase, K.; Gibson, L. H.; Yang–Feng, T. L.; Richards, J. E.; Thompson, D. A.: Molecular characterization of the human gene encoding an abundant 61 kDa protein specific to the retinal pigment epithelium. Hum. Molec. Genet. 4: 641–649, 1995.
- [42348] 15900. Adekun, A. M.; West, S. P.; Ellis, F. R.; Halsall, P. J.; Hopkins, P. M.; Foroughmand, A. M.; Iles, D. E.; Robinson, R. L.; Stewart, A. D.; Curran, J. L.: The G1021A substitution in the RYR1 gene does not cosegregate with malignant hy–

perthermia susceptibility in a British pedigree. *Am. J. Hum. Genet.* 60: 833–341, 1997.

[42349] 15901. Alestrom, A.; Fagerlund, T. H.; Berg, K.: A simple method to detect the RYR1 mutation G1021A, a cause of malignant hyperthermia susceptibility. *Clin. Genet.* 47: 274–275, 1995.

[42350] 15902. Avila, G.; O'Brien, J. J.; Dirksen, R. T.: Excitation–contraction uncoupling by a human central core disease mutation in the ryanodine receptor. *Proc. Nat. Acad. Sci.* 98: 4215–4220, 2001.

[42351] 15903. Barone, V.; Bertocchini, F.; Bottinelli, R.; Protasi, F.; Allen, P. D.; Armstrong, C. F.; Reggiani, C.; Sorrentino, V.: Contractile impairment and structural alterations of skeletal muscles from knockout mice lacking type 1 and type 3 ryanodine receptors. *FEBS Lett.* 422: 160–164, 1998.

[42352] 15904. Brandt, A.; Schleithoff, L.; Jurkat-Rott, K.; Klingler, W.; Baur, C.; Lehmann-Horn, F.: Screening of ryanodine receptor gene in 105 malignant hyperthermia families: novel mutations and concordance with the in vitro contracture test. *Hum. Molec. Genet.* 8: 2055–2062, 1999.

[42353] 15905. Brown, R. L.; Pollock, A. N.; Couchman, K. G.; Hodges, M.; Hutchinson, D. O.; Waaka, R.; Lynch, P.; McCarthy, T. V.; Stowell, K. M.: A novel ryanodine receptor

mutation and genotype–phenotype correlation in a large malignant hyperthermia New Zealand Maori pedigree. Hum. Molec. Genet. 9: 1515–1524, 2000.

[42354] 15906. Cavanna, J. S.; Greenfield, A. J.; Johnson, K. J.; Marks, A. R.; Nadal-Ginard, B.; Brown, S. D. M.: Establishment of the mouse chromosome 7 region with homology to the myotonic dystrophy region of human chromosome 19q. Genomics 7: 12–18, 1990.

[42355] 15907. Cheng, H.; Lederer, W. J.; Cannell, M. B.: Calcium sparks: elementary events underlying excitation–contraction coupling in heart muscle. Science 262: 740–744, 1993.

[42356] 15908. Deufel, T.; Sudbrak, R.; Feist, Y.; Rubsam, B.; Du Chesne, I.; Schafer, K.–L.; Roewer, N.; Grimm, T.; Lehmann–Horn, F.; Hartung, E. J.; Muller, C. R.: Discordance, in a malignant hyperthermia pedigree, between in vitro contracture–test phenotypes and haplotypes for the MHS1 region on chromosome 19q12–13.2, comprising the C1840T transition in the RYR1 gene. Am. J. Hum. Genet. 56: 1334–1342, 1995.

[42357] 15909. Eu, J. P.; Sun, J.; Xu, L.; Stamler, J. S.; Meissner, G.: The skeletal muscle calcium release channel: coupled O₂ sensor and NO signaling functions. Cell 102: 499–509,

2000.

- [42358] 15910.Fagerlund, T.; Ording, H.; Bendixen, D.; Berg, K.: Search for three known mutations in the RYR gene in 48 Danish families with malignant hyperthermia. Clin. Genet. 46: 401–404, 1994.
- [42359] 15911.Fagerlund, T.; Ording, H.; Bendixen, D.; Islander, G.; Ranklev-Twetman, E.; Berg, K.: RYR1 mutation G1021A (gly341-to-arg) is not frequent in Danish and Swedish families with malignant hyperthermia susceptibility. Clin. Genet. 49: 186–188, 1996.
- [42360] 15912.Fagerlund, T. H.; Islander, G.; Ranklev-Twetman, E.; Berg, K.: Recombination between the postulated CCD/MHE/MHS locus and RYR1 gene markers. Clin. Genet. 50: 455–458, 1996.
- [42361] 15913.Fagerlund, T. H.; Islander, G.; Twetman, E. R.; Berg, K.: A search for three known RYR1 gene mutations in 41 Swedish families with predisposition to malignant hyperthermia. Clin. Genet. 48: 12–16, 1995.
- [42362] 15914.Fagerlund, T. H.; Ording, H.; Bendixen, D.; Islander, G.; Ranklev-Twetman, E.; Berg, K.: Discordance between malignant hyperthermia susceptibility and RYR1 mutation C1840T in two Scandinavian MH families exhibiting this mutation. Clin. Genet. 52: 416–421, 1997.

- [42363] 15915.Ferreiro, A.; Monnier, N.; Romero, N. B.; Leroy, J.-P.; Bonnemann, C.; Haenggeli, C.-A.; Straub, V.; Voss, W. D.; Nivoche, Y.; Jungbluth, H.; Lemainque, A.; Voit, T.; Lunnardi, J.; Fardeau, M.; Guicheney, P.: A recessive form of central core disease, transiently presenting as multi-minicore disease, is associated with a homozygous mutation in the ryanodine receptor type 1 gene. *Ann. Neurol.* 51: 750–759, 2002.
- [42364] 15916.Fujii, J.; Otsu, K.; Zorzato, F.; de Leon, S.; Khanna, V. K.; Weiler, J. E.; O'Brien, P. J.; MacLennan, D. H.: Identification of a mutation in porcine ryanodine receptor associated with malignant hyperthermia. *Science* 253: 448–451, 1991.
- [42365] 15917.Gillard, E. F.; Otsu, K.; Fujii, J.; Duff, C.; de Leon, S.; Khanna, V. K.; Britt, B. A.; Worton, R. G.; MacLennan, D. H.: Polymorphisms and deduced amino acid substitutions in the coding sequence of the ryanodine receptor (RYR1) gene in individuals with malignant hyperthermia. *Genomics* 13:1247–1254, 1992.
- [42366] 15918.Gillard, E. F.; Otsu, K.; Fujii, J.; Khanna, V. K.; de Leon, S.; Derdemezi, J.; Britt, B. A.; Duff, C. L.; Worton, R. G.; MacLennan, D. H.: A substitution of cysteine for arginine 614 in the ryanodine receptor is potentially causative

of human malignant hyperthermia. Genomics
11:751–755, 1991.

[42367] 15919.Hall–Curran, J. L.; Stewart, A. D.; Ball, S. P.; Halsall, J. P.;Hopkins, P. M.; Ellis, F. R.: No C1840 to T mutation in RYR1 in malignanthyperthermia.(Letter) Hum. Mutat. 2: 330, 1993.

[42368] 15920.Quane, K. A.; Keating, K. E.; Manning, B. M.; Healy, J. M. S.;Monsieurs, K.; Heffron, J. J. A.; Lehane, M.; Heytens, L.; Krivosic–Horber,R.; Adnet, P.; Ellis, F. R.; Monnier, N.; Lunardi, J.; McCarthy, T.V.: Detection of a novel common mutation in the ryanodine receptorgene in malignant hyperthermia: implications for diagnosis and heterogeneitystudies. Hum. Molec. Genet. 3: 471–476, 1994.

[42369] 15921.Vincenti, V.; Cassano, C.; Rocchi, M.; Persico, M. G.: Assignmentof the vascular endothelial growth factor gene to human chromosome6p21.3. Circulation 93: 1493–1495, 1996.

[42370] 15922.Alonso, M.; Barton, D. E.; Francke, U.: MAL, a membrane proteinassociated with human T–cell differen–tiation, is encoded on humanchromosome 2, region cen–q13. (Abstract) Cytogenet. Cell Genet. 46:570 only, 1987.

[42371] 15923.Alonso, M. A.; Barton, D. E.; Francke, U.: Assign–

ment of the T-cell differentiation gene MAL to human chromosome 2, region cen-q13. Immunogenetics 27:91-95, 1988.

[42372] 15924. Alonso, M. A.; Weissman, S. M.: cDNA cloning and sequence of MAL, a hydrophobic protein associated with human T-cell differentiation. Proc. Nat. Acad. Sci. 84: 1997-2001, 1987.

[42373] 15925. Millan, J.; Puertollano, R.; Fan, L.; Rancano, C.; Alonso, M. A.: The MAL proteolipid is a component of the detergent-insoluble membranesubdomains of human T-lymphocytes. Biochem. J. 321: 247-252, 1997.

[42374] 15926. Rancano, C.; Rubio, T.; Alonso, M. A.: Alternative splicing of human T-cell-specific MAL mRNA and its correlation with the exon/intron organization of the gene. Genomics 21: 447-450, 1994.

[42375] 15927. Wong-Staal, F.; Dalla-Favera, R.; Franchini, G.; Gelmann, E. P.; Gallo, R. C.: Three distinct genes in human DNA related to the transforming genes of mammalian sarcoma retroviruses. Science 213: 226-228, 1981.

[42376] 15928. Aurias, A.; Rimbaut, C.; Buffe, D.; Dubousset, J.; Mazabraud, A.: Chromosomal translocations in Ewing's sarcoma. (Letter) New Eng. J. Med. 309: 496-497, 1983.

[42377] 15929. Bartram, C. R.; de Klein, A.; Hagemeijer, A.;

Grosveld, G.; Heisterkamp, N.; Groffen, J.: Localization of the human c-sis oncogene in Ph-1-positive and Ph-1-negative chronic myelocytic leukemia by in situ hybridization. *Blood* 63:223-225, 1984.

[42378] 15930. Bechet, J.-M.; Bornkamm, G.; Freese, U.-K.; Lenoir, G. M.: The c-sis oncogene is not activated in Ewing's sarcoma. (Letter) *New Eng. J. Med.* 310: 393 only, 1984.

[42379] 15931. Bishop, J. M.: Enemies within: the genesis of retrovirus oncogenes. *Cell* 23:5-6, 1981.

[42380] 15932. Bolger, G. B.; Stamberg, J.; Kirsch, I. R.; Hollis, G. F.; Schwarz, D. F.; Thomas, G. H.: Chromosomal translocation t(14;22) and oncogene (c-sis) variant in a pedigree with familial meningioma. *New Eng. J. Med.* 312: 564-567, 1985.

[42381] 15933. Cohen, J. B.; Levinson, A. D.: A point mutation in the last intron responsible for increased expression and transforming activity of the c-Ha-ras oncogene. *Nature* 334: 119-124, 1988.

[42382] 15934. Collins, T.; Ginsburg, D.; Boss, J. M.; Orkin, S. H.; Pober, J. S.: Cultured human endothelial cells express platelet-derived growth factor B chain: cDNA cloning and structural analysis. *Nature* 316:748-750, 1985.

[42383] 15935. Dalla-Favera, R.; Gallo, R. C.; Giallongo, A.; Croce,

C.: Chromosomal localization of the human homolog (c-sis) of the simian sarcoma virus onc gene. Science 218: 686–688, 1982.

[42384] 15936. Dalla-Favera, R.; Gelmann, E. P.; Gallo, R. C.; Wong-Staal, F.: A human onc gene homologous to the transforming gene (v-sis) of simian sarcoma virus. Nature 292: 31–35, 1981.

[42385] 15937. Deuel, T. F.; Huang, J. S.; Huang, S. S.; Stroobant, P.; Waterfield, M. D.: Expression of a platelet-derived growth factor-like protein in simian sarcoma virus transformed cells. Science 221: 1348–1350, 1983.

[42386] 15938. Devare, S. G.; Reddy, E. P.; Law, J. D.; Robbins, K. C.; Aaronson, S. A.: Nucleotide sequence of the simian sarcoma virus genome: demonstration that its acquired cellular sequences encode the transforming gene product p28-sis. Proc. Nat. Acad. Sci. 80: 731–735, 1983.

[42387] 15939. Charlet-B., N.; Logan, P.; Singh, G.; Cooper, T. A.: Dynamic antagonism between ETR-3 and PTB regulates cell type-specific alternative splicing. Molec. Cell 9: 649–658, 2002.

[42388] 15940. Borlum, A. D.; Flint, T.; Tommerup, N.; Fleckner, J.; Justesen, J.; Kruse, T. A.: Assignment of the human tryptophanyl-tRNA synthetase gene (WARS) to chromo-

some 14q32.2–q32.32. *Cytogenet. Cell Genet.* 73:99–103, 1996.

[42389] 15941.Denney, R. M.; Borgaonkar, D.; Ruddle, F. H.: Order of genes for NP and TRPRS on chromosome 14. *Cytogenet. Cell Genet.* 22: 493–497, 1978.

[42390] 15942.Francke, U.; Denney, R. M.; Ruddle, F. H.: Intra-chromosomal genemapping in man: the gene for tryptophanyl-tRNA synthetase maps in region q21–qter of chromosome 14. *Somat. Cell Genet.* 3: 381–389, 1977.

[42391] 15943.Benson, K. F.; Horwitz, M.; Wolff, J.; Friend, K.; Thompson, E.; White, S.; Richards, R. I.; Raskind, W. H.; Bird, T. D.: CAG repeat expansion in autosomal dominant familial spastic paraparesis: novel expansion in a subset of patients. *Hum. Molec. Genet.* 7: 1779–1786, 1998.

[42392] 15944.Fonknechten, N.; Mavel, D.; Byrne, P.; Davoine, C.–S.; Cruaud, C.; Boentsch, D.; Samson, D.; Coutinho, P.; Hutchinson, M.; McMonagle, P.; Burgunder, J.–M.; Tartaglione, A.; and 10 others: Spectrum of SPG4 mutations in autosomal dominant spastic paraplegia. *Hum. Molec. Genet.* 9: 637–644, 2000.

[42393] 15945.Nielsen, J. E.; Koefoed, P.; Abell, K.; Hasholt, L.; Eiberg, H.; Fenger, K.; Niebuhr, E.; Sorensen, S. A.: CAG repeat expansion in autosomal dominant pure spastic para–

plegia linked to chromosome 2p21–p24. Hum.Molec. Genet. 6: 1811–1816, 1997.

- [42394] 15946.Svenson, I. K.; Ashley–Koch, A. E.; Gaskell, P. C.; Riney, T.J.; Cumming, W. J. K.; Kingston, H. M.; Hogan, E. L.; Boustany, R.–M.N.; Vance, J. M.; Nance, M. A.; Pericak–Vance, M. A.; Marchuk, D.A.: Identification and expression analysis of spastin gene mutations in hereditary spastic paraplegia. Am. J. Hum. Genet. 68: 1077–1085, 2001.
- [42395] 15947.Ala–Kapee, M.; Nevanlinna, H.; Mali, M.; Jalkanen, M.; Schroder, J.: Localization of gene for human syndecan, an integral membrane proteoglycan and a matrix receptor, to chromosome 2. Somat. Cell Molec. Genet. 16: 501–505, 1990.
- [42396] 15948.Alexander, C. M.; Reichsman, F.; Hinkes, M. T.; Lincecum, J.; Becker, K. A.; Cumberledge, S.; Bernfield, M.: Syndecan–1 is required for Wnt–1–induced mammary tumorigenesis in mice. Nature Genet. 25: 329–332, 2000.
- [42397] 15949.Mali, M.; Jaakkola, P.; Arvilommi, A.–M.; Jalkanen, M.: Sequence of human syndecan indicates a novel gene family of integral membrane proteoglycans. J. Biol. Chem. 265: 6884–6889, 1990.
- [42398] 15950.Oettinger, H. F.; Streeter, H.; Lose, E.; Copeland, N. G.; Gilbert, D. J.; Justice, M. J.; Jenkins, N. A.; Mohandas, T.;

Bernfield, M.: Chromosome mapping of the murine syndecan gene. *Genomics* 11: 334–338,1991.

- [42399] 15951.Reizes, O.; Lincecum, J.; Wang, Z.; Goldberger, O.; Huang, L.;Kaksonen, M.; Ahima, R.; Hinkes, M. T.; Barsh, G. S.; Rauvala, H.;Bernfield, M.: Transgenic expression of syndecan–1 uncovers a physiological control of feeding behavior by syndecan–3. *Cell* 106: 105–116, 2001.
- [42400] 15952.Sanderson, R. D.; Lalor, P.; Bernfield, M.: B lymphocytes express and lose syndecan at specific stages of differentiation. *Cell Regulation* 1:27–35, 1989.
- [42401] 15953.Westman, P.; Hsieh, C.–L.; Mali, M.; Jalkanen, M.; Francke, U.;Schroder, J.: Assignment of the human syndecan (SDC) gene to shortarm of chromosome 2. (Abstract) *Cytogenet. Cell Genet.* 58: 1873–1874,1991.
- [42402] 15954.Asch, A. S.; Barnwell, J.; Silverstein, R. L.; Nachman, R. L.:Isolation of the thrombospondin membrane receptor. *J. Clin. Invest.* 79:1054–1061, 1987.
- [42403] 15955.de Fraipont, F.; El Atifi, M.; Gicquel, C.; Bertagna, X.; Chambaz,E. M.; Feige, J. J.: Expression of the angiogenesis markers vascular endothelial growth factor–A, thrombospondin–1, and platelet–derived endothelial cell growth factor in human sporadic adrenocortical tumors: correlation with genotypic alterations. *J. Clin. En–*

docr. Metab. 85:4734–4741, 2000.

- [42404] 15956.Dixit, V. M.; Hennessy, S. W.; Grant, G. A.; Rotwein, P.; Frazier,W. A.: Characterization of a cDNA encoding the heparin and collagenbinding domains of human thrombospondin. Proc. Nat. Acad. Sci. 83:5449–5453, 1986.
- [42405] 15957.Frazier, W. A.: Thrombospondin: a modular adhesive glycoproteinof platelets and nucleated cells. J. Cell Biol. 105: 625–632, 1987.
- [42406] 15958.Jaffe, E.; Bornstein, P.; Disteché, C. M.: Mapping of the thrombospondingene to human chromosome 15 and mouse chromosome 2 by in situ hybridization. Genomics 7:123–126, 1990.
- [42407] 15959.Lawler, J.; Sunday, M.; Thibert, V.; Duquette, M.; George, E. L.;Rayburn, H.; Hynes, R. O.: Thrombospondin–1 is required for normalmurine pulmonary homeostasis and its absence causes pneumonia. J.Clin. Invest. 101: 982–992, 1998.
- [42408] 15960.Rodriguez–Manzanéque, J. C.; Lane, T. F.; Ortega, M. A.; Hynes,R. O.; Lawler, J.; Iruela–Arispe, M. L.: Thrombospondin–1 suppressesspontaneous tumor growth and inhibits activation of matrix metalloproteinase–9and mobilization of vascular endothelial growth factor. Proc. Nat.Acad. Sci. 98: 12485–12490, 2001.

- [42409] 15961.Wolf, F. W.; Eddy, R. L.; Shows, T. B.; Dixit, V. M.: Structureand chromosomal localization of the human thrombospondin gene. *Genomics* 6:685–691, 1990.
- [42410] 15962.Imai, K.; Sarker, A. H.; Akiyama, K.; Ikeda, S.; Yao, M.; Tsutsui,K.; Shohmori, T.; Seki, S.: Genomic structure and sequence of a humanhomologue (NTHL1/NTH1) of *Escherichia coli* endonuclease III with thoseof the adjacent parts of TSC2 and SLC9A3R2 genes. *Gene* 222: 287–295,1998.
- [42411] 15963.Mira, J.–P.; Cariou, A.; Grall, F.; Delclaux, C.; Losser, M.–R.;Heshmati, F.; Cheval, C.; Monchi, M.; Teboul, J.–L.; Riche, F.; Leleu,G.; Arbibe, L.; Mignon, A.; Delpech, M.; Dhainaut, J.–F.: Associationof TNF2, a TNF–alpha promoter polymorphism, with septic shock suscep–tibilityand mortality: a multicenter study. *J.A.M.A.* 282: 561–568, 1999.
- [42412] 15964.Moffatt, M. F.; Cookson, W. O. C. M.: Tumour necrosis factorhaplotypes and asthma. *Hum. Molec. Genet.* 6: 551–554, 1997.
- [42413] 15965.Moraes, M. O.; Duppre, N. C.; Suffys, P. N.; Santos, A. R.; Almeida,A. S.; Nery, J. A. C.; Sampaio, E. P.; Sarno, E. N.: Tumor necrosisfactor–alpha promoter polymorphism TNF2 is associated with a strongerdelayed–type hypersen–

sitivity reaction in the skin of borderline tuberculoi-
dleprosy patients. Immunogenetics 53: 45–47, 2001.

- [42414] 15966. Mulcahy, B.; Waldron-Lynch, F.; McDermott, M. F.; Adams, C.; Amos, C. I.; Zhu, D. K.; Ward, R. H.; Clegg, D. O.; Shanahan, F.; Molloy, M. G.; O'Gara, F.: Genetic variability in the tumor necrosis factor–lymphotoxin region influences susceptibility to rheumatoid arthritis. Am. J. Hum. Genet. 59: 676–683, 1996.
- [42415] 15967. Muller, U.; Jongeneel, C. V.; Nedospasov, S. A.; Lindahl, K. F.; Steinmetz, M.: Tumour necrosis factor and lymphotoxin genes map close to H-2D in the mouse major histocompatibility complex. Nature 325: 265–267, 1987.
- [42416] 15968. Nadel, S.; Newport, M. J.; Booy, R.; Levin, M.: Variation in the tumor necrosis factor–alpha gene promoter region may be associated with death from meningococcal disease. J. Infect. Dis. 174: 878–880, 1996.
- [42417] 15969. Nedospasov, S. A.; Hirt, B.; Shakhov, A. N.; Dobrynin, V. N.; Kawashima, E.; Accolla, R. S.; Jongeneel, C. V.: The genes for tumor necrosis factor (TNF–alpha) and lymphotoxin (TNF–beta) are tandemly arranged on chromosome 17 of the mouse. Nucleic Acids Res. 14: 7713–7725, 1986.
- [42418] 15970. Nedwin, G. E.; Naylor, S. L.; Sakaguchi, A. Y.; Smith,

D.; Jarrett-Nedwin, J.; Pennica, D.; Goeddel, D. V.; Gray, P. W.: Human lymphotoxin and tumor necrosis factor genes: structure, homology and chromosomal localization. *Nucleic Acids Res.* 13: 6361–6373, 1985.

[42419] 15971. Norman, R. A.; Bogardus, C.; Ravussin, E.: Linkage between obesity and a marker near the tumor necrosis factor- α locus in Pima Indians. *J. Clin. Invest.* 96: 158–162, 1995.

[42420] 15972. Obayashi, H.; Hasegawa, G.; Fukui, M.; Kamiuchi, K.; Kitamura, A.; Ogata, M.; Kanatsuka, T.; Shigeta, H.; Kitagawa, Y.; Nakano, K.; Nishimura, M.; Ohta, M.; Nakamura, N.: Tumor necrosis factor microsatellite polymorphism influences the development of insulin dependency in adult-onset diabetes patients with the DRB1*1502–DQB1*0601 allele and anti-glutamic acid decarboxylase antibodies. *J. Clin. Endocr. Metab.* 85: 3348–3351, 2000.

[42421] 15973. Old, L. J.: Tumor necrosis factor (TNF). *Science* 230: 630–632, 1985.

[42422] 15974. Pennica, D.; Nedwin, G. E.; Hayflick, J. S.; Seeburg, P. H.; Derynck, R.; Palladino, M. A.; Kohr, W. J.; Aggarwal, B. B.; Goeddel, D. V.: Human tumour necrosis factor: precursor structure, expression and homology to lympho-

toxin. *Nature* 312: 724–729, 1984.

- [42423] 15975. Ragoussis, J.; Bloemer, K.; Weiss, E. H.; Ziegler, A.: Localization of the genes for tumor necrosis factor and lymphotoxin between the HLA class I and III regions by field inversion gel electrophoresis. *Immunogenetics* 27:66–69, 1988.
- [42424] 15976. Rasmussen, S. K.; Urhammer, S. A.; Jensen, J. N.; Hansen, T.; Borch-Johnsen, K.; Pedersen, O.: The –238 and –308 G6A polymorphisms of the tumor necrosis factor alpha gene promoter are not associated with features of the insulin resistance syndrome or altered birthweight in Danish Caucasians. *J. Clin. Endocr. Metab.* 85: 1731–1734, 2000.
- [42425] 15977. Rosmond, R.; Chagnon, M.; Bouchard, C.; Bjorn-torp, P.: G–308 A polymorphism of the tumor necrosis factor alpha gene promoter and salivary cortisol secretion. *J. Clin. Endocr. Metab.* 86: 2178–2180, 2001.
- [42426] 15978. Roy, S.; McGuire, W.; Mascie-Taylor, C. G.; Saha, B.; Hazra, S. K.; Hill, A. V.; Kwiatkowski, D.: Tumor necrosis factor promoter polymorphism and susceptibility to lepromatous leprosy. *J. Infect. Dis.* 176: 530–532, 1997.
- [42427] 15979. Ruuls, S. R.; Sedgwick, J. D.: Unlinking tumor necrosis factor biology from the major histocompatibility

complex: lessons from humangenetics and animal models. Am. J. Hum. Genet. 65: 294–301, 1999.

[42428] 15980.Spies, T.; Morton, C. C.; Nedospasov, S. A.; Fiers, W.; Pious,D.; Strominger, J. L.: Genes for the tumor necrosis factors alphaand beta are linked to the human major histocompatibility complex. Proc.Nat. Acad. Sci. 83: 8699–8702, 1986.

[42429] 15981.van Hensbroek, M. B.; Palmer, A.; Onyiorah, E.; Schneider, G.;Jaffar, S.; Dolan, G.; Memming, H.; Frenkel, J.; Enwere, G.; Bennett,S.; Kwiatkowski, D.; Greenwood, B.: The effect of a monoclonal antibodyto tumor necrosis factor on survival from childhood cerebral malaria. J.Infect. Dis. 174: 1091–1097, 1996.

[42430] 15982.Sashio, H.; Tamura, K.; Ito, R.; Yamamoto, Y.; Bamba, H.; Kosaka,T.; Fukui, S.; Sawada, K.; Fukuda, Y.; Tamura, K.; Satomi, M.; Shimoyama,T.; Furuyama, J.: Polymorphisms of the TNF gene and the TNF receptorsuperfamily member 1B gene are associated with susceptibility to ulcerativecolitis and Crohn's disease, respectively. Immunogenetics 53: 1020–1027,2002.

[42431] 15983.Van Ostade, X.; Vandenabeele, P.; Everaerd, B.; Loetscher, H.;Gentz, R.; Brockhaus, M.; Lesslauer, W.; Tavernier, J.; Brouckaert,P.; Fiers, W.: Human TNF mutants

with selective activity on the p55receptor. *Nature* 361: 266–269, 1993.

- [42432] 15984.Wang, A. M.; Creasey, A. A.; Ladner, M. B.; Lin, L. S.; Strickler,J.; Van Arsdell, J. N.; Yamamoto, R.; Mark, D. F.: Molecular cloningof the complementary DNA for human tumor necrosis factor. *Science* 228:149–154, 1985.
- [42433] 15985.Wilson, A. G.; Symons, J. A.; McDowell, T. L.; et al: Effectsof a polymorphism in the human tumor necrosis factor alpha promoter on transcriptional activation. *Proc. Nat. Acad. Sci.* 94: 3195–3199,1997.
- [42434] 15986.Winchester, E. C.; Millwood, I. Y.; Rand, L.; Penny, M. A.; Kessling,A. M.: Association of the TNF–alpha–308 (G–A) polymorphism with self–reportedhistory of childhood asthma. *Hum. Genet.* 107: 591–596, 2000.
- [42435] 15987.Witte, J. S.; Palmer, L. J.; O'Connor, R. D.; Hopkins, P. J.;Hall, J. M.: Relation between tumour necrosis factor polymorphismTNF–alpha–308 and risk of asthma. *Europ. J. Hum. Genet.* 10: 82–85,2002.
- [42436] 15988.Peter, D.; Finn, J. P.; Klisak, I.; Liu, Y.; Kojis, T.; Heinzmann,C.; Roghani, A.; Sparkes, R. S.; Edwards, R. H.: Chromosomal localizationof the human vesicular amine transporter genes. *Genomics* 18: 720–723,1993.
- [42437] 15989.Roghani, A.; Welch, C.; Xia, Y.–R.; Liu, Y.; Peter, D.;

Finn, J.P.; Edwards, R. H.; Lusk, A. J.: Assignment of the mouse vesicular monoamine transporter genes, Slc18a1 and Slc18a2, to chromosomes 8 and 19 by linkage analysis. *Mammalian Genome* 7: 393–394, 1996.

[42438] 1990. Lania, L.; Dotti, E.; Pannuti, A.; Pascucci, A.; Pengue, G.; Feliciello, I.; La Mantia, G.; Lanfranccone, L.; Pelicci, P.-G.: cDNA isolation, expression analysis, and chromosomal localization of two human zinc finger genes. *Genomics* 6: 333–340, 1990.

[42439] 1991. Thiesen, H.-J.: Multiple genes encoding zinc finger domains are expressed in human T cells. *New Biologist* 2: 363–374, 1990.

[42440] 1992. Seite, P.; Huebner, K.; Rousseau-Merck, M. F.; Berger, R.; Thiesen, H. J.: Two human genes encoding zinc finger proteins, ZNF12 (KOX3) and ZNF26 (KOX20), map to chromosomes 7p22–p21 and 12q24.33, respectively. *Hum. Genet.* 86: 585–590, 1991.

[42441] 1993. Gaynor, R. B.; Muchardt, C.; Diep, A.; Mohandas, T. K.; Sparkes, R. S.; Lusk, A. J.: Localization of the zinc finger DNA-binding protein HIV-EP1/MBP-1/PRDII-BF1 to human chromosome 6p22.3–p24. *Genomics* 9: 758–761, 1991.

[42442] 1994. Tripathi, R. K.; Droetto, S.; Spritz, R. A.: Many pa-

tients with 'tyrosinase-positive' oculocutaneous albinism have tyrosinase gene mutations. (Abstract) Am. J. Hum. Genet. 51 (suppl.): A179, 1992.

- [42443] 15995.Brilliant, M. H.: The mouse pink-eyed dilution locus: a model for aspects of Prader-Willi syndrome, Angelman syndrome, and a form of hypomelanosis of Ito. Mammalian Genome 3: 187-191, 1992.
- [42444] 15996.Castle, D.; Kromberg, J.; Kowalsky, R.; Moosa, R.; Gillman, N.; Zwane, E.; Fritz, V.: Visual evoked potentials in Negro carriers of the gene for tyrosinase positive oculocutaneous albinism. J. Med. Genet. 25: 835-837, 1988.
- [42445] 15997.Colman, M. A.; Stevens, G.; Ramsay, M.; Kwon, B.; Jenkins, T.: Exclusion of two candidate pigment loci, c and b, part of chromosome 11p, and 33 random polymorphic markers as the locus for tyrosinase-positive oculocutaneous albinism. Hum. Genet. 90: 556-560, 1993.
- [42446] 15998.Donlon, T. A.; Lalande, M.; Wyman, A.; Bruns, G.; Latt, S. A.: Isolation of molecular probes associated with the chromosome 15 instability in the Prader-Willi syndrome. Proc. Nat. Acad. Sci. 83: 4408-4412, 1986.
- [42447] 15999.Durham-Pierre, D.; Gardner, J. M.; Nakatsu, Y.; King, R. A.; Francke, U.; Ching, A.; Aquaron, R.; del Mar-

mol, V.; Brilliant, M. H.: African origin of an intragenic deletion of the human P gene in tyrosinase positive oculocutaneous albinism. *Nature Genet.* 7: 176–179, 1994.

[42448] 16000. Durham–Pierre, D.; King, R. A.; Naber, J. M.; Laken, S.; Brilliant, M. H.: Estimation of carrier frequency of a 2.7 kb deletion allele of the P gene associated with OCA2 in African–Americans. *Hum. Mutat.* 7: 370–373, 1996.

[42449] 16001. Gardner, J. M.; Nakatsu, Y.; Gondo, Y.; Lee, S.; Lyon, M. F.; King, R. A.; Brilliant, M. H.: The mouse pink-eyed dilution gene: association with human Prader–Willi and Angelman syndromes. *Science* 257: 1121–1124, 1992.

[42450] 16002. Gondo, Y.; Gardner, J. M.; Nakatsu, Y.; Durham–Pierre, D.; Deveau, S. A.; Kuper, C.; Brilliant, M. H.: High-frequency genetic reversion mediated by a DNA duplication: the mouse pink-eyed unstable mutation. *Proc. Nat. Acad. Sci.* 90: 297–301, 1993.

[42451] 16003. Hagiwara, N.; Klewer, S. E.; Samson, R. A.; Erickson, D. T.; Lyon, M. F.; Brilliant, M. H.: Sox6 is a candidate gene for p100H myopathy, heart block, and sudden neonatal death. *Proc. Nat. Acad. Sci.* 97: 4180–4185, 2000.

[42452] 16004. Hamabe, J.; Fukushima, Y.; Harada, N.; Abe, K.; Matsuo, N.; Nagai, T.; Yoshioka, A.; Tonoki, H.; Tsukino, R.; Niikawa, N.: Molecular study of the Prader–Willi syn–

drome: deletion, RFLP, and phenotype analyses of 50 patients. *Am. J. Med. Genet.* 41: 54–63, 1991.

[42453] 16005. Heim, R. A.; Dunn, D. S.; Candy, S. E.; Zwane, E.; Kromberg, J.G. R.; Jenkins, T.: The tyrosine-positive oculocutaneous albinism locus is not linked to the beta-globin locus in man. *Hum. Genet.* 79:89, 1988.

[42454] 16006. Kagore, F.; Lund, P. M.: Oculocutaneous albinism among schoolchildren in Harare, Zimbabwe. *J. Med. Genet.* 32: 859–861, 1995.

[42455] 16007. Kedda, M. A.; Stevens, G.; Manga, P.; Viljoen, C.; Jenkins, T.; Ramsay, M.: The tyrosinase-positive oculocutaneous albinism gene shows locus homogeneity on chromosome 15q11–q13 and evidence of multiple mutations in Southern African negroids. *Am. J. Hum. Genet.* 54: 1078–1084, 1994.

[42456] 16008. Kerr, R.; Stevens, G.; Manga, P.; Salm, S.; John, P.; Haw, T.; Ramsay, M.: Identification of P gene mutations in individuals with oculocutaneous albinism in sub-Saharan Africa. *Hum. Mutat.* 15: 166–172, 2000.

[42457] 16009. King, R. A.: Personal Communication. Minneapolis, Minn. 12/31/1992.

[42458] 16010. Ansari-Lari, M. A.; Muzny, D. M.; Lu, J.; Lu, F.; Lilley, C. E.; Spanos, S.; Malley, T.; Gibbs, R. A.: A gene-rich

cluster between the CD4 and triosephosphate isomerase genes at human chromosome 12p13. *Genome Res.* 6: 314–326, 1996.

[42459] 16011. Bach, M. A.; Phan-Dinh-Tuy, F.; Bach, J. F.; Wallach, D.; Biddison, W. E.; Sharrow, S. O.; Goldstein, G.; Kung, P. C.: Unusual phenotypes of human inducer T cells as measured by OKT4 and related monoclonal antibodies. *J. Immun.* 127: 980–982, 1981.

[42460] 16012. Browning, J.; Horner, J. W.; Pettoello-Mantovani, M.; Raker, C.; Yurasov, S.; DePinho, R. A.; Goldstein, H.: Mice transgenic for human CD4 and CCR5 are susceptible to HIV infection. *Proc. Nat. Acad. Sci.* 94:14637–14641, 1997.

[42461] 16013. Buttini, M.; Westland, C. E.; Masliah, E.; Yafeh, A. M.; Wyss-Coray, T.; Mucke, L.: Novel role of human CD4 molecule identified in neurodegeneration. *Nature Med.* 4: 441–446, 1998.

[42462] 16014. Committee on Human Leukocyte Differentiation Antigens, IUIS WHO Nomenclature Subcommittee: Proposed nomenclature for human leukocyte differentiation antigens. *Bull. WHO* 5: 809–811, 1984. Note: *Alternate: Immunology Today* 5: 280 only, 1984.

[42463] 16015. Fukuda, T.; Matsunaga, M.; Kurata, A.; Mine, M.;

Ikari, N.; Katamine, S.; Kanazawa, H.; Eguchi, K.; Nagataki, S.: Hereditary deficiency of OKT4-positive cells: studies for mode of inheritance and lymphocyte functions. *Immunology* 53: 643–649, 1984.

[42464] 16016. Fuller, T. C.; Trevithick, J. E.; Fuller, A. A.; Colvin, R. B.; Cosimi, A. B.; Kung, P. C.: Antigenic polymorphism of the T4 differentiation antigen expressed on human T helper/inducer lymphocytes. *Hum. Immun.* 9:89–102, 1984.

[42465] 16017. Gill, J. C.; Maples, J.; Nikaein, A.; Kirchner, P.; Lockhart, D.; Snyder, A. J.; Montgomery, R. R.; Casper, J. T.: Inherited absence of OKT4 lymphocyte antigen in a chronically transfused patient with homozygous sickle cell disease. *J. Pediat.* 107: 251–253, 1985.

[42466] 16018. Irvine, D. J.; Purbhoo, M. A.; Krosgaard, M.; Davis, M. M.: Direct observation of ligand recognition by T cells. *Nature* 419:845–849, 2002.

[42467] 16019. Isobe, M.; Huebner, K.; Maddon, P. J.; Littman, D. R.; Axel, R.; Croce, C. M.: The gene encoding the T-cell surface protein T4 is located on human chromosome 12. *Proc. Nat. Acad. Sci.* 83: 4399–4402, 1986.

[42468] 16020. Karol, R. A.; Eng, J.; Dennison, D. K.; Faris, E.; Marcus, D. M.: Hereditary abnormalities of the OKT4 human

lymphocyte epitope in two families. *J. Clin. Immun.* 4: 71–74, 1984.

[42469] 16021. Kozbor, D.; Finan, J.; Nowell, P. C.; Croce, C. M.: The gene encoding the T4 antigen maps to human chromosome 12. *J. Immun.* 136:1141–1143, 1986.

[42470] 16022. Maddon, P. J.; Molineaux, S. M.; Maddon, D. E.; Zimmerman, K. A.; Godfrey, M.; Alt, F. W.; Chess, L.; Axel, R.: Structure and expression of the human and mouse T4 genes. *Proc. Nat. Acad. Sci.* 84: 9155–9159, 1987.

[42471] 16023. Piguet, V.; Gu, F.; Foti, M.; Demarex, N.; Gruenberg, J.; Carpentier, J.-L.; Trono, D.: Nef-induced CD4 degradation: a diacidic-based motif in Nef functions as a lysosomal targeting signal through the binding of beta-COP in endosomes. *Cell* 97: 63–73, 1999.

[42472] 16024. Sato, M.; Hayashi, Y.; Yoshida, H.; Yanagawa, T.; Yura, Y.: A family with hereditary lack of T4⁺ inducer/helper T cell subsets in peripheral blood lymphocytes. *J. Immun.* 132: 1071–1073, 1984.

[42473] 16025. Sawada, S.; Scarborough, J. D.; Killeen, N.; Littman, D. R.: A lineage-specific transcriptional silencer regulates CD4 gene expression during T lymphocyte development. *Cell* 77: 917–929, 1994.

[42474] 16026. Zinman, B.; Hanley, A. J. G.; Harris, S. B.; Kwan, J.;

Fantus, I. G.: Circulating tumor necrosis factor- α concentrations in a Native Canadian population with high rates of type 2 diabetes mellitus. *J. Clin. Endocr. Metab.* 84: 272–278, 1999.

[42475] 16027. Spurr, N. K.; Goodfellow, P. N.; Sheer, D.; Bodmer, W. F.; Vennstrom, B.: Mapping of cellular oncogenes: ERBA1 is on chromosome 17. (Abstract) *Cytogenet. Cell Genet.* 37: 591 only, 1984.

[42476] 16028. Thompson, C. C.; Weinberger, C.; Lebo, R.; Evans, R. M.: Identification of a novel thyroid hormone receptor expressed in the mammalian central nervous system. *Science* 237: 1610–1614, 1987.

[42477] 16029. Weinberger, C.; Thompson, C. C.; Ong, E. S.; Lebo, R.; Gruol, D. J.; Evans, R. M.: The c-erb-A gene encodes a thyroid hormone receptor. *Nature* 324: 641–646, 1986.

[42478] 16030. Zabel, B. U.; Fournier, R. E. K.; Lalley, P. A.; Naylor, S. L.; Sakaguchi, A. Y.: Cellular homologs of the avian erythroblastosis virus erb-A and erb-B genes are syntenic in mouse but asyntenic in man. *Proc. Nat. Acad. Sci.* 81: 4874–4878, 1984.

[42479] 16031. Carraway, K. L., III; Sliwkowski, M. X.; Akita, R.; Platko, J. V.; Guy, P. M.; Nuijens, A.; Diamonti, A. J.; Vanden, R. L.; Cantley, L. C.; Cerione, R. A.: The erbB3 gene

product is a receptor for heregulin. J.Biol. Chem. 269: 14303–14306, 1994.

[42480] 16032.Cho, H.-S.; Leahy, D. J.: Structure of the extracellular region of HER3 reveals an interdomain tether. Science 297: 1330–1333, 2002.

[42481] 16033.Kraus, M. H.; Issing, W.; Miki, T.; Popescu, N. C.; Aaronson, S.A.: Isolation and characterization of ERBB3, a third member of the ERBB/epidermal growth factor receptor family: evidence for overexpression in a subset of human mammary tumors. Proc. Nat. Acad. Sci. 86: 9193–9197, 1989.

[42482] 16034.Plowman, G. D.; Whitney, G. S.; Neubauer, M. G.; Green, J. M.; McDonald, V. L.; Todaro, G. J.; Shoyab, M.: Molecular cloning and expression of an additional epidermal growth factor receptor-related gene. Proc. Nat. Acad. Sci. 87: 4905–4909, 1990.

[42483] 16035.Elchebly, M.; Payette, P.; Michaliszyn, E.; Cromlish, W.; Collins, S.; Loy, A. L.; Normandin, D.; Cheng, A.; Himms-Hagen, J.; Chan, C.-C.; Ramachandran, C.; Gresser, M. J.; Tremblay, M. L.; Kennedy, B. P.: Increased insulin sensitivity and obesity resistance in mice lacking the protein tyrosine phosphatase-1B gene. Science 283: 1544–1548, 1999.

- [42484] 16036.Forsell, P. K. A. L.; Boie, Y.; Montalibet, J.; Collins, S.; Kennedy, B. P.: Genomic characterization of the human and mouse protein tyrosinephosphatase-1B genes. *Gene* 260: 145-153, 2000.
- [42485] 16037.Gu, H. F.; Almgren, P.; Lindholm, E.; Frittitta, L.; Pizzuti, A.;Trischitta, V.; Groop, L. C.: Association between the human glycoproteinPC-1 gene and elevated glucose and insulin levels in paired-siblinganalysis. *Diabetes* 49: 1601-1603, 2000.
- [42486] 16038.Haj, F. G.; Verveer, P. J.; Squire, A.; Neel, B. G.; Bastiaens, P. I. H.: Imaging sites of receptor dephosphorylation by PTP1B onthe surface of the endoplasmic reticulum. *Science* 295: 1708-1711,2002.
- [42487] 16039.Jia, Z.; Barford, D.; Flint, A. J.; Tonks, N. K.: Structuralbasis for phosphotyrosine peptide recognition by protein tyrosinephosphatase 1B. *Science* 268: 1754-1758, 1995.
- [42488] 16040.Kennedy, B. P.; Ramachandran, C.: Protein tyrosine phosphatase-1Bin diabetes. *Biochem. Pharm.* 60: 877-883, 2000.
- [42489] 16041.Mok, A.; Cao, H.; Zinman, B.; Hanley, A. J. G.; Harris, S. B.;Kennedy, B. P.; Hegele, R. A.: A single nucleotide polymorphism inprotein tyrosine phosphatase PTP-1B is

associated with protection from diabetes or impaired glucose tolerance in Oji-Cree. *J. Clin. Endocr. Metab.* 87: 724–727, 2002.

[42490] 16042. Tonks, N. K.; Diltz, C. D.; Fischer, E. H.: Purification of the major protein-tyrosine-phosphatases of human placenta. *J. Biol. Chem.* 263:6722–6730, 1988.

[42491] 16043. Barnea, G.; Silvennoinen, O.; Shaanan, B.; Honegger, A. M.; Canoll, P. D.; D'Eustachio, P.; Morse, B.; Levy, J. B.; Laforgia, S.; Huebner, K.; Musacchio, J. M.; Sap, J.; Schlessinger, J.: Identification of a carbonic anhydrase-like domain in the extracellular region of RPTP-gamma defines a new subfamily of receptor tyrosine phosphatases. *Molec. Cell. Biol.* 13: 1497–1506, 1993.

[42492] 16044. Kastury, K.; Ohta, M.; Lasota, J.; Moir, D.; Dorman, T.; LaForgia, S.; Druck, T.; Huebner, K.: Structure of the human receptor tyrosine phosphatase gamma gene (PTPRG) and relation to the familial RCC t(3;8) chromosome translocation. *Genomics* 32: 225–235, 1996.

[42493] 16045. LaForgia, S.; Morse, B.; Levy, J.; Barnea, G.; Cannizzaro, L. A.; Li, F.; Nowell, P. C.; Boghosian-Sell, L.; Glick, J.; Weston, A.; Harris, C. C.; Drabkin, H.; Patterson, D.; Croce, C. M.; Schlessinger, J.; Huebner, K.: Receptor protein-tyrosine phosphatase gamma is a candidate tumor suppressor.

sor gene at human chromosome region 3p21. Proc.Nat. Acad. Sci. 88: 5036–5040, 1991.

- [42494] 16046.Latif, F.; Tory, K.; Modi, W.; Geil, L.; LaForgia, S.; Huebner,K.; Zbar, B.; Lerman, M. I.: A Mspl polymorphism and linkage mapping of the human protein–tyrosine phosphatase G (PTPRG) gene. Hum. Molec.Genet. 2: 91, 1993.
- [42495] 16047.Ariyama, T.; Hasegawa, K.; Inazawa, J.; Mizuno, K.; Ogimoto, M.;Katagiri, T.; Yakura, H.: Assignment of the human protein tyrosinephosphatase, receptor–type, zeta (PTPRZ) gene to chromosome band 7q31.3. Cytogenet.Cell Genet. 70: 52–54, 1995.
- [42496] 16048.Schols, L.; Szymanski, S.; Peters, S.; Przuntek, H.; Epplen, J.T.; Hardt, C.; Riess, O.: Genetic background of apparently idiopathicsporadic cerebellar ataxia. Hum. Genet. 107: 132–137, 2000.
- [42497] 16049.Chan, J. Y.; Cheung, M.–C.; Moi, P.; Chan, K.; Kan, Y. W.: Chromosomallocalization of the human NF–E2 family of bZIP transcription factorsby fluorescence in situ hybridization. Hum. Genet. 95: 265–269,1995.
- [42498] 16050.Chan, J. Y.; Han, X.–L.; Kan, Y. W.: Cloning of Nrf1, an NF–E2–relatedtranscription factor, by genetic selection in yeast. Proc. Nat. Acad.Sci. 90: 11371–11375, 1993.
- [42499] 16051.Chan, J. Y.; Kwong, M.; Lu, R.; Chang, J.; Wang, B.;

Yen, T. S.B.; Kan, Y. W.: Targeted disruption of the ubiquitous CNC-bZIP transcriptionfactor, Nrf-1, results in anemia and embryonic lethality in mice. EMBOJ. 17: 1779-1787, 1998.

[42500] 16052.Luna, L.; Johnsen, O.; Skartlien, A.; Pedeutour, F.; Turc-Carel,C.; Prydz, H.; Kolsto, A.-B.: Molecular cloning of a putative novelhuman bZIP transcription factor on chromosome 17q22. Genomics 22:553-562, 1994.

[42501] 16053.Luna, L.; Johnsen, O.; Skartlien, A. H.; Pedeutour, F.; Turc-Carel,C.; Prydz, H.; Kolsto, A.-B.: Molecular cloning of a putative novelhuman bZIP transcription factor on chromosome 17q22. Genomics 22:553-562, 1994.

[42502] 16054.McKie, J.; Johnstone, K.; Mattei, M.-G.; Scambler, P.: Cloningand mapping of murine Nfe2l1. Genomics 25: 716-719, 1995.

[42503] 16055.McKie, J.; Scambler, P. J.: The Nfe2l1 gene maps to distal mousechromosome 11. Mammalian Genome 7: 89-90, 1996.

[42504] 16056.Kas, K.; Wlodarska, I.; Meyen, E.; Van den Berghe, H.; Van de Ven,W. J. M.: Assignment of the gene encoding human Kruppel-related zincfinger protein 4 (GLI4) to 8q24.3 by fluorescent in situ hybridization. Cytogenet.Cell Genet. 72: 297-298, 1996.

- [42505] 16057. Goldblum, S. E.; Ding, X.; Funk, S. E.; Sage, E. H.: SPARC (secreted protein acidic and rich in cysteine) regulates endothelial cell shape and barrier function. *Proc. Nat. Acad. Sci.* 91: 3448–3452, 1994.
- [42506] 16058. Lossie, A. C.; Gordon, D. F.; Camper, S. A.: Localization of thyrotropin-releasing hormone receptor and thyrotroph embryonic factor on mouse chromosome 15. *Mammalian Genome* 4: 621–623, 1993.
- [42507] 16059. Matre, V.; Karlsen, H. E.; Wright, M. S.; Lundell, I.; Fjeldheim, A. K.; Gabrielsen, O. S.; Larhammar, D.; Gautvik, K. M.: Molecular cloning of a functional human thyrotropin-releasing hormone receptor. *Biochem. Biophys. Res. Commun.* 195: 179–185, 1993.
- [42508] 16060. Morrison, N.; Duthie, S. M.; Boyd, E.; Eidne, K. A.; Connor, J. M.: Assignment of the gene encoding the human thyrotropin-releasing hormone receptor to 8q23 by fluorescence in situ hybridization. *Hum. Genet.* 93: 716–718, 1994.
- [42509] 16061. Straub, R. E.; Frech, G. C.; Joho, R. H.; Gershengorn, M. C.: Expression cloning of a cDNA encoding the mouse pituitary thyrotropin-releasing hormone receptor. *Proc. Nat. Acad. Sci.* 87: 9514–9518, 1990.
- [42510] 16062. Yamada, M.; Monden, T.; Konaka, S.; Mori, M.: As-

signment of human thyrotropin-releasing hormone (TRH) receptor gene to chromosome 8. *Somat. Cell Molec. Genet.* 19: 577–580, 1993.

[42511] 16063. Zhao, D.; Yang, J.; Jones, K. E.; Gerald, C.; Suzuki, Y.; Hogan, P. G.; Chin, W. W.; Tashjian, A. H., Jr.: Molecular cloning of a complementary deoxyribonucleic acid encoding the thyrotropin-releasing hormone receptor and regulation of its messenger ribonucleic acid in rat GH cells. *Endocrinology* 130: 3529–3536, 1992.

[42512] 16064. Bongarzone, I.; Butti, M. G.; Coronelli, S.; Borrello, M. G.; Santoro, M.; Mondellini, P.; Pilotti, S.; Fusco, A.; Della Porta, G.; Pierotti, M. A.: Frequent activation of ret protooncogene by fusion with a new activating gene in papillary thyroid carcinomas. *Cancer Res.* 54: 2979–2985, 1994.

[42513] 16065. Bongarzone, I.; Butti, M. G.; Fugazzola, L.; Pacini, F.; Pinchera, A.; Vorontsova, T. V.; Demidchik, E. P.; Pierotti, M. A.: Comparison of the breakpoint regions of ELE1 and RET genes involved in the generation of RET/PTC3 oncogene in sporadic and in radiation-associated papillary thyroid carcinomas. *Genomics* 42: 252–259, 1997.

[42514] 16066. Klein, M.; Vignaud, J.-M.; Hennequin, V.; Toussaint,

B.; Bresler, L.; Plenat, F.; Leclere, J.; Duprez, A.; Weryha, G.: Increased expression of the vascular endothelial growth factor is a pejorative prognosis marker in papillary thyroid carcinoma. *J. Clin. Endocr. Metab.* 86:656–658, 2001.

[42515] 16067. Katyal, S. L.; Singh, G.; Locker, J.: Characterization of a second human pulmonary surfactant-associated protein SP-A gene. *Am. J. Resp. Cell Molec. Biol.* 6: 446–452, 1992.

[42516] 16068. Ramet, M.; Lofgren, J.; Albo, O.-P.; Hallman, M.: Surfactant protein-A gene locus associated with recurrent otitis media. *J. Pediat.* 138:266–268, 2001.

[42517] 16069. Robinson, P. R.; Cohen, G. B.; Zhukovsky, E. A.; Oprian, D. D.: Constitutively active mutants of rhodopsin. *Neuron* 9: 719–725, 1992.

[42518] 16070. Mason, I. J.; Murphy, D.; Munke, M.; Francke, U.; Elliott, R. W.; Hogan, B. L. M.: Developmental and transformation-sensitive expression of the SPARC gene on mouse chromosome 11. *EMBO J.* 5: 1831–1837, 1986.

[42519] 16071. Mason, I. J.; Taylor, A.; Williams, J. G.; Sage, H.; Hogan, B. L. M.: Evidence from molecular cloning that SPARC, a major product of mouse embryo parietal endoderm, is related to an endothelial cell 'culture shock' glycoprotein of Mr 43,000. *EMBO J.* 5: 1465–1472, 1986.

- [42520] 16072.Naylor, S. L.; Helen-Davis, D.; Villarreal, X. C.; Long, G. L.: The human osteonectin gene on chromosome 5 is polymorphic. (Abstract) Cytogenet.Cell Genet. 51: 1051 only, 1989.
- [42521] 16073.Schwartz, R. C.; Young, M. F.; Tsipouras, P.: Two RFLPs in the 5-prime end of the human osteonectin (ON) gene. Nucleic Acids Res. 16:9076 only, 1988.
- [42522] 16074.Stenner, D. D.; Tracy, R. P.; Riggs, B. L.; Mann, K. G.: Human platelets contain and secrete osteonectin, a major protein of mineralized bone. Proc. Nat. Acad. Sci. 83: 6892-6896, 1986.
- [42523] 16075.Swaroop, A.; Francke, U.: Molecular cloning, cDNA sequence, and expression of human SPARC (osteonectin). (Abstract) Am. J. Hum. Genet. 41:A240 only, 1987.
- [42524] 16076.Swaroop, A.; Hogan, B. L. M.; Francke, U.: Molecular analysis of the cDNA for human SPARC/osteonectin/BM-40: sequence, expression, and localization of the gene to chromosome 5q31-q33. Genomics 2:37-47, 1988.
- [42525] 16077.Termin, J. D.; Kleinman, H. K.; Whitson, S. W.; Conn, K. M.; McGarvey, M. L.; Martin, G. R.: Osteonectin, a bone-specific protein linking mineral to collagen. Cell 26: 99-105, 1981.

- [42526] 16078. Blau, N.; Thony, B.; Renneberg, A.; Arnold, L. A.; Hyland, K.: Dihydropteridine reductase deficiency localized to the central nervous system. *J. Inher. Metab. Dis.* 21: 433–434, 1998.
- [42527] 16079. Blau, N.; Thony, B.; Renneberg, A.; Penzien, J. M.; Hyland, K.; Hoffmann, G.: Variant of dihydropteridine reductase deficiency without hyperphenylalaninemia: effect of oral phenylalanine loading. *J. Inher. Metab. Dis.* 22: 216–220, 1999.
- [42528] 16080. Bonafe, L.; Thony, B.; Penzien, J. M.; Czarnecki, B.; Blau, N.: Mutations in the sepiapterin reductase gene cause a novel tetrahydrobiopterin-dependent-monoamine-neurotransmitter deficiency without hyperphenylalaninemia. *Am. J. Hum. Genet.* 69: 269–277, 2001.
- [42529] 16081. Ichinose, H.; Katoh, S.; Sueoka, T.; Titani, K.; Fujita, K.; Nagatsu, T.: Cloning and sequencing of cDNA encoding human sepiapterin reductase: an enzyme involved in tetrahydrobiopterin biosynthesis. *Biochem. Biophys. Res. Commun.* 179: 183–189, 1991.
- [42530] 16082. Murdoch, J. N.; Eddleston, J.; Stanier, P.; Copp, A. J.: Localization of the mouse gene encoding tyrosine kinase receptor type 10 on distal chromosome 1. *Mammalian Genome* 8: 941–952, 1997.

- [42531] 16083. Thony, B.; Heizmann, C. W.; Mattei, M.-G.: Human GTP-cyclohydrolase I gene and sepiapterin reductase gene map to region 14q21-q22 and 2p14-p12, respectively, by *in situ* hybridization. *Genomics* 26: 168-170, 1995.
- [42532] 16084. Jin, H.; Oksenberg, D.; Ashkenazi, A.; Peroutka, S. J.; Duncan, A. M. V.; Rozmahel, R.; Yang, Y.; Mengod, G.; Palacios, J. M.; O'Dowd, B. F.: Characterization of the human 5-hydroxytryptamine (1B) receptor. *J. Biol. Chem.* 267: 5735-5738, 1992.
- [42533] 16085. Levy, F. O.; Gudermann, T.; Birnbaumer, M.; Kaumann, A. J.; Birnbaumer, L.: Molecular cloning of a human gene (S31) encoding a novel serotonin receptor mediating inhibition of adenylyl cyclase. *FEBS Lett.* 296: 201-206, 1992.
- [42534] 16086. Levy, F. O.; Holtgreve-Grez, H.; Tasken, K.; Solberg, R.; Ried, T.; Gudermann, T.: Assignment of the gene encoding the 5-HT_{1E} serotonin receptor (S31) (locus HTR1E) to human chromosome 6q14-q15. *Genomics* 22: 637-640, 1994.
- [42535] 16087. McAllister, G.; Charlesworth, A.; Snodin, C.; Beer, M. S.; Noble, A. J.; Middlemiss, D. N.; Iversen, L. L.; Whiting, P.: Molecular cloning of a serotonin receptor from human brain (5HT_{1E}): a fifth 5HT₁-like subtype. *Proc. Nat.*

Acad. Sci. 89: 5517–5521, 1992.

[42536] 16088. Weinshank, R. L.; Zgombick, J. M.; Macchi, M. J.; Branchek, T.A.; Hartig, P. R.: Human serotonin 1D receptor is encoded by a subfamily of two distinct genes: 5-HT(1D-alpha) and 5-HT(1D-beta). Proc. Nat. Acad. Sci. 89: 3630–3634, 1992.

[42537] 16089. Evans, T.; Reitman, M.; Felsenfeld, G.: An erythrocyte-specific DNA-binding factor recognizes a regulatory sequence common to all chicken globin genes. Proc. Nat. Acad. Sci. 85: 5976–5980, 1988.

[42538] 16090. Chen, Z.-Y.; Battinelli, E. M.; Fielder, A.; Bunday, S.; Sims, K.; Breakefield, X. O.; Craig, I. W.: A mutation in the Norrie disease gene (NDP) associated with X-linked familial exudative vitreoretinopathy. Nature Genet. 5: 180–183, 1993.

[42539] 16091. Dudgeon, J.: Familial exudative vitreo-retinopathy. Trans. Ophthal. Soc. U.K. 99: 45–49, 1979.

[42540] 16092. Yang-Feng, T. L.; DeGennaro, L. J.; Francke, U.: Genes for synapsin I, a neuronal phosphoprotein, map to conserved regions of human and murine X chromosomes. Proc. Nat. Acad. Sci. 83: 8679–8683, 1986.

[42541] 16093. Sinke, R. J.; de Leeuw, B.; Janssen, H. A. P.; Olde Weghuis, D.; Suijkerbuijk, R. F.; Meloni, A. M.;

Gilgenkrantz, S.; Berger, W.; Ropers, H. H.; Sandberg, A. A.; Geurts van Kessel, A.: Localization of X chromosome short arm markers relative to synovial sarcoma–adrenocarcinoma–associated translocation breakpoints. *Hum. Genet.* 92:305–308, 1993.

[42542] 16094. Brennan, T. J.; Seeley, W. W.; Kilgard, M.; Schreiner, C. E.; Tecott, L. H.: Sound-induced seizures in serotonin 5-HT_{2C} receptor mutant mice. *Nature Genet.* 16: 387–390, 1997.

[42543] 16095. Gurevich, I.; Tamir, H.; Arango, V.; Dwork, A. J.; Mann, J. J.; Schmauss, C.: Altered editing of serotonin 2C receptor pre-mRNA in the prefrontal cortex of depressed suicide victims. *Neuron* 34: 349–356, 2002.

[42544] 16096. Hall, C. S.: Genetic differences in fatal audiogenic seizures between two inbred strains of house mice. *J. Hered.* 38: 3–6, 1947.

[42545] 16097. Lappalainen, J.; Zhang, L.; Dean, M.; Oz, M.; Ozaki, N.; Yu, D.; Virkkunen, M.; Weight, F.; Linnoila, M.; Goldman, D.: Identification, expression, and pharmacology of a cys(23)–ser(23) substitution in the human 5-HT_{2C} receptor gene (HTR2C). *Genomics* 27: 274–279, 1995.

[42546] 16098. Milatovich, A.; Hsieh, C.-L.; Bonaminio, G.; Tecott, L.; Julius, D.; Francke, U.: Serotonin receptor 1c gene as–

signed to X chromosome in human (band q24) and mouse (bands D–F4). *Hum. Molec. Genet.* 1:681–684, 1992.

[42547] 16099. Tecott, L. H.; Sun, L. M.; Akana, S. F.; Strack, A. M.; Lowenstein, D. H.; Dallman, M. F.; Julius, D.: Eating disorder and epilepsy in mice lacking 5-HT_{2C} serotonin receptors. *Nature* 374: 542–546, 1995.

[42548] 16100. Blaschke, R. J.; Monaghan, A. P.; Schiller, S.; Schechinger, B.; Rao, E.; Padilla–Nash, H.; Ried, T.; Rappold, G. A.: SHOT, a SHOX–related homeobox gene, is implicated in craniofacial, brain, heart, and limb development. *Proc. Nat. Acad. Sci.* 95: 2406–2411, 1998.

[42549] 16101. Clement–Jones, M.; Schiller, S.; Rao, E.; Blaschke, R. J.; Zuniga, A.; Zeller, R.; Robson, S. C.; Binder, G.; Glass, I.; Strachan, T.; Lindsay, S.; Rappold, G. A.: The short stature homeobox gene SHOX is involved in skeletal abnormalities in Turner syndrome. *Hum. Molec. Genet.* 9: 695–702, 2000.

[42550] 16102. Affara, N. A.; Chalmers, I. J.; Ferguson–Smith, M. A.: Analysis of the SRY gene in 22 sex–reversed XY females identifies four new point mutations in the conserved DNA binding domain. *Hum. Molec. Genet.* 2: 785–789, 1993.

[42551] 16103. Behlke, M. A.; Bogan, J. S.; Beer–Romero, P.; Page, D. C.: Evidence that the SRY protein is encoded by a single

exon on the human Y chromosome. *Genomics* 17:736–739, 1993.

- [42552] 16104. Berkovitz, G. D.; Fechner, P. Y.; Zacur, H. W.; Rock, J. A.; Snyder, H. M., III; Migeon, C. J.; Perlman, E. J.: Clinical and pathologic spectrum of 46,XY gonadal dysgenesis: its relevance to the understanding of sex differentiation. *Medicine* 70: 375–383, 1991.
- [42553] 16105. Bowles, J.; Cooper, L.; Berkman, J.; Koopman, P.: Sry requires a CAG repeat domain for male sex determination in *Mus musculus*. *Nature Genet.* 22: 405–408, 1999.
- [42554] 16106. Braun, A.; Kammerer, S.; Cleve, H.; Lohrs, U.; Schwarz, H.-P.; Kuhnle, U.: True hermaphroditism in a 46,XY individual, caused by a postzygotic somatic point mutation in the male gonadal sex-determining locus (SRY): molecular genetics and histological findings in a sporadic case. *Am. J. Hum. Genet.* 52: 578–585, 1993.
- [42555] 16107. Brown, S.; Yu, C. C.; Lanzano, P.; Heller, D.; Thomas, L.; Warburton, D.; Kitajewski, J.; Stadtmauer, L.: A de novo mutation (Gln2stop) at the 5-prime end of the SRY gene leads to sex reversal with partial ovarian function. (Letter) *Am. J. Hum. Genet.* 62: 189–192, 1998.
- [42556] 16108. Canto, P.; de la Chesnaye, E.; Lopez, M.; Cervantes, A.; Chavez, B.; Vilchis, F.; Reyes, E.; Ulloa-Aguirre, A.; Kof-

man-Alfaro, S.;Mendez, J. P.: A mutation in the 5-prime non-high mobility groupbox region of the SRY gene in patients with Turner syndrome and Ymosaicism. J. Clin. Endocr. Metab. 85: 1908–1911, 2000.

[42557] 16109.Cherfas, J.: Sex and the single gene. Science 252: 782, 1991.

[42558] 16110.Clepet, C.; Schafer, A. J.; Sinclair, A. H.; Palmer, M. S.; Lovell-Badge,R.; Goodfellow, P. N.: The human SRY transcript. Hum. Molec. Genet. 2:2007–2012, 1993.

[42559] 16111.Davis, R. M.: Localisation of male determining factors in man:a thorough review of structural anomalies of the Y chromosome. J.Med. Genet. 18: 161–195, 1981.

[42560] 16112.Dork, T.; Stuhrmann, M.; Miller, K.; Schmidtke, J.: Independentobservation of SRY mutation I90M in a patient with complete gonadaldysgenesis. (Letter) Hum. Mutat. 11: 90–91, 1998.

[42561] 16113.Eicher, E. M.; Washburn, L. L.: Genetic control of primary sexdetermination in mice. Annu. Rev. Genet. 20: 327–360, 1986.

[42562] 16114.Foster, J. W.; Brennan, F. E.; Hampikian, G. K.; Goodfellow, P.N.; Sinclair, A. H.; Lovell-Badge, R.; Selwood, L.; Renfree, M. B.;Cooper, D. W.; Graves, J. A. M.: Evolution of sex determination andthe Y chromosome:

SRY-related sequences in marsupials. *Nature* 359:531–533, 1992.

- [42563] 16115. Fuqua, J. S.; McLaughlin, J.; Perlman, E. J.; Berkowitz, G. D.: Analysis of the SRY gene in gonadal tissue of subjects with 46,XY gonadal dysgenesis (Letter) *J. Clin. Endocr. Metab.* 82: 701–702, 1997.
- [42564] 16116. Domenice, S.; Nishi, M. Y.; Billerbeck, A. E. C.; Latronico, A. C.; Medeiros, M. A.; Russell, A. J.; Vass, K.; Carvalho, F. M.; Frade, E. M. C.; Arnhold, I. J. P.; Mendonca, B. B.: A novel missense mutation (S18N) in the 5-prime non-HMG box region of the SRY gene in a patient with partial gonadal dysgenesis and his normal male relatives. *Hum. Genet.* 102: 213–215, 1998.
- [42565] 16117. Giese, K.; Pagel, J.; Grosschedl, R.: Distinct DNA-binding properties of the high mobility group domain of murine and human SRY sex-determining factors. *Proc. Nat. Acad. Sci.* 91: 3368–3372, 1994.
- [42566] 16118. Goodfellow, P. N.; Lovell-Badge, R.: SRY and sex determination in mammals. *Annu. Rev. Genet.* 27: 71–92, 1993.
- [42567] 16119. Gubbay, J.; Collignon, J.; Koopman, P.; Capel, B.; Economou, A.; Munsterberg, A.; Vivian, N.; Goodfellow, P. N.; Lovell-Badge, R.: A gene mapping to the sex-

determining region of the mouse Y chromosome is a member of a novel family of embryonically expressed genes. *Nature* 346:245–250, 1990.

- [42568] 16120. Haqq, C. M.; King, C.-Y.; Ukiyama, E.; Falsafi, S.; Haqq, T. N.; Donahoe, P. K.; Weiss, M. A.: Molecular basis of mammalian sexual determination: activation of Mullerian inhibiting substance gene expression by SRY. *Science* 266: 1494–1500, 1994.
- [42569] 16121. Clark, J.; Rocques, P. J.; Crew, A. J.; Gill, S.; Shipley, J.; Chan, A. M.-L.; Gusterson, B. A.; Cooper, C. S.: Identification of novel genes, SYT and SSX, involved in the t(X;18)(p11.2;q11.2) translocation found in human synovial sarcoma. *Nature Genet.* 7: 502–508, 1994.
- [42570] 16122. Yorifuji, T.; Wilson, R. W.; Beaudet, A. L.: Retroviral mediated expression of CD18 in normal and deficient human bone marrow progenitor cells. *Hum. Molec. Genet.* 2: 1443–1448, 1993.
- [42571] 16123. Ramulu, P.; Kennedy, M.; Xiong, W.-H.; Williams, J.; Cowan, M.; Blesh, D.; Yau, K.-W.; Hurley, J. B.; Nathans, J.: Normal light response, photoreceptor integrity, and rhodopsin dephosphorylation in mice lacking both protein phosphatases with EF hands (PPEF-1 and PPEF-2). *Molec. Cell. Biol.* 21: 8605–8614, 2001.

- [42572] 16124.van de Vosse, E.; Franco, B.; van der Bent, P.; Montini, E.; Orth,U.; Hanauer, A.; Tijmes, N.; van Ommen, G.-J. B.; Ballabio, A.; denDunnen, J. T.; Bergen, A. A. B.: Exclusion of PPEF as the gene causingX-linked juvenile retinoschisis. *Hum. Genet.* 101: 235–237, 1997.
- [42573] 16125.Fisher, S. E.; Ciccodicola, A.; Tanaka, K.; Curci, A.; Desicato,S.; D'Urso, M.; Craig, I. W.: Sequence-based exon prediction aroundthe synaptophysin locus reveals a gene-rich area containing novelgenes in human proximal Xp. *Genomics* 45: 340–347, 1997.
- [42574] 16126.Oliva, M. M.; Wu, T. C.; Yang, V. W.: Isolation and characterizationof a differentiation-dependent gene in the human colonic cell lineHT29–18. *Arch. Biochem. Biophys.* 302: 183–192, 1993.
- [42575] 16127.Prakash, S. K.; Paylor, R.; Jenna, S.; Lamarche–Vane, N.; Armstrong,D. L.; Xu, B.; Mancini, M. A.; Zoghbi, H. Y.: Functional analysisof ARHGAP6, a novel GTPase-activating protein for RhoA. *Hum. Molec.Genet.* 9: 477–488, 2000.
- [42576] 16128.Schaefer, L.; Prakash, S.; Zoghbi, H. Y.: Cloning and characterizationof a novel rho-type GTPase-activating protein gene (ARHGAP6) fromthe critical region for microphthalmia with linear skin defects. *Genomics*

46:268–277, 1997.

- [42577] 16129.Aman, M. J.; Tayebi, N.; Obiri, N. I.; Puri, R. K.; Modi, W. S.;Leonard, W. J.: cDNA cloning and characterization of the human interleukin13 receptor alpha chain. *J. Biol. Chem.* 271: 29265–29270, 1996.
- [42578] 16130.Guo, J.; Apiou , F.; Mellerin, M.–P.; Lebeau, B.; Jacques, Y.;Minvielle, S.: Chromosome mapping and expression of the human interleukin–13receptor. *Genomics* 42: 141–5, 1997.
- [42579] 16131.Hilton, D. J.; Zhang, J.–G.; Metcalf, D.; Alexander, W. S.; Nicola,N. A.; Willson, T. A.: Cloning and characterization of a bindingsubunit of the interleukin 13 receptor that is also a component ofthe interleukin 4 receptor. *Proc. Nat. Acad. Sci.* 93: 497–501, 1996.
- [42580] 16132.Kaye, F. J.; Modi, S.; Ivanovska, I.; Koonin, E. V.; Thress, K.;Kubo, A.; Kornbluth, S.; Rose, M. D.: A family of ubiquitin–likeproteins binds the ATPase domain of Hsp70–like Stch. *FEBS Lett.* 467:348–352, 2000.
- [42581] 16133.Kaye, F. J.; Shows, T. B.: Assignment of ubiquilin 2 (UBQLN2)to human chromosome xp11.23–p11.1 by Gene–Bridge radiation hybrids. *Cytogenet.Cell Genet.* 89: 116–117, 2000.
- [42582] 16134.Kleijnen, M. F.; Shih, A. H.; Zhou, P.; Kumar, S.;

Soccio, R. E.;Kedersha, N. L.; Gill, G.; Howley, P. M.: The hPLIC proteins mayprovide a link between the ubiquitination machinery and the proteasome. *Molec.Cell* 6: 409–419, 2000.

[42583] 16135.Marchand, J.–B.; Kaiser, D. A.; Pollard, T. D.; Higgs, H. N.:Interaction of WASP/Scar proteins with actin and vertebrate Arp2/3complex. *Nature Cell Biol.* 3: 76–82, 2001.

[42584] 16136.Lee, D. K.; Nguyen, T.; Lynch, K. R.; Cheng, R.; Vanti, W. B.;Arkhitko, O.; Lewis, T.; Evans, J. F.; George, S. R.; O'Dowd, B. F.: Discovery and mapping of ten novel G protein–coupled receptor genes. *Gene* 275:83–91, 2001.

[42585] 16137.Furneaux, H. M.; Rosenblum, M. K.; Dalmau, J.; Wong, E.; Woodruff,P.; Graus, F.; Posner, J. B.: Selective expression of Purkinje–cellantigens in tumor tissue from patients with paraneoplastic cerebellardegeneration. *New Eng. J. Med.* 322: 1844–1851, 1990.

[42586] 16138.Furneaux, H. M.; Wong, E.; Posner, J. B.: Isolation of cDNA clonesencoding the major Yo paraneoplastic anti–gen. (Abstract) *Neurology* 40(suppl. 1): 166 only, 1990.

[42587] 16139.Knight, J. C.; Renwick, P. J.; Downing, J. R.; Okuda, T.: Physicallinkage of the cdc2–related gene (PCTK1) and the ubiquitin–activatingenzyme E1 gene (UBE1) on human

Xp11.3. Cytogenet. Cell Genet. 71:155–157, 1995.

- [42588] 16140.Aarskog, D.: A familial syndrome of short stature associated with facial dysplasia and genital anomalies. J. Pediat. 77: 856–861, 1970.
- [42589] 16141.Baldellou, A.; Galve, L.; Bassecourt, M.: Risk of medullary damage in Aarskog–Scott syndrome. (Abstract) Clin. Genet. 23: 225 only, 1983.
- [42590] 16142.Bawle, E.; Tyrkus, M.; Lipman, S.; Bozimowski, D.: Aarskog syndrome: full male and female expression associated with an X–autosome translocation. Am.J. Med. Genet. 17: 595–602, 1984.
- [42591] 16143.Berman, P. A.; Desjardins, C.; Fraser, F. C.: Inheritance of the Aarskog syndrome. Birth Defects Orig. Art. Ser. X(7): 151–159, 1974.
- [42592] 16144.Buechner, M.; Hall, D. H.; Bhatt, H.; Hedgecock, E. M.: Cystic canal mutants in *Caenorhabditis elegans* are defective in the apical membrane domain of the renal (excretory) cell. Dev. Biol. 214: 227–241, 1999.
- [42593] 16145.Casteels, M.; Samain, H.; Penninckx, F.; Coremans, G.; Beirinckx, J.; Fryns, J. P.: Megadolichosigmoid in a young male with Aarskog syndrome. Genet. Counsel. 5: 81–83, 1994.
- [42594] 16146.Estrada, L.; Caron, E.; Gorski, J. L.: Fgd1, the Cdc42

guaninenucleotide exchange factor responsible for facio-genital dysplasia, is localized to the subcortical actin cytoskeleton and Golgi membrane. *Hum.Molec. Genet.* 10: 485–495, 2001.

- [42595] 16147. Fernandez, I.; Tsukahara, M.; Mito, H.; Yoshii, H.; Uchida, M.; Matsuo, K.; Kajii, T.: Congenital heart defects in Aarskog syndrome. *Am.J. Med. Genet.* 50: 318–322, 1994.
- [42596] 16148. Fryns, J.-P.: Dolichomegasigmoid in Aarskog syndrome. (Letter) *Am.J. Med. Genet.* 45: 122 only, 1993.
- [42597] 16149. Fryns, J. P.: Aarskog syndrome: the changing phenotype with age. *Am.J. Med. Genet.* 43: 420–427, 1992.
- [42598] 16150. Fryns, J. P.; Macken, J.; Vinken, L.; Igodt-Ameye, L.; van denBerghe, H.: The Aarskog syndrome. *Hum. Genet.* 42: 129–135, 1978.
- [42599] 16151. Funderburk, S. J.; Crandall, B. F.: The Aarskog syndrome in three brothers. *Clin. Genet.* 6: 119–124, 1974.
- [42600] 16152. Furukawa, C. T.; Hall, B. D.; Smith, D. W.: The Aarskog syndrome. *J.Pediat.* 81: 1117–1122, 1972.
- [42601] 16153. Gao, J.; Estrada, L.; Cho, S.; Ellis, R. E.; Gorski, J. L.: The *Caenorhabditis elegans* homolog of FGD1, the human Cdc42 GEF gene responsible for faciogenital dysplasia, is critical for excretory cell morphogenesis. *Hum.Molec.*

Genet. 10: 3049–3062, 2001.

- [42602] 16154.Glover, T. W.; Verga, V.; Rafael, J.; Barcroft, C.; Gorski, J.L.; Bawle, E. V.; Higgins, J. V.: Translocation breakpoint in Aarskogs syndrome maps to Xp11.21 between ALAS2 and DXS323. Hum. Molec. Genet. 2:1717–1718, 1993.
- [42603] 16155.Grier, R. E.; Farrington, F. H.; Kendig, R.; Mamunes, P.: Autosomaldominant inheritance of the Aarskog phenotype. (Abstract) Am. J.Hum. Genet. 33: 64A only, 1981.
- [42604] 16156.Hoo, J. J.: The Aarskog (facio-digito-genital) syndrome. Clin.Genet. 16: 269–276, 1979.
- [42605] 16157.Kodama, M.; Fujimoto, S.; Namikawa, T.; Matsuda, I.: Aarskogs syndrome with isolated growth hormone deficiency. Europ. J. Pediat. 135:273–276, 1981.
- [42606] 16158.Lebel, R. R.; May, M.; Pouls, S.; Lubs, H. A.; Stevenson, R. E.;Schwartz, C. E.: Non-syndromic X-linked mental retardation associated with a missense mutation (P312L) in the FGD1 gene. Clin. Genet. 61:139–145, 2002.
- [42607] 16159.Logie, L. J.; Porteous, M. E. M.: Intelligence and development in Aarskog syndrome. Arch. Dis. Child. 79: 359–360, 1998.
- [42608] 16160.Mikelsaar, R. V.–A.; Lurie, I. W.: Atypical case of Aarskog syndrome. J.Med. Genet. 29: 349–350, 1992.

- [42609] 16161.Nielsen, K. B.: Aarskog syndrome in a Danish family: an illustration of the need for dysmorphology in paediatrics. Clin. Genet. 33: 315–317, 1988.
- [42610] 16162.Oberiter, V.; Lovrencic, M. K.; Schmutzer, L.; Kraus, O.: The Aarskog syndrome. Acta Paediat. Scand. 69: 567–570, 1980.
- [42611] 16163.Orrico, A.; Galli, L.; Falciani, M.; Bracci, M.; Cavaliere, M.L.; Rinaldi, M. M.; Musacchio, A.; Sorrentino, V.: A mutation in the pleckstrin homology (PH) domain of the FGD1 gene in an Italian family with faciogenital dysplasia (Aarskog–Scott syndrome). FEBS Lett. 478: 216–220, 2000.
- [42612] 16164.Pasteris, N. G.; Cadle, A.; Logie, L. J.; Porteous, M. E. M.; Schwartz, C. E.; Stevenson, R. E.; Glover, T. W.; Wilroy, R. S.; Gorski, J. L.: Isolation and analysis of the faciogenital dysplasia (Aarskog–Scott syndrome) gene: a putative, rho/rac guanine nucleotide exchange factor. Cell 79:669–678, 1994.
- [42613] 16165.Pasteris, N. G.; de Gouyon, B.; Cadle A. B.; Campbell, K.; Herman, G. E.; Gorski, J. L.: Cloning and regional localization of the mouse faciogenital dysplasia (Fgd1) gene. Mammalian Genome 6: 658–661, 1995.
- [42614] 16166.Pedersen, J. C.; Fryns, J. P.; Bracke, P.; Geeraert, M.; Van Den Berghe, H.: The Aarskog syndrome. Ann. Genet.

23: 108–110, 1980.

- [42615] 16167. Porteous, M. E. M.; Goudie, D. R.: Aarskog syndrome. *J. Med. Genet.* 28: 44–47, 1991.
- [42616] 16168. Rafael, J.; Verga, V.; Hall, B.; Burright, E.; Gorski, J.; Bawle, J.; Higgins, J. V.; Glover, T. W.: Assignment of the translocation breakpoint in a patient with Aarskog syndrome to Xp11.21. (Abstract) *Am. J. Hum. Genet.* 51 (suppl.): A116 only, 1992.
- [42617] 16169. Schwartz, C. E.; Gillessen-Kaesbach, G.; May, M.; Cappa, M.; Gorski, J.; Steindl, K.; Neri, G.: Two novel mutations confirm FGD1 is responsible for the Aarskog syndrome. *Europ. J. Hum. Genet.* 8: 869–874, 2000.
- [42618] 16170. Scott, C. I., Jr.: Unusual facies, joint hypermobility, genital anomaly and short stature: a new dysmorphic syndrome. *Birth Defects Orig. Art. Ser.* VII(6): 240–246, 1971.
- [42619] 16171. Demarest, S. J.; Martinez-Yamout, M.; Chung, J.; Chen, H.; Xu, W.; Dyson, H. J.; Evans, R. M.; Wright, P. E.: Mutual synergistic folding in recruitment of CBP/p300 by p160 nuclear receptor coactivators. *Nature* 415: 549–553, 2002.
- [42620] 16172. Eckner, R.; Ewen, M. E.; Newsome, D.; Gerdes, M.; DeCaprio, J. A.; Lawrence, J. B.; Livingston, D. M.: Molecular cloning and functional analysis of the adenovirus

E1A-associated 300-kD protein (p300) reveals a protein with properties of a transcriptional adaptor. *Genes Dev.* 15:869–884, 1994.

- [42621] 16173. Lin, C. H.; Hare, B. J.; Wagner, G.; Harrison, S. C.; Maniatis, T.; Fraenkel, E.: A small domain of CBP/p300 binds diverse proteins: solution structure and functional studies. *Molec. Cell* 8: 581–590, 2001.
- [42622] 16174. Cohen–Haguenauer, O.; Picard, J. Y.; Mattei, M.–G.; Serero, S.; Van Cong, N.; de Tand, M.–F.; Guerrier, D.; Hors–Cayla, M.–C.; Josso, N.; Frezal, J.: Mapping of the gene for anti–mullerian hormone to the short arm of human chromosome 19. *Cytogenet. Cell Genet.* 44:2–6, 1987.
- [42623] 16175. Forest, M. G.: Serum Mullerian inhibiting substance assay -- a new diagnostic test for disorders of gonadal development. (Editorial) *New Eng. J. Med.* 336: 1519–1521, 1997.
- [42624] 16176. Cate, R. L.; Mattaliano, R. J.; Hession, C.; Tizard, R.; Farber, N. M.; Cheung, A.; Ninfa, E. G.; Frey, A. Z.; Gash, D. J.; Chow, E. P.; Fisher, R. A.; Bertonis, J. M.; Torres, G.; Wallner, B. P.; Ramachandran, K. L.; Ragin, R. C.; Mangano, T. F.; MacLaughlin, D. T.; Donahoe, P. K.: Isolation of the bovine and human genes for mullerian inhibiting sub–

stance and expression of the human gene in animal cells. Cell 45:685–698, 1986.

- [42625] 16177. Guerrier, D.; Tran, D.; Vanderwinden, J. M.; Hideux, S.; Van Outryve, L.; Legeai, L.; Bouchard, M.; Van Vliet, G.; De Laet, M. H.; Picard, J. Y.; Kahn, A.; Josso, N.: The persistent mullerian duct syndrome: a molecular approach. J. Clin. Endocr. Metab. 68: 46–52, 1989.
- [42626] 16178. Harbison, M. D.; Magid, M. L. S.; Josso, N.; Mininberg, D. T.; New, M. I.: Anti-Mullerian hormone in three intersex conditions. Ann. Genet. 34: 226–232, 1991.
- [42627] 16179. King, T. R.; Lee, B. K.; Behringer, R. R.; Eicher, E. M.: Mapping anti-Mullerian hormone (Amh) and related sequences in the mouse: identification of a new region of homology between MMU10 and HSA19p. Genomics 11: 273–283, 1991.
- [42628] 16180. Knebelmann, B.; Boussin, L.; Guerrier, D.; Legeai, L.; Kahn, A.; Josso, N.; Picard, J.-Y.: Anti-Mullerian hormone Bruxelles: a nonsense mutation associated with the persistent Mullerian duct syndrome. Proc. Nat. Acad. Sci. 88: 3767–3771, 1991.
- [42629] 16181. Lee, M. M.; Donahoe, P. K.; Silverman, B. L.; Hasegawa, T.; Hasegawa, Y.; Gustafson, M. L.; Chang, Y.; MacLaughlin, D. T.: Measurement of serum Mullerian in-

hibiting substance in the evaluation of children with non-palpable gonads. *New Eng. J. Med.* 336: 1480–1486, 1997.

[42630] 16182. Rey, R. A.; Belville, C.; Nihoul-Fekete, C.; Michel-Calemard, L.; Forest, M. G.; Lahlou, N.; Jaubert, F.; Mowszowicz, I.; David, M.; Saka, N.; Bouvattier, C.; Bertrand, A.-M.; and 16 others: Evaluation of gonadal function in 107 intersex patients by means of serum antimüllerian hormone measurement. *J. Clin. Endocr. Metab.* 84: 627–631, 1999.

[42631] 16183. Rogers, D. S.; Gallagher, D. S.; Womack, J. E.: Somatic cell mapping of the genes for anti-müllerian hormone and osteonectin in cattle: identification of a new bovine syntenic group. *Genomics* 9:298–300, 1991.

[42632] 16184. Riesewijk, A. M.; Blagitko, N.; Schinzel, A. A.; Hu, L.; Schulz, U.; Hamel, B. C. J.; Ropers, H.-H.; Kalscheuer, V. M.: Evidence against a major role of PEG1/MEST Silver-Russell syndrome. *Europ. J. Hum. Genet.* 6: 114–120, 1998.

[42633] 16185. Riesewijk, A. M.; Hu, L.; Schulz, U.; Tariverdian, G.; Hoglund, P.; Kere, J.; Ropers, H.-H.; Kalscheuer, V. M.: Monoallelic expression of human PEG1/MEST is paralleled by parent-specific methylation in fetuses. *Genomics* 42:

236–244, 1997.

- [42634] 16186.Sado, T.; Nakajima, N.; Tada, M.; Takagi, N.: A novel mesoderm-specific cDNA isolated from a mouse embryonal carcinoma cell line. *Dev. Growth Differ.* 35: 551–560, 1993.
- [42635] 16187.Jayakumar, A.; Chirala, S. S.; Chinault, A. C.; Baldini, A.; Abu-Elheiga, L.; Wakil, S. J.: Isolation and chromosomal mapping of genomic clones encoding the human fatty acid synthase gene. *Genomics* 23: 420–424, 1994.
- [42636] 16188.Jayakumar, A.; Tai, M.-H.; Huang, W.-Y.; Al-Feel, W.; Hsu, M.; Abu-Elheiga, L.; Chirala, S. S.; Wakil, S. J.: Human fatty acid synthase: properties and molecular cloning. *Proc. Nat. Acad. Sci.* 92: 8695–8699, 1995.
- [42637] 16189.Loftus, T. M.; Jaworsky, D. E.; Frehywot, G. L.; Townsend, C. A.; Ronnett, G. V.; Lane, M. D.; Kuhajda, F. P.: Reduced food intake and body weight in mice treated with fatty acid synthase inhibitors. *Science* 288: 2379–2381, 2000.
- [42638] 16190.Wakil, S. J.: Fatty acid synthase, a proficient multifunctional enzyme. *Biochemistry* 28: 4523–4530, 1989.
- [42639] 16191.Ye, Q.; Chung, L. W. K.; Li, S.; Zhau, H. E.: Identification of a novel FAS/ER- α fusion transcript expressed in human cancer cells. *Biochim. Biophys. Acta* 1493:

373–377, 2000.

- [42640] 16192.Hofmann, M. A.; Drury, S.; Fu, C.; Qu, W.; Taguchi, A.; Lu, Y.;Avila, C.; Kambham, N.; Bierhaus, A.; Nawroth, P.; Neurath, M. F.;Slattery, T.; Beach, D.; McClary, J.; Nagashima, M.; Morser, J.; Stern,D.; Schmidt, A. M.: RAGE mediates a novel proinflammatory axis: acentral cell surface receptor for S100/calgranulin polypeptides. *Cell* 97:889–901, 1999.
- [42641] 16193.Yan, S. D.; Chen, X.; Fu, J.; Chen, M.; Zhu, H.; Røher, A.; Slattery,T.; Zhao, L.; Nagashima, M.; Morser, J.; Migheli, A.; Nawroth, P.;Stern, D.; Schmidt, A. M.: RAGE and amyloid-beta peptide neurotoxicityin Alzheimer's disease. *Nature* 382: 685–691, 1996.
- [42642] 16194.Argeson, A. C.; Druck, T.; Veronese, M. L.; Knopf, J. L.; Buchberg,A. M.; Huebner, K.; Siracusa, L. D.: Phospholipase C gamma-2 (Plcg2)and phospholipase C gamma-1 (Plcg1) map to distinct regions in thehuman and mouse genomes. *Genomics* 25: 29–35, 1995.
- [42643] 16195.Hernandez, D.; Egan, S. E.; Yulug, I. G.; Fisher, E. M. C.: Mappingthe gene that encodes phosphatidylinositol-specific phospholipaseC-gamma-2 in the human and the mouse. *Genomics* 23: 504–507, 1994.
- [42644] 16196.Kang, J. S.; Kohlhuber, F.; Hug, H.; Marme, D.; Eick,

D.; Ueffing, M.: Cloning and functional analysis of the hematopoietic cell-specific phospholipase C-gamma-2 promoter. FEBS Lett. 399: 14-20, 1996.

- [42645] 16197. Dumont, D. J.; Anderson, L.; Breitman, M. L.; Duncan, A. M. V.: Assignment of the endothelial-specific protein receptor tyrosine kinase gene (TEK) to human chromosome 9p21. Genomics 23: 512-513, 1994.
- [42646] 16198. Delot, E.; King, L. M.; Briggs, M. D.; Wilcox, W. R.; Cohn, D. H.: Trinucleotide expansion mutations in the cartilage oligomeric matrix protein (COMP) gene. Hum. Molec. Genet. 8: 123-128, 1999.
- [42647] 16199. Back, A. L.; Kerkering, M.; Baker, D.; Bauer, T. R.; Embree, L. J.; Hickstein, D. D.: A point mutation associated with leukocyte adhesion deficiency type 1 of moderate severity. Biochem. Biophys. Res. Commun. 193: 912-918, 1993.
- [42648] 16200. Back, A. L.; Kwok, W. W.; Hickstein, D. D.: Identification of two molecular defects in a child with leukocyte adhesion deficiency. J. Biol. Chem. 267: 5482-5487, 1992.
- [42649] 16201. Barclay, A. N.; Birkeland, M. L.; Brown, M. H.; Beyers, A. D.; Davis, S. J.; Somoza, C.; Williams, A. F.: The Leukocyte Antigen Facts Book. New York: Academic Press

(pub.) 1993. Pp. 124–127and 140–141.

- [42650] 16202.Beatty, P. G.; Ochs, H. D.; Harlan, J. M.; Price, T. H.; Rosen,H.; Taylor, R. F.; Hansen, J. A.; Klebanoff, S. J.: Absence of monoclonal–antibody–definedprotein complex in a boy with abnormal leucocyte function. *Lancet* I:535–537, 1984.
- [42651] 16203.Bianchi, E.; Denti, S.; Granata, A.; Bossi, G.; Geginat, J.; Villa,A.; Rogge, L.; Pardi, R.: Integrin LFA–1 interacts with the transcriptionalco–activator JAB1 to modulate AP–1 activity. *Nature* 404: 617–621,2000.
- [42652] 16204.Bissenden, J. G.; Haeney, M. R.; Tarlow, M. J.; Thompson, R. A.: Delayed separation of the umbilical cord, severe widespread infections,and immunodeficiency. *Arch. Dis. Child.* 56: 397–399, 1981.
- [42653] 16205.Boucheix, C.: Personal Communication. Villejuif, France 1/31/1987.
- [42654] 16206.Bowen, T. J.; Ochs, H. D.; Altman, L. C.; Price, T. H.; Van Epps,D. E.; Brautigan, D. L.; Rosin, R. E.; Perkins, W. D.; Babior, B.M.; Klebanoff, S. J.; Wedgwood, R. J.: Severe recurrent bacterialinfections associated with defective adherence and chemotaxis in twopatients with neutrophils deficient in a cell–associated glycoprotein. *J.Pediat.* 101: 932–940, 1982.

- [42655] 16207. Bullard, D. C.; Scharffetter-Kochanek, K.; McArthur, M. J.; Chosay, J. G.; McBride, M. E.; Montgomery, C. A.; Beaudet, A. L.: A polygenic mouse model of psoriasiform skin disease in CD18-deficient mice. *Proc. Nat. Acad. Sci.* 93: 2116–2121, 1996.
- [42656] 16208. Crowley, C. A.; Curnutte, J. T.; Rosin, R. E.; Andre-Schwartz, J.; Gallin, J. I.; Klempner, M.; Snyderman, R.; Southwick, F. S.; Stossel, T. P.; Babior, B. M.: An inherited abnormality of neutrophil adhesion: its genetic transmission and its association with a missing protein. *New Eng. J. Med.* 302: 1163–1168, 1980.
- [42657] 16209. Dana, N.; Clayton, L. K.; Tennen, D. G.; Pierce, M. W.; Lachmann, P. J.; Law, S. A.; Arnaout, M. A.: Leukocytes from four patients with complete or partial Leu-CAM deficiency contain the common beta-subunit precursor and beta-subunit messenger RNA. *J. Clin. Invest.* 79: 1010–1015, 1987.
- [42658] 16210. Dana, N.; Todd, R. F., III; Pitt, J.; Springer, T. A.; Arnaout, M. A.: Deficiency of a surface membrane glycoprotein (Mo1) in man. *J. Clin. Invest.* 73: 153–159, 1984.
- [42659] 16211. Fischer, A.; Griscelli, C.; Friedrich, W.; Kubanek, B.; Levinsky, R.; Morgan, G.; Vossen, J.; Wagemaker, G.; Landais, P.: Bone-marrow transplantation for immunodeficiency.

ciencies and osteopetrosis: Europeansurvey, 1968–1985.
Lancet II: 1080–1084, 1986.

[42660] 16212.Fujita, K.; Kobayashi, K.; Kajii, T.: Impaired neutrophil adhesion:a new patient in a previously reported family. *Acta Paediat. Jpn.* 27:527–534, 1985.

[42661] 16213.Fujita, K.; Kobayashi, K.; Okino, F.: Juvenile rheumatoid arthritisin two siblings with congenital leukocyte adhesion deficiency. *Europ.J. Pediat.* 148: 118–119, 1988.

[42662] 16214.Harvath, L.; Andersen, B. R.: Defective initiation of oxidativemetabolism in polymorphonuclear leukocytes. *New Eng. J. Med.* 300:1130–1135, 1979.

[42663] 16215.Hayward, A. R.; Leonard, J.; Harvey, B. A. M.; Greenwood, M. C.;Wood, C. B. S.; Soothill, J. F.: Delayed separation of the umbilicalcord, widespread infections, and defective neutrophil mobility. *Lancet I*:1099–1101, 1979.

[42664] 16216.Hibbs, M. L.; Wardlaw, A. J.; Stacker, S. A.; Anderson, D. C.;Lee, A.; Roberts, T. M.; Springer, T. A.: Transfection of cells frompatients with leukocyte adhesion deficiency with an integrin betasubunit (CD18) restores lymphocyte function–associated antigen–1 expressionand function. *J. Clin. Invest.* 85: 674–681, 1990.

- [42665] 16217.Hogg, N.; Stewart, M. P.; Scarth, S. L.; Newton, R.; Shaw, J.M.; Law, S. K. A.; Klein, N.: A novel leukocyte adhesion deficiency caused by expressed but nonfunctional beta-2 integrins Mac-1 and LFA-1. *J.Clin. Invest.* 103: 97-106, 1999.
- [42666] 16218.Hynes, R. O.: Integrins: versatility, modulation and signaling in cell adhesion. *Cell* 69: 11-25, 1992.
- [42667] 16219.Kehrli, M. E., Jr.; Ackermann, M. R.; Shuster, D. E.; van derMaaten, M. J.; Schmalstieg, F. C.; Anderson, D. C.; Hughes, B. J.: Bovine leukocyte adhesion deficiency: beta(2) integrin deficiency in young Holstein cattle. *Am. J. Path.* 140: 1489-1492, 1992.
- [42668] 16220.Kishimoto, T. K.; Hollander, N.; Roberts, T. M.; Anderson, D.C.; Springer, T. A.: Heterogeneous mutations in the beta subunit common to the LFA-1, Mac-1, and p150,95 glycoproteins cause leukocyte adhesion deficiency. *Cell* 50: 193-202, 1987.
- [42669] 16221.Kishimoto, T. K.; O'Connor, K.; Lee, A.; Roberts, T. M.; Springer, T. A.: Cloning of the beta subunit of the leukocyte adhesion proteins: homology to an extracellular matrix receptor defines a novel supergene family. *Cell* 48: 681-690, 1987.
- [42670] 16222.Kobayashi, K.; Fujita, K.; Okino, F.; Kajii, T.: An ab-

normality of neutrophil adhesion: autosomal recessive inheritance associated with missing neutrophil glycoproteins. *Pediatrics* 73: 606–610, 1984.

- [42671] 16223. Krauss, J. C.; Mayo-Bond, L. A.; Rogers, C. E.; Weber, K. L.; Todd, R. F., III; Wilson, J. M.: An in vivo animal model of gene therapy for leukocyte adhesion deficiency. *J. Clin. Invest.* 88: 1412–1417, 1991.
- [42672] 16224. Matsuura, S.; Kishi, F.; Tsukahara, M.; Nunoi, H.; Matsuda, I.; Kobayashi, K.; Kajii, T.: Leukocyte adhesion deficiency: identification of novel mutations in two Japanese patients with a severe form. *Biochem. Biophys. Res. Commun.* 184: 1460–1467, 1992.
- [42673] 16225. Nelson, C.; Rabb, H.; Arnaout, M. A.: Genetic cause of leukocyte adhesion molecule deficiency: abnormal splicing and a missense mutation in a conserved region of CD18 impair cell surface expression of beta-2 integrins. *J. Biol. Chem.* 267: 3351–3357, 1992.
- [42674] 16226. Tini, M.; Benecke, A.; Um, S.-J.; Torchia, J.; Evans, R. M.; Chambon, P.: Association of CBP/p300 acetylase and thymine DNA glycosylase links DNA repair and transcription. *Molec. Cell* 9: 265–277, 2002.
- [42675] 16227. Weaver, B. K.; Kumar, K. P.; Reich, N. C.: Interferon regulatory factor 3 and CREB-binding protein/p300 are

subunits of double-stranded RNA-activated transcription factor DRAF1. *Molec. Cell. Biol.* 18:1359–1368, 1998.

[42676] 16228. Hansen, J. J.; Durr, A.; Cournu-Rebeix, I.; Georgopoulos, C.; Ang, D.; Nielsen, M. N.; Davoine, C.-S.; Brice, A.; Fontaine, B.; Gregersen, N.; Bross, P.: Hereditary spastic paraplegia SPG13 is associated with a mutation in the gene encoding the mitochondrial chaperonin Hsp60. *Am. J. Hum. Genet.* 70: 1328–1332, 2002.

[42677] 16229. Todd, M. J.; Viitanen, P. V.; Lorimer, G. H.: Dynamics of the chaperonin ATPase cycle: implications for facilitated protein folding. *Science* 265:659–666, 1994.

[42678] 16230. Hellevuo, K.; Berry, R.; Sikela, J. M.; Tabakoff, B.: Localization of the gene for a novel human adenylyl cyclase (ADCY7) to chromosome 16. *Hum. Genet.* 95: 197–200, 1995.

[42679] 16231. Hellevuo, K.; Yoshimura, M.; Kao, M.; Hoffman, P. L.; Cooper, D. M. F.; Tabakoff, B.: A novel adenylyl cyclase sequence cloned from the human erythroleukemia cell line. *Biochem. Biophys. Res. Commun.* 192:311–318, 1993.

[42680] 16232. Iavarone, A.; Garg, P.; Lasorella, A.; Hsu, J.; Israel, M. A.: The helix-loop-helix protein Id-2 enhances cell proliferation and binds to the retinoblastoma protein. *Genes Dev.* 8: 1270–1284, 1994.

- [42681] 16233.Lasorella, A.; Iavarone, A.; Israel, M. A.: Id2 specifically alters regulation of the cell cycle by tumor suppressor proteins. *Molec.Cell. Biol.* 16: 2570–2578, 1996.
- [42682] 16234.Lasorella, A.; Nosedà, M.; Beyna, M.; Iavarone, A.: Id2 is a retinoblastoma protein target and mediates signalling by Myc oncoproteins. *Nature* 407:592–598, 2000.
- [42683] 16235.Moerman, P.; Fryns, J.-P.; Sastrowijoto, S. H.; Vandenberghe, K.; Lauweryns, J. M.: Hereditary renal adysplasia: new observations and hypotheses. *Pediat. Path.* 14: 405–410, 1994.
- [42684] 16236.Stapleton, D.; Woollatt, E.; Mitchell, K. I.; Nicholl, J. K.; Fernandez, C. S.; Michell, B. J.; Witters, L. A.; Power, D. A.; Sutherland, G. R.; Kemp, B. E.: AMP-activated protein kinase isoenzyme family: subunit structure and chromosomal location. *FEBS Lett.* 409: 452–456, 1997.
- [42685] 16237.Budarf, M. L.; Perier, F.; Barnoski, B. L.; Bell, C. J.; Vandenberg, C. A.: Assignment of the human hippocampal inward rectifier potassium channel (HIR) gene to 22q13.1. *Genomics* 26: 625–629, 1995.
- [42686] 16238.Makhina, E. N.; Kelly, A. J.; Lopatin, A. N.; Mercer, R. W.; Nichols, C. G.: Cloning and expression of a novel human brain inward rectifier potassium channel. *J. Biol. Chem.* 269: 20468–20474, 1994.

- [42687] 16239.Perier, F.; Radeke, C. M.; Vandenberg, C. A.: Primary structure and characterization of a small-conductance inwardly rectifying potassium channel from human hippocampus. *Proc. Nat. Acad. Sci.* 91: 6240–6244, 1994.
- [42688] 16240.Derry, J. M.; Barnard, P. J.: The gene for tissue inhibitor of metalloproteinases (TIMP) is located within an intron of the synapsin I gene on the X chromosome. (Abstract) *Cytogenet. Cell Genet.* 58:2061–2062, 1991.
- [42689] 16241.Gal, A.; Wieringa, B.; Smeets, D. F. C. M.; Bleeker-Wagemakers, L.; Ropers, H. H.: Submicroscopic interstitial deletion of the X chromosome explains a complex genetic syndrome dominated by Norrie disease. *Cytogenet. Cell Genet.* 42: 219–224, 1986.
- [42690] 16242.Simpson, E.: The H-Y antigen and sex reversal. *Cell* 44: 813–814, 1986.
- [42691] 16243.Di Paola, R.; Frittitta, L.; Miscio, G.; Bozzali, M.; Baratta, R.; Centra, M.; Spampinato, D.; Santagati, M. G.; Ercolino, T.; Cisternino, C.; Soccio, T. Mastroianno, S.; Tassi, V.; Almgren, P.; Pizzuti, A.; Vigneri, R.; Trischitta, V.: A variation in 3-prime UTR of hPTP1B increases specific gene expression and associates with insulin resistance. *Am. J. Hum. Genet.* 70: 806–812, 2002.

- [42692] 16244.Eidne, K.; Taylor, P.; Connor, M.; Duthie, S.: Isolation, characterization and chromosomal localization of the human thyrotropin releasing hormone receptor. (Abstract) 75th Annual Meeting of the Endocr. Soc. 437only, 1993.
- [42693] 16245.Castedo, M.; Ferri, K. F.; Blanco, J.; Roumier, T.; Larochette, N.; Barretina, J.; Amendola, A.; Nardacci, R.; Metivier, D.; Este, J. A.; Piacentini, M.; Kroemer, G.: Human immunodeficiency virus 1 envelope glycoprotein complex-induced apoptosis involves mammalian target of rapamycin/FKBP12-rapamycin-associated protein-mediated p53 phosphorylation. *J. Exp. Med.* 194: 1097-1110, 2001.
- [42694] 16246.El-Deiry, W. S.; Tokino, T.; Velculescu, V. E.; Levy, D. B.; Parsons, R.; Trent, J. M.; Lin, D.; Mercer, E.; Kinzler, K. W.; Vogelstein, B.: WAF1, a potential mediator of p53 tumor suppression. *Cell* 75:817-825, 1993.
- [42695] 16247.Moore, K. J.; D'Amore-Bruno, M. A.; Korfhagen, T. R.; Glasser, S. W.; Whitsett, J. A.; Jenkins, N. A.; Copeland, N. G.: Chromosomal localization of three pulmonary surfactant protein genes in the mouse. *Genomics* 12:388-393, 1992.
- [42696] 16248.Nogee, L. M.; Dunbar, A. E., III; Wert, S. E.; Askin, F.; Hamvas, A.; Whitsett, J. A.: A mutation in the surfactant protein C gene associated with familial interstitial lung

disease. *New Eng. J. Med.* 344:573–579, 2001.

- [42697] 16249. Warr, R. G.; Hawgood, S.; Buckley, D. I.; Crisp, T. M.; Schilling, J.; Benson, B. J.; Ballard, P. L.; Clements, J. A.; White, R. T.: Low molecular weight human pulmonary surfactant protein (SP5): isolation, characterization, and cDNA and amino acid sequences. *Proc. Nat. Acad. Sci.* 84: 7915–7919, 1987.
- [42698] 16250. Wood, S.; Yaremko, M. L.; Schertzer, M.; Kelemen, P. R.; Minna, J.; Westbrook, C. A.: Mapping of the pulmonary surfactant SP5 (SFTP2) locus to 8p21 and characterization of a microsatellite repeat marker that shows frequent loss of heterozygosity in human carcinomas. *Genomics* 24:597–600, 1994.
- [42699] 16251. Kolble, K.; Lu, J.; Mole, S. E.; Kaluz, S.; Reid, K. B. M.: Assignment of the human pulmonary surfactant protein D gene (SFTP4) to 10q22–q23 close to the surfactant protein A gene cluster. *Genomics* 17: 294–298, 1993.
- [42700] 16252. Botas, C.; Poulain, F.; Akiyama, J.; Brown, C.; Allen, L.; Goerke, J.; Clements, J.; Carlson, E.; Gillespie, A. M.; Epstein, C.; Hawgood, S.: Altered surfactant homeostasis and alveolar type II cell morphology in mice lacking surfactant protein D. *Proc. Nat. Acad. Sci.* 95:11869–11874, 1998.
- [42701] 16253. Crouch, E.; Rust, K.; Veile, R.; Donis-Keller, H.;

Grosso, L.: Genomic organization of human surfactant protein D (SP-D): SP-D is encoded on chromosome 10q22.2–23.1. *J. Biol. Chem.* 268: 2976–2983, 1993.

- [42702] 16254. Holmskov, U.; Lawson, P.; Teisner, B.; Tornøe, I.; Willis, A. C.; Morgan, C.; Koch, C.; Reid, K. B. M.: Isolation and characterization of a new member of the scavenger receptor superfamily, glycoprotein-340 (gp-340), as a lung surfactant protein-D binding molecule. *J. Biol. Chem.* 272: 13743–13749, 1997.
- [42703] 16255. Holmskov, U.; Møllenhauer, J.; Madsen, J.; Vitved, L.; Grønlund, J.; Tornøe, I.; Kliem, A.; Reid, K. B. M.; Poustka, A.; Skjodt, K.: Cloning of gp-340, a putative opsonin receptor for lung surfactant protein D. *Proc. Nat. Acad. Sci.* 96: 10794–10799, 1999.
- [42704] 16256. LeVine, A. M.; Whitsett, J. A.; Hartshorn, K. L.; Crouch, E. C.; Korfhagen, T. R.: Surfactant protein D enhances clearance of influenza A virus from the lung in vivo. *J. Immun.* 167: 5868–5873, 2001.
- [42705] 16257. Lu, J.; Willis, A. C.; Reid, K. B. M.: Purification, characterization and cDNA cloning of human lung surfactant protein D. *Biochem. J.* 284: 795–802, 1992.
- [42706] 16258. Rust, K.; Grosso, L.; Zhang, V.; Chang, D.; Persson, A.; Longmore, W.; Cai, G.-Z.; Crouch, E.: Human surfactant

protein D: SP-D contains a C-type lectin carbohydrate recognition domain. Arch. Biochem. Biophys. 290:116–126, 1991.

[42707] 16259. Wert, S. E.; Yoshida, M.; LeVine, A. M.; Ikegami, M.; Jones, T.; Ross, G. F.; Fisher, J. H.; Korfhagen, T. R.; Whitsett, J. A.: Increased metalloproteinase activity, oxidant production, and emphysema in surfactant protein D gene-inactivated mice. Proc. Nat. Acad. Sci. 97: 5972–5977, 2000.

[42708] 16260. Gedde-Dahl, T., Jr.; Olaisen, B.; Teisberg, P.; Wilhelmy, M. C.; Mevag, B.; Helland, R.: The locus for apolipoprotein E (apoE) is close to the Lutheran (Lu) blood group locus on chromosome 19. Hum. Genet. 67: 178–182, 1984.

[42709] 16261. Grubb, R.: Zur Genetik des Lewis-Systems. Naturwissenschaften 21:560–561, 1953.

[42710] 16262. Kolanus, W.; Nagel, W.; Schiller, B.; Zeitlmann, L.; Godar, S.; Stockinger, H.; Seed, B.: Alpha-L-beta-2 integrin/LFA-1 binding to ICAM-1 induced by cytohesin-1, a cytoplasmic regulatory molecule. Cell 86:233–242, 1996.

[42711] 16263. Ogasawara, M.; Kim, S.-C.; Adamik, R.; Togawa, A.; Ferrans, V.J.; Takeda, K.; Kirby, M.; Moss, J.; Vaughan, M.: Similarities in function and gene structure of cytohesin-4

and cytohesin-1, guaninenucleotide-exchange proteins for ADP-ribosylation factors. *J. Biol.Chem.* 275: 3221-3230, 2000.

- [42712] 16264.Findlay, D. M.; Fisher, L. W.; McQuillan, C. I.; Ter-
mine, J. D.;Young, M. F.: Isolation of the osteonectin gene:
evidence that avariable region of the osteonectin molecule
is encoded within oneexon. *Biochemistry* 27: 1483-1489,
1988.
- [42713] 16265.Gilmour, D. T.; Lyon, G. J.; Carlton, M. B. L.; Sanes,
J. R.; Cunningham,J. M.; Anderson, J. R.; Hogan, B. L. M.;
Evans, M. J.; Colledge, W.H.: Mice deficient for the se-
creted glycoprotein SPARC/osteonectin/BM40develop
normally but show severe age-onset cataract formation
anddisruption of the lens. *EMBO J.* 17: 1860-1870, 1998.
- [42714] 16266.Sanyanusin, P.; McNoe, L. A.; Sullivan, M. J.;
Weaver, R. G.;Eccles, M. R.: Mutation of PAX2 in two sib-
lings with renal-colobomasymndrome. *Hum. Molec. Genet.*
4: 2183-2184, 1995.
- [42715] 16267.Sanyanusin, P.; Norrish, J. H.; Ward, T. A.; Nebel,
A.; McNoe,L. A.; Eccles, M. R.: Genomic structure of the
human PAX2 gene. *Genomics* 35:258-261, 1996.
- [42716] 16268.Sanyanusin, P.; Schimmenti, L. A.; McNoe, L. A.;
Ward, T. A.;Pierpont, M. E. M.; Sullivan, M. J.; Dobyns, W.

B.; Eccles, M. R.: Mutation of the PAX2 gene in a family with optic nerve colobomas, renal anomalies and vesicoureteral reflux. *Nature Genet.* 9: 358–364, 1995.

[42717] 16269. Schimmenti, L. A.; Cunliffe, H. E.; McNoe, L. A.; Ward, T. A.; French, M. C.; Shim, H. H.; Zhang, Y.-H.; Proesmans, W.; Leys, A.; Byerly, K. A.; Braddock, S. R.; Masuno, M.; Imaizumi, K.; Devriendt, K.; Eccles, M. R.: Further delineation of renal-coloboma syndrome in patients with extreme variability of phenotype and identical PAX2 mutations. *Am. J. Hum. Genet.* 60: 869–878, 1997.

[42718] 16270. Schimmenti, L. A.; Pierpont, M. E.; Carpenter, B. L. M.; Kashtan, C. E.; Johnson, M. R.; Dobyns, W. B.: Autosomal dominant optic nerve colobomas, vesicoureteral reflux, and renal anomalies. *Am. J. Med. Genet.* 59: 204–208, 1995.

[42719] 16271. Schimmenti, L. A.; Shim, H. H.; Wirtschafter, J. D.; Panzarino, V. A.; Kashtan, C. E.; Kirkpatrick, S. J.; Wargowski, D. S.; France, T. D.; Michel, E.; Dobyns, W. B.: Homonucleotide expansion and contraction mutations of PAX2 and inclusion of Chiari 1 malformation as part of renal-coloboma syndrome. *Hum. Mutat.* 14: 369–376, 1999.

[42720] 16272. Stapleton, P.; Weith, A.; Urbanek, P.; Kozmik, Z.;

Busslinger,M.: Chromosomal localization of seven PAX genes and cloning of anovel family member, PAX-9. Nature Genet. 3: 292-298, 1993.

[42721] 16273.Tellier, A.-L.; Amiel, J.; Delezoide, A.-L.; Audollent, S.; Auge,J.; Esnault, D.; Encha-Razavi, F.; Munnich, A.; Lyonnet, S.; Vekemans,M.; Attie-Bitach, T.: Expression of the PAX2 gene in human embryosand exclusion in the CHARGE syndrome. Am. J. Med. Genet. 93: 85-88,2000.

[42722] 16274.Tellier, A.-L.; Amiel, J.; Salomon, R.; Jolly, D.; Delezoide,A.-L.; Auge, J.; Gubler, M.-C.; Munnich, A.; Lyonnet, S.; Antignac,C.; Vekemans, M.; Broyer, M.; Attie-Bitach, T.: PAX2 expression duringearly human development and its mutations in renal hypoplasia withor without coloboma. (Abstract) Am. J. Hum. Genet. 63 (suppl.):A7 only, 1998.

[42723] 16275.Ward, T. A.; Nebel, A.; Reeve, A. E.; Eccles, M. R.: Alternativemessenger RNA forms and open reading frames within an additional conservedregion of the human PAX-2 gene. Cell Growth Differ. 5: 1015-1021,1994.

[42724] 16276.Weaver, R. G.; Cashwell, L. F.; Lorentz, W.; White-man, D.; Geisinger,K. R.; Ball, M.: Optic nerve coloboma associated with renal disease. Am.J. Med. Genet. 29: 597-605, 1988.

[42725] 16277.Barr, F. G.; Nauta, L. E.; Davis, R. J.; Schafer, B. W.;

Nycum, L. M.; Biegel, J. A.: In vivo amplification of the PAX3-FKHR and PAX7-FKHR fusion genes in alveolar rhabdomyosarcoma. *Hum. Molec. Genet.* 5: 15–21, 1996.

[42726] 16278. Burri, M.; Tromvoukis, Y.; Bopp, D.; Frigerio, G.; Noll, M.: Conservation of the paired domain in metazoans and its structure in three isolated human genes. *EMBO J.* 8: 1183–1190, 1989.

[42727] 16279. Gruss, P.; Walther, C.: Pax in development. *Cell* 69: 719–722, 1992.

[42728] 16280. Schafer, B. W.; Mattei, M. G.: The human paired domain gene PAX7 (Hup1) maps to chromosome 1p35–1p36.2. *Genomics* 17: 249–251, 1993.

[42729] 16281. Seale, P.; Sabourin, L. A.; Girgis-Gabardo, A.; Mansouri, A.; Gruss, P.; Rudnicki, M. A.: Pax7 is required for the specification of myogenic satellite cells. *Cell* 102: 777–786, 2000.

[42730] 16282. Shapiro, D. N.; Sublett, J. E.; Li, B.; Valentine, M. B.; Morris, S. W.; Noll, M.: The gene for PAX7, a member of the paired-box-containing genes, is localized on human chromosome arm 1p36. *Genomics* 17: 767–769, 1993.

[42731] 16283. Helwig, U.; Imai, K.; Schmahl, W.; Thomas, B. E.; Varnum, D. S.; Nadeau, J. H.; Balling, R.: Interaction between undulated and Patch leads to an extreme form of

spina bifida in double-mutant mice. *NatureGenet.* 11: 60–63, 1995.

[42732] 16284.Sun, J.; Rose, J. B.; Bird, P.: Gene structure, chromosomal localization, and expression of the murine homologue of human proteinase inhibitor6 (PI-6) suggests divergence of PI-6 from the ovalbumin serpins. *J.Biol. Chem.* 270: 16089–16096, 1995.

[42733] 16285.Aberle, H.; Bierkamp, C.; Torchard, D.; Serova, O.; Wagner, T.; Natt, E.; Wirsching, J.; Heidkamper, C.; Montagna, M.; Lynch, H. T.; Lenoir, G. M.; Scherer, G.; Feunteun, J.; Kemler, R.: The human plakoglobingene localizes on chromosome 17q21 and is subjected to loss of heterozygosity in breast and ovarian cancers. *Proc. Nat. Acad. Sci.* 92: 6384–6388, 1995.

[42734] 16286.Bierkamp, C.; Mclaughlin, K. J.; Schwarz, H.; Huber, O.; Kemler, R.: Embryonic heart and skin defects in mice lacking plakoglobin. *Dev.Biol.* 180: 780–785, 1996.

[42735] 16287.Cowley, C. M. E.; Simrak, D.; Spurr, N. K.; Arneemann, J.; Buxton, R. S.: The plakophilin 1 (PKP1) and plakoglobin (JUP) genes map to human chromosomes 1q and 17, respectively. *Hum. Genet.* 100: 486–488, 1997.

[42736] 16288.Franke, W. W.; Goldschmidt, M. D.; Zimbelmann, R.; Mueller, H.M.; Schiller, D. L.; Cowin, P.: Molecular cloning

and amino acid sequence of human plakoglobin, the common junctional plaque protein. *Proc. Nat. Acad. Sci.* 86: 4027–4031, 1989.

[42737] 16289. Guenet, J.-L.; Simon-Chazottes, D.; Ringwald, M.; Kemler, R.: The genes coding for alpha and beta catenin (Catna1 and Catnb) and plakoglobin (Jup) map to mouse chromosomes 18, 9, and 11, respectively. *Mammalian Genome* 6: 363–366, 1995.

[42738] 16290. Knudsen, K. A.; Wheelock, M. J.: Plakoglobin, or an 83-kD homolog distinct from beta-catenin, interacts with E-cadherin and N-cadherin. *J. Cell Biol.* 118: 671–679, 1992.

[42739] 16291. Kolligs, F. T.; Kolligs, B.; Hajra, K. M.; Hu, G.; Tani, M.; Cho, K. R.; Fearon, E. R.: Gamma-catenin is regulated by the APC tumor suppressor and its oncogenic activity is distinct from that of beta-catenin. *Genes Dev.* 14: 1319–1331, 2000.

[42740] 16292. Mathur, M.; Goodwin, L.; Cowin, P.: Interactions of the cytoplasmic domain of the desmosomal cadherin Dsg1 with plakoglobin. *J. Biol. Chem.* 269: 14075–14080, 1994.

[42741] 16293. McKoy, G.; Protonotarios, N.; Crosby, A.; Tsatsopoulou, A.; Anastasakis, A.; Coonar, A.; Norman, M.; Baboonian, C.; Jeffery, S.; McKenna, W. J.: Identification of a

deletion in plakoglobin in arrhythmogenic right ventricular cardiomyopathy with palmoplantar keratoderma and woolly hair (Naxos disease). *Lancet* 355: 2119–2124, 2000.

[42742] 16294. Ruiz, P.; Brinkmann, V.; Ledermann, B.; Behrend, M.; Grund, C.; Thalhammer, C.; Vogel, F.; Birchmeier, C.; Gunthert, U.; Franke, W.W.; Birchmeier, W.: Targeted mutation of plakoglobin in mice reveals essential functions of desmosomes in the embryonic heart. *J. Cell Biol.* 135: 215–225, 1996.

[42743] 16295. Buckley, M. F.; Goding, J. W.: Plasma cell membrane glycoprotein gene Pca-1 (alkaline phosphodiesterase I) is linked to the proto-oncogene Myb on mouse chromosome 10. *Immunogenetics* 36: 199–201, 1992.

[42744] 16296. Buckley, M. F.; Loveland, K. A.; McKinstry, W. J.; Garson, O. M.; Goding, J. W.: Plasma cell membrane glycoprotein PC-1: cDNA cloning of the human molecule, amino acid sequence, and chromosomal location. *J. Biol. Chem.* 265: 17506–17511, 1990.

[42745] 16297. Frittitta, L.; Baratta, R.; Spampinato, D.; Di Paola, R.; Pizzuti, A.; Vigneri, R.; Trischitta, V.: The Q121 PC-1 variant and obesity have additive and independent effects in causing insulin resistance. *J. Clin. Endocr. Metab.* 86:

5888–5891, 2001.

- [42746] 16298. Harahap, A. R.; Goding, J. W.: Distribution of the murine plasmacell antigen PC-1 in non-lymphoid tissues. *J. Immun.* 141: 2317–2320, 1988.
- [42747] 16299. Huang, R.; Rosenbach, M.; Vaughn, R.; Provvedini, D.; Rebbe, N.; Hickman, S.; Goding, J.; Terkeltaub, R.: Expression of the murine plasma cell nucleotide pyrophosphohydrolase PC-1 is shared by human liver, bone, and cartilage cells: regulation of PC-1 expression in osteosarcoma cells by transforming growth factor-beta. *J. Clin. Invest.* 94: 560–567, 1994.
- [42748] 16300. Kahn, C. R.: Causes of insulin resistance. *Nature* 373: 384–385, 1995.
- [42749] 16301. Maddux, B. A.; Goldfine, I. D.: Membrane glycoprotein PC-1 inhibition of insulin receptor function occurs via direct interaction with the receptor alpha-subunit. *Diabetes* 49: 13–19, 2000.
- [42750] 16302. Maddux, B. A.; Sbraccia, P.; Kumakura, S.; Sasson, S.; Youngren, J.; Fisher, A.; Spencer, S.; Grupe, A.; Henzel, W.; Stewart, T. A.; Reaven, G. M.; Goldfine, I. D.: Membrane glycoprotein PC-1 and insulin resistance in non-insulin-dependent diabetes mellitus. *Nature* 373: 448–451, 1995.

- [42751] 16303. Nakamura, I.; Ikegawa, S.; Okawa, A.; Okuda, S.; Koshizuka, Y.; Kawaguchi, H.; Nakamura, K.; Koyama, T.; Goto, S.; Toguchida, J.; Matsushita, M.; Ochi, T.; Takaoka, K.; Nakamura, Y.: Association of the human NPPS gene with ossification of the posterior longitudinal ligament of the spine (OPLL). *Hum. Genet.* 104: 492–497, 1999.
- [42752] 16304. Okawa, A.; Nakamura, I.; Goto, S.; Moriya, H.; Nakamura, Y.; Ikegawa, S.: Mutation in *Npps* in a mouse model of ossification of the posterior longitudinal ligament of the spine. *Nature Genet.* 19: 271–273, 1998.
- [42753] 16305. Pizzuti, A.; Frittitta, L.; Argiolas, A.; Baratta, R.; Goldfine, I. D.; Bozzali, M.; Ercolino, T.; Scarlato, G.; Iacoviello, L.; Vigneri, R.; Tassi, V.; Trischitta, V.: A polymorphism (K121Q) of the human glycoprotein PC-1 gene coding region is strongly associated with insulin resistance. *Diabetes* 48: 1881–1884, 1999.
- [42754] 16306. Rebbe, N. F.; Tong, B. D.; Finley, E. M.; Hickman, S.: Identification of nucleotide pyrophosphatase/alkaline phosphodiesterase I activity associated with the mouse plasma cell differentiation antigen PC-1. *Proc. Nat. Acad. Sci.* 88: 5192–5196, 1991.
- [42755] 16307. Sakaguchi, A. Y.; Lalley, P. A.; Zabel, B. U.; Ellis, R. W.; Scolnick, E. M.; Naylor, S. L.: Chromosome assignments

of four mouse cellular homologs of sarcoma and leukemia virus oncogenes. *Proc. Nat. Acad. Sci.* 81: 525–529, 1984.

- [42756] 16308. Takahashi, T.; Old, L. J.; Boyse, E. A.: Surface alloantigen of plasma cells. *J. Exp. Med.* 131: 1325–1341, 1970.
- [42757] 16309. van Driel, I. R.; Goding, J. W.: Plasma cell membrane glycoprotein PC-1: primary structure deduced from cDNA clones. *J. Biol. Chem.* 262:4882–4887, 1987.
- [42758] 16310. Frank, S. L.; Klisak, I.; Sparkes, R. S.; Lusk, A. J.: A gene homologous to plasminogen located on human chromosome 2q11–p11. *Genomics* 4:449–451, 1989.
- [42759] 16311. Westergaard, J. G.; Chemnitz, J.; Teisner, B.; Poulsen, H. K.; Ipsen, L.; Beck, B.; Grudzinski, J. G.: Pregnancy-associated plasma protein A: a possible marker in the classification and prenatal diagnosis of Cornelia de Lange syndrome. *Prenatal Diag.* 3: 225–232, 1983.
- [42760] 16312. Barnett, T.; Pickle, W., II; Rae, P. M. M.; Hart, J.; Kamarck, M.; Elting, J.: Pregnancy-specific beta-1-glycoproteins are related to carcinoembryonic antigens and map to chromosome 19. (Abstract) *Cytogenet. Cell Genet.* 51: 958, 1989.
- [42761] 16313. Barnett, T. R.; Pickle, W., II; Rae, P. M. M.; Hart, J.; Kamarck, M.; Elting, J.: Human pregnancy-specific

beta(1)-glycoproteins are coded within chromosome 19.

Am. J. Hum. Genet. 44: 890-893, 1989.

[42762] 16314. Bartels, I.; Lindemann, A.: Maternal levels of pregnancy-specific beta-1-glycoprotein (SP-1) are elevated in pregnancies affected by Down's syndrome. Hum. Genet. 80: 46-48, 1988.

[42763] 16315. Brandriff, B. F.; Gordon, L. A.; Tynan, K. T.; Olsen, A. S.; Mohrenweiser, H. W.; Fertitta, A.; Carrano, A. V.; Trask, B. J.: Order and genomic distances among members of the carcinoembryonic antigen (CEA) gene family determined by fluorescence in situ hybridization. Genomics 12: 773-779, 1992.

[42764] 16316. Chan, W.-Y.; Qiu, W.-R.: Human pregnancy-specific beta-1 glycoprotein is encoded by multiple genes localized on two chromosomes. Am. J. Hum. Genet. 43: 152-159, 1988.

[42765] 16317. Khan, W. N.; Teglund, S.; Bremer, K.; Hammarstrom, S.: The pregnancy-specific glycoprotein family of the immunoglobulin superfamily: identification of new members and estimation of family size. Genomics 12: 780-787, 1992.

[42766] 16318. Niemann, S. C.; Flake, A.; Bohn, H.; Bartels, I.: Pregnancy-specific beta-1-glycoprotein: cDNA cloning,

tissue expression, and species specificity of one member of the PSBG family. Hum. Genet. 82: 239–243, 1989.

[42767] 16319. Niemann, S. C.; Schonk, D.; van Dijk, P.; Wieringa, B.; Grzeschik, K.-H.; Bartels, I.: Regional localization of the gene encoding pregnancy-specific beta-1-glycoprotein 1 (PSBG1) to human chromosome 19q13.1. Cytogenet. Cell Genet. 52: 95–97, 1989.

[42768] 16320. Niemann, S. C.; Schonk, D.; van Dijk, P. E.; Grzeschik, K.-H.; Bartels, I.: Chromosomal assignment of a cDNA clone encoding pregnancy-specific-beta-1-glycoprotein to chromosome 19. (Abstract) Cytogenet. Cell Genet. 51: 1053, 1989.

[42769] 16321. Olsen, A.; Teglund, S.; Nelson, D.; Gordon, L.; Copeland, A.; Georgescu, A.; Carrano, A.; Hammarstrom, S.: Gene organization of the pregnancy-specific glycoprotein region on human chromosome 19: assembly and analysis of a 700-kb cosmid contig spanning the region. Genomics 23: 659–668, 1994.

[42770] 16322. Streydio, C.; Swillens, S.; Georges, M.; Szpirer, C.; Vassart, G.: Structure, evolution and chromosomal localization of the human pregnancy-specific beta-1 glycoprotein gene family. Genomics 6: 579–592, 1990. Note: Erratum: Genomics 7: 661–662, 1990.

- [42771] 16323.Teglund, S.; Olsen, A.; Khan, W. N.; Frangsmyr, L.; Hammarstrom,S.: The pregnancy-specific glycoprotein (PSG) gene cluster on human chromosome 19: fine structure of the 11 PSG genes and identification of 6 new genes forming a third subgroup within the carcinoembryonic antigen (CEA) family. *Genomics* 23: 669–684, 1994.
- [42772] 16324.Thompson, J.; Koumari, R.; Wagner, K.; Barnert, S.; Schleussner,C.; Schrewe, H.; Zimmermann, W.; Muller, G.; Schempp, W.; Zaninetta,D.; Ammaturo, D.; Hardman, N.: The human pregnancy-specific glycoprotein genes are tightly linked on the long arm of chromosome 19 and are coordinately expressed. *Biochem. Biophys. Res. Commun.* 167: 848–859,1990.
- [42773] 16325.Watanabe, S.; Chou, J. Y.: Isolation and characterization of complementary DNAs encoding human pregnancy-specific beta-1-glycoprotein. *J.Biol. Chem.* 263: 2049–2054, 1988.
- [42774] 16326.Leslie, K. K.; Watanabe, S.; Lei, K.-J.; Chou, D. Y.; Plouzek,C. A.; Deng, H.-C.; Torres, J.; Chou, J. Y.: Linkage of two human pregnancy-specific beta-1-glycoprotein genes: one is associated with hydatidiform mole. *Proc. Nat. Acad. Sci.* 87: 5822–5826, 1990.
- [42775] 16327.Olsen, A.; Teglund, S.; Nelson, D.; Gordon, L.;

Copeland, A.; Georgescu, A.; Carrano, A.; Hammarstrom, S.: Gene organization of the pregnancy-specific glycoprotein region on human chromosome 19: assembly and analysis of a 700-kb cosmid contig spanning the region. *Genomics* 23: 659–668, 1994.

[42776] 16328. Thompson, J.; Zimmermann, W.; Osthus-Bugat, P.; Schleussner, C.; Eades-Perner, A.-M.; Barnert, S.; von Kleist, S.; Willcocks, T.; Craig, I.; Tynan, K.; Olsen, A.; Mohrenweiser, H.: Long-range chromosomal mapping of the carcinoembryonic antigen (CEA) gene family cluster. *Genomics* 12: 761–772, 1992.

[42777] 16329. Tynan, K.; Olsen, A.; Trask, B.; de Jong, P.; Thompson, J.; Zimmermann, W.; Carrano, A.; Mohrenweiser, H.: Assembly and analysis of cosmid contigs in the CEA-gene family region of human chromosome 19. *Nucleic Acids Res.* 20: 1629–1636, 1992.

[42778] 16330. Sheer, D.; Sheppard, D. M.; Le Beau, M.; Rowley, J. D.; San Roman, C.; Solomon, E.: Localization of the oncogene c-erbA1 immediately proximal to the acute promyelocytic leukaemia breakpoint on chromosome 17. *Ann. Hum. Genet.* 49: 167–171, 1985.

[42779] 16331. Yoshitake, H.; Rittling, S. R.; Denhardt, D. T.; Noda, M.: Osteopontin-deficient mice are resistant to ovarian

tomy-induced bone resorption. Proc.Nat. Acad. Sci. 96: 8156–8160, 1999.

- [42780] 16332.Edelmann, W.; Zervas, M.; Costello, P.; Roback. L.; Fischer, I.;Hammarback, J. A.; Cowan, N.; Davies, P.; Wainer, B.; Kucherlapati,R.: Neuronal abnormalities in microtubule-associated protein 1B mutantmice. Proc. Nat. Acad. Sci. 93: 1270–1275, 1996.
- [42781] 16333.Hammarback, J. A.; Obar, R. A.; Hughes, S. M.; Vallee, R. B.:MAP1B is encoded as a polyprotein that is processed to form a complexN-terminal microtubule-binding domain. Neuron 7: 129–139, 1991.
- [42782] 16334.Lien, L. L.; Boyce, F. M.; Kleyn, P.; Brzustowicz, L. M.; Menninger,J.; Ward, D. C.; Gilliam, T. C.; Kunkel, L. M.: Mapping of humanmicrotubule-associated protein 1B in proximity to the spinal muscularatrophy locus at 5q13. Proc. Nat. Acad. Sci. 88: 7873–7876, 1991.
- [42783] 16335.Lien, L. L.; Feener, C. A.; Fischbach, N.; Kunkel, L. M.: Cloningof human microtubule-associated protein 1B and the identificationof a related gene on chromosome 15. Genomics 22: 273–280, 1994.
- [42784] 16336.Wirth, B.; Voosen, B.; Rohrig, D.; Knapp, M.; Piechaczek, B.; Rudnik-Schoneborn,S.; Zerres, K.: Fine mapping and narrowing of the genetic intervalof the

spinal muscular atrophy region by linkage studies. *Genomics* 15:113–118, 1993.

- [42785] 16337. Zhang, Y. Q.; Bailey, A. M.; Matthies, H. J. G.; Renden, R. B.; Smith, M. A.; Speese, S. D.; Rubin, G. M.; Broadie, K.: *Drosophila* fragile X-related gene regulates the MAP1B homolog Futsch to control synaptic structure and function. *Cell* 107: 591–603, 2001.
- [42786] 16338. Garner, C. C.; Tucker, R. P.; Matus, A.: Selective localization of messenger RNA for cytoskeletal protein MAP2 in dendrites. *Nature* 336:674–677, 1988.
- [42787] 16339. Kalcheva, N.; Albala, J.; O'Guin, K.; Rubino, H.; Garner, C.; Shafit-Zagardo, B.: Genomic structure of human microtubule-associated protein 2 (MAP-2) and characterization of additional MAP-2 isoforms. *Proc. Nat. Acad. Sci.* 92: 10894–10898, 1995.
- [42788] 16340. Kindler, S.; Garner, C. C.: Four repeat MAP2 isoforms in human and rat brain. *Molec. Brain Res.* 26: 218–224, 1994.
- [42789] 16341. Marsden, K. M.; Doll, T.; Ferralli, J.; Botteri, F.; Matus, A.: Transgenic expression of embryonic MAP2 in adult mouse brain: implications for neuronal polarization. *J. Neurosci.* 16: 3265–3273, 1996.
- [42790] 16342. Neve, R. L.; Harris, P.; Kosik, K. S.; Kurnit, D. M.;

Donlon, T.A.: Identification of cDNA clones for the human microtubule-associated protein tau and chromosomal localization of the genes for tau and microtubule-associated protein 2. *Molec. Brain Res.* 1: 271–280, 1986.

[42791] 16343. Chapin, S. J.; Bulinski, J. C.: Non-neuronal 210 x 10(3) M(r) microtubule-associated protein (MAP4) contains a domain homologous to the microtubule-binding domains of neuronal MAP2 and tau. *J. Cell Sci.* 98: 27–36, 1991.

[42792] 16344. Adriaansen, H. J.; Geurts Van Kessel, A. H. M.; Wijdenes–De Bresser, J. H. F. M.; Van Drunen–Schoenmaker, E.; Van Dongen, J. J. M.: Expression of the myeloid differentiation antigen CD33 depends on the presence of human chromosome 19 in human–mouse hybrids. *Ann. Hum. Genet.* 54:115–119, 1990.

[42793] 16345. Bonthron, D. T.; Dunlop, N.; Barr, D. G. D.; El Sanousi, A. A.; Al–Gazali, L. I.: Organisation of the human PAX4 gene and its exclusion as a candidate for the Wolcott–Rallison syndrome. *J. Med. Genet.* 35:288–292, 1998.

[42794] 16346. Inoue, H.; Nomiyama, J.; Nakai, K.; Matsutani, A.; Tanizawa, Y.; Oka, Y.: Isolation of full-length cDNA of mouse PAX4 gene and identification of its human homologue. *Biochem. Biophys. Res. Commun.* 243:

628–633,1998.

[42795] 16347.Mansouri, A.; St-Onge, L.; Gruss, P.: Role of Pax genes in endoderm-derivedorgans. Trends Endocr. Metab. 10: 164–167, 1999.

[42796] 16348.Matsushita, T.; Yamaoka, T.; Otsuka, S.; Moritani, M.; Matsumoto,T.; Itakura, M.: Molecular cloning of mouse paired-box-containinggene (Pax)–4 from an islet beta cell line and deduced sequence ofhuman Pax–4. Biochem. Bio-phys. Res. Commun. 242: 176–180, 1998.

[42797] 16349.Sosa–Pineda, B.; Chowdhury, K.; Torres, M.; Oliver, G.; Gruss,P.: The Pax4 gene is essential for differentiation of insulin–producingbeta cells in the mammalian pan-creas. Nature 386: 399–402, 1997.

[42798] 16350.St-Onge, L.; Sosa–Pineda, B.; Chowdhury, K.; Man-souri, A.; Gruss,P.: Pax6 is required for differentiation of glucagon–producing alpha–cells in mouse pancreas. Na-ture 387: 406–409, 1997.

[42799] 16351.Tamura, T.; Izumikawa, Y.; Kishino, T.; Soejima, H.; Jinno, Y.;Niikawa, N.: Assignment of the human PAX4 gene to chromosome band7q32 by fluorescence in situ hy-bridization. Cytogenet. Cell Genet. 66:132–134, 1994.

[42800] 16352.Adams, B.; Dorfler, P.; Aguzzi, A.; Kozmik, Z.; Ur-banek, P.; Maurer–Fogy,I.; Busslinger, M.: Pax–5 encodes

the transcription factor BSAP and is expressed in B lymphocytes, the developing CNS, and adult testis. *Genes Dev.* 6: 1589–1607, 1992.

[42801] 16353. Busslinger, M.; Klix, N.; Pfeffer, P.; Graninger, P. G.; Kozmik, Z.: Deregulation of PAX-5 by translocation of the E- μ enhancer of the IgH locus adjacent to two alternative PAX-5 promoters in a diffuse large-cell lymphoma. *Proc. Nat. Acad. Sci.* 93: 6129–6134, 1996.

[42802] 16354. Mikkola, I.; Heavey, B.; Horcher, M.; Busslinger, M.: Reversion of B cell commitment upon loss of Pax5 expression. *Science* 297:110–113, 2002.

[42803] 16355. Nutt, S. L.; Heavey, B.; Rolink, A. G.; Busslinger, M.: Commitment to the B-lymphoid lineage depends on the transcription factor Pax5. *Nature* 401:556–562, 1999.

[42804] 16356. Nutt, S. L.; Vambrie, S.; Steinlein, P.; Kozmik, Z.; Rolink, A.; Weith, A.; Busslinger, M.: Independent regulation of the two Pax5 alleles during B-cell development. *Nature Genet.* 21: 390–395, 1999.

[42805] 16357. Ohno, H.; Furukawa, T.; Fukuhara, S.; Zong, S. Q.; Kamesaki, H.; Shows, T. B.; Le Beau, M. M.; McKeithan, T. W.; Kawakami, T.; Honjo, T.: Molecular analysis of a chromosomal translocation, t(9;14)(p13;q32), in a diffuse large-cell lymphoma cell line expressing the Ki-1 antigen.

Proc.Nat. Acad. Sci. 87: 628–632, 1990.

[42806] 16358.Rolink, A. G.; Nutt, S. L.; Melchers, F.; Busslinger, M.: Long-term in vivo reconstitution of T-cell development by Pax5-deficient B-cell progenitors. *Nature* 401: 603–606, 1999.

[42807] 16359.Vorechovsky, I.; Koskinen, S.; Paganelli, R.; Smith, C. I. E.; Busslinger, M.; Hammarstrom, L.: The PAX5 gene: a linkage and mutation analysis in candidate human primary immunodeficiencies. *Immunogenetics* 42:149–152, 1995.

[42808] 16360.Walther, C.; Guenet, J.-L.; Simon, D.; Deutsch, U.; Jostes, B.; Goulding, M. D.; Plachov, D.; Balling, R.; Gruss, P.: Pax: a murine multigene family of paired box containing genes. *Genomics* 11: 424–434, 1991.

[42809] 16361.Koseki, H.; Zachgo, J.; Mizutani, Y.; Simon-Chazottes, D.; Guenet, J.-L.; Balling, R.; Gossler, A.: Fine genetic mapping of the proximal part of mouse chromosome 2 excludes Pax-8 as a candidate gene for Danforth's short tail (Sd). *Mammalian Genome* 4: 324–327, 1993.

[42810] 16362.Kroll, T. G.; Sarraf, P.; Pecciarini, L.; Chen, C.-J.; Mueller, E.; Splegeman, B. M.; Fletcher, J. A.: PAX8-PPAR-gamma-1 fusion in oncogene human thyroid carcinoma. *Science* 289: 1357–1360, 2000.

[42811] 16363.Macchia, P. E.; Lapi, P.; Krude, H.; Pirro, M. T.; Mis-

sero, C.;Chiovato, L.; Souabni, A.; Baserga, M.; Tassi, V.; Pinchera, A.; Fenzi,G.; Gruters, A.; Busslinger, M.; Di Lauro, R.: PAX8 mutations associatedwith congenital hypothyroidism caused by thyroid dysgenesis. NatureGenet. 19: 83–86, 1998.

[42812] 16364.Mansouri, A.; Chowdhury, K.; Gruss, P.: Follicular cells of thethyroid gland require Pax8 gene function. Nature Genet. 19: 87–90,1998.

[42813] 16365.Pasca di Magliano, M.; Di Lauro, R.; Zannini, M.: Pax8 has a keyrole in thyroid cell differentiation. Proc. Nat. Acad. Sci. 97:13144–13149, 2000.

[42814] 16366.Plachov, D.; Chowdhury, K.; Walther, C.; Simon, D.; Guenet, J.–L.;Gruss, P.: Pax–8, a murine paired box gene expressed in the developingexcretory system and thyroid gland. Development 110: 643–651, 1990.

[42815] 16367.Tell, G.; Pellizzari, L.; Esposito, G.; Pucillo, C.; Macchia,P. E.; Di Lauro, R.; Damante, G.: Structural defects of a Pax8 mutantthat give rise to congenital hypothyroidism. Biochem. J. 341: 89–93,1999.

[42816] 16368.Bongarzone, I.; Vigano, E.; Alberti, L.; Borrello, M. G.; Pasini,B.; Greco, A.; Mondellini, P.; Smith, D. P.; Ponder, B. A. J.; Romeo,G.; Pierotti, M. A.: Full activation of MEN2B mutant RET by an additionalMEN2A mutation or by

ligand GDNF stimulation. *Oncogene* 16: 2295–2301,1998.

[42817] 16369. Carlson, K. M.; Bracamontes, J.; Jackson, C. E.; Clark, R.; Lacroix, A.; Wells, S. A., Jr.; Goodfellow, P. J.: Parent-of-origin effects in multiple endocrine neoplasia type 2B. *Am. J. Hum. Genet.* 55:1076–1082, 1994.

[42818] 16370. Carlson, K. M.; Dou, S.; Chi, D.; Scavarda, N.; Toshima, K.; Jackson, C. E.; Wells, S. A., Jr.; Goodfellow, P. J.; Donis-Keller, H.: Single missense mutation in the tyrosine kinase catalytic domain of the RET protooncogene is associated with multiple endocrine neoplasia type 2B. *Proc. Nat. Acad. Sci.* 91: 1579–1583, 1994.

[42819] 16371. Mulligan, L. M.; Eng, C.; Healey, C. S.; Clayton, D.; Kwok, J. B. J.; Gardner, E.; Ponder, M. A.; Frilling, A.; Jackson, C. E.; Lehnert, H.; Neumann, H. P. H.; Thibodeau, S. N.; Ponder, B. A. J.: Specific mutations of the RET protooncogene are related to disease phenotype in MEN 2A and FMTC. *Nature Genet.* 6: 70–74, 1994.

[42820] 16372. Bruss, M.; Kunz, J.; Lingen, B.; Bonisch, H.: Chromosomal mapping of the human gene for the tricyclic antidepressant-sensitive noradrenaline transporter. *Hum. Genet.* 91: 278–280, 1993.

[42821] 16373. Esler, M.; Jennings, G.; Lambert, G.; Meredith, I.; Horne, M.; Eisenhofer, G.: Overflow of catecholamine neu-

rotransmitters to the circulation: source, fate, and functions. *Physiol. Rev.* 70: 963–985, 1990.

- [42822] 16374. Fritz, J. D.; Jayanthi, L. D.; Thoreson, M. A.; Blakely, R. D.: Cloning and chromosomal mapping of the murine norepinephrine transporter. *J. Neurochem.* 70: 2241–2251, 1998.
- [42823] 16375. Gelernter, J.; Kruger, S.; Kidd, K. K.; Amara, S.: TaqI RFLP at norepinephrine transporter protein (NET) locus. *Hum. Molec. Genet.* 2:820 only, 1993.
- [42824] 16376. Gelernter, J.; Kruger, S.; Pakstis, A. J.; Pacholczyk, T.; Sparkes, R. S.; Kidd, K. K.; Amara, S.: Assignment of the norepinephrine transporter protein (NET1) locus to chromosome 16. *Genomics* 18: 690–692, 1993.

- [42825] 16377.Kaye, W. H.; Jimerson, D. C.; Lake, C. R.; Ebert, M. H.: Altered norepinephrine metabolism following long-term weight recovery in patients with anorexia nervosa. *Psychiat. Res.* 14: 333–342, 1985.
- [42826] 16378.Pacholczyk, T.; Blakely, R. D.; Amara, S. G.: Expression cloning of a cocaine- and antidepressant-sensitive human noradrenaline transporter. *Nature* 350:350–354, 1991.
- [42827] 16379.Paczkowski, F. A.; Bonisch, H.; Bryan-Lluka, L. J.: Pharmacological properties of the naturally occurring ala457pro variant of the human norepinephrine transporter. *Pharmacogenetics* 12: 165–173, 2002.
- [42828] 16380.Pirke, K. M.; Kellner, M.; Philipp, E.; Laessle, R.; Krieg, J.C.; Fichter, M. M.: Plasma norepinephrine after a standardized test meal in acute and remitted patients with anorexia nervosa and in healthy controls. *Biol. Psychiat.* 31: 1074–1077, 1992.
- [42829] 16381.Porzgen, P.; Bonisch, H.; Bruss, M.: Molecular cloning and organization of the coding region of the human norepinephrine transporter gene. *Biochem.Biophys. Res. Commun.* 215: 1145–1150, 1995. Notes: Erratum: *Biochem.Biophys. Res. Commun.* 227: 642–643, 1996.
- [42830] 16382.Shannon, J. R.; Flattem, N. L.; Jordan, J.; Jacob, G.;

Black, B. K.; Biaggioni, I.; Blakely, R. D.; Robertson, D.: Orthostatic intolerance and tachycardia associated with norepinephrine-transporter deficiency. *New Eng. J. Med.* 342: 541–549, 2000.

[42831] 16383. Urwin, R. E.; Bennetts, B.; Wilcken, B.; Lampropoulos, B.; Beumont, P.; Clarke, S.; Russell, J.; Tanner, S.; Nunn, K. P.: Anorexia nervosa (restrictive subtype) is associated with a polymorphism in the novel norepinephrine transporter gene promoter polymorphic region. *Molec. Psychiat.* 7: 652–657, 2002.

[42832] 16384. Tomilin, A.; Remenyi, A.; Lins, K.; Bak, H.; Leidel, S.; Vriend, G.; Wilmanns, M.; Scholer, H. R.: Synergism with the coactivator OBF-1 (OCA-B, BOB-1) is mediated by a specific POU dimer configuration. *Cell* 103: 853–864, 2000.

[42833] 16385. Ko, H.-S.; Fast, P.; McBride, W.; Staudt, L. M.: A human protein specific for the immunoglobulin octamer DNA motif contains a functional homeobox domain. *Cell* 55: 135–144, 1988.

[42834] 16386. Schubart, K.; Massa, S.; Schubart, D.; Corcoran, L. M.; Rolink, A. G.; Matthias, P.: B cell development and immunoglobulin gene transcription in the absence of Oct-2 and OBF-1. *Nature Immun.* 2: 69–74, 2001.

- [42835] 16387. Staudt, L. M.; Clerc, R. G.; Singh, H.; LeBowitz, J. H.; Sharp, P. A.; Baltimore, D.: Cloning of a lymphoid-specific cDNA encoding a protein binding the regulatory octamer DNA motif. *Science* 241:577–580, 1988.
- [42836] 16388. Crouau-Roy, B.; Amadou, C.; Bouissou, C.; Clayton, J.; Vernet, C.; Ribouchon, M.-T.; Pontarotti, P.: Localization of the OTF3 gene within the human MHC class I region by physical and meiotic mapping. *Genomics* 21:241–243, 1994.
- [42837] 16389. Guillaudeux, T.; Mattei, M. G.; Depetris, D.; Le Bouteiller, P.; Pontarotti, P.: In situ hybridization localizes the human OTF3 to chromosome 6p21.3–p22 and OTF3L to 12p13. *Cytogenet. Cell Genet.* 63:212–214, 1993.
- [42838] 16390. Nichols, J.; Zevnik, B.; Anastassiadis, K.; Niwa, H.; Klewe-Nebenius, D.; Chambers, I.; Scholer, H.; Smith, A.: Formation of pluripotent stem cells in the mammalian embryo depends on the POU transcription factor Oct4. *Cell* 95: 379–391, 1998.
- [42839] 16391. Niwa, H.; Miyazaki, J.; Smith, A. G.: Quantitative expression of Oct-3/4 defines differentiation, dedifferentiation or self-renewal of ES cells. *Nature Genet.* 24: 372–376, 2000.
- [42840] 16392. Rosner, M. H.; De Santo, R. J.; Arnheiter, H.; Staudt,

L. M.:Oct-3 is a maternal factor required for the first mouse embryonic division. *Cell* 64: 1103–1110, 1991.

Note: Retraction: *Cell* 69: 724only, 1992.

[42841] 16393.Takeda, J.; Seino, S.; Bell, G. I.: Human Oct3 gene family: cDNA sequences, alternative splicing, gene organization, chromosomal location, and expression at low levels in adult tissues. *Nucleic Acids Res.* 20:4613–4620, 1992.

[42842] 16394.Parker, R. C.; Mardon, G.; Lebo, R. V.; Varmus, H. E.; Bishop, J. M.: Isolation of duplicated human c-src genes located on chromosomes 1 and 20. *Molec. Cell. Biol.* 5: 831–838, 1985.

[42843] 16395.Tronick, S. R.; Popescu, N. C.; Cheah, M. S. C.; Swan, D. C.; Amsbaugh, S. C.; Lengel, C. R.; DiPaolo, J. A.; Robbins, K. C.: Isolation and chromosomal localization of the human fgr protooncogene, a distinct member of the tyrosine kinase gene family. *Proc. Nat. Acad. Sci.* 82:6595–6599, 1985.

[42844] 16396.Krebs, L. T.; Xue, Y.; Norton, C. R.; Shutter, J. R.; Maguire, M.; Sundberg, J. P.; Gallahan, D.; Closson, V.; Kitajewski, J.; Callahan, R.; Smith, G. H.; Stark, K. L.; Gridley, T.: Notch signaling is essential for vascular morphogenesis in mice. *Genes Dev.* 14: 1343–1352, 2000.

[42845] 16397.Li, L.; Huang, G. M.; Banta, A. B.; Deng, Y.; Smith,

T.; Dong,P.; Friedman, C.; Chen, L.; Trask, B. J.; Spies, T.; Rowen, L.; Hood,L.: Cloning, characterization, and the complete 56.8–kilobase DNAsequence of the human NOTCH4 gene. *Genomics* 51: 45–58, 1998.

[42846] 16398.McGinnis, R. E.; Fox, H.; Yates, P.; Cameron, L.–A.; Barnes, M.R.; Gray, I. C.; Spurr, N. K.; Hurko, O.; St. Clair, D.: Failureto confirm NOTCH4 association with schizophrenia in a large population–basedsample from Scotland. *Nature Genet.* 28: 128–129, 2001.

[42847] 16399.Sklar, P.; Schwab, S. G.; Williams, N. M.; Daly, M.; Schaffner,S.; Maier, W.; Albus, M.; Trixler, M.; Eichhammer, P.; Lerer, B.;Hallmayer, J.; Norton, N.; and 11 others: Association analysisof NOTCH4 loci in schizophrenia using family and population–basedcontrols. *Nature Genet.* 28: 126–128, 2001.

[42848] 16400.Sugaya, K.; Sasanuma, S.; Nohata, J.; Kimura, T.; Fukagawa, T.;Nakamura, Y.; Ando, A.; Inoko, H.; Ikemura, T.; Mita, K.: Gene organizationof human NOTCH4 and (CTG)_n polymorphism in this human counterpartgene of mouse proto–oncogene Int3. *Gene* 189: 235–244, 1997.

[42849] 16401.Uyttendaele, H.; Marazzi, G.; Wu, G.; Yan, Q.; Sassoon, D.; Kitajewski,J.: Notch4/int–3, a mammary proto–oncogene, is an endothelial cell–specificmammalian Notch

gene. Development 122: 2251–2259, 1996.

- [42850] 16402.Wei, J.; Hemmings, G. P.: The NOTCH4 locus is associated with susceptibility to schizophrenia. (Letter) Nature Genet. 25: 376–377, 2000.
- [42851] 16403.Bork, P.: The modular architecture of a new family of growth regulators related to connective tissue growth factor. FEBS Lett. 327: 125–130, 1993.
- [42852] 16404.Sugaya, K.; Fukagawa, T.; Matsumoto, K.; Mita, K.; Takahashi, E.; Ando, A.; Inoko, H.; Ikemura, T.: Three genes in the human MHC class III region near the junction with the class II: gene for receptor of advanced glycosylation end products, PBX2 homeobox gene and a Notch homolog, human counterpart of mouse mammary tumor gene int-3. Genomics 23: 408–419, 1994.
- [42853] 16405.Joliot, V.; Martinerie, C.; Dambrine, G.; Plassiart, G.; Brisac, M.; Crochet, J.; Perbal, B.: Proviral rearrangements and overexpression of a new cellular gene (nov) in myeloblastosis-associated virus type 1-induced nephroblastomas. Molec. Cell. Biol. 12: 10–21, 1992.
- [42854] 16406.Kim, H.-S.; Nagalla, S. R.; Oh, Y.; Wilson, E.; Roberts, C. T., Jr.; Rosenfeld, R. G.: Identification of a family of low-affinity insulin-like growth factor binding proteins (IFGBPs): characterization of connective tissue growth

factor as a member of the IGFBP superfamily. *Proc.Nat. Acad. Sci.* 94: 12981–12986, 1997.

[42855] 16407.Martinerie, C.; Perbal, B.: Expression of a gene encoding a novel potential IGF binding protein in human tissues. *C. R. Acad. Sci.(Paris)* 313 (ser. 3): 345–351, 1991.

[42856] 16408.Snaith, M. R.; Natarajan, D.; Taylor, L. B.; Choi, C.-P.; Martinerie, C.; Perbal, B.; Schofield, P. N.; Boulter, C. A.: Genomic structure and chromosomal mapping of the mouse nov gene. *Genomics* 38: 425–428, 1996.

[42857] 16409.Soret, J.; Dambrine, G.; Perbal, B.: Induction of nephroblastoma by myeloblastosis-associated virus type 1: state of proviral DNA in tumor cells. *J. Virol.* 63: 1803–1807, 1989.

[42858] 16410.Amson, R.; Sigaux, F.; Przedborski, S.; Flandrin, G.; Givol, D.; Telerman, A.: The human protooncogene product p33pim is expressed during fetal hematopoiesis and in diverse leukemias. *Proc. Nat. Acad.Sci.* 86: 8857–8861, 1989.

[42859] 16411.Ark, B.; Gummere, G.; Bennett, D.; Artzt, K.: Mapping of the Pim-1 oncogene in mouse t-haplotypes and its use to define the relative map positions of the tcl loci t-0(t-6) and t-w12 and the marker tf(tufted). *Genomics* 10: 385–389, 1991.

- [42860] 16412.Cuypers, H. T.; Selten, G.; Berns, A.; Geurts van Kessel, A. H.M.: Assignment of the human homologue of Pim-1, a mouse gene implicated in leukemogenesis, to the pter-q12 region of chromosome 6. Hum. Genet. 72:262-265, 1986.
- [42861] 16413.Domen, J.; von Lindern, M.; Hermans, A.; Breuer, M.; Grosveld, G.; Berns, A.: Comparison of the human and mouse PIM-1 cDNAs: nucleotide sequence and immunological identification of the in vitro synthesized PIM-1 protein. Oncogene Res. 1: 103-112, 1987.
- [42862] 16414.Boitard, C.; Villa, M. C.; Becourt, C.; Pham Gia, H.; Huc, C.; Sempe, P.; Portier, M. M.; Bach, J. F.: Peripherin: an islet antigen that is cross-reactive with nonobese diabetic mouse class II gene products. Proc. Nat. Acad. Sci. 89: 172-176, 1992.
- [42863] 16415.Leonard, D. G.; Gorham, J. D.; Cole, P.; Greene, L. A.; Ziff, E.B.: A nerve growth factor-regulated messenger RNA encodes a new intermediate filament protein. J. Cell Biol. 106: 181-193, 1988.
- [42864] 16416.Moncla, A.; Landon, F.; Mattei, M.-G.; Portier, M.-M.: Chromosomal localisation of the mouse and human peripherin genes. Genet. Res. 59:125-129, 1992.
- [42865] 16417.Pendleton, J. W.; Violette, S. M.; Hunihan, L. W.;

Greene, L. A.;Ruddle, F. H.: The peripherin gene maps to mouse chromosome 15. *Genomics* 9:369–372, 1991.

[42866] 16418.Portier, M.–M.; de Nechaud, B.; Gros, F.: Peripherin, a new memberof the intermediate filament protein family. *Dev. Neurosci.* 6:335–344, 1984.

[42867] 16419.Thompson, M. A.; Ziff, E. B.: Structure of the gene encoding peripherin,an NGF–regulated neuronal–specific type III intermediate filamentprotein. *Neuron* 2: 1043–1053, 1989.

[42868] 16420.Ben Othmane, K.; Loeb, D.; Hayworth–Hodgte, R.; Hentati, F.; Rao,N.; Roses, A. D.; Ben Hamida, M.; Pericak–Vance, M. A.; Vance, J.M.: Physical and genetic mapping of the CMT4A locus and exclusionof PMP–2 as the defect in CMT4A. *Genomics* 28: 286–290, 1995.

[42869] 16421.Hayasaka, K.; Himoro, M.; Takada, G.; Takahashi, E.; Minoshima,S.; Shimizu, N.: Structure and localization of the gene encodinghuman peripheral myelin protein 2 (PMP2). *Genomics* 18: 244–248,1993.

[42870] 16422.Hayasaka, K.; Nanao, K.; Tahara, M.; Sato, W.; Takada, G.; Miura,M.; Uyemura, K.: Isolation and sequence determination of cDNA encodingP2 protein of human peripheral myelin. *Biochem. Biophys. Res. Commun.* 181:204–207, 1991.

- [42871] 16423.Narayanan, V.; Ripepi, B.; Jabs, E. W.; Hawkins, A.; Griffin, C.;Tennekoon, G.: Partial structure and mapping of the human myelinP2 protein gene. J. Neurochem. 63: 2010–2013, 1994.
- [42872] 16424.Manser, E.; Loo, T.–H.; Koh, C.–G.; Zhao, Z.–S.; Chen, X.–Q.; Tan,L.; Tan, I.; Leung, T.; Lim, L.: PAK kinases are directly coupledto the PIX family of nucleotide ex–change factors. Molec. Cell 1:183–192, 1998.
- [42873] 16425.Nomura, N.; Miyajima, N.; Sazuka, T.; Tanaka, A.; Kawarabayashi,Y.; Sato, S.; Nagase, T.; Seki, N.; Ishikawa, K.; Tabata, S.: Predictionof the coding sequences of unidentified human genes. I. The codingsequences of 40 new genes (KIAA0001–KIAA0040) deduced by analysisof randomly sampled cDNA clones from human immature myeloid cell line,KG–1. DNA Res. 1: 27–35, 1994.
- [42874] 16426.Buggy, J. J.; Sideris, M. L.; Mak, P.; Lorimer, D. D.; McIntosh,B.; Clark, J. M.: Cloning and characterization of a novel human histonedeacetylase, HDAC8. Biochem. J. 350: 199–205, 2000.
- [42875] 16427.Hu, E.; Chen, Z.; Fredrickson, T.; Zhu, Y.; Kirk–patrick, R.; Zhang,G.–F.; Johanson, K.; Sung, C.–M.; Liu, R.; Winkler, J.: Cloning andcharacterization of a novel human class I histone deacetylase thatfunctions as a transcription

repressor. J. Biol. Chem. 275: 15254–15264,2000.

[42876] 16428. Van den Wyngaert, I.; de Vries, W.; Kremer, A.; Neefs, J.-M.; Verhasselt, P.; Luyten, W. H. M. L.; Kass, S. U.: Cloning and characterization of human histone deacetylase 8. FEBS Lett. 478: 77–83, 2000.

[42877] 16429. Grozinger, C. M.; Hassig, C. A.; Schreiber, S. L.: Three proteins define a class of human histone deacetylases related to yeast Hda1p. Proc. Nat. Acad. Sci. 96: 4868–4873, 1999.

[42878] 16430. Hubbert, C.; Guardiola, A.; Shao, R.; Kawaguchi, Y.; Ito, A.; Nixon, A.; Yoshida, M.; Wang, X.-F.; Yao, T.-P.: HDAC6 is a microtubule-associated deacetylase. Nature 417: 455–458, 2002.

[42879] 16431. Mahlke, U.; Schnittger, S.; Landgraf, F.; Schoch, C.; Ottmann, O. G.; Hiddemann, W.; Hoelzer, D.: Assignment of the human histone deacetylase 6 gene (HDAC6) to X chromosome p11.23 by in situ hybridization. Cytogenet. Cell Genet. 93: 135–136, 2001.

[42880] 16432. Nagase, T.; Isikawa, K.; Suyama, M.; Kikuno, R.; Hirose, M.; Miyajima, N.; Tanaka, A.; Kotani, H.; Nomura, N.; Ohara, O.: Prediction of the coding sequences of unidentified human genes. XII. The complete sequences of 100 new cDNA clones from brain which code for large

proteins *in vitro*. DNA Res. 5: 355–364, 1998.

- [42881] 16433. Disteche, C. M.; Brannan, C. I.; Larsen, A.; Adler, D. A.; Schorderet, D. F.; Gearing, D.; Copeland, N. G.; Jenkins, N. A.; Park, L. S.: The human pseudoautosomal GM-CSF receptor alpha subunit gene is autosomal in mouse. Nature Genet. 1: 333–336, 1992.
- [42882] 16434. Sugarman, G. I.; Rimoin, D. L.; Lachman, R. S.: The facial–digital–genital (Aarskog) syndrome. Am. J. Dis. Child. 126: 248–252, 1973.
- [42883] 16435. Tsukahara, M.; Fernandez, G. I.: Umbilical findings in Aarskog syndrome. Clin. Genet. 45: 260–265, 1994.
- [42884] 16436. Tyrkus, M.; Bawle, E.; Lipman, S.; Bozimowski, D.; Woolley, P. V., Jr.: Aarskog–Scott syndrome inherited as an X-linked dominant with full male–female expression. (Abstract) Am. J. Hum. Genet. 32:134A only, 1980.
- [42885] 16437. van den Bergh, P.; Fryns, J. P.; Wilms, G.; Piot, R.; Dralands, G.; van den Bergh, R.: Anomalous cerebral venous drainage in Aarskog syndrome. Clin. Genet. 25: 288–294, 1984.
- [42886] 16438. Zheng, Y.; Fischer, D. J.; Santos, M. F.; Tigyi, G.; Pasteris, N. G.; Gorski, J. L.; Xu, Y.: The faciogenital dysplasia gene product FGD1 functions as a Cdc42Hs-specific guanine–nucleotide exchange factor. J. Biol. Chem. 271:

33169–33172, 1996.

- [42887] 16439.Inaba, H.; Fujimaki, M.; Kazazian, H. H., Jr.; Antonarakis, S.E.: MspI polymorphic site in intron 22 of the factor VIII gene in the Japanese population. *Hum. Genet.* 84: 214–215, 1990.
- [42888] 16440.Levinson, B.; Bermingham, J. R., Jr.; Metzenberg, A.; Kenwrick, S.; Chapman, V.; Gitschier, J.: Sequence of the human factor VIII-associated gene is conserved in mouse. *Genomics* 13: 862–865, 1992.
- [42889] 16441.Levinson, B.; Kenwrick, S.; Lakich, D.; Hammonds, G.; Gitschier, J.: A transcribed gene in an intron of the human factor VIII gene. *Genomics* 7:1–11, 1990.
- [42890] 16442.Peters, M. F.; Ross, C. A.: Isolation of a 40-kDa huntingtin-associated protein. *J. Biol. Chem.* 276: 3188–3194, 2001.
- [42891] 16443.Berra, B.; Gornati, R.; Rapelli, S.; Gatti, R.; Mancini, G. M.S.; Ciana, G.; Bembi, B.: Infantile sialic acid storage disease: biochemical studies. *Am. J. Med. Genet.* 58: 24–31, 1995.
- [42892] 16444.Cameron, P. D.; Dubowitz, V.; Besley, G. T. N.; Fensom, A. H.: Sialic acid storage disease. *Arch. Dis. Child.* 65: 314–315, 1990.
- [42893] 16445.Haataja, L.; Schleutker, J.; Laine, A.-P.; Renlund, M.;

Savontaus, M.-L.; Dib, C.; Weissenbach, J.; Peltonen, L.; Aula, P.: The genetic locus for free sialic acid storage disease maps to the long arm of chromosome 6. *Am. J. Hum. Genet.* 54: 1042–1049, 1994.

[42894] 16446. Fojo, A.; Lebo, R.; Shimizu, N.; Chin, J. E.; Roninson, I. B.; Merlino, G. T.; Gottesman, M. M.; Pastan, I.: Localization of multidrug resistance-associated DNA sequences to human chromosome 7. *Somat. Cell Molec. Genet.* 12: 415–420, 1986.

[42895] 16447. Fojo, A. T.; Ueda, K.; Slamon, D. J.; Poplack, D. G.; Gottesman, M. M.; Pastan, I.: Expression of a multidrug-resistance gene in human tumors and tissues. *Proc. Nat. Acad. Sci.* 84: 265–269, 1987.

[42896] 16448. Fujii, J.; Zarain-Herzberg, A.; Willard, H. F.; Tada, M.; MacLennan, D. H.: Structure of the rabbit phospholamban gene, cloning of the human cDNA, and assignment of the gene to human chromosome 6. *J. Biol. Chem.* 266: 11669–11675, 1991.

[42897] 16449. McTiernan, C. F.; Frye, C. S.; Lemster, B. H.; Kinder, E. A.; Ogletree-Hughes, M. L.; Moravec, C. S.; Feldman, A. M.: The human phospholamban gene: structure and expression. *J. Molec. Cell Cardiol.* 31: 679–692, 1999.

[42898] 16450. Otsu, K.; Fujii, J.; Periasamy, M.; DiFilippantonio,

M.; Uppender, M.; Ward, D. C.; MacLennan, D. H.: Chromosome mapping of five human cardiac and skeletal muscle sarcoplasmic reticulum protein genes. *Genomics* 17:507–509, 1993.

[42899] 16451. Barhanin, J.; Lesage, F.; Guillemare, E.; Fink, M.; Lazdunski, M.; Romey, G.: K(v)LQT1 and IsK (minK) proteins associate to form the I(Ks) cardiac potassium current. *Nature* 384: 78–80, 1996.

[42900] 16452. Marx, S. O.; Kurokawa, J.; Reiken, S.; Motoike, H.; D'Armiento, J.; Marks, A. R.; Kass, R. S.: Requirement of a macromolecular signaling complex for beta adrenergic receptor modulation of the KCNQ1–KCNE1 potassium channel. *Science* 295: 496–499, 2002.

[42901] 16453. Sanguinetti, M. C.; Curran, M. E.; Zou, A.; Shen, J.; Spector, P. S.; Atkinson, D. L.; Keating, M. T.: Coassembly of K(v)LQT1 and minK (IsK) proteins to form cardiac I(Ks) potassium channel. *Nature* 384:80–83, 1996.

[42902] 16454. Coleman, R. A.; Smith, W. L.; Narumiya, S.: VIII. International union of pharmacology classification of prostanoid receptors: properties, distribution, and structure of the receptors and their subtypes. *Pharm.Rev.* 46: 205–229, 1994.

[42903] 16455. Duncan, A. M. V.; Anderson, L. L.; Funk, C. D.;

Abramovitz, M.;Adam, M.: Chromosomal localization of the human prostanoid receptorgene family. *Genomics* 25: 740–742, 1995.

- [42904] 16456.Nagata, A.; Suzuki, Y.; Igarashi, M.; Eguchi, N.; Toh, H.; Urade,Y.; Hayaishi, O.: Human brain prostaglandin D synthase has been evolutionarilydifferentiated from lipophilic–ligand carrier proteins. *Proc. Nat.Acad. Sci.* 88: 4020–4024, 1991.
- [42905] 16457.Onoe, H.; Ueno, R.; Fujita, I.; Nishino, H.; Oomura, Y.; Hayashi,O.:*Proc. Nat. Acad. Sci.* 85: 4082–4086, 1988.
- [42906] 16458.Pinzar, E.; Kanaoka, Y.; Inui, T.; Eguchi, N.; Urade, Y.; Hayaishi,O.: Prostaglandin D synthase gene is involved in the regulation ofnon–rapid eye movement sleep. *Proc. Nat. Acad. Sci.* 97: 4903–4907,2000.
- [42907] 16459.Tanaka, T.; Urade, Y.; Kimura, H.; Eguchi, N.; Nishikawa, A.; Hayaishi,O.: Lipocalin–type prostaglandin D synthase (beta–trace) is a newlyrecognized type of retinoid transporter. *J. Biol. Chem.* 272: 15789–15795,1997.
- [42908] 16460.White, D. M.; Mikol, D. D.; Espinosa, R.; Weimer, B.; Le Beau,M. M.; Stefansson, K.: Structure and chromosomal localization ofthe human gene for a brain form of prostaglandin D–2 synthase. *J.Biol. Chem.* 267:

23202–23208, 1992.

- [42909] 16461.Sales, K. J.; Katz, A. A.; Davis, M.; Hinz, S.; Soeters, R. P.; Hofmeyr, M. D.; Millar, R. P.; Jabbour, H. N.: Cyclooxygenase-2 expression and prostaglandin E2 synthesis are up-regulated in carcinomas of the cervix: a possible autocrine/paracrine regulation of neoplastic cell function via EP2/EP4 receptors. *J. Clin. Endocr. Metab.* 86: 2243–2249, 2001.
- [42910] 16462.Smock, S. L.; Pan, L. C.; Castleberry, T. A.; Lu, B.; Mather, R.J.; Owen, T. A.: Cloning, structural characterization, and chromosomal localization of the gene encoding the human prostaglandin E2 receptor EP2 subtype. *Gene* 237: 393–402, 1999.
- [42911] 16463.Taketo, M.; Rochelle, J. M.; Sugimoto, Y.; Namba, T.; Honda, A.; Negishi, M.; Ichikawa, A.; Narumiya, S.; Seldin, M. F.: Mapping of the genes encoding mouse thromboxane A2 receptor and prostaglandin E receptor subtypes EP2 and EP3. *Genomics* 19: 585–588, 1994.
- [42912] 16464.Tilley, S. L.; Audoly, L. P.; Hicks, E. H.; Kim, H.-S.; Flannery, P. J.; Coffman, T. M.; Koller, B. H.: Reproductive failure and reduced blood pressure in mice lacking the EP2 prostaglandin E-2 receptor. *J. Clin. Invest.* 103: 1539–1545, 1999.

- [42913] 16465.Catella-Lawson, F.; Reilly, M. P.; Kapoor, S. C.; Cucchiara, A.J.; DeMarco, S.; Tournier, B.; Vyas, S. N.; FitzGerald, G. A.: Cyclooxygenaseinhibitors and the antiplatelet effects of aspirin. *New Eng. J. Med.* 345:1809–1817, 2001.
- [42914] 16466.Dube, J.-N.; Drouin, J.; Aminian, M.; Plant, M. H.; Laneuville,O.: Characterization of a partial prostaglandin endoperoxide H synthase-1deficiency in a patient with a bleeding disorder. *Brit. J. Haemat.* 113:878–885, 2001.
- [42915] 16467.Funk, C. D.; Funk, L. B.; Kennedy, M. E.; Pong, A. S.; Fitzgerald,G. A.: Human platelet/erythro leukemia cell prostaglandin G/H synthase:cDNA cloning, expression, and gene chromosomal assignment. *FASEBJ.* 5: 2304–2312, 1991.
- [42916] 16468.Gavett, S. H.; Madison, S. L.; Chulada, P. C.; Scarborough, P.E.; Qu, W.; Boyle, J. E.; Tiano, H. F.; Lee, C. A.; Langenbach, R.;Roggli, V. L.; Zeldin, D. C.: Allergic lung responses are increasedin prostaglandin H synthase-deficient mice. *J. Clin. Invest.* 104:721–732, 1999.
- [42917] 16469.Kirschenbaum, A.; Liotta, D. R.; Yao, S.; Liu, X.-H.; Klausner,A. P.; Unger, P.; Shapiro, E.; Leav, I.; Levine, A. C.: Immunohistochemicallocalization of cyclooxygenase-1 and cyclooxygenase-2 in the humanfetal and adult male

reproductive tracts. J. Clin. Endocr. Metab. 85:3436–3441, 2000.

[42918] 16470. Langenbach, R.; Morham, S. G.; Tiano, H. F.; Loftin, C. D.; Ghanayem, B. I.; Chulada, P. C.; Mahler, J. F.; Lee, C. A.; Goulding, E. H.; Kluckman, K. D.; Kim, H. S.; Smithies, O.: Prostaglandin synthase 1 gene disruption in mice reduces arachidonic acid-induced inflammation and indomethacin-induced gastric ulceration. Cell 83: 483–492, 1995.

[42919] 16471. Malkowski, M. G.; Ginell, S. L.; Smith, W. L.; Garavito, R. M.: The productive conformation of arachidonic acid bound to prostaglandin synthase. Science 289: 1933–1937, 2000.

[42920] 16472. Pareti, F. I.; Manucci, P. M.; D'Angelo, A.; Smith, J. B.; Sautebin, L.; Galli, G.: Congenital deficiency of thromboxane and prostacyclin. Lancet I: 898–900, 1980.

[42921] 16473. Picot, D.; Loll, P. J.; Garavito, R. M.: The x-ray crystal structure of the membrane protein prostaglandin H-2 synthase-1. Nature 367: 243–249, 1994.

[42922] 16474. Allen, J. M.; Bloom, S. R.: Neuropeptide Y: a putative neurotransmitter. Neurochem. Int. 8: 1–8, 1986.

[42923] 16475. Niswender, K. D.; Morton, G. J.; Stearns, W. H.; Rhodes, C. J.; Myers, M. G., Jr.; Schwartz, M. W.: Key en-

zyme in leptin-induced anorexia. *Nature* 413: 794–795, 2001.

[42924] 16476. Shimomura, I.; Matsuda, M.; Hammer, R. E.; Bashmakov, Y.; Brown, M. S.; Goldstein, J. L.: Decreased IRS-2 and increased SREBP-1c lead to mixed insulin resistance and sensitivity in livers of lipodystrophic and ob/ob mice. *Molec. Cell* 6: 77–86, 2000.

[42925] 16477. Siracusa, L. D.; Rosner, M. H.; Vigano, M. A.; Gilbert, D. J.; Staudt, L. M.; Copeland, N. G.; Jenkins, N. A.: Chromosomal location of the octamer transcription factors, Otf-1, Otf-2, and Otf-3, defines multiple Otf-3-related sequences dispersed in the mouse genome. *Genomics* 10: 313–326, 1991.

[42926] 16478. Sturm, R. A.; Eyre, H. J.; Baker, E.; Sutherland, G. R.: The human OTF1 locus which overlaps the CD3Z gene is located at 1q22–q23. *Cytogenet. Cell Genet.* 68: 231–232, 1995.

[42927] 16479. Marshall, J. B.; Diaz-Arias, A. A.; Bochna, G. S.; Voge, K. A.: Achalasia due to diffuse esophageal leiomyomatosis and inherited as an autosomal dominant disorder. *Gastroenterology* 98: 1358–1365, 1990.

[42928] 16480. Mattei, M. G.; d'Auriol, L.; Andre, C.; Passage, E.; Mattei, J. F.; Galibert, F.: Assignment of the human c-kit

proto-oncogene to the q11–q12 region of chromosome 4, using in situ hybridization. (Abstract) *Cytogenet. Cell Genet.* 46: 657, 1987.

- [42929] 16481. Nagata, H.; Worobec, A. S.; Oh, C. K.; Chowdhury, B. A.; Tannenbaum, S.; Suzuki, Y.; Metcalfe, D. D.: Identification of a point mutation in the catalytic domain of the protooncogene c-kit in peripheral blood mononuclear cells of patients who have mastocytosis with an associated hematologic disorder. *Proc. Nat. Acad. Sci.* 92: 10560–10564, 1995.
- [42930] 16482. Nishida, T.; Hirota, S.; Taniguchi, M.; Hashimoto, K.; Isozaki, K.; Nakamura, H.; Kanakura, Y.; Tanaka, T.; Takabayashi, A.; Matsuda, H.; Kitamura, Y.: Familial gastrointestinal stromal tumours with germline mutation of the KIT gene. (Letter) *Nature Genet.* 19: 323–324, 1998.
- [42931] 16483. Nocka, K.; Tan, J. C.; Chiu, E.; Chu, T. Y.; Ray, P.; Traktman, P.; Besmer, P.: Molecular bases of dominant negative and loss of function mutations at the murine c-kit/white spotting locus: W-37, W-v, W-41 and W. *EMBO J.* 9: 1805–1813, 1990.
- [42932] 16484. Nomura, K.; Hatayama, I.; Narita, T.; Kaneko, T.; Shiraishi, M.: A novel KIT gene missense mutation in a Japanese family with piebaldism. (Letter) *J. Invest. Derm.*

111: 337–338, 1998.

[42933] 16485. Pignon, J.-M.; Giraudier, S.; Duquesnoy, P.; Jouault, H.; Imbert, M.; Vainchenker, W.; Vernant, J.-P.; Tulliez, M.: A new c-kit mutation in a case of aggressive mast cell disease. *Brit. J. Haemat.* 96:374–376, 1997.

[42934] 16486. Reinsch, N.; Thomsen, H.; Xu, N.; Brink, M.; Looft, C.; Kalm, E.; Brockmann, G. A.; Grupe, S.; Kuhn, C.; Schwerin, M.; Leyhe, B.; Hiendleder, S.; Erhardt, G.; Medjugorac, I.; Russ, I.; Forster, M.; Reents, R.; Averdunk, G.: A QTL for the degree of spotting in cattle shows synteny with the KIT locus on chromosome 6. *J. Hered.* 90:629–634, 1999.

[42935] 16487. Selmanowitz, V. J.; Rabinowitz, A. D.; Orentreich, N.; Wenk, E.: Pigmentary correction of piebaldism by autografts. I. Procedures and clinical findings. *J. Derm. Surg. Oncol.* 3: 615–622, 1977.

[42936] 16488. Spritz, R. A.; Beighton, P.: Piebaldism with deafness: molecular evidence for an expanded syndrome. *Am. J. Med. Genet.* 75: 101–103, 1998.

[42937] 16489. Spritz, R. A.; Droetto, S.; Fukushima, Y.: Deletion of the KIT and PDGFRA genes in a patient with piebaldism. *Am. J. Med. Genet.* 44:492–495, 1992.

[42938] 16490. Spritz, R. A.; Giebel, L. B.: Mutation of the c-kit (mast/stem cell growth factor receptor) proto-oncogene in

human piebaldism. (Abstract) Am.J. Hum. Genet. 49 (suppl.): 38, 1991.

[42939] 16491.Spritz, R. A.; Giebel, L. B.; Holmes, S. A.: Dominant negative and loss of function mutations of the c-kit (mast/stem cell growth factor receptor) proto-oncogene in human piebaldism. Am. J. Hum. Genet. 50: 261–269, 1992.

[42940] 16492.Spritz, R. A.; Holmes, S. A.; Ramesar, R.; Greenberg, J.; Curtis, D.; Beighton, P.: Mutations of the KIT (mast/stem cell growth factor receptor) proto-oncogene account for a continuous range of phenotypes in human piebaldism. Am. J. Hum. Genet. 51: 1058–1065, 1992.

[42941] 16493.Syrris, P.; Malik, N. M.; Murday, V. A.; Patton, M. A.; Carter, N. D.; Hughes, H. E.; Metcalfe, K.: Three novel mutations of the proto-oncogene KIT cause human piebaldism. (Letter) Am. J. Med. Genet. 95:79–81, 2000.

[42942] 16494.Tan, J. C.; Nocka, K.; Ray, P.; Traktman, P.; Besmer, P.: The dominant W42 spotting phenotype results from a missense mutation in the c-kit receptor kinase. Science 247: 209–212, 1990.

[42943] 16495.Taylor, M. L.; Dastych, J.; Sehgal, D.; Sundstrom, M.; Nilsson, G.; Akin, C.; Mage, R. G.; Metcalfe, D. D.: The Kit-activating mutation D816V enhances stem cell factor-

dependent chemotaxis. *Blood* 98:1195–1199, 2001.

[42944] 16496. Thomsen, L.; Robinson, T. L.; Lee, J. C. F.; Farraway, L. A.; Hughes, M. J. G.; Andrews, D. W.; Huizinga, J. D.: Interstitial cells of Cajal generate a rhythmic pacemaker current. *Nature Med.* 4: 848–851, 1998.

[42945] 16497. Tian, Q.; Frierson, H. F., Jr.; Krystal, G. W.; Moskaluk, C. A.: Activating c-kit gene mutations in human germ cell tumors. *Am. J. Path.* 154: 1643–1647, 1999.

[42946] 16498. Tsujimura, T.; Furitsu, T.; Morimoto, M.; Isozaki, K.; Nomura, S.; Matsuzawa, Y.; Kitamura, Y.; Kanakura, Y.: Ligand-independent activation of c-kit receptor tyrosine kinase in a murine mastocytoma cell line P-815 generated by a point mutation. *Blood* 83: 2619–2626, 1994.

[42947] 16499. Vandenbark, G. R.; deCastro, C. M.; Taylor, H.; Dew-Knight, S.; Kaufman, R. E.: Cloning and structural analysis of the human c-kit gene. *Oncogene* 7: 1259–1266, 1992.

[42948] 16500. Worobec, A. S.; Semere, T.; Nagata, H.; Metcalfe, D. D.: Clinical correlates of the presence of the asp816val c-kit mutation in the peripheral blood mononuclear cells of patients with mastocytosis. *Cancer* 83:2120–2129, 1998.

[42949] 16501. Yarden, Y.; Kuang, W.-J.; Yang-Feng, T.; Coussens, L.; Munemitsu, S.; Dull, T. J.; Chen, E.; Schlessinger, J.;

Francke, U.; Ullrich,A.: Human proto-oncogene c-kit: a new cell surface receptor tyrosinekinase for an unidentified ligand. EMBO J. 6: 3341–3351, 1987.

[42950] 16502.Cheah, M. S. C.; Ley, T. J.; Tronick, S. R.; Robbins, K. C.: Fgrproto-oncogene mRNA induced in B lymphocytes by Epstein–Barr virusinfection. Nature 319: 238–240, 1986.

[42951] 16503.Dracopoli, N. C.; Stanger, B. Z.; Lager, M.; Housman, D. E.: Localizationof the FGR protooncogene on the genetic linkage map of human chromosome1p. Genomics 3: 124–128, 1988.

[42952] 16504.Le Beau, M. M.; Westbrook, C. A.; Diaz, M. O.; Rowley, J. D.:Evidence for two distinct c-src loci on human chromosomes 1 and 20. Nature 312:70–71, 1984.

[42953] 16505.Lebo, R. V.; Cheung, M.–C.; Bruce, B. D.: Rapid gene mapping bydual laser chromosome sorting and spot blot DNA analysis. (Abstract) Am.J. Hum. Genet. 36: 101S, 1984.

[42954] 16506.Nishizawa, M.; Semba, K.; Yoshida, M. C.; Yamamoto, T.; Sasaki,M.; Toyoshima, K.: Structure, expression, and chromosomal locationof the human c-fgr gene. Molec. Cell. Biol. 6: 511–517, 1986.

[42955] 16507.Muyan, M.; Furuhashi, M.; Sugahara, T.; Boime, I.:

The carboxy-terminal region of the beta-subunits of luteinizing hormone and chorionic gonadotropin differentially influence secretion and assembly of the heterodimers. *Molec. Endocr.* 10: 1678–1687, 1996.

[42956] 16508. Ye, K.; Aghdasi, B.; Luo, H. R.; Moriarty, J. L.; Wu, F. Y.; Hong, J. J.; Hurt, K. J.; Bae, S. S.; Suh, P.-G.; Snyder, S. H.: Phospholipase C-gamma-1 is a physiological guanine nucleotide exchange factor for the nuclear GTPase PIKE. *Nature* 415: 541–544, 2002.

[42957] 16509. Tsujii, M.; Kawano, S.; Tsuji, S.; Sawaoka, H.; Hori, M.; DuBois, R. N.: Cyclooxygenase regulates angiogenesis induced by colon cancer cells. *Cell* 93: 705–716, 1998.

[42958] 16510. Vane, J. R.; Mitchell, J. A.; Appleton, I.; Tomlinson, A.; Bishop-Bailey, D.; Croxtall, J.; Willoughby, D. A.: Inducible isoforms of cyclooxygenase and nitric-oxide synthase in inflammation. *Proc. Nat. Acad. Sci.* 91: 2046–2050, 1994.

[42959] 16511. Yokoyama, C.; Tanabe, T.: Cloning of human gene encoding prostaglandin endoperoxide synthase and primary structure of the enzyme. *Biochem. Biophys. Res. Commun.* 165: 888–894, 1989.

[42960] 16512. Adam, M.; Boie, Y.; Rushmore, T. H.; Muller, G.; Bastien, L.; McKee, K. T.; Metters, K. M.; Abramovitz, M.:

Cloning and expression of three isoforms of the human EP(3) prostanoïd receptor. FEBS Lett. 338:170–174, 1994.

[42961] 16513. Kotani, M.; Tanaka, I.; Ogawa, Y.; Suganami, T.; Matsumoto, T.; Muro, S.; Yamamoto, Y.; Sugawara, A.; Yoshimasa, Y.; Sagawa, N.; Narumiya, S.; Nakao, K.: Multiple signal transduction pathways through two prostaglandin E receptor EP3 subtype isoforms expressed in human uterus. J. Clin. Endocr. Metab. 85: 4315–4322, 2000.

[42962] 16514. Kotani, M.; Tanaka, I.; Ogawa, Y.; Usui, T.; Mori, K.; Ichikawa, A.; Narumiya, S.; Yoshimi, T.; Nakao, K.: Molecular cloning and expression of multiple isoforms of human prostaglandin E receptor EP(3) subtype generated by alternative messenger RNA splicing: multiple second messenger systems and tissue-specific distributions. Molec. Pharm. 48: 869–879, 1995.

[42963] 16515. Kotani, M.; Tanaka, I.; Ogawa, Y.; Usui, T.; Tamura, N.; Mori, K.; Narumiya, S.; Yoshimi, T.; Nakao, K.: Structural organization of the human prostaglandin EP(3) receptor subtype gene (PTGER3). Genomics 40:425–434, 1997.

[42964] 16516. Schmid, A.; Thierach, K.-H.; Schleuning, W.-D.; Dinter, H.: Splice variants of the human EP(3) receptor for prostaglandin E(2). Europ. J. Biochem. 228: 23–30, 1995.

- [42965] 16517.Ushikubi, F.; Segi, E.; Sugimoto, Y.; Murata, T.; Matsuoka, T.;Kobayashi, T.; Hizaki, H.; Tuboi, K.; Katsuyama, M.; Ichikawa, A.;Tanaka, T.; Yoshida, N.; Narumiya, S.: Impaired febrile response in mice lacking the prostaglandin E receptor subtype EP(3). *Nature* 395:281–284, 1998.
- [42966] 16518.Wurz, H.; Geiger, W.; Kunzig, H. J.; Jabs–Lehmann, A.; Bohn, H.;Luben, G.: Radioimmunoassay of SP1 (pregnancy–specific beta–1–glycoprotein) in maternal blood and in amniotic fluid in normal and pathologic pregnancies. *J.Perinat. Med.* 2: 67–78, 1981.
- [42967] 16519.Kaplan, R.; Morse, B.; Huebner, K.; Croce, C.; Howk, R.; Ravera,M.; Ricca, G.; Jaye, M.; Schlessinger, J.: Cloning of three humantyrrosine phosphatases reveals a multigene family of receptor–linkedprotein–tyrosine–phosphatases expressed in brain. *Proc. Nat. Acad.Sci.* 87: 7000–7004, 1990.
- [42968] 16520.Banville, D.; Stocco, R.; Shen, S.–H.: Human protein tyrosinephosphatase 1C (PTPN6) gene structure: alternate promoter usage and exon skipping generate multiple transcripts. *Genomics* 27: 165–173,1995.
- [42969] 16521.Beghini, A.; Ripamonti, C. B.; Peterlongo, P.; Roversi, G.; Cairoli,R.; Morra, E.; Larizza, L.: RNA hypered–

iting and alternative splicing of hematopoietic cell phosphatase (PTPN6) gene in acute myeloid leukemia.

Hum.Molec. Genet. 9: 2297–2304, 2000.

[42970] 16522. Matthews, R. J.; Bowne, D. B.; Flores, E.; Thomas, M. L.: Characterization of hematopoietic intracellular protein tyrosine phosphatases: description of a phosphatase containing an SH2 domain and another enriched in proline-, glutamic acid-, serine-, and threonine-rich sequences. Molec. Cell. Biol. 12: 2396–2405, 1992.

[42971] 16523. Plutzky, J.; Neel, B. G.; Rosenberg, R. D.; Eddy, R. L.; Byers, M. G.; Jani-Sait, S.; Shows, T. B.: Chromosomal localization of an SH2-containing tyrosine phosphatase (PTPN6). Genomics 13: 869–872, 1992.

[42972] 16524. Shen, S.-H.; Bastien, L.; Posner, B. I.; Chretien, P.: A protein-tyrosine phosphatase with sequence similarity to the SH2 domain of the protein-tyrosine kinases. Nature 352: 736–739, 1991.

[42973] 16525. Shultz, L. D.; Schweitzer, P. A.; Rajan, T. V.; Yi, T.; Ihle, J. N.; Matthews, R. J.; Thomas, M. L.; Beier, D. R.: Mutations at the murine *hemo* locus are within the hematopoietic cell protein-tyrosine phosphatase (*Hcph*) gene. Cell 73: 1445–1454, 1993.

[42974] 16526. Tsui, H. W.; Siminovitch, K. A.; de Souza, L.; Tsui, F.

W. L.: Motheaten and viable motheaten mice have mutations in the haematopoietic cell phosphatase gene. *Nature Genet.* 4: 124–129, 1993.

[42975] 16527. Yi, T.; Cleveland, J. L.; Ihle, J. N.: Identification of novel protein tyrosine phosphatases of hematopoietic cells by PCR amplification. *Blood* 78: 2222–2228, 1991.

[42976] 16528. Yi, T.; Cleveland, J. L.; Ihle, J. N.: Protein tyrosine phosphatase containing SH2 domains: characterization, preferential expression in hematopoietic cells, and localization to human chromosome 12p12–p13. *Molec. Cell. Biol.* 12: 836–846, 1992.

[42977] 16529. Yi, T.; Gilbert, D. J.; Jenkins, N. A.; Copeland, N. G.; Ihle, J. N.: Assignment of a novel protein tyrosine phosphatase gene (Hcph) to mouse chromosome 6. *Genomics* 14: 793–795, 1992.

[42978] 16530. Jirik, F. R.; Anderson, L. L.; Duncan, A. M. V.: The human protein-tyrosine phosphatase PTP-alpha/LRP gene (PTPA) is assigned to chromosome 20p13. *Cytogenet. Cell Genet.* 60: 117–118, 1992.

[42979] 16531. Jirik, F. R.; Janzen, N. M.; Melhado, I. G.; Harder, K. W.: Cloning and chromosomal assignment of a widely expressed human receptor-like protein-tyrosine phosphatase. *FEBS Lett.* 273: 239–242, 1990.

- [42980] 16532. Matthews, R. J.; Cahir, E. D.; Thomas, M. L.: Identification of an additional member of the protein-tyrosine-phosphatase family: evidence for alternative splicing in the tyrosine phosphatase domain. *Proc. Nat. Acad. Sci.* 87: 4444–4448, 1990.
- [42981] 16533. Rao, V. V. N. G.; Loffler, C.; Sap, J.; Schlessinger, J.; Hansmann, I.: The gene for receptor-linked protein-tyrosine-phosphatase (PTPA) is assigned to human chromosome 20p12-pter by in situ hybridization (ISH and FISH). *Genomics* 13: 906–907, 1992.
- [42982] 16534. Schnittger, S.; Rao, V. V. N. G.; Deutsch, U.; Gruss, P.; Balling, R.; Hansmann, I.: PAX1, a member of the paired box-containing class of developmental control genes, is mapped to human chromosome 20p11.2 by in situ hybridization (ISH and FISH). *Genomics* 14: 740–744, 1992.
- [42983] 16535. Ahmad, F.; Azevedo, J. L., Jr.; Cortright, R.; Dohm, G. L.; Goldstein, B. J.: Alterations in skeletal muscle protein-tyrosine phosphatase activity and expression in insulin-resistant human obesity and diabetes. *J. Clin. Invest.* 100: 449–458, 1997.
- [42984] 16536. Brown-Shimer, S.; Johnson, K. A.; Lawrence, J. B.; Johnson, C.; Bruskin, A.; Green, N. R.; Hill, D. E.: Molecular cloning and chromosomal mapping of the human gene en-

coding protein phosphotyrosyl phosphatase1B. Proc. Nat. Acad. Sci. 87: 5148–5152, 1990.

[42985] 16537.Charbonneau, H.; Tonks, N. K.; Kumar, S.; Diltz, C. D.; Harrylock,M.; Cool, D. E.; Krebs, E. G.; Fischer, E. H.; Walsh, K. A.: Humanplacenta protein-tyrosine-phosphatase: amino acid sequence and relationship to a family of receptor-like proteins. Proc. Nat. Acad. Sci. 86:5252–5256, 1989.

[42986] 16538.Chernoff, J.; Schievella, A. R.; Jost, C. A.; Erikson, R. L.; Neel,B. G.: Cloning of a cDNA for a major human protein-tyrosine-phosphatase. Proc.Nat. Acad. Sci. 87: 2735–2739, 1990.

[42987] 16539.Matilla, A.; Gorbea, C.; Einum, D. D.; Townsend, J.; Michalik,A.; van Broeckhoven, C.; Jensen, C. C.; Murphy, K. J.; Ptacek, L.J.; Fu, Y.–H.: Association of ataxin–7 with the proteasome subunitS4 of the 19S regulatory complex. Hum. Molec. Genet. 10: 2821–2831,2001.

[42988] 16540.Cazzaniga, G.; Tosi, S.; Aloisi, A.; Giudici, G.; Daniotti, M.;Pioltelli, P.; Kearney, L.; Biondi, A.: The tyrosine kinase Abl-relatedgene ARG is fused to ETV6 in an AML–M4Eo patient with a t(1;12)(q25;p13):molecular cloning of both reciprocal transcripts. Blood 94: 4370–4373,1999.

[42989] 16541.Iijima, Y.; Ito, T.; Oikawa, T.; Eguchi, M.; Eguchi–

Ishimae, M.; Kamada, N.; Kishi, K.; Asano, S.; Sakaki, Y.; Sato, Y.: A new ETV6/TEL partner gene, ARG (ABL-related gene or ABL2), identified in an AML-M3 cell line with a t(1;12)(q25;p13) translocation. Blood 95: 2126–2131, 2000.

[42990] 16542. Joazeiro, C. A. P.; Wing, S. S.; Huang, H.; Levenson, J. D.; Hunter, T.; Liu, Y.-C.: The tyrosine kinase negative regulator c-Cbl as a RING-type, E2-dependent ubiquitin-protein ligase. Science 286: 309–312, 1999.

[42991] 16543. Langdon, W. Y.; Hartley, J. W.; Klinken, S. P.; Ruscetti, S. K.; Morse, H. C., III: v-cbl, an oncogene from a dual-recombinant murine retrovirus that induces early B-lineage lymphomas. Proc. Nat. Acad. Sci. 86: 1168–1172, 1989.

[42992] 16544. Mastick, C. C.; Brady, M. J.; Saltiel, A. R.: Insulin stimulates the tyrosine phosphorylation of caveolin. J. Cell Biol. 129: 1523–1531, 1995.

[42993] 16545. Petrelli, A.; Gilestro, G. F.; Lanzardo, S.; Comoglio, P. M.; Migone, N.; Giordano, S.: The endophilin-CIN85-Cbl complex mediates ligand-dependent downregulation of c-Met. Nature 416: 187–190, 2002.

[42994] 16546. Savage, P. D.; Shapiro, M.; Langdon, W. Y.; Geurts van Kessel, A. D.; Seunemann, H. N.; Akao, Y.; Croce, C.;

Morse, H. C., III; Kersey, J. H.: Relationship of the human protooncogene CBL2 on 11q23 to the t(4;11), t(11;22), and t(11;14) breakpoints. *Cytogenet. Cell Genet.* 56:112–115, 1991.

[42995] 16547. Soubeyran, P.; Kowanetz, K.; Szymkiewicz, I.; Langdon, W. Y.; Dikic, I.: Cbl–CIN85–endophilin complex mediates ligand–induced downregulation of EGF receptors. *Nature* 416: 183–187, 2002.

[42996] 16548. Thien, C. B. F.; Walker, F.; Langdon, W. Y.: RING finger mutations that abolish c–Cbl–directed polyubiquitination and downregulation of the EGF receptor are insufficient for cell transformation. *Molec. Cell* 7: 355–365, 2001.

[42997] 16549. Wei, S.; Rocchi, M.; Archidiacono, N.; Sacchi, N.; Romeo, G.; Gatti, R. A.: Physical mapping of the human chromosome 11q23 region containing the ataxia–telangiectasia locus. *Cancer Genet. Cytogenet.* 46:1–8, 1990.

[42998] 16550. Yoon, C. H.; Lee, J.; Jongeward, G. D.; Sternberg, P. W.: Similarity of sli–1, a regulator of vulval development in *C. elegans*, to the mammalian proto–oncogene c–cbl. *Science* 269: 1102–1105, 1995.

[42999] 16551. Cannizzaro, L. A.; Madaule, P.; Hecht, F.; Axel, R.;

Croce, C.M.; Huebner, K.: Chromosome localization of human ARH genes, a ras-related gene family. *Genomics* 6: 197–203, 1990.

[43000] 16552.Chardin, P.; Madaule, P.; Tavitian, A.: Coding sequence of human rho cDNAs clone 6 and clone 9. *Nucleic Acid Res.* 16: 2717 only, 1988.

[43001] 16553.Madaule, P.; Axel, R.: A novel ras-related gene family. *Cell* 41:31–40, 1985.

[43002] 16554.Maekawa, M.; Ishizaki, T.; Boku, S.; Watanabe, N.; Fujita, A.; Iwamatsu, A.; Obinata, T.; Ohashi, K.; Mizuno, K.; Narumiya, S.: Signaling from Rho to the actin cytoskeleton through protein kinases ROCK and LIM-kinase. *Science* 285: 895–898, 1999.

[43003] 16555.Ridley, A. J.; Hall, A.: The small GTP-binding protein rho regulates the assembly of focal adhesions and actin stress fibers in response to growth factors. *Cell* 70: 389–399, 1992.

[43004] 16556.Fagan, K. P.; Oliveira, L.; Pittler, S. J.: Sequence of rho small GTP-binding protein cDNAs from human retina and identification of novel 5-prime end cloning artifacts. *Exp. Eye Res.* 59: 235–237, 1994.

[43005] 16557.Morris, S. W.; Valentine, M. B.; Kirstein, M. N.; Huebner, K.: Reassignment of the human ARH9 RAS-re-

lated gene to chromosome 1p13–p21. *Genomics* 15:677–679, 1993.

- [43006] 16558. Maesaki, R.; Ihara, K.; Shimizu, T.; Kuroda, S.; Kaibuchi, K.; Hakoshima, T.: The structural basis of Rho effector recognition revealed by the crystal structure of human RhoA complexed with the effector domain of PKN/PRK1. *Molec. Cell* 4: 793–803, 1999.
- [43007] 16559. Nakamura, M.; Nagano, T.; Chikama, T.; Nishida, T.: Role of the small GTP-binding protein Rho in epithelial cell migration in the rabbit cornea. *Invest. Ophthalmol. Vis. Sci.* 42: 941–947, 2001.
- [43008] 16560. Rao, P. V.; Deng, P.-F.; Kumar, J.; Epstein, D. L.: Modulation of aqueous humor outflow facility by the Rho kinase-specific inhibitor Y-27632. *Invest. Ophthalmol. Vis. Sci.* 42: 1029–1037, 2001.
- [43009] 16561. Akiyama, T.; Takasawa, S.; Nata, K.; Kobayashi, S.; Abe, M.; Shervani, N. J.; Ikeda, T.; Nakagawa, K.; Unno, M.; Matsuno, S.; Okamoto, H.: Activation of Reg gene, a gene for insulin-producing beta-cell regeneration: poly(ADP-ribose) polymerase binds Reg promoter and regulates the transcription by autopoly(ADP-ribosylation). *Proc. Nat. Acad. Sci.* 98: 48–53, 2001.
- [43010] 16562. Bartoli, C.; Gharib, B.; Giorgi, D.; Sansonetti, A.;

Dagorn, J.-C.;Berge-Lefranc, J.-L.: A gene homologous to the reg gene is expressed in human pancreas. FEBS Lett. 327: 289-293, 1993.

[43011] 16563.Gharib, B.; Fox, M. F.; Bartoli, C.; Giorgi, D.; Sansonetti, A.;Swallow, D. M.; Dagorn, J. C.; Berge-Lefranc, J. L.: Human regenerationprotein/lithostathine genes map to chromosome 2p12. Ann. Hum. Genet. 57:9-16, 1993.

[43012] 16564.Giorgi, D.; Bernard, J.-P.; Rouquier, S.; Iovanna, J.; Sarles,H.; Dagorn, J.-C.: Secretory pancreatic stone protein messenger RNA:nucleotide sequence and expression in chronic calcifying pancreatitis. J.Clin. Invest. 84: 100-106, 1989.

[43013] 16565.Miyashita, H.; Nakagawara, K.; Mori, M.; Narushima, Y.; Noguchi,N.; Moriizumi, S.; Takasawa, S.; Yonekura, H.; Takeuchi, T.; Okamoto,H.: Human REG family genes are tandemly ordered in a 95-kilobaseregion of chromosome 2p12. FEBS Lett. 377: 429-433, 1995.

[43014] 16566.Perfetti, R.; Hawkins, A. L.; Griffin, C. A.; Egan, J. M.; Zenilman,M. E.; Shuldiner, A. R.: Assignment of the human pancreatic regenerating(REG) gene to chromosome 2p12. Genomics 20: 305-307, 1994.

[43015] 16567.Sarles, H.; Dagorn, J. C.; Giorgi, D.; Bernard, J. P.: Renamingpancreatic stone protein as 'lithostathine'.

(Letter) Gastroenterology 99:900–901, 1990.

[43016] 16568.Stewart, T. A.: The human REG gene encodes pancreatic stone protein.(Letter) Biochem. J. 260: 622–623, 1989.

[43017] 16569.Terazono, K.; Yamamoto, H.; Takasawa, S.; Shiga, K.; Yonemura,Y.; Tochino, Y.; Okamoto, H.: A novel gene activated in regeneratingislets. J. Biol. Chem. 263: 2111–2114, 1988.

[43018] 16570.Unno, M.; Yonekura, H.; Nakagawara, K.; Watanabe, T.; Miyashita,H.; Moriizumi, S.; Okamoto, H.: Structure, chromosomal localization,and expression of mouse reg genes, reg I and reg II: a novel typeof reg gene, reg II, exists in the mouse genome. J. Biol. Chem. 268:15974–15982, 1993.

[43019] 16571.Verdier, J. M.; Dussol, B.; Casanova, P.; Daudon, M.; Dupuy, P.;Berthezene, P.; Boistelle, R.; Berland, Y.; Dagorn, J. C.: Evidencethat human kidney produces a protein similar to lithostathine, thepancreatic inhibitor of CaCO_3 crystal growth. Europ. J. Clin. Invest. 22:469–474, 1992.

[43020] 16572.Moriizumi, S.; Watanabe, T.; Unno, M.; Nakagawara, K.; Suzuki,Y.; Miyashita, H.; Yonekura, H.; Okamoto, H.: Isolation, structuraldetermination and expression of a novel reg gene, human regI-beta.

Biochim.Biophys. Acta 1217: 199–202, 1994.

- [43021] 16573.Boel, E.; Schwartz, T. W.; Norris, K. E.; Fiil, N. P.: A cDNA encoding a small common precursor for human pancreatic polypeptide and pancreatic icosapeptide. EMBO J. 3: 909–912, 1984.
- [43022] 16574.Chandrasekharappa, S. C.; Friedman, L.; King, S. E.; Lee, Y.–H.; Welsch, P.; Bowcock, A. M.; Weber, B. L.; King, M.–C.; Collins, F.S.: The gene for pancreatic polypeptide (PPY) and the anonymous marker D17S78 are within 45 kb of each other on chromosome 17q21. Genomics 21:458–460, 1994.
- [43023] 16575.Hort, Y.; Baker, E.; Sutherland, G. R.; Shine, J.; Herzog, H.: Gene duplication of the human peptide YY gene (PYY) generated the pancreatic polypeptide gene (PPY) on chromosome 17q21.1. Genomics 26:77–83, 1995.
- [43024] 16576.Kimmel, J. R.; Hayden, L. J.; Pollock, H. G.: Isolation and characterization of a new pancreatic polypeptide hormone. J. Biol. Chem. 250: 9369–9376, 1975.
- [43025] 16577.Larsson, L.–I.; Sundler, F.; Hakanson, R.: Pancreatic polypeptide—a postulated new hormone: identification of its cellular storage site by light and electron microscopic immunocytochemistry. Diabetologia 12:211–216, 1976.
- [43026] 16578.Schwartz, T. W.: Pancreatic polypeptide: a hormone

under vagal control. *Gastroenterology* 85: 1411–1425, 1983.

[43027] 16579. Schwartz, T. W.; Tager, H. S.: Isolation and biogenesis of a new peptide from pancreatic islets. *Nature* 294: 589–591, 1981.

[43028] 16580. Takeuchi, T.; Gumucio, D.; Eddy, R.; Meisler, M.; Minth, C.; Dixon, J.; Yamada, T.; Shows, T.: Assignment of the related pancreatic polypeptide (PNP or PPY) and neuropeptide Y (NPY) genes to regions on human chromosomes 17 and 7. (Abstract) *Cytogenet. Cell Genet.* 40: 759 only, 1985.

[43029] 16581. Zipf, W. B.; O'Dorisio, T. M.; Cataland, S.; Sotos, J.: Blunted pancreatic polypeptide responses in children with obesity of Prader–Willi syndrome. *J. Clin. Endocr. Metab.* 52: 1264–1265, 1981.

[43030] 16582. Chen, K.; Yang, W.; Grimsby, J.; Shih, J. C.: The human 5-HT₂ receptor is encoded by a multiple intron–exon gene. *Brain Res. Molec. Brain Res.* 14: 20–26, 1992.

[43031] 16583. Holmes, C.; Arranz, M. J.; Powell, J. F.; Collier, D. A.; Lovestone, S.: 5-HT_{2A} and 5-HT_{2C} receptor polymorphisms and psychopathology in late onset Alzheimer's disease. *Hum. Molec. Genet.* 7: 1507–1509, 1998.

[43032] 16584. Hsieh, C.-L.; Bowcock, A. M.; Farrer, L. A.; Hebert,

J. M.; Huang, K. N.; Cavalli-Sforza, L. L.; Julius, D.; Francke, U.: The serotonin receptor subtype 2 locus HTR2 is on human chromosome 13 near genes for esterase D and retinoblastoma-1 and on mouse chromosome 14. *Somat. Cell Molec. Genet.* 16: 567–574, 1990.

[43033] 16585. Kato, M. V.; Shimizu, T.; Nagayoshi, M.; Kaneko, A.; Sasaki, M. S.; Ikawa, Y.: Genomic imprinting of the human serotonin-receptor (HTR2) gene involved in development of retinoblastoma. *Am. J. Hum. Genet.* 59: 1084–1090, 1996.

[43034] 16586. Liu, J.; Chen, Y.; Kozak, C. A.; Yu, L.: The 5-HT(2) serotonin receptor gene Htr-2 is tightly linked to Es-10 on mouse chromosome 14. *Genomics* 11: 231–234, 1991.

[43035] 16587. Peroutka, S. J.: 5-Hydroxytryptamine receptor subtypes. *Ann. Rev. Neurosci.* 11: 45–60, 1988.

[43036] 16588. Sparkes, R. S.; Lan, N.; Klisak, I.; Mohandas, T.; Diep, A.; Kojis, T.; Heinzmann, C.; Shih, J. C.: Assignment of a serotonin 5HT-2 receptor gene (HTR2) to human chromosome 13q14–q21 and mouse chromosome 14. *Genomics* 9: 461–465, 1991.

[43037] 16589. Williams, J.; McGuffin, P.; Nothen, M.; Owen, M. J.; EMAS Collaborative Group: Meta-analysis of association between the 5-HT (2A) receptor T102C polymorphism and

schizophrenia. (Letter) Lancet 349: 1221only, 1997.

- [43038] 16590. Williams, J.; Spurlock, G.; McGuffin, P.; Mallet, J.; Nothen, M.M.; Gill, M.; Aschauer, H.; Nylander, P. O.; Mac-
ciardi, F.; Owen, M. J.: Association between schizophrenia
and T102C polymorphism of the 5-hydroxytryptophan
type 2a-receptor gene. European Multicentre Association
Study of Schizophrenia (EMASS) Group. Lancet 347:
1294–1296, 1996.
- [43039] 16591. Philipson, L. H.; Hice, R. E.; Schaefer, K.; LaMen-
dola, J.; Bell, G. I.; Neldon, D. J.; Steiner, D. F.: Sequence
and functional expression in *Xenopus* oocytes of a human
insulinoma and islet potassium channel. Proc. Nat. Acad.
Sci. 88: 53–57, 1991.
- [43040] 16592. Ghanshani, S.; Pak, M.; McPherson, J. D.; Strong,
M.; Dethlefs, B.; Wasmuth, J. J.; Salkoff, L.; Gutman, G. A.;
Chandy, K. G.: Genomic organization, nucleotide sequence,
and cellular distribution of a Shaw-related potassium
channel gene, Kv3.3, and mapping of Kv3.3 and Kv3.4 to
human chromosomes 19 and 1. Genomics 12: 190–196,
1992.
- [43041] 16593. McPherson, J. D.; Wasmuth, J. J.; Chandy, K. G.;
Swanson, R.; Dethlefs, B.; Chandy, G.; Wymore, R.; Ghan-
shani, S.: Chromosomal localization of 7 potassium chan-

nel genes. (Abstract) Cytogenet. Cell Genet. 58:1979 only, 1991.

[43042] 16594.Gessler, M.; Grupe, A.; Grzeschik, K.-H.; Pongs, O.: The potassiumchannel gene HK1 maps to human chromosome 11p14.1, close to the FSHBgene. Hum. Genet. 90: 319-321, 1992.

[43043] 16595.Grandy, D.; Mathew, M. K.; Ramaswami, M.; Tanouye, M.; Sheffield, V.; Jones, C. A.; Al-Dhalimi, M.; Zhang, Y.; Saez, C.; Litt, M.: A human voltage-gated potassium channel gene, HuKII, maps to chromosome 11p14-p13. (Abstract) Am. J. Hum. Genet. 51 (suppl.): A396 only, 1992.

[43044] 16596.Philipson, L. H.; Eddy, R. L.; Shows, T. B.; Bell, G. I.: Assignment of human potassium channel gene KCNA4 (Kv1.4, PCN2) to chromosome 11q13.4-q14.1. Genomics 15: 463-464, 1993.

[43045] 16597.Philipson, L. H.; Schaefer, K.; LaMendola, J.; Bell, G. I.; Steiner, D. F.: Sequence of a human fetal skeletal muscle potassium channel cDNA related to RCK4. Nucleic Acids Res. 18: 7160 only, 1990.

[43046] 16598.Stuehmer, W.; Ruppertsberg, J. P.; Schroter, K. H.; Sakmann, B.; Stocker, M.; Giese, K. P.; Perschke, A.; Baumann, A.; Pongs, O.: Molecular basis of functional diversity

of voltage-gated potassium channels in mammalian brain. EMBO J. 8: 3235–3244, 1989.

- [43047] 16599. Tamkun, M. M.; Knoth, K. M.; Walbridge, J. A.; Kroemer, H.; Roden, D. M.; Glover, D. M.: Molecular cloning and characterization of two voltage-gated K⁺ channel cDNAs from human ventricle. FASEB J. 5:331–337, 1991.
- [43048] 16600. Phromchotikul, T.; Browne, D. L.; Curran, M. E.; Keating, M. T.; Litt, M.: Dinucleotide repeat polymorphism at the KCNA5 locus. Hum. Molec. Genet. 2: 1512 only, 1993.
- [43049] 16601. Bardien-Kruger, S.; Wulff, H.; Arieff, Z.; Brink, P.; Chandy, K.G.; Corfield, V.: Characterisation of the human voltage-gated potassium channel gene, KCNA7, a candidate gene for inherited cardiac disorders, and its exclusion as cause of progressive familial heart block I (PFHBI). Europ. J. Hum. Genet. 10: 36–43, 2002.
- [43050] 16602. Day, J. R.; Albers, J. J.; Lofton-Day, C. E.; Gilbert, T. L.; Ching, A. F. T.; Grant, F. J.; O'Hara, P. J.; Marcovina, S. M.; Adolphson, J. L.: Complete cDNA encoding human phospholipid transfer protein from human endothelial cells. J. Biol. Chem. 269: 9388–9391, 1994.
- [43051] 16603. Jiang, X.; Bruce, C.; Mar, J.; Lin, M.; Ji, Y.; Francone, O. L.; Tall, A. R.: Targeted mutation of plasma phospho-

lipid transfer proteingene markedly reduces high-density lipoprotein levels. *J. Clin. Invest.* 103:907–914, 1999.

[43052] 16604.LeBoeuf, R. C.; Caldwell, M.; Tu, A.-Y.; Albers, J. J.: Phospholipidtransfer protein maps to distal mouse chromosome 2. *Genomics* 34:259–260, 1996.

[43053] 16605.Tu, A.-Y.; Wolfbauer, G.; Albers, J. J.: Functional characterizationof the promoter region of the human phospholipid transfer proteingene. *Biochem. Biophys. Res. Commun.* 217: 705–711, 1995.

[43054] 16606.Whitmore, T. E.; Day, J. R.; Albers, J. J.: Localization of thehuman phospholipid transfer protein gene to chromosome 20q12–q13.1. *Genomics* 28:599–600, 1995.

[43055] 16607.Comings, D. E.: Evidence for ancient tetraploidy and conservationof linkage groups in mammalian chromosomes. *Nature* 238: 455–457,1972.

[43056] 16608.D'Ancona, G. G.; Chern, C. J.; Benn, P.; Croce, C. M.: Assignmentof the human gene for enolase 1 to region pter–p36 of chromosome 1. *Cytogenet.Cell Genet.* 18: 327–332, 1977.

[43057] 16609.D'Ancona, G. G.; Croce, C. M.: Assignment of the gene for enolaseto mouse chromosome 4 using somatic cell hybrids. *Cytogenet. CellGenet.* 19: 1–6, 1977.

[43058] 16610.Feo, S.; Oliva, D.; Arico, B.; Barba, G.; Cali, L.; Gial-

longo,A.: The human genome contains a single processed pseudogene for alphaenolase located on chromosome 1. DNA Sequence 1: 79–83, 1990.

- [43059] 16611.Giallongo, A.; Feo, S.; Moore, R.; Croce, C. M.; Showe, L. C.:Molecular cloning and nucleotide sequence of a full-length cDNA forhuman alpha enolase. Proc. Nat. Acad. Sci. 83: 6741–6745, 1986.
- [43060] 16612.Giblett, E. R.; Chen, S.–H.; Anderson, J. E.; Lewis, M.: A familystudy suggesting genetic linkage of phospho-pyruvate hydratase (enolase)to the Rh blood group system. Cytogenet. Cell Genet. 13: 91–92,1974.
- [43061] 16613.Lachant, N. A.; Jennings, M. A.; Tanaka, K. R.: Partial erythrocyteenolase deficiency: a hereditary disorder with variable clinical expression.(Abstract) Blood 68: 55a only, 1986.
- [43062] 16614.Lachant, N. A.; Tanaka, K. R.: Enolase kinetic properties inpartial erythrocyte enolase deficiency. (Abstract) Clin. Res. 35:426A only, 1987.
- [43063] 16615.Lalley, P. A.; Francke, U.; Minna, J. D.: Homologous genes forenolase, phosphogluconate dehydrogenase, phosphoglucomutase, and adenylatekinase are syntenic on mouse chromosome 4 and human chromosome 1p. Proc.Nat. Acad. Sci. 75: 2382–2386, 1978.

- [43064] 16616. Ribaud, M. R.; Di Leonardo, A.; Rubino, P.; Giallongo, A.; Feo, S.: Assignment of enolase processed pseudogene (ENO1P) to human chromosome 1 bands 1q41-q42. *Cytogenet. Cell Genet.* 74: 201-202, 1996.
- [43065] 16617. Van Cong, N.; Weil, D.; Rebourcet, R.; Frezal, J.: Localisation des enolases 1 et 2 respectivement sur les chromosomes 1 et 12 par l'analyse des hybrides homme-souris. *Ann. Genet.* 20: 153-157, 1977.
- [43066] 16618. Wistow, G. J.; Lietman, T.; Williams, L. A.; Stapel, S. O.; de Jong, W. W.; Horwitz, J.; Piatigorsky, J.: Tau-crystallin/alpha-enolase: one gene encodes both an enzyme and a lens structural protein. *J. Cell Biol.* 107: 2729-2736, 1988.
- [43067] 16619. Barton, J. W.; Hart, I. M.; Patterson, D.: Mapping of a locus correcting lack of phosphoribosylaminoimidazole carboxylase activity in Chinese hamster ovary cell Ade(-) mutants to human chromosome 4. *Genomics* 9: 314-321, 1991.
- [43068] 16620. Brayton, K. A.; Chen, Z.; Zhou, G.; Nagy, P. L.; Gavalas, A.; Trent, J. M.; Deaven, L. L.; Dixon, J. E.; Zalkin, H.: Two genes for de novo purine nucleotide synthesis on human chromosome 4 are closely linked and divergently transcribed. *J. Biol. Chem.* 269: 5313-5321, 1994.

- [43069] 16621.Torban, E.; Pelletier, J.; Goodyer, P.: F329L polymorphism in the human PAX8 gene. *Am. J. Med. Genet.* 72: 186–187, 1997.
- [43070] 16622.Gartner, J.; Kearns, W.; Pearson, P.; Valle, D.: Characterization and localization of the human 70-kD peroxisomal membrane protein (PMP70) gene. (Abstract) *Am. J. Hum. Genet.* 51 (suppl.): A168, 1992.
- [43071] 16623.Gartner, J.; Kearns, W.; Rosenberg, C.; Pearson, P.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Valle, D.: Localization of the 70-kDa peroxisomal membrane protein to human 1p21–p22 and mouse
- [43072] 16624.*Genomics* 15: 412–414, 1993.3. Gartner, J.; Moser, H.; Valle, D.: Mutations in the 70K peroxisomal membrane protein gene in Zellweger syndrome. *Nature Genet.* 1: 16–23, 1992.
- [43073] 16625.Vilain, C.; Rydlewski, C.; Duprez, L.; Heinrichs, C.; Abramowicz, M.; Malvaux, P.; Renneboog, B.; Parma, J.; Costagliola, S.; Vassart, G.: Autosomal dominant transmission of congenital thyroid hypoplasia due to loss-of-function mutation of PAX8. *J. Clin. Endocr. Metab.* 86:234–238, 2001.
- [43074] 16626.Walther, C.; Guenet, J.-L.; Simon, D.; Deutsch, U.; Jostes, B.; Goulding, M.; Plachov, D.; Balling, R.; Gruss, P.:

Pax: a murinemultigene family of paired box-containing genes. *Genomics* 11: 424–434,1991.

- [43075] 16627.Das, P.; Stockton, D. W.; Bauer, C.; Shaffer, L. G.; D'Souza, R.N.; Wright, J. T.; Patel, P. I.: Haploinsufficiency of PAX9 is associatedwith autosomal dominant hypodontia. *Hum. Genet.* 110: 371–376, 2002.
- [43076] 16628.Nieminen, P.; Arte, S.; Tanner, D.; Paulin, L.; Alalu-usua, S.;Thesleff, I.; Pirinen, S.: Identification of a non-sense mutationin the PAX9 gene in molar oligodontia. *Europ. J. Hum. Genet.* 9:743–746, 2001.
- [43077] 16629.Peters, H.; Neubuser, A.; Kratochwil, K.; Balling, R.: Pax9-deficientmice lack pharyngeal pouch derivatives and teeth and exhibit craniofacialand limb abnormalities. *Genes Dev.* 12: 2735–2747, 1998.
- [43078] 16630.Santagati, F.; Gerber, J.–K.; Blusch, J. H.; Kokubu, C.; Peters,H.; Adamski, J.; Werner, T.; Balling, R.; Imai, K.: Comparative analysisof the genomic organization of Pax9 and its conserved physical associationwith Nkx2–9 in the human, mouse, and pufferfish genomes. *MammalianGenome* 12: 232–237, 2001.
- [43079] 16631.Stockton, D. W.; Das, P.; Goldenberg, M.; D'Souza, R. N.; Patel,P. I.: Mutation of PAX9 is associated with oligodontia. (Letter) *NatureGenet.* 24: 18–19, 2000.

- [43080] 16632.Wallin, J.; Mizutani, Y.; Imai, K.; Miyashita, N.; Moriwaki, K.; Taniguchi, M.; Koseki, H.; Balling, R.: A new Pax gene, Pax-9, maps to mouse chromosome 12. *Mammalian Genome* 4: 354-358, 1993.
- [43081] 16633.Grueneberg, D. A.; Natesan, S.; Alexandre, C.; Gilman, M. Z.: Human and Drosophila homeodomain proteins that enhance the DNA-binding activity of serum response factor. *Science* 257: 1089-1095, 1992.
- [43082] 16634.Kern, M. J.; Argao, E. A.; Birkenmeier, E. H.; Rowe, L. B.; Potter, S. S.: Genomic organization and chromosome localization of the murine homeobox gene Pmx. *Genomics* 19: 334-340, 1994.
- [43083] 16635.Nakamura, T.; Yamazaki, Y.; Hatano, Y.; Miura, I.: NUP98 is fused to PMX1 homeobox gene in human acute myelogenous leukemia with chromosome translocation t(1;11)(q23;p15). *Blood* 94: 741-747, 1999.
- [43084] 16636.Norris, R. A.; Scott, K. K.; Moore, C. S.; Stetten, G.; Brown, C. R.; Jabs, E. W.; Wulfsberg, E. A.; Yu, J.; Kern, M. J.: Human PRRX1 and PRRX2 genes: cloning, expression, genomic localization, and exclusion as disease genes for Nager syndrome. *Mammalian Genome* 11:1000-1005, 2000.
- [43085] 16637.Riazuddin, S.; Castelein, C. M.; Ahmed, Z. M.; Lal-

wani, A. K.;Mastroianni, M. A.; Naz, S.; Smith, T. N.;
Liburd, N. A.; Friedman,T. B.; Griffith, A. J.; Riazuddin, S.;
Wilcox, E. R.: Dominant modifierDFNM1 suppresses recessive deafness DFNB26. *Nature Genet.* 26: 431–434,2000.

[43086] 16638.Hebrok, M.; Kim, S. K.; St Jacques, B.; McMahon, A. P.; Melton,D. A.: Regulation of pancreas development by hedgehog signaling. *Development* 127:4905–4913, 2000.

[43087] 16639.Reidenberg, M. M.; Levy, M.; Drayer, D. E.; Zylber-Katz, E.; Robbins,W. C.: Acetylator phenotype in idiopathic systemic lupus erythematosus. *ArthritisRheum.* 23: 569–573, 1980.

[43088] 16640.Stohl, W.; Crow, M. K.; Kunkel, H. G.: Systemic lupus erythematosuswith deficiency of the T4 epitope on T helper/inducer cells. *NewEng. J. Med.* 312: 1671–1678, 1985.

[43089] 16641.Theofilopoulos, A. N.; Dixon, F. J.: Murine models of systemiclupus erythematosus. *Adv. Immun.* 37: 269–390, 1985.

[43090] 16642.Friedl, W.; Uhlhaas, S.; Schulmann, K.; Stolte, M.; Loff, S.; Back,W.; Mangold, E.; Stern, M.; Knaebel, H. P.; Sutter, C.; Weber, R.G.; Pistorius, S.; Burger, B.; Propping, P.: Juvenile polyposis:massive gastric polyposis is more common in MADH4 mutation carriersthan in BMPR1A mu–

tation carriers. Hum. Genet. 111: 108–111, 2002.

- [43091] 16643.Houlston, R.; Bevan, S.; Williams, A.; Young, J.; Dunlop, M.; Rozen,P.; Eng, C.; Markie, D.; Woodford–Richens, K.; Rodriguez–Bigas, M.A.; Leggett, B.; Neale, K.; Phillips, R.; Sheridan, E.; Hodgson, S.;Iwama, T.; Eccles, D.; Bodmer, W.; Tomlinson, I.: Mutations in DPC4(SMAD4) cause juvenile polyposis syndrome, but only account for aminority of cases. Hum. Molec. Genet. 7: 1907–1912, 1998.
- [43092] 16644.Howe, J. R.; Ringold, J. C.; Summers, R. W.; Mitros, F. A.; Nishimura,D. Y.; Stone, E. M.: A gene for familial juvenile polyposis mapsto chromosome 18q21.1. Am. J. Hum. Genet. 62: 1129–1136, 1998.
- [43093] 16645.Howe, J. R.; Roth, S.; Ringold, J. C.; Summers, R. W.; Jarvinen,H. J.; Sistonen, P.; Tomlinson, I. P. M.; Houlston, R. S.; Bevan,S.; Mitros, F. A.; Stone, E. M.; Aaltonen, L. A.: Mutations in theSMAD4/DPC4 gene in juvenile polyposis. Science 280: 1086–1088, 1998.
- [43094] 16646.Leslie, K. K.; Watanabe, S.; Lei, K.–J.; Chou, D. Y.; Plouzek,C. A.; Deng, H.–C.; Torres, J.; Chou, J. Y.: Linkage of two humanpregnancy–specific beta 1–glycoprotein genes: one is associated withhydatidiform mole. Proc. Nat. Acad. Sci. 87: 5822–5826, 1990.

- [43095] 16647.Tatarinov, Y. S.; Falaleeva, D. M.; Kalashnikov, V. V.: Humanpregnancy-specific beta-1-globulin and its relation to chorioepithelioma. *Int.J. Cancer* 17: 626-632, 1976.
- [43096] 16648.Berry, C.; Cree, J.; Mann, T.: Aarskog's syndrome. *Arch. Dis.Child.* 55: 706-710, 1980.
- [43097] 16649.Escobar, V.; Weaver, D. D.: Aarskog syndrome: new findings andgenetic analysis. *J.A.M.A.* 240: 2638-2641, 1978.
- [43098] 16650.Hanley, W. B.; McKusick, V. A.; Barranco, F. T.: Osteochondritisdissecans and associated malformations in brothers: a review of familialaspects. *J. Bone Joint Surg.* 49A: 925-937, 1967.
- [43099] 16651.Nagase, T.; Seki, N.; Tanaka, A.; Ishikawa, K.; Nomura, N.: Predictionof the coding sequences of unidentified human genes. IV. The codingsequences of 40 new genes (KIAA0121-KIAA0160) deduced by analysisof cDNA clones from human cell line KG-1. *DNA Res.* 2: 167-174, 1995.
- [43100] 16652.Parrish, J. E.; Ciccodicola, A.; Wehnert, M.; Cox, G. F.; Chen,E.; Nelson, D. L.: A muscle-specific DNase I-like gene in human Xq28. *Hum.Molec. Genet.* 4: 1557-1564, 1995.

- [43101] 16653.Pergolizzi, R.; Appierto, V.; Bosetti, A.; DeBellis, G. L.; Rovidia,E.; Biunno, I.: Cloning of a gene encoding a DNase I-like endonucleasein the human Xq28 region. Gene 168: 267–270, 1996.
- [43102] 16654.Klink, A.; Schiebel, K.; Winkelmann, M.; Rao, E.; Horsthemke, B.;Ludecke, H.–J.; Claussen, U.; Scherer, G.; Rappold, G.: The humanprotein kinase gene PKX1 on Xp22.3 displays Xp/Yp homology and isa site of chromosomal instability. Hum. Molec. Genet. 4: 869–878,1995.
- [43103] 16655.Schiebel, K.; Mertz, A.; Winkelmann, B.; Glaser, B.; Schempp, W.;Rappold, G.: FISH localization of the human Y–homolog of proteinkinase PRKX (PRKY) to Yp11.2 and two pseudogenes to 15q26 and Xq12–q13. Cytogenet.Cell Genet. 76: 49–52, 1997.
- [43104] 16656.Brown, C. J.; Carrel, L.; Willard, H. F.: Expression of genesfrom the human active and inactive X chromosomes. Am. J. Hum. Genet. 60:1333–1343, 1997.
- [43105] 16657.Dong, B.; Horowitz, D. S.; Kobayashi, R.; Krainer, A. R.: Purificationand cDNA cloning of HeLa cell p54(nrb), a nuclear protein with twoRNA recognition motifs and extensive homology to human splicing factorPSF and Drosophila NONA/BJ6. Nucleic Acids Res. 21: 4085–4092, 1993.

- [43106] 16658.Nelson, D. L.; Ballabio, A.; Cremers, F.; Monaco, A. P.; Schlessinger,D.: Report of the sixth international workshop on X chromosome mapping1995. Cytogenet. Cell Genet. 71: 308–342, 1995.
- [43107] 16659.Janssens, R.; Boeynaems, J.–M.; Godart, M.; Comuni, D.: Cloningof a human heptahelical receptor closely related to the P2Y(5) receptor. Biochem.Biophys. Res. Commun. 236: 106–112, 1997.
- [43108] 16660.O'Dowd, B. F.; Nguyen, T.; Jung, B. P.; Marchese, A.; Cheng, R.;Heng, H. H. Q.; Kolakowski, L. F., Jr.; Lynch, K. R.; George, S. R.: Cloning and chromosomal mapping of four putative novel human G–protein–coupledreceptor genes. Gene 187: 75–81, 1997.
- [43109] 16661.Knight, S. W.; Heiss, N. S.; Vulliamy, T. J.; Aalfs, C. M.; McMahon,C.; Richmond, P.; Jones, A.; Hennekam, R. C. M.; Poustka, A.; Mason,P. J.; Dokal, I.: Unexplained aplastic anaemia, immunodeficiency,and cerebellar hypoplasia (Hoyeraal–Hreidarsson syndrome) due to mutationsin the dyskeratosis congenita gene, DKC1. Brit. J. Haemat. 107:335–339, 1999.
- [43110] 16662.Knight, S. W.; Heiss, N. S.; Vulliamy, T. J.; Greschner, S.; Stavrides,G.; Pai, G. S.; Lestringant, G.; Varma, N.; Mason, P. J.; Dokal, I.;Poustka, A.: X–linked

dyskeratosis congenita is predominantly caused by missense mutations in the DKC1 gene. *Am. J. Hum. Genet.* 65: 50–58, 1999.

[43111] 16663. Knight, S. W.; Vulliamy, T. J.; Morgan, B.; Devriendt, K.; Mason, P. J.; Dokal, I.: Identification of novel DKC1 mutations in patients with dyskeratosis congenita: implications for pathophysiology and diagnosis. *Hum. Genet.* 108: 299–303, 2001.

[43112] 16664. Luzzatto, L.; Karadimitris, A.: Dyskeratosis and ribosomal rebellion. *Nature Genet.* 19: 6–7, 1998.

[43113] 16665. McGrath, J. A.: Dyskeratosis congenita: new clinical and molecular insights into ribosome function. *Lancet* 353: 1204–1205, 1999.

[43114] 16666. Mitchell, J. R.; Wood, E.; Collins, K.: A telomerase component is defective in the human disease dyskeratosis congenita. *Nature* 402: 551–555, 1999.

[43115] 16667. Salowsky, R.; Heiss, N. S.; Benner, A.; Wittig, R.; Poustka, A.: Basal transcription activity of the dyskeratosis congenita gene is mediated by Sp1 and Sp3 and a patient mutation in a Sp1 binding site is associated with decreased promoter activity. *Gene* 293: 9–19, 2002.

[43116] 16668. Vulliamy, T. J.; Knight, S. W.; Heiss, N. S.; Smith, O. P.; Poustka, A.; Dokal, I.; Mason, P. J.: Dyskeratosis con–

genita caused by a 3–primedeletion: germline and somatic mosaicism in a female carrier. *Blood* 94:1254–1260, 1999.

- [43117] 16669.Yaghmai, R.; Kimyai–Asadi, A.; Rostamiani, K.; Heiss, N. S.; Poustka,A.; Eyaid, W.; Bodurtha, J.; Nousari, H. C.; Hamosh, A.; Metzenberg,A.: Overlap of dyskeratosis congenita with the Hoyeraal–Hreidarssonsyndrome. *J. Pediat.* 136: 390–393, 2000.
- [43118] 16670.Greenfield, A.; Carrel, L.; Pennisi, D.; Philippe, C.; Quaderi,N.; Siggers, P.; Steiner, K.; Tam, P. P. L.; Monaco, A. P.; Willard,H. F.; Koopman, P.: The UTX gene escapes X inactivation in mice andhumans. *Hum. Molec. Genet.* 7: 737–742, 1998.
- [43119] 16671.Greenfield, A. J.; Scott, D.; Pennisi, D.; Ehrmann, I.; Ellis,P.; Cooper, L.; Simpson, E.; Koopman, P.: An H–YDb epitope is encodedby a novel mouse Y chromosome gene. *Nature Genet.* 14: 474–478, 1996.
- [43120] 16672.Caput, D.; Laurent, P.; Kaghad, M.; Lelias, J.–M.; Lefort, S.;Vita, N.; Ferrara, P.: Cloning and characterization of a specificinterleukin (IL)–13 binding protein structurally related to the IL–5receptor alpha chain. *J. Biol. Chem.* 271: 16921–16926, 1996.
- [43121] 16673.Guo, J.; Apiou, F.; Mellerin, M.–P.; Lebeau, B.;

Jacques, Y.; Minvielle, S.: Chromosome mapping and expression of the human interleukin-13 receptor. *Genomics* 42: 141–145, 1997.

[43122] 16674. Lin, C.-S.; Aebersold, R. H.; Leavitt, J.: Correction of the N-terminal sequences of the human plastin isoforms by using anchored polymerase chain reaction: identification of a potential calcium-binding domain. *Molec. Cell. Biol.* 10: 1818–1821, 1990.

[43123] 16675. Fukuda, M. N.; Sato, T.; Nakayama, J.; Klier, G.; Mikami, M.; Aoki, D.; Nozawa, S.: Trophinin and tasin, a novel cell adhesion molecule complex with potential involvement in embryo implantation. *Genes Dev.* 9: 1199–1210, 1995.

[43124] 16676. Pack, S. D.; Tanigami, A.; Ledbetter, D. H.; Sato, T.; Fukuda, M. N.: Assignment of trophoblast/endometrial epithelium cell adhesion molecule trophinin gene TRO to human chromosome bands Xp11.22–p11.21 by *in situ* hybridization. *Cytogenet. Cell Genet.* 79: 123–124, 1997.

[43125] 16677. Suzuki, N.; Nakayama, J.; Shih, I. M.; Aoki, D.; Nozawa, S.; Fukuda, M. N.: Expression of trophinin, tasin, and bystin by trophoblast and endometrial cells in human placenta. *Biol. Reprod.* 60: 621–627, 1999.

[43126] 16678. Sana, T. R.; Debets, R.; Timans, J. C.; Bazan, J. F.;

Kastelein, R. A.: Computational identification, cloning, and characterization of IL1R9, a novel interleukin-1 receptor-like gene encoded over an unusually large interval of human chromosome Xq22.2-q22.3. *Genomics* 69:252-262, 2000.

[43127] 16679. Bech-Hansen, N. T.; Naylor, M. J.; Maybaum, T. A.; Sparkes, R. L.; Koop, B.; Birch, D. G.; Bergen, A. A. B.; Prinsen, C. F. M.; Polomeno, R. C.; Gal, A.; Drack, A. V.; Musarella, M. A.; Jacobson, S. G.; Young, R. S. L.; Weleber, R. G.: Mutations in NYX, encoding the leucine-rich proteoglycan nyctalopin, cause X-linked complete congenital stationary night blindness. *Nature Genet.* 26: 319-323, 2000.

[43128] 16680. Pusch, C. M.; Zeitz, C.; Brandau, O.; Pesch, K.; Achatz, H.; Feil, S.; Scharfe, C.; Maurer, J.; Jacobi, F. K.; Pinckers, A.; Andreasson, S.; Hardcastle, A.; Wissinger, B.; Berger, W.; Meindl, A.: The complete form of X-linked congenital stationary night blindness is caused by mutations in a gene encoding a leucine-rich repeat protein. *Nature Genet.* 26: 324-327, 2000.

[43129] 16681. Isbrandt, D.; Leicher, T.; Waldschutz, R.; Zhu, X.; Luhmann, U.; Michel, U.; Sauter, K.; Pongs, O.: Gene structures and expression profiles of three human KCND (Kv4)

potassium channels mediating A-type currents $I_{(to)}$ and $I_{(sa)}$. *Genomics* 64: 144–154, 2000.

- [43130] 16682. Davies, J. P.; Cotter, P. D.; Ioannou, Y. A.: Cloning and mapping of human Rab7 and Rab9 cDNA sequences and identification of a Rab9 pseudogene. *Genomics* 41: 131–134, 1997.
- [43131] 16683. Brinkmann, U.; Vasmatazis, G.; Lee, B.; Pastan, I.: Novel genes in the PAGE and GAGE family of tumor antigens found by homology walking in the dbEST database. *Cancer Res.* 59: 1445–1448, 1999.
- [43132] 16684. Liu, X. F.; Helman, L. J.; Yeung, C.; Bera, T. K.; Lee, B.; Pastan, I.: XAGE-1, a new gene that is frequently expressed in Ewing's sarcoma. *Cancer Res.* 60: 4752–4755, 2000.
- [43133] 16685. Lahn, B. T.; Page, D. C.: Functional coherence of the human Y chromosome. *Science* 278: 675–680, 1997.
- [43134] 16686. Chung, J.; Lee, S.-G.; Song, K.: Identification of a human homolog of a putative RNA helicase gene (mDEAD3) expressed in mouse erythroid cells. *Korean J. Biochem.* 27: 193–197, 1995.
- [43135] 16687. Park, S. H.; Lee, S.-G.; Kim, Y.; Song, K.: Assignment of a human putative RNA helicase gene, DDX3, to human X chromosome bands p11.3–p11.23. *Cyto-*

genet.Cell Genet. 81: 178–179, 1998.

- [43136] 16688.Gasper, N. J.; Kinzy, T. G.; Scherer, B. J.; Humbelin, M.; Hershey, J. W. B.; Merrick, W. C.: Translation initiation factor eIF-2: cloning and expression of the human cDNA encoding the gamma-subunit. J. Biol.Chem. 269: 3415–3422, 1994.
- [43137] 16689.Ehrmann, I. E.; Ellis, P. S.; Mazeyrat, S.; Duthie, S.; Brockdorff, N.; Mattei, M. G.; Gavin, M. A.; Affara, N. A.; Brown, G. M.; Simpson, E.; Mitchell, M. J.; Scott, D. M.: Characterization of genes encoding translation initiation factor eIF-2-gamma in mouse and human: sex chromosome localization, escape from X-inactivation and evolution. Hum.Molec. Genet. 7: 1725–1737, 1998.
- [43138] 16690.Lee, S. M. Y.; Tsui, S. K. W.; Chan, K. K.; Garcia-Barcelo, M.; Waye, M. M. Y.; Fung, K. P.; Liew, C. C.; Lee, C. Y.: Chromosomal mapping, tissue distribution and cDNA sequence of four-and-a-half LIM domain protein 1 (FHL1). Gene 216: 163–170, 1998.
- [43139] 16691.Morgan, M. J.; Madgwick, A. J.; Charleston, B.; Pell, J. M.; Loughna, P. T.: The developmental regulation of a novel muscle LIM-protein. Biochem.Biophys. Res. Commun. 212: 840–846, 1995.
- [43140] 16692.Morgan, M. J.; Madgwick, A. J. A.: Slim defines a

novel family of LIM-proteins expressed in skeletal muscle.

Biochem. Biophys. Res. Commun. 225: 632–638, 1996.

[43141] 16693. Bard, J. A.; Zgombick, J.; Adham, N.; Vaysse, P.; Branchek, T.A.; Weinshank, R. L.: Cloning of a novel human serotonin receptor(5-HT-7) positively linked to adenylate cyclase. J. Biol. Chem. 268:23422–23426, 1993.

[43142] 16694. Gelernter, J.; Rao, P. A.; Pauls, D. L.; Hamblin, M. W.; Sibley, D. R.; Kidd, K. K.: Assignment of the 5HT7 receptor gene (HTR7) to chromosome 10q and exclusion of genetic linkage with Tourette syndrome. Genomics 26:207–209, 1995.

[43143] 16695. Kenakin, T. P.; Bond, R. A.; Bonner, T. I.: II. Definition of pharmacological receptors. Pharm. Rev. 44: 351–362, 1992.

[43144] 16696. Lassig, J. P.; Vachirasomtoon, K.; Hartzell, K.; Leventhal, M.; Courchesne, E.; Courchesne, R.; Lord, C.; Leventhal, B. L.; Cook, E. H., Jr.: Physical mapping of the serotonin 5-HT(7) receptor gene(HTR7) to chromosome 10 and pseudogene (HTR7P) to chromosome 12, and testing of linkage disequilibrium between HTR7 and autistic disorder. Am. J. Med. Genet. 88: 472–475, 1999.

[43145] 16697. Ruat, M.; Traiffort, E.; Leurs, R.; Tardivel-Lacombe, J.; Diaz, J.; Arrang, J.-M.; Schwartz, J.-C.: Molecular

cloning, characterization, and localization of a high-affinity serotonin receptor (5-HT-7) activating cAMP formation. *Proc. Nat. Acad. Sci.* 90: 8547–8551, 1993.

[43146] 16698. Tsou, A.; Kosaka, A.; Bach, C.; Zuppan, P.; Yee, C.; Tom, L.; Alvarez, R.; Ramsey, S.; Bonhaus, D. W.; Stefanich, E.; Jakeman, L.; Eglen, R. M.; Chan, H. W.: Cloning and expression of a 5-hydroxytryptamine-7 receptor positively coupled to adenylyl cyclase. *J. Neurochem.* 63:456–464, 1994.

[43147] 16699. Gelernter, J.; Kruger, S.; Pakstis, A. J.; Amara, S.; Kidd, K. K.: Linkage mapping of norepinephrine transporter protein and serotonin transporter protein genes. (Abstract) *Am. J. Hum. Genet.* 53 (suppl.):A1003, 1993.

[43148] 16700. Gelernter, J.; Pakstis, A. J.; Kidd, K. K.: Linkage mapping of serotonin transporter protein gene SLC6A4 on chromosome 17. *Hum. Genet.* 95: 677–680, 1995.

[43149] 16701. Gregor, P.; Patel, A.; Shimada, S.; Lin, C.-L.; Rochelle, J. M.; Kitayama, S.; Seldin, M. F.; Uhl, G. R.: Murine serotonin transporter: sequence and localization to chromosome 11. *Mammalian Genome* 4:283–284, 1993.

[43150] 16702. Hariri, A. R.; Mattay, V. S.; Tessitore, A.; Kolachana, B.; Fera, F.; Goldman, D.; Egan, M. F.; Weinberger, D. R.: Serotonin transporter genetic variation and the response of

the human amygdala. *Science* 297:400–403, 2002.

[43151] 16703.Heils, A.; Teufel, A.; Petri, S.; Seemann, M.; Bengel, D.; Balling,U.; Riederer, P.; Lesch, K. P.: Functional promoter and polyadenylationsite mapping of the human serotonin (5-HT) transporter gene. *J. NeuralTransm.* 102: 247–254, 1995.

[43152] 16704.Heils, A.; Teufel, A.; Petri, S.; Stober, G.; Riederer, P.; Bengel,D.; Lesch, K. P.: Allelic variation of human serotonin transporter gene expression. *J. Neurochem.* 66: 2621–2624, 1996.

[43153] 16705.Kim, S.-J.; Cox, N.; Courchesne, R.; Lord, C.; Corsello, C.; Akshoomoff,N.; Guter, S.; Leventhal, B. L.; Courchesne, E.; Cook, E. H., Jr.: Transmission disequilibrium mapping at the serotonin transporter gene (SLC6A4) region in autistic disorder. *Molec. Psychiat.* 7: 278–288,2002.

[43154] 16706.Lesch, K. P.; Balling, U.; Gross, J.; Strauss, K.; Wolozin, B.L.; Murphy, D. L.: Organisation of the human serotonin transporter gene. *J. Neural Trans.* 95: 157–162, 1994.

[43155] 16707.Chern, C. J.; Beutler, E.: Biochemical and electrophoretic studies of erythrocyte pyridoxine kinase in white and black Americans. *Am.J. Hum. Genet.* 28: 9–17,

1976.

- [43156] 16708.Hanna, M. C.; Turner, A. J.; Kirkness, E. F.: Human pyridoxalkinase: cDNA cloning, expression, and modulation by ligands of the benzodiazepine receptor. *J. Biol. Chem.* 272: 10756–10760, 1997.
- [43157] 16709.Dougherty, K. M.; Brandriss, M. C.; Valle, D.: Cloning human pyrroline–5–carboxylate reductase cDNA by complementation in *Saccharomyces cerevisiae*. *J. Biol. Chem.* 267: 871–875, 1992.
- [43158] 16710.Merrill, M. J.; Yeh, G. C.; Phang, J. M.: Purified human erythrocyte pyrroline–5–carboxylate reductase: preferential oxidation of NADPH. *J. Biol. Chem.* 264: 9352–9358, 1989.
- [43159] 16711.Brissenden, J. E.; Caras, I.; Thelander, L.; Francke, U.: The structural gene for the M1 subunit of human ribonucleotide reductase maps to chromosome 11, band p15, in human and to chromosome 7 in mouse. *Exp. Cell Res.* 174: 302–308, 1988.
- [43160] 16712.Byrne, J.; Smith, P.: Human polymorphic probe pE1.8 detects a polymorphism in the ribonucleotide reductase M1 subunit gene. *Hum. Genet.* 87: 376 only, 1991.
- [43161] 16713.Barletta, C.; Druck, T.; LaForgia, S.; Calabretta, B.;

Drabkin,H.; Patterson, D.; Croce, C. M.; Huebner, K.:
Chromosome locationsof the MYB related genes, AMYB
and BMYB. Cancer Res. 51: 3821–3824,1991.

[43162] 16714.Nomura, N.; Takahashi, M.; Matsui, M.; Ishii, S.;
Date, T.; Sasamoto,S.; Ishizaki, R.: Isolation of human
cDNA clones of MYB–related genes,A–MYB and B–MYB.
Nucleic Acids Res. 16: 11075–11089, 1988.

[43163] 16715.Takahashi, T.; Nakagoshi, H.; Sarai, A.; Nomura, N.;
Yamamoto,T.; Ishii, S.: Human A–myb gene encodes a
transcriptional activatorcontaining the negative regulatory
domains. FEBS Lett. 358: 89–96,1995.

[43164] 16716.Boylan, K. B.; Takahashi, N.; Diamond, M.; Hood, L.
E.; Prusiner,S. B.: DNA length polymorphism located
5–prime to the human myelinbasic protein gene. Am. J.
Hum. Genet. 40: 387–400, 1987.

[43165] 16717.Dayhoff, M. O.: Atlas of Protein Sequence and
Structure. Myelinmembrane encephalitogenic protein.
Washington: National BiomedicalResearch Foundation
(pub.) 5: 1972. Pp. D324 only.

[43166] 16718.Eylar, E. H.; Brostoff, S.; Hashim, G.; Westall, F. C.:
BasicA1 protein of the myelin membrane: the complete
amino acid sequence. J.Biol. Chem. 246: 5770–5784,
1971.

- [43167] 16719.Gomez, C. M.; Muggleton-Harris, A. L.; Whittingham, D. G.; Hood, L. E.; Readhead, C.: Rapid preimplantation detection of mutant (shiverer) and normal alleles of the mouse myelin basic protein gene allowing selective implantation and birth of live young. *Proc. Nat. Acad. Sci.* 87: 4481–4484, 1990.
- [43168] 16720.Kamholz, J.; de Ferra, F.; Puckett, C.; Lazzarini, R.: Identification of three forms of human myelin basic protein by cDNA cloning. *Proc. Nat. Acad. Sci.* 83: 4962–4966, 1986.
- [43169] 16721.Kamholz, J.; Spielman, R.; Gogolin, K.; Modi, W.; O'Brien, S.; Lazzarini, R.: The human myelin–basic–protein gene: chromosomal localization and RFLP analysis. *Am. J. Hum. Genet.* 40: 365–373, 1987.
- [43170] 16722.Lalley, P. A.; McKusick, V. A.: Report of the committee on comparative mapping (HGM8). *Cytogenet. Cell Genet.* 40: 536–566, 1985.
- [43171] 16723.Martensen, R. E.: Myelin basic protein speciation. *Prog. Clin. Biol. Res.* 146: 511–521, 1984.
- [43172] 16724.Marty, M. C.; Alliot, F.; Rutin, J.; Fritz, R.; Trisler, D.; Pessac, B.: The myelin basic protein gene is expressed in differentiated blood cell lineages and in hemopoietic progenitors. *Proc. Nat. Acad. Sci.* 99: 8856–8861, 2002.

- [43173] 16725.Molineaux, S. M.; Engh, H.; de Ferra, F.; Hudson, L.; Lazzarini,R. A.: Recombination within the myelin basic protein gene createdthe dysmyelinating shiverer mouse mutation. Proc. Nat. Acad. Sci. 83:7542–7546, 1986.
- [43174] 16726.Popko, B.; Puckett, C.; Lai, E.; Shine, H. D.; Readhead, C.; Takahashi,N.; Hunt, S. W., III; Sidman, R. L.; Hood, L.: Myelin deficient mice:expression of myelin basic protein and generation of mice with varyinglevels of myelin. Cell 48: 713–721, 1987.
- [43175] 16727.Readhead, C.; Popko, B.; Takahashi, N.; Shine, H. D.; Saavedra,R. A.; Sidman, R. L.; Hood, L.: Expression of a myelin basic proteingene in transgenic shiverer mice: correction of the dysmyelinatingphenotype. Cell 48: 703–712, 1987.
- [43176] 16728.Roach, A.; Boylan, K.; Horvath, S.; Prusiner, S. B.; Hood, L.E.: Characterization of cloned cDNA representing rat myelin basicprotein: absence of expression in shiverer mutant mice. Cell 34:799–806, 1983.
- [43177] 16729.Roach, A.; Takahashi, N.; Pravtcheva, D.; Ruddle, F.; Hood, L.: Chromosomal mapping of mouse myelin basic protein gene and structureand transcription of the partially deleted gene in shiverer mutantmice. Cell 42: 149–155, 1985.

- [43178] 16730.Saxe, D. F.; Takahashi, N.; Hood, L.; Simon, M. I.: Localization of the human myelin basic protein gene (MBP) to region 18q22–qter by in situ hybridization. *Cytogenet. Cell Genet.* 39: 246–249, 1985.
- [43179] 16731.Sheremata, W.; Cosgrove, J. B. R.; Hylar, E. H.: Cellular hypersensitivity to basic myelin (A1) protein and clinical multiple sclerosis. *New Eng. J. Med.* 291: 14–17, 1974.
- [43180] 16732.Sidman, R.: Personal Communication. Boston, Mass. 1983.
- [43181] 16733.Sidman, R. L.; Conover, C. S.; Carson, J. H.: Shiverer gene maps near the distal end of chromosome 18 in the house mouse. *Cytogenet. Cell Genet.* 39: 241–245, 1985.
- [43182] 16734.Sparkes, R. S.; Mohandas, T.; Heinzmann, C.; Roth, H. J.; Klisak, I.; Campagnoni, A. T.: Assignment of the myelin basic protein gene to human chromosome 18q22–qter. *Hum. Genet.* 75: 147–150, 1987.
- [43183] 16735.Takahashi, N.; Roach, A.; Teplow, D. B.; Prusiner, S. B.; Hood, L.: Cloning and characterization of the myelin basic protein gene from mouse: one gene can encode both 14 kd and 18.5 kd MBPs by alternate use of exons. *Cell* 42: 139–148, 1985.
- [43184] 16736.Barouch, L. A.; Harrison, R. W.; Skaf, M. W.; Rosas, G. O.; Cappola, T. P.; Kobeissi, Z. A.; Hobai, I. A.; Lemmon,

C. A.; Burnett, A. L.; O'Rourke, B.; Rodriguez, E. R.; Huang, P. L.; Lima, J. A. C.; Berkowitz, D. E.; Hare, J. M.: Nitric oxide regulates the heart by spatial confinement of nitric oxide synthase isoforms. *Nature* 416: 337–340, 2002.

[43185] 16737. Dimmeler, S.; Fleming, I.; Fisslthaler, B.; Hermann, C.; Busse, R.; Zeiher, A. M.: Activation of nitric oxide synthase in endothelial cells by Akt-dependent phosphorylation. *Nature* 399: 601–605, 1999.

[43186] 16738. Fulton, D.; Gratton, J.-P.; McCabe, T. J.; Fontana, J.; Fujio, Y.; Walsh, K.; Franke, T. F.; Papapetropoulos, A.; Sessa, W. C.: Regulation of endothelium-derived nitric oxide production by the protein kinase Akt. *Nature* 399: 597–601, 1999.

[43187] 16739. Kruh, G. D.; King, C. R.; Kraus, M. H.; Popescu, N. C.; Amsbaugh, S. C.; McBride, W. O.; Aaronson, S. A.: A novel human gene closely related to the abl proto-oncogene. *Science* 234: 1545–1548, 1986.

[43188] 16740. Abe, T.; Kikuchi, T.; Shinohara, T.: The sequence of the human phosphodiesterase-3 gene (PDC) and its 5-prime-flanking region. *Genomics* 19: 369–372, 1994.

[43189] 16741. Craft, C. M.; Lolley, R. N.; Seldin, M. F.; Lee, R. H.: Rat pineal gland phosphodiesterase-3: cDNA isolation, nucleotide sequence, and chromosomal assignment in the mouse. *Ge-*

nomics 10: 400–409, 1991.

- [43190] 16742.Ding, C.; Li, X.; Griffin, C. A.; Jabs, E. W.; Hawkins, A. L.; Levine, M. A.: The gene for human phosducin (PDC), a soluble protein that binds G-protein beta gamma dimers, maps to 1q25–q31.1. *Genomics* 18:457–459, 1993.
- [43191] 16743.Lee, R. H.; Fowler, A.; McGinnis, J. F.; Lolley, R. N.; Craft, C. M.: Amino acid and cDNA sequence of bovine phosducin, a soluble phosphoprotein from photoreceptor cells. *J. Biol. Chem.* 265: 15867–15873, 1990.
- [43192] 16744.Reiter, R. J.: The mammalian pineal gland: structure and function. *Am.J. Anat.* 162: 287–313, 1981.
- [43193] 16745.Sparkes, R. S.; Lee, R. H.; Shinohara, T.; Craft, C. M.; Kojis, T.; Klisak, I.; Heinzmann, C.; Bateman, J. B.: Assignment of the phosducin (PDC) gene to human chromosome 1q25–1q32.1 by somatic cell hybridization and in situ hybridization. *Genomics* 18: 426–428, 1993.
- [43194] 16746.Arnaud, J.; Vavrusa, B.; Sevin, J.; Constans, J.: Human red-cell acid phosphatase (ACP1): a new mutant (ACP1*KUK) detected by isoelectric focusing, kinetics of thermostability and substrate activity. *Hum.Hered.* 39: 288–293, 1989.
- [43195] 16747.Arnaud, J.; Vavrusa, B.; Wiederanders, G.; Constans, J.: Human red-cell acid phosphatase (ACP1): kinetic

and thermodynamic characterization of the KUK variant.

Hum. Hered. 42: 140–142, 1992.

[43196] 16748. Beemer, F. A.; van der Heiden, C.; Van Hemel, J. O.; Jansen, M.: Letter to the editors. (Letter) Clin. Genet. 24: 151, 1983.

[43197] 16749. Berg, K.: Close linkage between APOB and ACP1 excluded. (Abstract) Cytogenet. Cell Genet. 46: 580, 1987.

[43198] 16750. Bottini, E.; Carapella, E.; Orzalesi, M.; Lucarelli, P.; Pascone, R.; Gloria-Bottini, F.; Coccia, M.: Is there a role of erythrocyte acid phosphatase polymorphism in intrauterine development? (Letter) Am. J. Hum. Genet. 32: 764–767, 1980.

[43199] 16751. Seldin, M. F.; Kruh, G. D.: Mapping of Abll within a conserved linkage group on distal mouse chromosome 1 syntenic with human chromosome 1 using an interspecific cross. Genomics 4: 221–223, 1989.

[43200] 16752. Bello, M. J.; Salagnon, N.; Rey, J. A.; Guichaoua, M. R.; Berge-Lefranc, J. L.; Jordan, B. R.; Luciani, J. M.: Precise in situ localization of NCAM, ETS1, and D11S29 on human meiotic chromosomes. Cytogenet. Cell Genet. 52: 7–10, 1989.

[43201] 16753. Gold, D. P.; van Dongen, J. J. M.; Morton, C. C.; Bruns, G. A. P.; van den Elsen, P.; Geurts van Kessel, A. H.

M.; Terhorst, C.:The gene encoding the epsilon subunit of the T3/T-cell receptor complex maps to chromosome 11 in humans and to chromosome 9 in mice. *Proc.Nat. Acad. Sci.* 84: 1664–1668, 1987.

[43202] 16754.Ohtani, N.; Zebedee, Z.; Huot, T. J. G.; Stinson, J. A.; Sugimoto, M.; Ohashi, Y.; Sharrocks, A. D.; Peters, G.; Hara, E.: Opposing effects of Ets and Id proteins on p16(INK4A) expression during cellular senescence. *Nature* 409: 1067–1070, 2001.

[43203] 16755.Sacchi, N.; Watson, D. K.; Geurts van Kessel, A. H. M.; Hagemeijer, A.; Kersey, J.; Drabkin, H. D.; Patterson, D.; Papas, T. S.: Hu-ets-1 and Hu-ets-2 genes are transposed in acute leukemias with (4;11) and (8;21) translocations. *Science* 231: 379–382, 1986.

[43204] 16756.Watson, D. K.; McWilliams-Smith, M. J.; Kozak, C.; Reeves, R.; Gearhart, J.; Nunn, M. F.; Nash, W.; Fowle, J. R., III; Duesberg, P.; Papas, T. S.; O'Brien, S. J.: Conserved chromosomal positions of dual domains of the ets protooncogene in cats, mice, and humans. *Proc.Nat. Acad. Sci.* 83: 1792–1796, 1986.

[43205] 16757.Watson, D. K.; McWilliams-Smith, M. J.; Nunn, M. F.; Duesberg, P. H.; O'Brien, S. J.; Papas, T. S.: The ets sequence from the transforming gene of avian erythroblasto-

sis virus, E26, has unique domains on human chromosomes 11 and 21: both loci are transcriptionally active. Proc.Nat. Acad. Sci. 82: 7294–7298, 1985.

- [43206] 16758. Bellacosa, A.; Franke, T. F.; Gonzalez-Portal, M. E.; Datta, K.; Taguchi, T.; Gardner, J.; Cheng, J. Q.; Testa, J. R.; Tsichlis, P.N.: Structure, expression and chromosomal mapping of c-akt: relationship to v-akt and its implications. *Oncogene* 8: 745–754, 1993.
- [43207] 16759. Brunet, A.; Bonni, A.; Zigmond, M. J.; Lin, M. Z.; Juo, P.; Hu, L. S.; Anderson, M. J.; Arden, K. C.; Blenis, J.; Greenberg, M. E.: Akt promotes cell survival by phosphorylating and inhibiting a Forkhead transcription factor. *Cell* 96: 857–868, 1999.
- [43208] 16760. Chen, W. S.; Xu, P.-Z.; Gottlob, K.; Chen, M.-L.; Sokol, K.; Shiyanova, T.; Roninson, I.; Weng, W.; Suzuki, R.; Tobe, K.; Kadowaki, T.; Hay, N.: Growth retardation and increased apoptosis in mice with homozygous disruption of the akt1 gene. *Genes Dev.* 15: 2203–2208, 2001.
- [43209] 16761. Condorelli, G.; Drusco, A.; Stassi, G.; Bellacosa, A.; Roncarati, R.; Iaccarino, G.; Russo, M. A.; Gu, Y.; Dalton, N.; Chung, C.; Latronico, M. V. G.; Napoli, C.; Sadoshima, J.; Croce, C. M.; Ross, J., Jr.: Akt induces enhanced myocardial contractility and cell size in vivo in transgenic mice. *Proc.*

Nat. Acad. Sci. 99: 12333–12338, 2002.

- [43210] 16762.Ellis, N. A.: Ecce Ohno. Nature Genet. 10: 373–375, 1995.
- [43211] 16763.Haldane, J. B. S.: Sex ratio and unisexual sterility in hybrid animals. J. Genet. 12: 101–109, 1922.
- [43212] 16764.Milatovich, A.; Kitamura, T.; Miyajima, A.; Francke, U.: Gene for the alpha-subunit of the human interleukin-3 receptor (IL3RA) localized to the X-Y pseudoautosomal region. Am. J. Hum. Genet. 53:1146–1153, 1993.
- [43213] 16765.Ohno, S.: Sex Chromosomes and Sex-linked Genes. Berlin and New York: Springer (pub.) 1967.
- [43214] 16766.Palmer, S.; Perry, J.; Ashworth, A.: A contravention of Ohno's law in mice. Nature Genet. 10: 472–476, 1995.
- [43215] 16767.Rugarli, E. I.; Adler, D. A.; Borsani, G.; Tsuchiya, K.; Franco, B.; Hauge, X.; Disteche, C.; Chapman, V.; Ballabio, A.: Different chromosomal localization of the Clcn4 gene in Mus spretus and C57BL/6J mice. Nature Genet. 10: 466–471, 1995.
- [43216] 16768.Schnur, R. E.; Wick, P. A.: Intragenic TaqI restriction fragment length polymorphism (RFLP) in CLCN4, between the loci for X-linked ocular albinism (OA1) and microphthalmia with linear skin defect syndrome (MLS). Hum. Genet. 95: 594–595, 1995.

- [43217] 16769.van Slegtenhorst, M. A.; Bassi, M. T.; Borsani, G.; Wapenaar, M.C.; Ferrero, G. B.; de Conciliis, L.; Rugarli, E. I.; Grillo, A.;Franco, B.; Zoghbi, H. Y.; Ballabio, A.: A gene from the Xp22.3 regionshares homology with voltage-gated chloride channels. *Hum. Molec.Genet.* 3: 547–552, 1994.
- [43218] 16770.Curry, C. J. R.; Magenis, R. E.; Brown, M.; Lanman, J. T., Jr.;Tsai, J.; O'Lague, P.; Goodfellow, P.; Mohandas, T.; Bergner, E. A.;Shapiro, L. J.: Inherited chondrodysplasia punctata due to a deletionof the terminal short arm of an X chromosome. *New Eng. J. Med.* 311:1010–1015, 1984.
- [43219] 16771.Maslen, G. L.; Boyd, Y.: Comparative mapping of the Grpr locuson the X chromosomes of man and mouse. *Genomics* 17: 106–109, 1993.
- [43220] 16772.Schantz, L. J.; Naylor, S. L.; Giladi, E.; Spindel, E. R.: Assignmentof the GRP receptor gene to the human X chromosome. (Abstract) *Cytogenet.Cell Genet.* 58: 2085–2086, 1991.
- [43221] 16773.Shiraishi, M.; Alitalo, T.; Sekiya, T.: The chromosomal organizationof the human endogenous retrovirus-like sequence HERV-H: clusteringof the HERV-H sequences in a 300-kb region close to the GRPR locuson the X chromosome. *DNA Res.* 3: 425–429, 1996.

- [43222] 16774. Shriver, S. P.; Bourdeau, H. A.; Gubish, C. T.; Tirpak, D. L.; Davis, A. L. G.; Luketich, J. D.; Siegfried, J. M.: Sex-specific expression of gastrin-releasing peptide receptor: relationship to smoking history and risk of lung cancer. *J. Nat. Cancer Inst.* 92:24–33, 2000.
- [43223] 16775. Spindel, E. R.; Giladi, E.; Brehm, P.; Goodman, R. H.; Segerson, T. P.: Cloning and functional characterization of a complementary DNA encoding the murine fibroblast bombesin/gastrin-releasing peptide receptor. *Molec. Endocr.* 4: 1956–1963, 1990.
- [43224] 16776. Ma, K.; Sharkey, A.; Kirsch, S.; Vogt, P.; Keil, R.; Hargreave, T. B.; McBeath, S.; Chandley, A. C.: Towards the molecular localisation of the AZF locus: mapping of microdeletions in azoospermic men within 14 subintervals of interval 6 of the human Y chromosome. *Hum. Molec. Genet.* 1: 29–33, 1992.
- [43225] 16777. Adam, A.; Tippet, P.; Gavin, J.; Noades, J.; Sanger, R.; Race, R. R.: The linkage relation of Xg to G6PD in Israelis: the evidence of a second series of families. *Am. J. Hum. Genet.* 30: 211–218, 1966.
- [43226] 16778. Ahluwalia, A.; Corcoran, C. M.; Vulliamy, T. J.; Ishwad, C. S.; Naidu, J. M.; Argusti, A.; Stevens, D. J.; Mason, P. J.; Luzzatto, L.: G6PD Kalyan and G6PD Kerala; two defi-

cient variants in India caused by the same 317 glu-to-lys mutation. Hum. Molec. Genet. 3:209–210, 1992.

[43227] 16779. Aksoy, K.; Yuregir, G. T.; Dikmen, N.; Unlukurt, I.: Three new G6PD variants, G6PD Adana, G6PD Samandag, and G6PD Balcali in Cukurova, Turkey. Hum. Genet. 76: 199–201, 1987.

[43228] 16780. Alfinito, F.; Cimmino, A.; Ferraro, F.; Cubellis, M. V.; Vitagliano, L.; Francese, M.; Zagari, A.; Rotoli, B.; Filosa, S.; Martini, G.: Molecular characterization of G6PD deficiency in Southern Italy: heterogeneity, correlation genotype–phenotype and description of a new variant (G6PD Neapolis). Brit. J. Haemat. 98: 41–46, 1997.

[43229] 16781. Alperin, J. B.; Mills, G. C.: New variants of glucose-6-phosphate dehydrogenase (G6PD). Clin. Res. 20: 76, 1972.

[43230] 16782. Azevedo, E.; Kirkman, H. N.; Morrow, A. C.; Motulsky, A. G.: Variants of red cell glucose-6-phosphate dehydrogenase among Asiatic Indians. Ann. Hum. Genet. 31: 373–379, 1968.

[43231] 16783. Azevedo, E. S.; Yoshida, A.: Brazilian variant of glucose-6-phosphate dehydrogenase (GD Minas Gerais). Nature 222: 380–382, 1969.

[43232] 16784. Babalola, A. O. G.; Beetlestone, J. G.; Luzzatto, L.:

Genetic variants of human erythrocyte glucose-6-phosphate dehydrogenase: kinetic and thermodynamic parameters of variants A, B, and A- in relation to quaternary structure. *J. Biol. Chem.* 251: 2993-3002, 1976.

[43233] 16785. Balinsky, D.; Cayanis, E.; Carter, G.; Jenkins, T.; Bersohn, I.: A new variant of human erythrocyte glucose-6-phosphate dehydrogenase: G6PD Port Elizabeth. *Int. J. Biochem.* 4: 235-244, 1973.

[43234] 16786. Balinsky, D.; Gomperts, E.; Cayanis, E.; Jenkins, T.; Bryer, D.; Bersohn, I.; Metz, J.: Glucose 6-phosphate dehydrogenase Johannesburg: a new variant with reduced activity in a patient with congenital non-spherocytic haemolytic anaemia. *Brit. J. Haemat.* 25: 385-392, 1973.

[43235] 16787. Balinsky, D.; Rootman, A. J.; Nurse, G. T.; Cayanis, E.; Lane, A.; Jenkins, T.; Bersohn, I.: G6PD Kuanyama: a new variant of human erythrocyte glucose 6-phosphate dehydrogenase showing slower than normal electrophoretic mobility. *S. Afr. J. Med. Sci.* 39: 5-13, 1974.

[43236] 16788. Barretto, O. C.; Nonoyama, K.: Gd(+) Cuiaba, a new rare glucose-6-phosphate dehydrogenase variant presenting normal activity. *Hum. Genet.* 77: 201-202, 1987.

[43237] 16789. Kornak, U.; Kasper, D.; Bosl, M. R.; Kaiser, E.;

Schweizer, M.;Schulz, A.; Friedrich, W.; Delling, G.;
Jentsch, T. J.: Loss of theClC-7 chloride channel leads to
osteopetrosis in mice and man. Cell 104:205–215, 2001.

[43238] 16790.Kornak, U.; Schulz, A.; Friedrich, W.; Uhlhaas, S.;
Kremens, B.;Voit, T.; Hasan, C.; Bode, U.; Jentsch, T. J.;
Kubisch, C.: Mutationsin the $\alpha 3$ subunit of the vacuolar
H(+)-ATPase cause infantile malignantosteopetrosis.
Hum. Molec. Genet. 9: 2059–2063, 2000.

[43239] 16791.Rosenberg, C. L.; Wong, E.; Petty, E. M.; Bale, A. E.;
Tsujimoto,Y.; Harris, N. L.; Arnold, A.: PRAD1, a candidate
BCL1 oncogene:mapping and expression in centrocytic
lymphoma. Proc. Nat. Acad.Sci. 88: 9638–9642, 1991.

[43240] 16792.Tsujimoto, Y.; Jaffe, E.; Cossman, J.; Gorham, J.;
Nowell, P.C.; Croce, C. M.: Clustering of breakpoints on
chromosome 11 in humanB-cell neoplasms with the
t(11;14) chromosome translocation. Nature 315:340–343,
1985.

[43241] 16793.Tsujimoto, Y.; Yunis, J.; Onorato–Showe, L.; Erik–
son, J.; Nowell,P. C.; Croce, C. M.: Molecular cloning of the
chromosomal breakpointof B-cell lymphomas and
leukemias with the t(11;14) chromosome translocation.
Science 224:1403–1406, 1984.

[43242] 16794.Barbany, G.; Hoglund, M.; Simonsson, B.: Complete

molecular remission in chronic myelogenous leukemia after imatinib therapy. (Letter) *New Eng. J. Med.* 347: 539–540, 2002.

[43243] 16795. Budarf, M.; Canaani, E.; Emanuel, B. S.: Linear order of the four BCR-related loci in 22q11. *Genomics* 3: 168–171, 1988.

[43244] 16796. Castellanos, A.; Pintado, B.; Weruaga, E.; Arevalo, R.; Lopez, A.; Orfao, A.; Sanchez-Garcia, I.: A BCR-ABL(p190) fusion gene made by homologous recombination causes B-cell acute lymphoblastic leukemia in chimeric mice with independence of the endogenous bcr product. *Blood* 90:2168–2174, 1997. 1. Brunning, R. D.: Philadelphia chromosome positive leukemia. *Hum. Path.* 11: 307–309, 1980.

[43245] 16797. Chisoe, S. L.; Bodenteich, A.; Wang, Y.-F.; Wang, Y.-P.; Burian, D.; Clifton, S. W.; Crabtree, J.; Freeman, A.; Iyer, K.; Jian, L.; Ma, Y.; McLaury, H.-J.; Pan, H.-Q.; Sarhan, O. H.; Toth, S.; Wang, Z.; Zhang, G.; Heisterkamp, N.; Groffen, J.; Roe, B. A.: Sequence and analysis of the human ABL gene, the BCR gene, and regions involved in the Philadelphia chromosomal translocation. *Genomics* 27: 67–82, 1995.

[43246] 16798. Court Brown, W. M.; Doll, R.: Mortality from cancer

and other causes after radiotherapy for ankylosing spondylitis. *Brit. Med.J.* 2: 1327–1332, 1965.

- [43247] 16799. Croce, C. M.; Huebner, K.; Isobe, M.; Fainstain, E.; Lifshitz, B.; Shtivelman, E.; Canaani, E.: Mapping of four distinct BCR-related loci to chromosome region 22q11: order of BCR loci relative to chronic myelogenous leukemia and acute lymphoblastic leukemia breakpoints. *Proc. Nat. Acad. Sci.* 84: 7174–7178, 1987.
- [43248] 16800. Daley, G. Q.; Van Etten, R. A.; Baltimore, D.: Induction of chronic myelogenous leukemia in mice by the P210(bcr/abl) gene of the Philadelphia chromosome. *Science* 247: 824–830, 1990.
- [43249] 16801. de Klein, A.; Geurts van Kessel, A.; Grosveld, G.; Bartram, C.R.; Hagemeijer, A.; Bootsma, D.; Spurr, N. K.; Heisterkamp, N.; Groffen, J.; Stephenson, J. R.: A cellular oncogene is translocated to the Philadelphia chromosome in chronic myelocytic leukaemia. *Nature* 300: 765–767, 1982.
- [43250] 16802. Diekmann, D.; Brill, S.; Garrett, M. D.; Totty, N.; Hsuan, J.; Monfries, C.; Hall, C.; Lim, L.; Hall, A.: Bcr encodes a GTPase-activating protein for p21(rac). *Nature* 351: 400–402, 1991.
- [43251] 16803. Druker, B. J.; Sawyers, C. L.; Kantarjian, H.; Resta,

D. J.; Reese, S. F.; Ford, J. M.; Capdeville, R.; Talpaz, M.: Activity of a specific inhibitor of the BCR-ABL tyrosine kinase in the blast crisis of chronic myeloid leukemia and acute lymphoblastic leukemia with the Philadelphia chromosome. *New Eng. J. Med.* 344: 1038-1042, 2001.

[43252] 16804. Druker, B. J.; Talpaz, M.; Resta, D. J.; Peng, B.; Buchdunger, E.; Ford, J. M.; Lydon, N. B.; Kantarjian, H.; Capdeville, R.; Ohno-Jones, S.; Sawyers, C. L.: Efficacy and safety of a specific inhibitor of the BCR-ABL tyrosine kinase in chronic myeloid leukemia. *New Eng. J. Med.* 344: 1031-1037, 2001.

[43253] 16805. Emanuel, B. S.; Selden, J. R.; Wang, E.; Nowell, P. C.; Croce, C. M.: In situ hybridization and translocation breakpoint mapping. I. Non-identical 22q11 breakpoints for the t(9;22) of CML and the t(8;22) of Burkitt lymphoma. *Cytogenet. Cell Genet.* 38: 127-131, 1984.

[43254] 16806. Grandori, C.; Mac, J.; Siebelt, F.; Ayer, D. E.; Eisenman, R. N.: Myc-Max heterodimers activate a DEAD box gene and interact with multiple E box-related sites in vivo. *EMBO J.* 15: 4344-4357, 1996.

[43255] 16807. Gallagher, A. R.; Cedzich, A.; Gretz, N.; Somlo, S.; Witzgall, R.: The polycystic kidney disease protein PKD2 interacts with Hax-1, a protein associated with the actin

cytoskeleton. *Proc. Nat. Acad.Sci.* 97: 4017–4022, 2000.

[43256] 16808.Cleutjens, K. B. J. M.; van Eekelen, C. C. E. M.; van der Korput,H. A. G. M.; Brinkmann, A. O.; Trapman, J.: Two androgen responseregions cooperate in steroid hormone regulated activity of the prostate–specificantigen promoter. *J. Biol. Chem.* 271: 6379–6388, 1996.

[43257] 16809.Evans, B. A.; Drinkwater, C. C.; Richards, R. I.: Mouse glandularkallikrein genes: structure and partial sequence analysis of the kallikreingene locus. *J. Biol. Chem.* 262: 8027–8034, 1987.

[43258] 16810.Lundwall, A.; Lilja, H.: Molecular cloning of human prostate specificantigen cDNA. *FEBS Lett.* 214: 317–322, 1987.

[43259] 16811.Melegos, D. N.; Yu, H.; Ashok, M.; Wang, C.; Stanczyk, F.; Diamandis,E. P.: Prostate–specific antigen in female serum, a potential newmarker of androgen excess. *J. Clin. Endocr. Metab.* 82: 777–780,1997.

[43260] 16812.Prendergast, G. C.; Lawe, D.; Ziff, E. B.: Association of Myn,the murine homolog of Max, with c–Myc stimulates methylation–sensitiveDNA binding and Ras cotransformation. *Cell* 65: 395–408, 1991.

[43261] 16813.Wagner, A. J.; Le Beau, M. M.; Diaz, M. O.; Hay, N.: Expression,regulation, and chromosomal localization of

the Max gene. Proc. Nat.Acad. Sci. 89: 3111–3115, 1992.

- [43262] 16814.Zervos, A. S.; Faccio, L.; Gatto, J. P.; Kyriakis, J. M.; Brent,R.: Mxi2, a mitogen–activated protein kinase that recognizes andphosphorylates Max protein. Proc. Nat. Acad. Sci. 92: 10531–10534,1995.
- [43263] 16815.Emi, M.; Katagiri, T.; Harada, Y.; Saito, H.; Inazawa, J.; Ito,I.; Kasumi, F.; Nakamura, Y.: A novel metallopro–tease/disintegrin–likegene at 17q21.3 is somatically rear–ranged in two primary breast cancers. NatureGenet. 5: 151–157, 1993.
- [43264] 16816.Katagiri, T.; Harada, Y.; Emi, M.; Nakamura, Y.: Hu–man metalloprotease/disintegrin–like(MDC) gene: exon–intron organization and alternative splicing. Cyto–genet.Cell Genet. 68: 39–44, 1995.
- [43265] 16817.Begley, C. G.; Lipkowitz, S.; Gobel, V.; Mahon, K. A.; Bertness,V.; Green, A. R.; Gough, N. M.; Kirsch, I. R.: Molecular characterizationof NSCL, a gene encoding a he–lix–loop–helix protein expressed in thedeveloping ner–vous system. Proc. Nat. Acad. Sci. 89: 38–42, 1992.
- [43266] 16818.Brown, L.; Espinosa, R., III; Le Beau, M. M.; Siciliano, M. J.;Baer, R.: HEN1 and HEN2: a subgroup of basic helix–loop–helix genesthat are coexpressed in a human neu–roblastoma. Proc. Nat. Acad. Sci. 89:8492–8496, 1992.

- [43267] 16819.Cogliati, T.; Good, D. J.; Haigney, M.; Delgado-Romero, P.; Eckhaus, M. A.; Koch, W. J.; Kirsch, I. R.: Pre-disposition to arrhythmia and autonomic dysfunction in Nhlh1-deficient mice. *Molec. Cell. Biol.* 22:4977–4983, 2002.
- [43268] 16820.Lipkowitz, S.; Gobel, V.; Varterasian, M. L.; Nakahara, K.; Tchorz, K.; Kirsch, I. R.: A comparative structural characterization of the human NSCL-1 and NSCL-2 genes: two basic helix-loop-helix genes expressed in the developing nervous system. *J. Biol. Chem.* 267: 21065–21071, 1992.
- [43269] 16821.Mullick, A.; Groulx, N.; Trasler, D.; Gros, P.: Nhlh1, a basic helix-loop-helix transcription factor, is very tightly linked to the mouse looptail (Lp) mutation. *Mammalian Genome* 6: 700–704, 1995.
- [43270] 16822.Meeker, T. C.; Nagarajan, L.; ar-Rushdi, A.; Rovera, G.; Huebner, K.; Croce, C. M.: Characterization of the human PIM-1 gene: a putative proto-oncogene coding for a tissue specific member of the protein kinase family. *Oncogene Res.* 1: 87–101, 1987.
- [43271] 16823.Pasqualucci, L.; Neumeister, P.; Goossens, T.; Nangud, G.; Chaganti, R. S. K.; Kuppers, R.; Dalla-Favera, R.: Hypermutation of multiple proto-oncogenes in B-cell

diffuse large-cell lymphomas. *Nature* 412:341–346, 2001.

- [43272] 16824. Ragoussis, J.; Senger, G.; Mockridge, I.; Sanseau, P.; Ruddy, S.; Dudley, K.; Sheer, D.; Trowsdale, J.: A testis-expressed Zn finger gene (ZNF76) in human 6p21.3 centromeric to the MHC is closely linked to the human homolog of the t-complex gene *tcp-11*. *Genomics* 14:673–679, 1992.
- [43273] 16825. Saris, C. J. M.; Domen, J.; Berns, A.: The *pim-1* oncogene encodes two related protein-serine/threonine kinases by alternative initiation at AUG and CUG. *EMBO J.* 10: 655–664, 1991.
- [43274] 16826. Selten, G.; Cuypers, H. T.; Boelens, W.; Robanus-Maandag, E.; Verbeek, J.; Domen, J.; van Beveren, C.; Berns, A.: The primary structure of the putative oncogene *pim-1* shows extensive homology with protein kinases. *Cell* 46: 603–611, 1986.
- [43275] 16827. Zoghbi, H. Y.; Ballantyne, C. M.; O'Brien, W. E.; McCall, A. E.; Kwiatkowski, T. J., Jr.; Ledbetter, S. A.; Beaudet, A. L.: Deletion and linkage mapping of eight markers from the proximal short arm of chromosome 6. *Genomics* 6: 352–357, 1990.
- [43276] 16828. Chapin, S. J.; Lue, C.-M.; Yu, M. T.; Bulinski, J. C.: Differential expression of alternatively spliced forms of

MAP4: a repertoire of structurally different microtubule-binding domains. *Biochemistry* 34:2289–2301, 1995.

- [43277] 16829. Laird, P. W.; van der Lugt, N. M. T.; Clarke, A.; Domen, J.; Linders, K.; McWhir, J.; Berns, A.; Hooper, M.: In vivo analysis of Pim-1 deficiency. *Nucleic Acids Res.* 21: 4750–4755, 1993.
- [43278] 16830. Gartner, J.; Obie, C.; Watkins, P.; Valle, D.: Restoration of peroxisome biogenesis in a peroxisome-deficient mammalian cell line by expression of either the 35 kDa or the 70 kDa peroxisomal membrane proteins. *J. Inherit. Metab. Dis.* 17: 327–329, 1994.
- [43279] 16831. Maric, S. C.; Crozat, A.; Janne, O. A.: Structure and organization of the human S-adenosylmethionine decarboxylase gene. *J. Biol. Chem.* 267:18915–18923, 1992.
- [43280] 16832. Maric, S. C.; Crozat, A.; Louhimo, J.; Knuutila, S.; Janne, O. A.: The human S-adenosylmethionine decarboxylase gene: nucleotide sequence of a pseudogene and chromosomal localization of the active gene (AMD1) and the pseudogene (AMD2). *Cytogenet. Cell Genet.* 70:195–199, 1995.
- [43281] 16833. Mangan, M. E.; Olmsted, J. B.: The gene for microtubule-associated protein 4 (Mtap4) maps to the distal region of mouse chromosome 9. *Mammalian Genome* 7:

918–925, 1996.

- [43282] 16834. West, R. R.; Tenbarger, K. M.; Olmsted, J. B.: A model for microtubule-associated protein 4 structure: domains defined by comparisons of human, mouse, and bovine sequences. *J. Biol. Chem.* 266: 21886–21896, 1991.
- [43283] 16835. Abel, K. J.; Boehnke, M.; Prahalad, M.; Ho, P.; Flejter, W. L.; Watkins, M.; VanderStoep, J.; Chandrasekharappa, S. C.; Collins, F.S.; Glover, T. W.; Weber, B. L.: A radiation hybrid map of the BRCA1 region of chromosome 17q12–q21. *Genomics* 17: 632–641, 1993.
- [43284] 16836. Alonso, A. D. C.; Grundke-Iqbal, I.; Iqbal, K.: Alzheimer's disease hyperphosphorylated tau sequesters normal tau into tangles of filaments and disassembles microtubules. *Nature Med.* 2: 783–787, 1996.
- [43285] 16837. Arima, K.; Kowalska, A.; Hasegawa, M.; Mukoyama, M.; Watanabe, R.; Kawai, M.; Takahashi, K.; Iwatsubo, T.; Tabira, T.; Sunohara, N.: Two brothers with frontotemporal dementia and parkinsonism with an N279K mutation of the tau gene. *Neurology* 54: 1787–1795, 2000.
- [43286] 16838. Baker, M.; Litvan, I.; Houlden, H.; Adamson, J.; Dickson, D.; Perez-Tur, J.; Hardy, J.; Lynch, T.; Bigio, E.; Hutton, M.: Association of an extended haplotype in the

tau gene with progressive supranuclear palsy. Hum.Molec. Genet. 8: 711–715, 1999.

- [43287] 16839.Brown, J.; Lantos, P. L.; Roques, P.; Fidani, L.; Rossor, M. N.: Familial dementia with swollen achromatic neurons and corticobasal inclusion bodies: a clinical and pathological study. J. Neurol. Sci. 135:21–30, 1996.
- [43288] 16840.Clark, L. N.; Poorkaj, P.; Wszolek, Z.; Geschwind, D. H.; Nasreddine, Z. S.; Miller, B.; Li, D.; Payami, H.; Awert, F.; Markopoulou, K.; Andreadis, A.; D'Souza, I.; Lee, V. M.–Y.; Reed, L.; Trojanowski, J. Q.; Zhukareva, V.; Bird, T.; Schellenberg, G.; Wilhelmsen, K. C.: Pathogenic implications of mutations in the tau gene in pallido–ponto–nigral degeneration and related neurodegenerative disorders linked to chromosome 17. Proc. Nat. Acad. Sci. 95: 13103–13107, 1998.
- [43289] 16841.Connell, J. W.; Gibb, G. M.; Betts, J. C.; Blackstock, W. P.; Gallo, J.–M.; Lovestone, S.; Hutton, M.; Anderton, B. H.: Effects of FTDP–17 mutations on the in vitro phosphorylation of tau by glycogen synthase kinase 3–beta identified by mass spectrometry demonstrate certain mutations exert long–range conformational changes. FEBS Lett. 493:40–44, 2001.
- [43290] 16842.Conrad, C.; Andreadis, A.; Trojanowski, J. Q.; Dick–

son,D. W.; Kang,D.; Chen, X.; Weiderholt, W.; Hansen, L.; Masliah, E.; Thal, L. J.;Katzman, R.; Xia, Y.; Saitoh, T.: Genetic evidence for the involvementof tau in progressive supranuclear palsy. *Ann. Neurol.* 41: 277–281,1997.

[43291] 16843.Conrad, C.; Vianna, C.; Freeman, M.; Davies, P.: A polymorphicgene nested within an intron of the tau gene: implications for Alzheimer'sdisease. *Proc. Nat. Acad. Sci.* 99: 7751–7756, 2002.

[43292] 16844.Delisle, M. B.; Murrell, J. R.; Richardson, R.; Trofatter, J.A.; Rascol, O.; Soulages, X.; Mohr, M.; Calvas, P.; Ghetti, B.: A mutation at codon 279 (N279K) in exon 10 of the tau gene causes a tauopathy with dementia and supranuclear palsy. *Acta Neuropath.* 98:62–77, 1999.

[43293] 16845.Donlon, T. A.; Harris, P.; Neve, R. L.: Localization of microtubule-associated protein tau (MTBT1) to chromosome 17q21. (Abstract) *Cytogenet. CellGenet.* 46: 607, 1987.

[43294] 16846.Goedert, M.; Crowther, R. A.; Spillantini, M. G.: Tau mutations cause frontotemporal dementias. *Neuron* 21: 955–958, 1998.

[43295] 16847.Goedert, M.; Spillantini, M. G.; Potier, M. C.; Ulrich, J.; Crowther, R. A.: Cloning and sequencing of the cDNA encoding an isoform of microtubule-associated protein tau

containing four tandem repeats:differential expression of tau protein mRNAs in human brain. EMBOJ. 8: 393–399, 1989.

- [43296] 16848.Goedert, M.; Wischik, C. M.; Crowther, R. A.; Walker, J. E.; Klug,A.: Cloning and sequencing of the cDNA encoding a core protein ofthe paired helical filament of Alzheimer disease: identification asthe microtubule–associated protein tau. Proc. Nat. Acad. Sci. 85:4051–4055, 1988.
- [43297] 16849.Goode, B. L.; Chau, M.; Denis, P. E.; Feinstein, S. C.: Structuraland functional differences between 3–repeat and 4–repeat tau isoforms:implications for normal tau function and the onset of neurodegenerativedisease. J. Biol. Chem. 275: 38182–38189, 2000.
- [43298] 16850.Heutink, P.: Untangling tau–related dementia. Hum. Molec. Genet. 9:979–986, 2000.
- [43299] 16851.Heutink, P.; Stevens, M.; Rizzu, P.; Bakker, E.; Kros, J. M.;Tibben, A.; Niermeijer, M. F.; van Duijn, C. M.; Oostra, B. A.; vanSwieten, J. C.: Hereditary frontotemporal dementia is linked to chromosome17q21–q22: a genetic and clinicopathological study of three Dutchfamilies. Ann. Neurol. 41: 150–159, 1997.
- [43300] 16852.Hiesberger, T.; Trommsdorff, M.; Howell, B. W.;

Goffinet, A.;Mumby, M. C.; Cooper, J. A.; Herz, J.: Direct binding of reelin toVLDL receptor and apoE receptor 2 induces tyrosine phosphorylationof disabled-1 and modulates tau phosphorylation. Neuron 24: 481–489,1999.

[43301] 16853.Hong, M.; Zhukareva, V.; Vogelsberg–Ragaglia, V.; Wszolek, Z.;Reed, L.; Miller, B. I.; Geschwind, D. H.; Bird, T. D.; McKeel, D.;Goate, A.; Morris, J. C.; Wilhelmsen, K. C.; Schellenberg, G. D.;Trojanowski, J. Q.; Lee, V. M.–Y.: Mutation–specific functional impairmentsin distinct tau isoforms of hereditary FTDP–17. Science 282: 1914–1917,1998.

[43302] 16854.Hutton, M.: Missense and splice site mutations in tau associatedwith FTDP–17: multiple pathogenic mechanisms. Neurology 56 (suppl.4): S21–S25, 2001.

[43303] 16855.Hutton, M.; Lendon, C. L.; Rizzu, P.; Baker, M.; Froelich, S.;Houlden, H.; Pickering–Brown, S.; Chakraverty, S.; Isaacs, A.; Grover,A.; Hackett, J.; Adamson, J.; and 39 others: Association of missenseand 5–prime–splice–site mutations in tau with the inherited dementiaFTDP–17. Nature 393: 702–705, 1998.

[43304] 16856.Iijima, M.; Tabira, T.; Poorkaj, P.; Schellenberg, G. D.; Trojanowski,J. Q.; Lee, V. M.; Schmidt, M. L.; Takahashi, K.; Nabika, T.; Matsumoto,T.; Yamashita, Y.; Yosh–

ioka, S.; Ishino, H.: A distinct familialpresenile dementia with a novel missense mutation in the tau gene. *Neuroreport* 10:497–501, 1999.

- [43305] 16857. Ishihara, T.; Hong, M.; Zhang, B.; Nakagawa, Y.; Lee, M. K.; Trojanowski, J. Q.; Lee, V. M.-Y.: Age-dependent emergence and progression of a tauopathy in transgenic mice overexpressing the shortest human tau isoform. *Neuron* 24: 751–762, 1999.
- [43306] 16858. Janssen, J. C.; Warrington, E. K.; Morris, H. R.; Lantos, P.; Brown, J.; Revesz, T.; Wood, N.; Khan, M. N.; Cipolotti, L.; Fox, N. C.; Rossor, M. N.: Clinical features of frontotemporal dementia due to the intronic tau 10 + 16 mutation. *Neurology* 58: 1161–1168, 2002.
- [43307] 16859. Bergstrom, D. A.; Penn, B. H.; Strand, A.; Perry, R. L. S.; Rudnicki, M. A.; Tapscott, S. J.: Promoter-specific regulation of MyoD binding and signal transduction cooperate to pattern gene expression. *Molec. Cell* 9: 587–600, 2002.
- [43308] 16860. Braun, T.; Grzeschik, K.-H.; Bober, E.; Arnold, H.-H.: The MYF genes, a group of human muscle determining factors, are localized on different human chromosomes. (Abstract) *Cytogenet. Cell Genet.* 51:969 only, 1989.

- [43309] 16861.Davis, R. L.; Weintraub, H.; Lassar, A. B.: Expression of a singletransfected cDNA converts fibroblasts to myoblasts. *Cell* 51: 987–1000,1987.
- [43310] 16862.de la Serna, I. L.; Carlson, K. A.; Imbalzano, A. N.: MammalianSWI/SNF complexes promote MyoD-mediated muscle differentiation. *NatureGenet.* 27: 187–190, 2001.
- [43311] 16863.Gessler, M.; Hameister, H.; Henry, I.; Junien, C.; Braun, T.; Arnold,H. H.: The human MyoD1 (MYF3) gene maps on the short arm of chromosome11 but is not associated with the WAGR locus or the region for theBeckwith-Wiedemann syndrome. *Hum. Genet.* 86: 135–138, 1990.
- [43312] 16864.Guttridge, D. C.; Mayo, M. W.; Madrid, L. V.; Wang, C.-Y.; Baldwin,A. S, Jr.: NF-kappa-B-induced loss of MyoD messenger RNA: possiblerole in muscle decay and cachexia. *Science* 289: 2363–2366, 2000.
- [43313] 16865.Henry, I.; Puech, A.; Antignac, C.; Couillin, P.; Jean-pierre, M.;Ahnine, L.; Barichard, F.; Boehm, T.; Augereau, P.; Scrable, H.; Rabbitts,T. H.; Rochefort, H.; Cavenee, W.; Junien, C.: Subregional mappingof BWS, CTSD, MYOD1, and a T-ALL breakpoint in 11p15. (Abstract) *Cytogenet.Cell Genet.* 51: 1013 only, 1989.
- [43314] 16866.Kim, Y.-J.; Noguchi, S.; Hayashi, Y. K.; Tsukahara, T.; Shimizu,T.; Arahata, K.: The product of an oculopha-

ryngeal muscular dystrophy gene, poly(A)-binding protein 2, interacts with SKIP and stimulates muscle-specific gene expression. *Hum. Molec. Genet.* 10: 1129–1139, 2001.

[43315] 16867. Olson, E. N.: MyoD family: a paradigm for development. *Genes Dev.* 4: 1454–1461, 1990.

[43316] 16868. Sartorelli, V.; Puri, P. L.; Hamamori, Y.; Ogryzko, V.; Chung, G.; Nakatani, Y.; Wang, J. Y. J.; Kedes, L.: Acetylation of MyoD directed by PCAF is necessary for the execution of the muscle program. *Molec. Cell* 4: 725–734, 1999.

[43317] 16869. Tapscott, S. J.; Davis, R. L.; Thayer, M. J.; Cheng, P. F.; Weintraub, H.; Lassar, A. B.: MyoD1: a nuclear phosphoprotein requiring a Myc homology region to convert fibroblasts to myoblasts. *Science* 242: 405–411, 1988.

[43318] 16870. Bonner, T. I.; Young, A. C.; de Miguel, C.; Detera-Wadleigh, S.; Modi, W. S.; O'Brien, S. J.: The chromosomal location of the two human tachykinin (sic) genes: human substance P and neurokinin K. (Abstract) *Cytogenet. Cell Genet.* 46: 584 only, 1987.

[43319] 16871. Cao, Y. Q.; Mantyh, P. W.; Carlson, E. J.; Gillespie, A.-M.; Epstein, C. J.; Basbaum, A. I.: Primary afferent tachykinins are required to experience moderate to intense pain. *Nature* 392: 390–393, 1998.

[43320] 16872. Krause, J. E.; Chirgwin, J. M.; Carter, M. S.; Xu, Z. S.;

Hershey,A. D.: Three rat preprotachykinin mRNAs encode the neuropeptidesubstance P and neurokinin A. Proc. Nat. Acad. Sci. 84: 881–885,1987.

- [43321] 16873.Liu, H.; Cao, Y.; Basbaum, A. I.; Mazarati, A. M; Sankar, R.; Wasterlain,C. G.: Resistance to excitotoxin–induced seizures and neuronal deathin mice lacking the preprotachykinin A gene. Proc. Nat. Acad. Sci. 96:12096–12101, 1999.
- [43322] 16874.Troger, J.; Neyer, S.; Heufler, C.; Huemer, H.; Schmid, E.; Griesser,U.; Kralinger, M.; Kremser, B.; Baldissera, I.; Kieselbach, G.: SubstanceP and vasoactive intestinal polypeptide in the streptozotocin–induceddiabetic rat retina. Invest. Ophthal. Vis. Sci. 42: 1045–1050, 2001.
- [43323] 16875.Zimmer, A.; Zimmer, A. M.; Baffi, J.; Usdin, T.; Reynolds, K.;Konig, M.; Palkovits, M.; Mezey, E.: Hypoalgesia in mice with a targeteddeletion of the tachykinin 1 gene. Proc. Nat. Acad. Sci. 95: 2630–2635,1998.
- [43324] 16876.Gregory, C. A.; Schwartz, J. S.: The cDNA of the human neuromedinB gene (NMB) mapped to 15q11–qter recognizes an XbaI RFLP. NucleicAcids Res. 19: 1167 only, 1991.
- [43325] 16877.Krane, I. M.; Naylor, S. L.; Helin–Davis, D.; Chin, W. W.; Spindel,E. R.: Molecular cloning of cDNAs encoding the

human bombesin-like peptide neuromedin B: chromosomal localization and comparison to cDNA encoding its amphibian homolog ranatensin. *J. Biol. Chem.*

263:13317–13323, 1988. Note: Erratum: *J. Biol. Chem.*

265: 7091 only, 1990.

[43326] 16878. Mattei, M.-G.; Riviere, M.; Krust, A.; Ingvarsson, S.; Vennstrom, B.; Islam, M. Q.; Levan, G.; Kautner, P.; Zelent, A.; Chambon, P.; Szpirer, J.; Szpirer, C.: Chromosomal assignment of retinoic acid receptor (RAR) genes in the human, mouse, and rat genomes. *Genomics* 10:1061–1069, 1991.

[43327] 16879. Abramson, D. H.; Ellsworth, R. M.; Zimmerman, L. E.: Monocular cancer in retinoblastoma survivors. *Trans. Am. Acad. Ophthalm. Otolaryng.* 81:454–457, 1976.

[43328] 16880. Aherne, G. E. S.; Roberts, D. F.: Retinoblastoma—a clinical survey and its genetic implications. *Clin. Genet.* 8: 275–290, 1975.

[43329] 16881. Alonso, J.; Garcia-Miguel, P.; Abelairas, J.; Mendiola, M.; Sarret, E.; Vendrell, M. T.; Navajas, A.; Pestana, A.: Spectrum of germline RB1 gene mutations in Spanish retinoblastoma patients: phenotypic and molecular epidemiological implications. *Hum. Mutat.* 17: 412–422, 2001.

- [43330] 16882.Amoaku, W. M. K.; Willshaw, H. E.; Parkes, S. E.; Shah, K. J.;Mann, J. R.: Trilateral retinoblastoma: a report of five patients. *Cancer* 78:858–863, 1996.
- [43331] 16883.Bader, J. L.; Meadows, A. T.; Zimmerman, L. E.; Rorke, L. B.; Voute,P. A.; Champion, L. A. A.; Miller, R. W.: Bilateral retinoblastomawith ectopic intracranial retinoblastoma: trilateral retinoblastoma. *CancerGenet. Cytogenet.* 5: 203–213, 1982.
- [43332] 16884.Balaban–Malenbaum, G.; Gilbert, F.; Nichols, W. W.; Hill, R.; Shields,J.; Meadows, A. T.: A deleted chromosome no. 13 in human retinoblastomacells: relevance to tumorigenesis. *Cancer Genet. Cytogenet.* 3: 243–250,1981.
- [43333] 16885.Bandara, L. R.; Adamczewski, J. P.; Hunt, T.; La Thangue, N. B.: Cyclin A and the retinoblastoma gene product complex with a commontranscription factor. *Nature* 352: 249–251, 1991.
- [43334] 16886.Benedict, W. F.; Murphree, A. L.; Banerjee, A.; Spina, C. A.; Sparkes,M. C.; Sparkes, R. S.: Patient with 13 chromosome deletion: evidencethat the retinoblastoma gene is a recessive cancer gene. *Science* 219:973–975, 1983.
- [43335] 16887.Benedict, W. F.; Xu, H.–J.; Hu, S.–X.; Takahashi, R.: Role ofthe retinoblastoma gene in the initiation and pro–

gression of humancancer. J. Clin. Invest. 85: 988–993, 1990.

[43336] 16888.Bia, B.; Cowell, J. K.: Independent constitutional germline mutationsoccurring in the RB1 gene in cousins with bilateral retinoblastoma. Oncogene 11:977–979, 1995.

[43337] 16889.Blanquet, V.; Turleau, C.; de Grouchy, J.; Creau–Goldberg, N.: Physical map around the retinoblastoma gene: possible genomic imprintingsuggested by Nrul digestion. Genomics 10: 350–355, 1991.

[43338] 16890.Blanquet, V.; Turleau, C.; Gross–Morand, M. S.; Senamaud–Beaufort,C.; Doz, F.; Besmond, C.: Spectrum of germline mutations in the RB1gene: a study of 232 patients with hereditary and non hereditary retinoblastoma. Hum.Molec. Genet. 4: 383–388, 1995.

[43339] 16891.Jiang, Z.; Cote, J.; Kwon, J. M.; Goate, A. M.; Wu, J. Y.: Aberrantsplicing of tau pre–mRNA caused by intronic mutations associated withthe inherited dementia from–totemporal dementia with Parkinsonism linkedto chromosome 17. Molec. Cell. Biol. 20: 4036–4048, 2000.

[43340] 16892.Tapscott, S. J.; Weintraub, H.: MyoD and the regulation of myogenesisby helix–loop–helix proteins. J. Clin. Invest. 87: 1133–1138, 1991.

- [43341] 16893.Futterweit, W.; Ritch, R.; Teekhasaenee, C.; Nelson, E. S.: Coexistence of Prader-Willi syndrome, congenital ectropion uveae with glaucoma, and factor XI deficiency. J.A.M.A. 255: 3280-3282, 1986.
- [43342] 16894.Trapani, J. A.; Browne, K. A.; Dawson, M. J.; Ramsay, R. G.; Eddy, R. L.; Shows, T. B.; White, P. C.; Dupont, B.: A novel gene constitutively expressed in human lymphoid cells is inducible with interferon-gamma in myeloid cells. Immunogenetics 36: 369-376, 1992.
- [43343] 16895.Bowcock, A. M.; Kidd, J. R.; Lathrop, G. M.; Daneshvar, L.; May, L. T.; Ray, A.; Sehgal, P. B.; Kidd, K. K.; Cavalli-Sforza, L. L.: The human 'interferon-beta-2/hepatocyte stimulating factor/interleukin-6' gene: DNA polymorphism studies and localization to chromosome 7p21. Genomics 3:8-16, 1988.
- [43344] 16896.Chen, Y.; Ferguson-Smith, A. C.; Newman, M. S.; May, L. T.; Sehgal, P. B.; Ruddle, F. H.: Regional localization of the human beta 2-interferon gene. (Abstract) Am. J. Hum. Genet. 41: A161, 1987.
- [43345] 16897.Chow, D.; He, X.; Snow, A. L.; Rose-John, S.; Garcia, K. C.: Structure of an extracellular gp130 cytokine receptor signaling complex. Science 291:2150-2155, 2001.
- [43346] 16898.Chung, U.; Tanaka, Y.; Fujita, T.: Association of in-

terleukin-6 and hypoaldosteronism in patients with cancer.
(Letter) New Eng. J. Med. 334: 473, 1996.

- [43347] 16899. Cressman, D. E.; Greenbaum, L. E.; DeAngelis, R. A.; Ciliberto, G.; Furth, E. E.; Poli, V.; Taub, R.: Liver failure and defective hepatocyte regeneration in interleukin-6-deficient mice. Science 274:1379-1382, 1996.
- [43348] 16900. De Benedetti, F.; Alonzi, T.; Moretta, A.; Lazzaro, D.; Costa, P.; Poli, V.; Martini, A.; Ciliberto, G.; Fattori, E.: Interleukin-6 causes growth impairment in transgenic mice through a decrease in insulin-like growth factor-I. J. Clin. Invest. 99: 643-650, 1997.
- [43349] 16901. Ferguson-Smith, A. C.; Chen, Y.-F.; Newman, M. S.; May, L. T.; Sehgal, P. B.; Ruddle, F. H.: Regional localization of the interferon-beta-2/B-cell stimulatory factor 2/hepatocyte stimulating factor gene to human chromosome 7p15-p21. Genomics 2: 203-208, 1988.
- [43350] 16902. Fernandez-Real, J.-M.; Broch, M.; Vendrell, J.; Richart, C.; Ricart, W.: Interleukin-6 gene polymorphism and lipid abnormalities in healthy subjects. J. Clin. Endocr. Metab. 85: 1334-1339, 2000.
- [43351] 16903. Fishman, D.; Faulds, G.; Jeffery, R.; Mohamed-Ali, V.; Yudkin, J. S.; Humphries, S.; Woo, P.: The effect of novel polymorphisms in the interleukin-6 (IL-6) gene on IL-6

transcription and plasma IL-6 levels, and an association with systemic-onset juvenile chronic arthritis. *J. Clin. Invest.* 102: 1369–1376, 1998.

[43352] 16904. Foster, C. B.; Lehrnbecher, T.; Samuels, S.; Stein, S.; Mol, F.; Metcalf, J. A.; Wyvill, K.; Steinberg, S. M.; Kovacs, J.; Blauvelt, A.; Yarchoan, R.; Chanock, S. J.: An IL6 promoter polymorphism is associated with a lifetime risk of development of Kaposi sarcoma in men infected with human immunodeficiency virus. *Blood* 96: 2562–2567, 2000.

[43353] 16905. Funatsu, H.; Yamashita, H.; Noma, H.; Mimura, T.; Yamashita, T.; Hori, S.: Increased levels of vascular endothelial growth factor and interleukin-6 in the aqueous humor of diabetics with macular edema. *Am. J. Ophthalmol.* 133: 70–77, 2002.

[43354] 16906. Hirano, T.; Yasukawa, K.; Harada, H.; Taga, T.; Watanabe, Y.; Matsuda, T.; Kashiwamura, S.; Nakajima, K.; Koyama, K.; Iwamatsu, A.; Tsunasawa, S.; Sakiyama, F.; Matsui, H.; Takahara, Y.; Taniguchi, T.; Kishimoto, T.: Complementary DNA for a novel human interleukin (BSF-2) that induces B lymphocytes to produce immunoglobulin. *Nature* 324: 73–76, 1986.

[43355] 16907. Kawano, M.; Hirano, T.; Matsuda, T.; Taga, T.; Horii, Y.; Iwato, K.; Asaoku, H.; Tang, B.; Tanabe, O.;

Tanaka, H.; Kuramoto, A.; Kishimoto, T.: Autocrine generation and requirement of BSF-2/IL-6 for human multiple myelomas. *Nature* 332: 83–85, 1988.

[43356] 16908. Kovalchuk, A. L.; Kim, J. S.; Park, S. S.; Coleman, A. E.; Ward, J. M.; Morse, H. C, III; Kishimoto, T.; Potter, M.; Janz, S.: IL-6 transgenic mouse model for extraosseous plasmacytoma. *Proc. Nat. Acad. Sci.* 99: 1509–1514, 2002.

[43357] 16909. May, L. T.; Ghrayeb, J.; Santhanam, U.; Tatter, S. B.; Sthoeger, Z.; Helfgott, D. C.; Chiorazzi, N.; Grieninger, G.; Sehgal, P. B.: Synthesis and secretion of multiple forms of beta-2-interferon/B-cell differentiation factor 2/hepatocyte-stimulating factor by human fibroblasts and monocytes. *J. Biol. Chem.* 263: 7760–7766, 1988.

[43358] 16910. Ota, N.; Hunt, S. C.; Nakajima, T.; Suzuki, T.; Hosoi, T.; Orimo, H.; Shirai, Y.; Emi, M.: Linkage of interleukin 6 locus to human osteopenia by sibling pair analysis. *Hum. Genet.* 105: 253–257, 1999.

[43359] 16911. Ota, N.; Nakajima, T.; Nakazawa, I.; Suzuki, T.; Hosoi, T.; Orimo, H.; Inoue, S.; Shirai, Y.; Emi, M.: A nucleotide variant in the promoter region of the interleukin-6 gene associated with decreased bone mineral density. *J. Hum. Genet.* 46: 267–272, 2001.

[43360] 16912. Redwine, L.; Hauger, R. L.; Gillin, J. C.; Irwin, M.:

Effects of sleep and sleep deprivation on interleukin-6, growth hormone, cortisol, and melatonin levels in humans. *J. Clin. Endocr. Metab.* 85: 3597–3603, 2000.

- [43361] 16913. Roodman, G. D.; Kurihara, N.; Ohsaki, Y.; Kukita, A.; Hosking, D.; Demulder, A.; Smith, J. F.; Singer, F. R.: Interleukin 6: a potential autocrine/paracrine factor in Paget's disease of bone. *J. Clin. Invest.* 89:46–52, 1992.
- [43362] 16914. Rooney, M.; David, J.; Symons, J.; Di Giovine, F.; Varsani, H.; Woo, P.: Inflammatory cytokine responses in juvenile chronic arthritis. *Brit. J. Rheum.* 34: 454–460, 1995.
- [43363] 16915. Santhanam, U.; Ray, A.; Sehgal, P. B.: Repression of the interleukin 6 gene promoter by p53 and the retinoblastoma susceptibility gene product. *Proc. Nat. Acad. Sci.* 88: 7605–7609, 1991.
- [43364] 16916. Scheidt-Nave, C.; Bismar, H.; Leidig-Bruckner, G.; Woitge, H.; Seibel, M. J.; Ziegler, R.; Pfeilschifter, J.: Serum interleukin 6 is a major predictor of bone loss in women specific to the first decade past menopause. *J. Clin. Endocr. Metab.* 86: 2032–2042, 2001.
- [43365] 16917. Sehgal, P. B.; May, L. T.; Tamm, I.; Vilcek, J.: Human beta-2 interferon and B-cell differentiation factor BSF-2 are identical. *Science* 235:731–732, 1987.

- [43366] 16918. Sehgal, P. B.; Walther, Z.; Tamm, I.: Rapid enhancement of beta(2)-interferon/B-cell differentiation factor BSF-2 gene expression in human fibroblasts by diacylglycerols and the calcium ionophore A23187. *Proc. Nat. Acad. Sci.* 84: 3663–3667, 1987.
- [43367] 16919. Sehgal, P. B.; Zilberstein, A.; Ruggieri, R.-M.; May, L. T.; Ferguson-Smith, A.; Slate, D. L.; Revel, M.; Ruddle, F. H.: Human chromosome 7 carries the beta-2 interferon gene. *Proc. Nat. Acad. Sci.* 83: 5219–5222, 1986.
- [43368] 16920. Lewis, J.; McGowan, E.; Rockwood, J.; Melrose, H.; Nacharaju, P.; Van Slegtenhorst, M.; Gwinn-Hardy, K.; Murphy, M. P.; Baker, M.; Yu, X.; Duff, K.; Hardy, J.; Corral, A.; Lin, W.-L.; Yen, S.-H.; Dickson, D. W.; Davies, P.; Hutton, M.: Neurofibrillary tangles, amyotrophy and progressive motor disturbance in mice expressing mutant (P301L) tau protein. *Nature Genet.* 25: 402–405, 2000. Note: Erratum: *Nature Genet.* 26: 127 only, 2000.
- [43369] 16921. Lippa, C. F.; Zhukareva, V.; Kawarai, T.; Uryu, K.; Shafiq, M.; Nee, L. E.; Grafman, J.; Liang, Y.; St George-Hyslop, P. H.; Trojanowski, J. Q.; Lee, V. M.-Y.: Frontotemporal dementia with novel tau pathology and a glu342val tau mutation. *Ann. Neurol.* 48: 850–858, 2000.
- [43370] 16922. Litvan, I.; Baker, M.; Hutton, M.: Tau genotype: no

effect on onset, symptom severity, or survival in progressive supranuclear palsy. *Neurology* 57:138–140, 2001.

[43371] 16923. Martin, E. R.; Scott, W. K.; Nance, M. A.; Watts, R. L.; Hubble, J. P.; Koller, W. C.; Lyons, K.; Pahwa, R.; Stern, M. B.; Colcher, A.; Hiner, B. C.; Jankovic, J.; and 20 others: Association of single-nucleotide polymorphisms of the Tau gene with late-onset Parkinson disease. *J.A.M.A.* 286:2245–2250, 2001.

[43372] 16924. Moser, A. B.; Rasmussen, M.; Naidu, S.; Watkins, P. A.; McGuinness, M.; Hajra, A. K.; Chen, G.; Raymond, G.; Liu, A.; Gordon, D.; Garnaas, K.; Walton, D. S.; Skjedal, O. H.; Guggenheim, M. A.; Jackson, L. G.; Elias, E. R.; Moser, H. W.: Phenotype of patients with peroxisomal disorders subdivided into sixteen complementation groups. *J. Pediatr.* 127:13–22, 1995.

[43373] 16925. Paton, B. C.; Heron, S. E.; Nelson, P. V.; Morris, C. P.; Poulos, A.: Absence of mutations raises doubts about the role of the 70-kD peroxisomal membrane protein in Zellweger syndrome. (Letter) *Am. J. Hum. Genet.* 60: 1535–1539, 1997.

[43374] 16926. Aljada, A.; Ghanim, H.; Friedman, J.; Garg, R.; Mohanty, P.; Dandona, P.: Troglitazone reduces the expression of PPAR- γ while stimulating that of PPAR- α

in mononuclear cells in obese subjects. *J. Clin. Endocr. Metab.* 86: 3130–3133, 2001.

- [43375] 16927. Costet, P.; Legendre, C.; More, J.; Edgar, A.; Galtier, P.; Pineau, T.: Peroxisome proliferator-activated receptor alpha-isoform deficiency leads to progressive dyslipidemia with sexually dimorphic obesity and steatosis. *J. Biol. Chem.* 273: 29577–29585, 1998.
- [43376] 16928. Djouadi, F.; Weinheimer, C. J.; Saffitz, J. E.; Pitchford, C.; Bastin, J.; Gonzalez, F. J.; Kelly, D. P.: A gender-related defect in lipid metabolism and glucose homeostasis in peroxisome proliferator-activated receptor alpha-deficient mice. *J. Clin. Invest.* 102: 1083–1091, 1998.
- [43377] 16929. Kersten, S.; Desvergne, B.; Wahli, W.: Roles of PPARs in health and disease. *Nature* 405: 421–424, 2000.
- [43378] 16930. Kersten, S.; Seydoux, J.; Peters, J. M.; Gonzalez, F. J.; Desvergne, B.; Wahli, W.: Peroxisome proliferator-activated receptor alpha mediates the adaptive response to fasting. *J. Clin. Invest.* 103: 1489–1498, 1999.
- [43379] 16931. Lee, S. S.-T.; Pineau, T.; Drago, J.; Lee, E. J.; Owens, J. W.; Kroetz, D. L.; Fernandez-Salguero, P. M.; Westphal, H.; Gonzalez, F. J.: Targeted disruption of the alpha isoform of the peroxisome proliferator-activated receptor gene in mice results in abolishment of the pleiotropic ef-

fects of peroxisome proliferators. *Molec. Cell.Biol.* 15: 3012–3022, 1995.

[43380] 16932.Lee, Y.; Yu, X.; Gonzales, F.; Mangelsdorf, D. J.; Wang, M.–Y.;Richardson, C.; Witters, L. A.; Unger, R. H.: PPAR–alpha is necessaryfor the lipopenic action of hyper–leptinemia on white adipose and livertissue. *Proc. Nat. Acad. Sci.* 99: 11848–11853, 2002.

[43381] 16933.Leone, T. C.; Weinheimer, C. J.; Kelly, D. P.: A critical rolefor the peroxisome proliferator–activated receptor alpha (PPAR–alpha)in the cellular fasting response: the PPAR–alpha–null mouse as a modelof fatty acid oxidation disorders. *Proc. Nat. Acad. Sci.* 96: 7473–7478,1999.

[43382] 16934.Michalik, L.; Desvergne, B.; Tan, N. S.; Basu–Modak, S.; Escher,P.; Rieusset, J.; Peters, J. M.; Kaya, G.; Gonzalez, F. J.; Zakany,J.; Metzger, D.; Chambon, P.; Duboule, D.; Wahli, W.: Impaired skinwound healing in peroxisome proliferator–activated receptor (PPAR)–alphaand PPAR–beta mutant mice. *J. Cell Biol.* 154: 799–814, 2001.

[43383] 16935.Sher, T.; Yi, H.–F.; McBride, O. W.; Gonzalez, F. J.: cDNA cloning,chromosomal mapping, and functional characterization of the humanperoxisome proliferator ac–tivated receptor. *Biochemistry* 32: 5598–5604,1993.

[43384] 16936.Xu, H. E.; Stanley, T. B.; Montana, V. G.; Lambert,

M. H.; Shearer, B. G.; Cobb, J. E.; McKee, D. D.; Galardi, C. M.; Plunket, K. D.; Nolte, R. T.; Parks, D. J.; Moore, J. T.; Kliewer, S. A.; Willson, T. M.; Stimmel, J. B.: Structural basis for antagonist-mediated recruitment of nuclear co-repressors by PPAR- α . *Nature* 415: 813–817, 2002.

[43385] 16937. Ames, G. F.-L.: The basis of multidrug resistance in mammalian cells: homology with bacterial transport. *Cell* 47: 323–324, 1986.

[43386] 16938. Baldini, N.; Scotlandi, K.; Barbanti-Brodano, G.; Manara, M. C.; Maurici, D.; Bacci, G.; Bertoni, F.; Picci, P.; Sottili, S.; Campanacci, M.; Serra, M.: Expression of P-glycoprotein in high-grade osteosarcoma in relation to clinical outcome. *New Eng. J. Med.* 333: 1380–1385, 1995.

[43387] 16939. Bell, D. R.; Trent, J. M.; Willard, H. F.; Riordan, J. R.; Ling, V.: Chromosomal location of human P-glycoprotein gene sequences. *Cancer Genet. Cytogenet.* 25: 141–148, 1987.

[43388] 16940. Callen, D. F.; Baker, E.; Simmers, R. N.; Seshadri, R.; Roninson, I. B.: Localization of the human multiple drug resistance gene, MDR1, to 7q21.1. *Hum. Genet.* 77: 142–144, 1987.

[43389] 16941. Chan, H. S. L.; Haddad, G.; Thorner, P. S.; DeBoer, G.; Lin, Y. P.; Ondrusek, N.; Yeager, H.; Ling, V.: P-

glycoprotein expression as a predictor of the outcome of therapy for neuroblastoma. *New Eng. J. Med.* 325: 1608–1614, 1991.

[43390] 16942. Chang, G.; Roth, C. B.: Structure of MsbA from *E. coli*: a homolog of the multidrug resistance ATP binding cassette (ABC) transporters. *Science* 293:1793–1800, 2001.

[43391] 16943. Chen, C.; Chin, J. E.; Ueda, K.; Clark, D. P.; Pastan, I.; Gottesman, M. M.; Roninson, I. B.: Internal duplication and homology with bacterial transport proteins in the *mdr1* (P-glycoprotein) gene from multidrug-resistant human cells. *Cell* 47: 381–389, 1986.

[43392] 16944. Choi, K. H.; Chen, C.-J.; Kriegler, M.; Roninson, I. B.: An altered pattern of cross-resistance in multidrug-resistant human cells results from spontaneous mutations in the *mdr1* (P-glycoprotein) gene. *Cell* 53:519–529, 1988.

[43393] 16945. Croop, J. M.; Gros, P.; Housman, D. E.: Genetics of multidrug resistance. *J. Clin. Invest.* 81: 1303–1309, 1988.

[43394] 16946. de Lannoy, I. A. M.; Silverman, M.: The MDR1 gene product, P-glycoprotein, mediates the transport of the cardiac glycoside, digoxin. *Biochem. Biophys. Res. Commun.* 189: 551–557, 1992.

[43395] 16947. Cameron, P.; Limjoco, G.; Rodkey, J.; Bennett, C.;

Schmidt, J.A.: Amino acid sequence analysis of human interleukin 1 (IL-1): evidence for biochemically distinct forms of IL-1. *J. Exp. Med.* 162: 790–801, 1985.

[43396] 16948.D'Eustachio, P.; Jadidi, S.; Fuhlbrigge, R. C.; Gray, P. W.; Chaplin, D. D.: Interleukin-1 alpha and beta genes: linkage on chromosome 2 in the mouse. *Immunogenetics* 26: 339–343, 1987.

[43397] 16949.Dinarello, C. A.: An update on human interleukin-1: from molecular biology to clinical relevance. *J. Clin. Immun.* 5: 287–297, 1985.

[43398] 16950.El-Omar, E. M.; Carrington, M.; Chow, W.-H.; McColl, K. E. L.; Bream, J. H.; Young, H. A.; Herrera, J.; Lisowska, J.; Yuan, C.-C.; Rothman, N.; Lanyon, G.; Martin, M.; Fraumeni, J. F., Jr.; Rabkin, C. S.: Interleukin-1 polymorphisms associated with increased risk of gastric cancer. *Nature* 404: 398–402, 2000. Note: Erratum: *Nature* 412: 99 only, 2001.

[43399] 16951.Furutani, Y.; Notake, M.; Yamayoshi, M.; Yamagishi, J.; Nomura, H.; Ohue, M.; Furuta, R.; Fukui, T.; Yamada, M.; Nakamura, S.: Cloning and characterization of the cDNAs for human and rabbit interleukin-1 precursor. *Nucleic Acids Res.* 13: 5869–5882, 1985.

[43400] 16952.Hamajima, N.; Matsuo, K.; Saito, T.; Tajima, K.;

Okuma, K.; Yamao, K.; Tominaga, S.: Interleukin 1 polymorphisms, lifestyle factors, and helicobacter pylori infection. *Jpn. J. Cancer Res.* 92: 383–389, 2001.

[43401] 16953. Le Beau, M. M.; Rowley, J. D.: Personal Communication. Chicago, Ill. 6/18/1986.

[43402] 16954. March, C. J.; Mosley, B.; Larsen, A.; Cerretti, D. P.; Braedt, G.; Price, V.; Gillis, S.; Henney, C. S.; Kronheim, S. R.; Grabstein, K.; Conlon, P. J.; Hopp, T. P.; Cosman, D.: Cloning, sequence and expression of two distinct human interleukin-1 complementary DNAs. *Nature* 315:641–647, 1985.

[43403] 16955. Nicklin, M. J. H.; Weith, A.; Duff, G. W.: A physical map of the region encompassing the human interleukin-1- α , interleukin-1- β , and interleukin-1 receptor antagonist genes. *Genomics* 19: 382–384, 1994.

[43404] 16956. Samad, T. A.; Moore, K. A.; Sapirstein, A.; Billet, S.; Allchorne, A.; Poole, S.; Bonventre, J. V.; Woolf, C. J.: Interleukin-1- β -mediated induction of Cox-2 in the CNS contributes to inflammatory pain hypersensitivity. *Nature* 410:471–475, 2001.

[43405] 16957. Vidal-Vanaclocha, F.; Fantuzzi, G.; Mendoza, L.; Fuentes, A. M.; Anasagasti, M. J.; Martin, J.; Carrascal, T.; Walsh, P.; Reznikov, L. L.; Kim, S.-H.; Novick, D.; Rubin-

stein, M.; Dinarello, C. A.:IL-18 regulates IL-1-beta-dependent hepatic melanoma metastasis via vascular cell adhesion molecule-1. Proc. Nat. Acad. Sci. 97: 734-739,2000.

[43406] 16958.Webb, A.; Collins, K.; Auron, P.; Eddy, R.; Nakai, H.; Byers,M.; Shows, T. B.: Genetics of acute phase response: a gene for interleukin-1is on chromosome 2. (Abstract) Am. J. Hum. Genet. 37: A142 only,1985.

[43407] 16959.Webb, A. C.; Collins, K. L.; Auron, P. E.; Eddy, R. L.; Nakai,H.; Byers, M.; Shows, T. B.: The gene for interleukin-1 (IL1) ison human chromosome 2. (Abstract) Cytogenet. Cell Genet. 40: 774only, 1985.

[43408] 16960.Webb, A. C.; Collins, K. L.; Auron, P. E.; Eddy, R. L.; Nakai,H.; Byers, M. G.; Haley, L. L.; Henry, W. M.; Shows, T. B.: Interleukin-1gene (IL1) assigned to long arm of human chromosome 2. LymphokineRes. 5: 77-85, 1986.

[43409] 16961.Ansano, M.; Toda, M.; Sakaguchi, N.; Sakaguchi, S.: Autoimmunedisease as a consequence of developmental abnormality of a T cellsubpopulation. J. Exp. Med. 184: 387-396, 1996.

[43410] 16962.Cosman, D.; Cerretti, D. P.; Larsen, A.; Park, L.; March, C.; Dower,S.; Gillis, S.; Urdal, D.: Cloning, sequence and expression of humaninterleukin-2 receptor. Nature

312: 768–771, 1984.

- [43411] 16963.Ferrari, S.; Cannizzaro, L. A.; Battini, R.; Huebner, K.; Baserga,R.: The gene encoding human vimentin is located on the short arm of chromosome 10. *Am. J. Hum. Genet.* 41: 616–626, 1987.
- [43412] 16964.Greene, W. C.; Leonard, W. J.; Depper, J. M.; Nelson, D. L.; Waldmann,T. A.: The human interleukin–2 receptor: normal and abnormal expression in T cells and in leukemias induced by the human T–lymphotropic retroviruses. *Ann.Intern. Med.* 105: 560–572, 1986.
- [43413] 16965.Hatakeyama, M.; Minamoto, S.; Taniguchi, T.: Intracytoplasmic phosphorylation sites of Tac antigen (p55) are not essential for the conformation, function, and regulation of the human interleukin 2 receptor. *Proc. Nat. Acad. Sci.* 83: 9650–9654, 1986.
- [43414] 16966.Hatakeyama, M.; Minamoto, S.; Uchiyama, T.; Hardy, R. R.; Yamada,G.; Taniguchi, T.: Reconstitution of functional receptor for human interleukin–2 in mouse cells. *Nature* 318: 467–470, 1985.
- [43415] 16967.Ihle, J. N.; Kerr, I. M.: Jaks and Stats in signaling by the cytokine receptor superfamily. *Trends Genet.* 11: 69–74, 1995.
- [43416] 16968.Ishida, N.; Kanamori, H.; Noma, T.; Nikaido, T.;

Sabe, H.; Suzuki, N.; Shimizu, A.; Honjo, T.: Molecular cloning and structure of the human interleukin 2 receptor gene. *Nucleic Acids Res.* 13: 7579–7589, 1985.

[43417] 16969. Kondo, S.; Shimizu, A.; Maeda, M.; Tagaya, Y.; Yodoi, J.; Honjo, T.: Expression of functional human interleukin-2 receptor in mouse T cells by cDNA transfection. *Nature* 320: 75–77, 1986.

[43418] 16970. Leonard, W. J.; Depper, J. M.; Crabtree, G. R.; Rudikoff, S.; Pumphrey, J.; Robb, R. J.; Kronke, M.; Svetlik, P. B.; Pfeffer, N. J.; Waldmann, T. A.; Greene, W. C.: Molecular cloning and expression of cDNAs for the human interleukin-2 receptor. *Nature* 311: 626–631, 1984.

[43419] 16971. Dahia, P. L. M.; Aguiar, R. C. T.; Alberta, J.; Kum, J. B.; Caron, S.; Sill, H.; Marsh, D. J.; Ritz, J.; Freedman, A.; Stiles, C.; Eng, C.: PTEN is inversely correlated with the cell survival factor Akt/PKB and is inactivated via multiple mechanisms in haematological malignancies. *Hum. Molec. Genet.* 8: 185–193, 1999.

[43420] 16972. Bryson, G. L. M.; Massa, H.; Trask, B. J.; Van Etten, R. L.: Gene structure, sequence, and chromosomal localization of the human red cell-type low molecular weight acid phosphotyrosyl phosphatase gene, ACP1. *Genomics* 30: 133–140, 1995.

- [43421] 16973.Scrable, H. J.; Johnson, D. K.; Rinchik, E. M.; Cave-
nee, W. K.: Rhabdomyosarcoma-associated locus and
MYOD1 are syntenic but separateloci on the short arm of
human chromosome 11. *Proc. Nat. Acad. Sci.*
87:2182-2186, 1990.
- [43422] 16974.Chen, T.-R.; McMorris, F. A.; Creagan, R.; Ricciuti,
F. C.; Tischfield,J.; Ruddle, F. H.: Assignment of the genes
for malate oxidoreductasedecarboxylating to chromosome
6 and peptidase B and lactate dehydrogenaseB to chromo-
some 12 in man. *Am. J. Hum. Genet.* 25: 200-207, 1973.
- [43423] 16975.Hamerton, J. L.; Mohandas, T.; McAlpine, P. J.;
Douglas, G. R.: Localization of human gene loci using
spontaneous chromosome rearrangementsin human-
Chinese hamster somatic cell hybrids. *Am. J. Hum. Genet.*
27:595-608, 1975.
- [43424] 16976.Herbschleb-Voogt, E.; Meera Khan, P.: Defining the
locus of originof a genetically determined electrophoretic
variant of a multilocusenzyme system; the Calcutta-1 of
human LDH system is a B-locus variant. *Hum.Genet.* 57:
290-295, 1981.
- [43425] 16977.Houki, N.; Matsushima, Y.; Kitamura, M.; Tukada,
T.; Nishina, T.;Nakayama, T.: A case of deficiency of lac-
tate dehydrogenase H-subunit. *Jpn.J. Clin. Chem.* 15:

85–90, 1986.

- [43426] 16978. Kitamura, M.; Iijima, N.; Hashimoto, F.; Hiratsuka, A.: Hereditary deficiency of subunit H of lactate dehydrogenase. *Clin. Chim. Acta* 34:419–423, 1971.
- [43427] 16979. Maekawa, M.; Sudo, K.; Kitajima, M.; Matsuura, Y.; Li, S. S.–L.; Kanno, T.: Analysis of a genetic mutation in an electrophoretic variant of the human lactate dehydrogenase–B(H) subunit. *Hum. Genet.* 91:423–426, 1993.
- [43428] 16980. Maekawa, M.; Sudo, K.; Nagura, K.; Li, S. S.–L.; Kanno, T.: Population screening of lactate dehydrogenase deficiencies in Fukuoka Prefecture in Japan and molecular characterization of three independent mutations in the lactate dehydrogenase–B(H) gene. *Hum. Genet.* 93: 74–76, 1994.
- [43429] 16981. Malpuech, G.; Kaplan, J. C.; Rethore, M. O.; Junien, C.; Geneix, A.: Une observation de deletion partielle du bras court du chromosome 12: localisation du gene de la lactate dehydrogenase B. *Lyon Med.* 233:275–279, 1975.
- [43430] 16982. Mayeda, K.; Weiss, L.; Lindahl, R.; Dully, M.: Localization of the human lactate dehydrogenase B gene on the short arm of chromosome 12. *Am. J. Hum. Genet.* 26: 59–64, 1974.
- [43431] 16983. Miwa, S.; Nishima, T.; Kanehashi, Y.; Kitamura, M.;

Hiratsuka,A.; Shizume, K.: Studies on erythrocyte metabolism in a case withhereditary deficiency of H-subunit of lactate dehydrogenase. *ActaHaemat. Jpn.* 34: 228–232, 1971.

[43432] 16984.Bell, D. W.; Varley, J. M.; Szydlo, T. E.; Kang, D. H.; Wahrer,D. C. R.; Shannon, K. E.; Lubratovich, M.; Verselis, S. J.; Isselbacher,K. J.; Fraumeni, J. F.; Birch, J. M.; Li, F. P.; Garber, J. E.; Haber,D. A.: Heterozygous germ line hCHK2 mutations in Li–Fraumeni syndrome. *Science* 286:2528–2531, 1999.

[43433] 16985.Goy, A.; Gilles, F.; Remache, Y.; Zelenetz, A. D.: Physical linkageof the lysyl oxidase–like (LOXL1) gene to the PML gene on human chromosome15q22. *Cytogenet. Cell Genet.* 88: 22–24, 2000.

[43434] 16986.Kenyon, K.; Modi, W. S.; Contente, S.; Friedman, R. M.: A novelhuman cDNA with a predicted protein similar to lysyl oxidase mapsto chromosome 15q24–q25. *J. Biol. Chem.* 268: 18435–18437, 1993.

[43435] 16987.Kim, Y.; Boyd, C. D.; Csiszar, K.: A new gene with sequence andstructural similarity to the gene encoding human lysyl oxidase. *J.Biol. Chem.* 270: 7176–7182, 1995.

[43436] 16988.Wydner, K. S.; Kim, Y.; Csiszar, K.; Boyd, C. D.;

Passmore, H.C.: An intron capture strategy used to identify and map a lysyl oxidase-like gene on chromosome 9 in the mouse. *Genomics* 40: 342–345, 1997.

[43437] 1989. Dudek, H.; Datta, S. R.; Franke, T. F.; Birnbaum, M. J.; Yao, R.; Cooper, G. M.; Segal, R. A.; Kaplan, D. R.; Greenberg, M. E.: Regulation of neuronal survival by the serine–threonine protein kinase Akt. *Science* 275:661–663, 1997.

[43438] 1990. Franke, T. F.; Kaplan, D. R.; Cantley, L. C.; Toker, A.: Direct regulation of the Akt proto-oncogene product by phosphatidylinositol–3,4–bisphosphate. *Science* 275:665–667, 1997.

[43439] 1991. Franke, T. F.; Yang, S.-I.; Chan, T. O.; Datta, K.; Kaziauskas, A.; Morrison, D. K.; Kaplan, D. R.; Tsichlis, P. N.: The protein kinase encoded by the Akt proto-oncogene is a target of the PDGF–activated phosphatidylinositol 3–kinase. *Cell* 81: 727–736, 1995.

[43440] 1992. Furnari, F. B.; Huang, H. J.; Cavenee, W. K.: The phosphoinositol phosphatase activity of PTEN mediates a serum–sensitive G1 growth arrest in glioma cells. *Cancer Res.* 58: 5002–5008, 1998.

[43441] 1993. Hajduch, E.; Litherland, G. J.; Hundal, H. S.: Protein kinase B (PKB/Akt)—a key regulator of glucose transport?

FEBS Lett. 492:199–203, 2001.

[43442] 16994.Hemmings, B. A.: Akt signaling: linking membrane events to lifeand death decisions. Science 275: 628–630, 1997.

[43443] 16995.Jones, P. F.; Jakubowicz, T.; Pitossi, F. J.; Maurer, F.; Hemmings,B. A.: Molecular cloning and identification of a serine/threonineprotein kinase of the second-messenger subfamily. Proc. Nat. Acad.Sci. 88: 4171–4175, 1991.

[43444] 16996.Lee, M.-J.; Thangada, S.; Paik, J.-H.; Sapkota, G. P.; Ancellin,N.; Chae, S.-S.; Wu, M.; Morales–Ruiz, M.; Sessa, W. C.; Alessi, D.R.; Hla, T.: Akt-mediated phosphorylation of the G protein-coupledreceptor EDG–1 is required for endothelial cell chemotaxis. Molec.Cell 8: 693–704, 2001.

[43445] 16997.Maira, S.-M.; Galetic, I.; Brazil, D. P.; Kaech, S.; Ingley, E.;Thelen, M.; Hemmings, B. A.: Carboxyl-terminal modulator protein(CTMP), a negative regulator of PKB/Akt and v-Akt at the plasma membrane. Science 294: 374–380, 2001.

[43446] 16998.Powell, D. W.; Rane, M. J.; Chen, Q.; Singh, S.; McLeish, K. R.: Identification of 14–3–3-zeta as a protein kinase B/Akt substrate. J.Biol. Chem. 277: 21639–21642, 2002.

[43447] 16999.Staal, S. P.: Molecular cloning of the akt oncogene

and its human homologues AKT1 and AKT2: amplification of AKT1 in a primary human gastric adenocarcinoma. *Proc. Nat. Acad. Sci.* 84: 5034–5037, 1987.

- [43448] 17000. Staal, S. P.; Huebner, K.; Croce, C. M.; Parsa, N. Z.; Testa, J. R.: The AKT1 proto-oncogene maps to human chromosome 14, band q32. *Genomics* 2: 96–98, 1988.
- [43449] 17001. Vanhaesebroeck, B.; Alessi, D. R.: The PI3K–PDK1 connection: more than just a road to PKB. *Biochem. J.* 346: 561–576, 2000.
- [43450] 17002. Weng, L.-P.; Brown, J. L.; Eng, C.: PTEN induces apoptosis and cell cycle arrest through phosphoinositol-3-kinase/Akt-dependent and -independent pathways. *Hum. Molec. Genet.* 10: 237–242, 2001.
- [43451] 17003. Yang, J.; Cron, P.; Thompson, V.; Good, V. M.; Hess, D.; Hemmings, B. A.; Barford, D.: Molecular mechanism for the regulation of protein kinase B/Akt by hydrophobic motif phosphorylation. *Molec. Cell* 9: 1227–1240, 2002.
- [43452] 17004. Le Beau, M. M.; Rowley, J. D.; Sacchi, N.; Watson, D. K.; Papas, T. S.; Diaz, M. O.: *hu-ets-2* is translocated to chromosome 8 in t(8;21) in acute myelogenous leukemia. *Cancer Genet. Cytogenet.* 23: 269–274, 1986.
- [43453] 17005. Mavrothalassitis, G. J.; Watson, D. K.; Papas, T. S.:

Molecular and functional characterization of the promoter of ETS2, the human *c-ets-2* gene. *Proc. Nat. Acad. Sci.* 87: 1047–1051, 1990.

[43454] 17006. Sacchi, N.; Cheng, S. V.; Tanzi, R. E.; Gusella, J. F.; Drabkin, H. A.; Patterson, D.; Haines, J. H.; Papas, T. S.: The ETS genes on chromosome 21 are distal to the breakpoint of the acute myelogenous leukemia translocation (8;21). *Genomics* 3: 110–116, 1988.

[43455] 17007. Sumarsono, S. H.; Wilson, T. J.; Tymms, M. J.; Venter, D. J.; Corrick, C. M.; Kola, R.; Lahoud, M. H.; Papas, T. S.; Seth, A.; Kola, I.: Down's syndrome-like skeletal abnormalities in *Ets2* transgenic mice. *Nature* 379: 534–540, 1996.

[43456] 17008. Chu, E. H. Y.; Chang, C. C.; Sun, N. C.: Synteny of the human genes for gal-1-PT, ACP-1, MDH-1, and gal-plus-activator and assignment to chromosome 2. *Birth Defects Orig. Art. Ser.* XI(3): 103–106, 1975. Note: Alternate: *Cytogenet. Cell Genet.* 14: 273–276, 1975...

[43457] 17009. Rasmussen, S. K.; Lautier, C.; Hansen, L.; Echwald, S. M.; Hansen, T.; Ekstrom, C. T.; Urhammer, S. A.; Borch-Johnsen, K.; Grigorescu, F.; Smith, R. J.; Pedersen, O.: Studies of the variability of the genes encoding the insulin-like growth factor I receptor and its ligand in relation to

type 2 diabetes mellitus. J. Clin. Endocr. Metab.
85:1606–1610, 2000.

- [43458] 17010. Dissing, J.; Johnsen, A. H.: Human red cell acid phosphatase (ACP1): the primary structure of the two pairs of isozymes encoded by the ACP1*A and ACP1*C alleles. Biochim. Biophys. Acta 1121: 261–268, 1992.
- [43459] 17011. Dissing, J.; Johnsen, A. H.; Sensabaugh, G. F.: Human red cell acid phosphatase (ACP1): the amino acid sequence of the two isozymes Bf and Bs encoded by the ACP1*B allele. J. Biol. Chem. 266: 20619–20625, 1991.
- [43460] 17012. Emanuel, B. S.; Zackai, E. H.; Van Dyke, D. C.; Swallow, D. M.; Allen, F. H.; Mellman, W. J.: Deletion mapping: further evidence for the location of acid phosphatase (ACP-1) within 2p23. Am. J. Med. Genet. 4: 167–172, 1979.
- [43461] 17013. Ferguson-Smith, M. A.; Newman, B. F.; Ellis, P. M.; Thomson, D. M. G.; Riley, I. D.: Assignment by deletion of human red cell acid phosphatase gene locus to the short arm of chromosome 2. Nature 243: 271–273, 1973.
- [43462] 17014. Fisher, R. A.; Harris, H.: Studies on the separate isoenzymes of red cell acid phosphatase phenotypes A and B: chromatographic separation of the isoenzymes. Ann. Hum. Genet. 34: 431–438, 1971.

- [43463] 17015.Fuhrmann, W.; Lichte, K. H.: Human red cell acid phosphatase polymorphism: a study on gene frequency and forensic use of the system in cases of disputed paternity. *Humangenetik* 3: 121–126, 1966.
- [43464] 17016.Giblett, E. R.; Scott, N. M.: Red cell acid phosphatase: racial distribution and report of a new phenotype. *Am. J. Hum. Genet.* 17:425–432, 1965.
- [43465] 17017.Herbich, J.; Fisher, R. A.; Hopkinson, D. A.: Atypical segregation of human red cell acid phosphatase phenotypes: evidence for a rare 'silent' allele P(O). *Ann. Hum. Genet.* 34: 145–152, 1970.
- [43466] 17018.Herbich, J.; Meinhart, K.: The rare 'silent' allele P(O) or P(V)(P Vienna) of human red cell acid phosphatase, typed in a second family. *Humangenetik* 15:345–348, 1972.
- [43467] 17019.Hopkinson, D. A.; Spencer, N.; Harris, H.: Red cell acid phosphatase variants: a new human polymorphism. *Nature* 199: 969–971, 1963.
- [43468] 17020.Hulten, M.; Lindsten, J.; Pen-Ming, L. M.; Fraccaro, M.; Mannini, A.; Tiepolo, L.; Robson, E. B.; Heiken, A.; Tillinger, K. G.: Possible localization of the genes for the Kidd blood group on an autosome involved in a reciprocal translocation. *Nature* 211: 1067–1068, 1966.

- [43469] 17021.Junien, C.; Kaplan, J.-C.; Bernheim, A.; Berger, R.: Regional assignment of red cell acid phosphatase locus to band 2p25. *Hum.Genet.* 48: 17–21, 1979.
- [43470] 17022.Karp, G. W., Jr.; Sutton, H. E.: Some new phenotypes of human red cell acid phosphatase. *Am. J. Hum. Genet.* 19: 54–62, 1967.
- [43471] 17023.Lothe, R. A.; Gedde-Dahl, T.; Olaisen, B.; Bakker, E.; Pearson, P.: Very close linkage between D2S1 and ACP1 on chromosome 2p. *Ann.Hum. Genet.* 50: 361–367, 1986.
- [43472] 17024.Mace, M. A.; Cook, P. J. L.; Robson, E. B.: Linkage data on red cell acid phosphatase from family studies. *Ann. Hum. Genet.* 38:471–477, 1975.
- [43473] 17025.Mace, M. A.; Robson, E. B.: Linkage data on ACP-1 and MNSs. *Cytogenet.Cell Genet.* 13: 123–125, 1974.
- [43474] 17026.Magenis, R. E.; Koler, R. D.; Lovrien, E. W.; Bigley, R. H.; Duval, M. C.; Overton, K. M.: Gene dosage: evidence for assignment of erythrocyte acid phosphatase locus to chromosome 2. *Proc. Nat. Acad. Sci.* 72:4526–4530, 1975.
- [43475] 17027.Mayr, W. R.: No close linkage between MNSs and red cell acid phosphatase. *Hum. Hered.* 26: 1–3, 1976.
- [43476] 17028.Miller, S. A.; Nelson, M. S.; Dykes, D. D.; Polesky, H. F.: Comparison of acid phosphatase ACP1 variants by iso-

electric focusing and conventional electrophoresis: identification of three new alleles, ACP1*N, ACP1*P and ACP1*S. Hum. Hered. 37: 371–375, 1987.

- [43477] 17029. Mohrenweiser, H. W.; Novotny, J. E.: ACP-1-GUA-1: a low-activity variant of human erythrocyte acid phosphatase: association with increased glutathione reductase activity. Am. J. Hum. Genet. 34: 425–433, 1982.
- [43478] 17030. Nezbeda, P.: Occurrence of the ACP-1 null allele in Czechoslovakia. Hum. Genet. 46: 227–229, 1979.
- [43479] 17031. Palmarino, R.; Agostino, R.; Gloria, F.; Lucarelli, P.; Businco, L.; Antognoni, G.; Maggioni, G.; Workman, P. L.; Bottini, E.: Red cell acid phosphatase: another polymorphism correlated with malaria? Am. J. Phys. Anthropol. 43: 177–186, 1975.
- [43480] 17032. Povey, S.; Swallow, D. M.; Bobrow, M.; Craig, I.; Van Heyningen, V.: Probable assignment of the locus determining human red cell acid phosphatase ACP(1) to chromosome 2 using somatic cell hybrids. Ann. Hum. Genet. 38: 1–5, 1974.
- [43481] 17033. Renwick, J. H.: Assignment and map-positioning of human loci using chromosomal variation. Ann. Hum. Genet. 35: 79–97, 1971.
- [43482] 17034. Sensabaugh, G. F.; Golden, V. L.: Phenotype de-

pendence in the inhibition of red cell acid phosphatase (ACP) by folates. *Am. J. Hum. Genet.* 30: 553–560, 1978.

[43483] 17035. Swallow, D. M.; Povey, S.; Harris, H.: Activity of the 'red cell' acid phosphatase locus in other tissues. *Ann. Hum. Genet.* 37: 31–38, 1973.

[43484] 17036. Wu, R. M.; Cheng, C. W.; Chen, K. H.; Lu, S. L.; Shan, D. E.; Ho, Y. F.; Chern, H. D.: The COMT L allele modifies the association between MAOB polymorphism and PD in Taiwanese. *Neurology* 56: 375–382, 2001.

[43485] 17037. Levy, J. B.; Canoll, P. D.; Silvennoinen, O.; Barnea, G.; Morse, B.; Honegger, A. M.; Huang, J.-T.; Cannizzaro, L. A.; Park, S.-H.; Druck, T.; Huebner, K.; Sap, J.; Ehrlich, M.; Musacchio, J. M.; Schlessinger, J.: The cloning of a receptor-type protein tyrosine phosphatase expressed in the central nervous system. *J. Biol. Chem.* 268: 10573–10581, 1993.

[43486] 17038. Berube, D.; Simard, J.; Sandberg, M.; Grzeschik, K.-H.; Gagne, R.; Hansson, V.; Jahnsen, T.: Assignment of the gene encoding the catalytic subunit C(beta) of cAMP-dependent protein kinase to the p36 band on chromosome 1. (Abstract) *Cytogenet. Cell Genet.* 58: 1850 only, 1991.

[43487] 17039. Leonard, W. J.; Depper, J. M.; Kanehisa, M.; Kronke, M.; Pfeffer, N. J.; Svetlik, P. B.; Sullivan, M.; Greene, W. C.:

Structure of the human interleukin-2 receptor gene. *Science* 230: 633-639, 1985.

- [43488] 17040. Leonard, W. J.; Depper, J. M.; Robb, R. J.; Waldmann, T. A.; Greene, W. C.: Characterization of the human receptor for T-cell growth factor. *Proc. Nat. Acad. Sci.* 80: 6957-6961, 1983.
- [43489] 17041. Marx, J. L.: The interleukin-2 receptor gene is cloned. *Science* 226: 1064-1065, 1985.
- [43490] 17042. Nikaido, T.; Shimizu, A.; Ishida, N.; Sabe, H.; Teshigawara, K.; Maeda, M.; Uchiyama, T.; Yodoi, J.; Honjo, T.: Molecular cloning of a cDNA encoding human interleukin-2 receptor. *Nature* 311: 631-635, 1984.
- [43491] 17043. Robb, R. J.; Rusk, C. M.; Neeper, M. P.: Structure-function relationships for the interleukin 2 receptor: location of ligand and antibody binding sites on the Tac receptor chain by mutational analysis. *Proc. Nat. Acad. Sci.* 85: 5654-5658, 1988.
- [43492] 17044. Sharfe, N.; Dadi, H. K.; Shahr, M.; Roifman, C. M.: Human immunodeficiency disorder arising from mutation of the alpha chain of the interleukin-2 receptor. *Proc. Nat. Acad. Sci.* 94: 3168-3171, 1997.
- [43493] 17045. Shevach, E. M.: Certified professionals: CD4(+)CD25(+) suppressor T cells. *J. Exp. Med.* 193:

F41–F45, 2001.

- [43494] 17046. Tsudo, M.; Kozak, R. W.; Goldman, C. K.; Waldmann, T. A.: Demonstration of a non-Tac peptide that binds interleukin 2: a potential participant in a multichain interleukin 2 receptor complex. *Proc. Nat. Acad. Sci.* 83: 9694–9698, 1986.
- [43495] 17047. Urdal, D. L.; March, C. J.; Gillis, S.; Larsen, A.; Dower, S.K.: Purification and chemical characterization of the receptor for interleukin 2 from activated human T lymphocytes and from a human T-cell lymphoma cell line. *Proc. Nat. Acad. Sci.* 81: 6481–6485, 1984.
- [43496] 17048. Celestin, J.; Rotschke, O.; Falk, K.; Ramesh, N.; Jabara, H.; Strominger, J.; Geha, R. S.: IL-3 induces B7.2 (CD86) expression and costimulatory activity in human eosinophils. *J. Immun.* 167: 6097–6104, 2001.
- [43497] 17049. Mohrenweiser, H. W.; Neel, J. V.: Frequency of thermostability variants: estimation of total 'rare' variant frequency in human populations. *Proc. Nat. Acad. Sci.* 78: 5729–5733, 1981.
- [43498] 17050. Gervais, F. G.; Singaraja, R.; Xanthoudakis, S.; Gutekunst, C.-A.; Leavitt, B. R.; Metzler, M.; Hackam, A. S.; Tam, J.; Vaillancourt, J. P.; Houtzager, V.; Rasper, D. M.; Roy, S.; Hayden, M. R.; Nicholson, D. W.: Recruitment and

activation of caspase-8 by the huntingtin-inter-acting protein Hip-1 and a novel partner Hippi. *Nature Cell Biol.* 4: 95-105, 2002.

- [43499] 17051. Abbott, A. M.; Bueno, R.; Pedrini, M. T.; Murray, J. M.; Smith, R. J.: Insulin-like growth factor I receptor gene structure. *J. Biol. Chem.* 267: 10759-10763, 1992.
- [43500] 17052. All-Ericsson, C.; Girnita, L.; Seregard, S.; Bartolazzi, A.; Jager, M. J.; Larsson, O.: Insulin-like growth factor-1 receptor in uveal melanoma: a predictor for metastatic disease and a potential therapeutic target. *Invest. Ophthalmol. Vis. Sci.* 43: 1-8, 2002.
- [43501] 17053. Cooke, D. W.; Bankert, L. A.; Roberts, C. T., Jr.; LeRoith, D.; Casella, S. J.: Analysis of the human type I insulin-like growth factor receptor promoter region. *Biochem. Biophys. Res. Commun.* 177: 1113-1120, 1991.
- [43502] 17054. Dey, B. R.; Furlanetto, R. W.; Nissley, S. P.: Cloning of human p55-gamma, a regulatory subunit of phosphatidylinositol 3-kinase, by a yeast two-hybrid library screen with the insulin-like growth factor-I receptor. *Gene* 209: 175-183, 1998.
- [43503] 17055. Fernandez, A. M.; Kim, J. K.; Yakar, S.; Dupont, J.; Hernandez-Sanchez, C.; Castle, A. L.; Filmore, J.; Shulman, G. I.; LeRoith, D.: Functional inactivation of the IGF-I and

insulin receptors in skeletal muscle causes type 2 diabetes. Genes Dev. 15: 1926–1934, 2001.

- [43504] 17056. Flier, J. S.; Usher, P.; Moses, A. C.: Monoclonal antibody to the type I insulin-like growth factor (IGF-I) receptor blocks IGF-I receptor-mediated DNA synthesis: clarification of the mitogenic mechanisms of IGF-I and insulin in human skin fibroblasts. Proc. Nat. Acad. Sci. 83: 664–668, 1986.
- [43505] 17057. Francke, U.; Yang-Feng, T. L.; Brissenden, J. E.; Ullrich, A.: Chromosomal mapping of genes involved in growth control. Cold Spring Harbor Symp. Quant. Biol. 51: 855–866, 1986.
- [43506] 17058. Grant, E. S.; Ross, M. B.; Ballard, S.; Naylor, A.; Habib, F. K.: The insulin-like growth factor type I receptor stimulates growth and suppresses apoptosis in prostatic stromal cells. J. Clin. Endocr. Metab. 83: 3252–3257, 1998.
- [43507] 17059. Herskowitz, I.: Functional inactivation of genes by dominant negative mutations. Nature 329: 219–222, 1987.
- [43508] 17060. Howard, T. K.; Algar, E. M.; Glatz, J. A.; Reeve, A. E.; Smith, P. J.: The insulin-like growth factor 1 receptor gene is normally biallelically expressed in human juvenile tissue and tumours. Hum. Molec. Genet. 2: 2089–2092, 1993.

- [43509] 17061.Kulkarni, R. N.; Holzenberger, M.; Shih, D. Q.; Ozcan, U.; Stoffel,M.; Magnuson, M. A.; Kahn, C. R.: Beta-cell-specific deletion of the Igf1 receptor leads to hyperinsulinemia and glucose intolerance but does not alter beta-cell mass. *Nature Genet.* 31: 111–115, 2002.
- [43510] 17062.Maor, S. B.; Abramovitch, S.; Erdos, M. R.; Brody, L. C.; Werner,H.: BRCA1 suppresses insulin-like growth factor-I receptor promoter activity: potential interaction between BRCA1 and Sp1. *Molec. Genet.Metab.* 69: 130–136, 2000.
- [43511] 17063.Poduslo, S. E.; Dean, M.; Kolch, U.; O'Brien, S. J.: Detecting high-resolution polymorphisms in human coding loci by combining PCR and single-strand conformation polymorphism (SSCP) analysis. *Am.J. Hum. Genet.* 49: 106–111, 1991.
- [43512] 17064.Prager, D.; Yamasaki, H.; Weber, M. M.; Gebremedhin, S.; Melmed,S.: Human insulin-like growth factor I receptor function in pituitary cells is suppressed by a dominant negative mutant. *J. Clin. Invest.* 90:2117–2122, 1992.
- [43513] 17065.Roback, E. W.; Barakat, A. J.; Dev, V. G.; Mbikay, M.; Chretien,M.; Butler, M. G.: An infant with deletion of the distal long arm of chromosome 15 (q26.1–qter) and loss of

insulin-like growth factor1 receptor gene. Am. J. Med. Genet. 38: 74–79, 1991.

- [43514] 17066.Okumura, N.; Terasawa, F.; Ueno, I.; Oki, K.; Yamauchi, K.; Hidaka,H.; Tozuka, M.; Okura, M.; Katsuyama, T.: Genetic analyses in homozygous and heterozygous variants of lactate dehydrogenase-B (H) subunit:LD-B Matsumoto I and II (LD-B W323R). Clin. Chim. Acta 287: 163–171,1999.
- [43515] 17067.Rethore, M.-O.; Junien, C.; Malpuech, G.; Baccichetti, C.; Tenconi,R.; Kaplan, J. C.; de Romeuf, J.; Lejeune, J.: Localisation du gene de la glyceraldehyde 3-phosphate dehydrogenase (G3PD) sur le segment distal du bras court du chromosome 12. Ann. Genet. 19: 140–142,1976.
- [43516] 17068.Rethore, M.-O.; Kaplan, J.-C.; Junien, C.; Cruveiller, J.; Dutrillaux,B.; Aurias, A.; Carpentier, S.; Lafourcade, J.; Lejeune, J.: Augmentation de l'activite de la LDH-B chez un garcon trisomique 12p par malsegregation d'une translocation maternelle t(12;14)(q12;p11). Ann. Genet. 18:81–87, 1975.
- [43517] 17069.Sakai, I.; Sharief, F. S.; Pan, Y.-C. E.; Li, S. S.-L.: The cDNA and protein sequences of human lactate dehydrogenase B. Biochem.J. 248: 933–936, 1987.
- [43518] 17070.Santachiara, A. S.; Nabholz, M.; Miggiano, V.; Dar-

lington, A.J.; Bodmer, W. F.: Linkage between human lactate dehydrogenase Band peptidase B genes. *Nature* 227: 248–251, 1970.

[43519] 17071.Steinbach, P.; Rehder, H.: Tetrasomy for the short arm of chromosome12 with accessory isochromosome (+i[12p]) and a marked LDH–B gened dosage effect. *Clin. Genet.* 32: 1–4, 1987.

[43520] 17072.Sudo, K.: Personal Communication. Komae City, Japan 7/12/1993.

[43521] 17073.Sudo, K.; Maekawa, M.; Ikawa, S.; Machida, K.; Kitamura, M.; Li, S. S.–L.: A missense mutation found in human lactate dehydrogenase–B(H) variant gene. *Biochem. Biophys. Res. Commun.* 168: 672–676, 1990.

[43522] 17074.Sudo, K.; Maekawa, M.; Luedemann, M. M.; Deaven, L. L.; Li, S.S.–L.: Human lactate dehydrogenase–B processed pseudogene: nucleotide sequence analysis and assignment to the X–chromosome. *Biochem. Biophys. Res. Commun.* 171: 67–74, 1990.

[43523] 17075.Sudo, K.; Maekawa, M.; Tomonaga, A.; Tsukada, T.; Nakayama, T.; Kitamura, M.; Li, S. S.–L.; Kanno, T.; Toriumi, J.: Molecular characterization of genetic mutations in human lactate dehydrogenase (LDH)B (H) variant. *Hum. Genet.* 89: 158–162, 1992.

- [43524] 17076. Van Someren, H.; Meera Khan, P.; Westerveld, A.; Bootsma, D.: Human genetics--two new linkage groups carrying different loci for LDH and glutamic-pyruvic transaminase found. *Nature* 240: 221-222, 1972.
- [43525] 17077. Weiss, L.; Mayeda, K.; Lindahl, R.; Dully, M.: Localization of human LDH-B gene of the short arm of chromosome 12. (Abstract) *Am. J. Hum. Genet.* 25: 85A only, 1973.
- [43526] 17078. Davies, H.; Bignell, G. R.; Cox, C.; Stephens, P.; Edkins, S.; Clegg, S.; Teague, J.; Woffendin, H.; Garnett, M. J.; Bottomley, W.; Davis, N.; Dicks, E.; and 40 others: Mutations of the BRAF gene in human cancer. *Nature* 417: 949-954, 2002.
- [43527] 17079. Eychene, A.; Barnier, J. V.; Apiou, F.; Dutrillaux, B.; Calothy, G.: Chromosomal assignment of two human B-raf(Rmil) proto-oncogene loci: B-raf-1 encoding the p94(Braf/Rmil) and B-raf-2, a processed pseudogene. *Oncogene* 7: 1657-1660, 1992.
- [43528] 17080. Rajagopalan, H.; Bardelli, A.; Lengauer, C.; Kinzler, K. W.; Vogelstein, B.; Velculescu, V. E.: RAF/RAS oncogenes and mismatch-repair status. (Letter) *Nature* 418: 934 only, 2002.
- [43529] 17081. Sithanandam, G.; Druck, T.; Cannizzaro, L. A.;

Leuzzi, G.; Huebner, K.; Rapp, U. R.: B-raf and a B-raf pseudogene are located on 7q in man. *Oncogene* 7: 795–799, 1992.

[43530] 17082. Wojnowski, L.; Zimmer, A. M.; Beck, T. W.; Hahn, H.; Bernal, R.; Rapp, U. R.; Zimmer, A.: Endothelial apoptosis in Braf-deficient mice. *Nature Genet.* 16: 293–297, 1997.

[43531] 17083. Zhong, S.; Delva, L.; Rachez, C.; Cenciarelli, C.; Gandini, D.; Zhang, H.; Kalantry, S.; Freedman, L. P.; Pandolfi, P. P.: A RA-dependent, tumour-growth suppressive transcription complex is the target of the PML-RAR-alpha and T18 oncoproteins. *Nature Genet.* 23: 287–295, 1999.

[43532] 17084. Fukuyama, R.; Ichijoh, Y.; Minoshima, S.; Kitamura, N.; Shimizu, N.: Assignment of hepatocyte growth factor (HGF) gene to chromosome 7q21.1. (Abstract) *Cytogenet. Cell Genet.* 58: 1921 only, 1991.

[43533] 17085. Fukuyama, R.; Ichijoh, Y.; Minoshima, S.; Kitamura, N.; Shimizu, N.: Regional localization of the hepatocyte growth factor (HGF) gene to human chromosome 7 band q21.1. *Genomics* 11: 410–415, 1991.

[43534] 17086. Gherardi, E.; Stoker, M.: Hepatocytes and scatter factor. *Nature* 346:228 only, 1990.

[43535] 17087. Gohda, E.; Tsubouchi, H.; Nakayama, H.; Hirono, S.;

Sakiyama, O.;Takahashi, K.; Miyazaki, H.; Hashimoto, S.; Daikuhara, Y.: Purificationand partial characterization of hepatocyte growth factor from plasmaof a patient with fulminant hepatic failure. J. Clin. Invest. 81:414–419, 1988.

- [43536] 17088.Kilby, M. D.; Afford, S.; Li, X. F.; Strain, A. J.; Ahmed, A.;Whittle, M. J.: Localisation of hepatocyte growth factor and itsreceptor (c-met) protein and mRNA in human term placenta. GrowthFactors 13: 133–139, 1996.
- [43537] 17089.Lai, L.; Goldschneider, I.: Cutting edge: identification of ahybrid cytokine consisting of IL-7 and the beta-chain of the hepatocytegrowth factor/scatter factor. J. Immun. 167: 3550–3554, 2001.
- [43538] 17090.Maina, F.; Casagrande, F.; Audero, E.; Simeone, A.; Comoglio, P.M.; Klein, R.; Ponzetto, C.: Uncoupling of Grb2 from the Met receptorin vivo reveals complex roles in muscle development. Cell 87: 531–542,1996.
- [43539] 17091.Miyazawa, K.; Tsubouchi, H.; Naka, D.; Takahashi, K.; Okigaki,M.; Arakaki, N.; Nakayama, H.; Hirono, S.; Sakiyama, O.; Takahashi,K.; Gohda, E.; Daikuhara, Y.; Kitamura, N.: Molecular cloning andsequence analysis of cDNA for human hepatocyte growth factor. Biochem.Biophys. Res. Commun. 163: 967–973, 1989.

- [43540] 17092.Nakamura, T.; Nishizawa, T.; Hagiya, M.; Seki, T.; Shimonishi,M.; Sugimura, A.; Tashiro, K.; Shimizu, S.: Molecular cloning and expression of human hepatocyte growth factor. *Nature* 342: 440–443,1989.
- [43541] 17093.Noonan, F. P.; Recio, J. A.; Takayama, H.; Duray, P.; Anver, M.R.; Rush, W. L.; De Fabo, E. C.; Merlino, G.: Neonatal sunburn and melanoma in mice: severe sunburn in newborn, but not adult, mice is linked with melanoma in later life. *Nature* 413: 271–272, 2001.
- [43542] 17094.Powell, E. M.; Mars, W. M.; Levitt, P.: Hepatocyte growth factor/scatterfactor is a motogen for interneurons migrating from the ventral to dorsal telencephalon. *Neuron* 30: 79–89, 2001.
- [43543] 17095.Rubin, J. S.; Chan, A. M.–L.; Bottaro, D. P.; Burgess, W. H.; Taylor, W. G.; Cech, A. C.; Hirschfield, D. W.; Wong, J.; Miki, T.; Finch, P. W.; Aaronson, S. A.: A broad–spectrum human lung fibroblast–derived mitogen is a variant of hepatocyte growth factor. *Proc. Nat. Acad.Sci.* 88: 415–419, 1991.
- [43544] 17096.Saccone, S.; Narsimhan, R. P.; Gaudino, G.; Dalpra, L.; Comoglio, P. M.; Della Valle, G.: Regional mapping of the human hepatocyte growth factor (HGF)–scatter factor gene to chromosome 7q21.1. *Genomics* 13:912–914,

1992.

- [43545] 17097. Schmidt, C.; Bladt, F.; Goedecke, S.; Brinkmann, V.; Zschiesche, W.; Sharpe, M.; Gherardi, E.; Birchmeier, C.: Scatter factor/hepatocyte growth factor is essential for liver development. *Nature* 373: 699–702, 1995.
- [43546] 17098. Uehara, Y.; Minowa, O.; Mori, C.; Shiota, K.; Kuno, J.; Noda, T.; Kitamura, N.: Placental defect and embryonic lethality in mice lacking hepatocyte growth factor/scatter factor. *Nature* 373: 702–705, 1995.
- [43547] 17099. Weidner, K. M.; Arakaki, N.; Hartmann, G.; Vandekerckhove, J.; Weingart, S.; Rieder, H.; Fonatsch, C.; Tsubouchi, H.; Hishida, T.; Daikuhara, Y.; Birchmeier, W.: Evidence for the identity of human scatter factor and human hepatocyte growth factor. *Proc. Nat. Acad. Sci.* 88: 7001–7005, 1991.
- [43548] 17100. Zarnegar, R.; Petersen, B.; DeFrances, M. C.; Michalopoulos, G.: Localization of hepatocyte growth factor (HGF) gene on human chromosome 7. *Genomics* 12: 147–150, 1992.
- [43549] 17101. Calandro, L. M.; Baer, D. M.; Sensibaugh, G. F.: Characterization of a recombinant that locates the hereditary hemochromatosis gene telomeric to HLA-F. *Hum. Genet.* 96: 339–342, 1995.

- [43550] 17102.Geraghty, D. E.; Wei, X. H.; Orr, H. T.; Koller, B. H.: Humanleukocyte antigen F (HLA-F): an expressed HLA gene composed of a class I coding sequence linked to a novel transcribed repetitive element. *J.Exp. Med.* 171: 1-18, 1990.
- [43551] 17103.Shiina, T.; Tamiya, G.; Oka, A.; Takishima, N.; Yamagata, T.; Kikkawa, E.; Iwata, K.; Tomizawa, M.; Okuaki, N.; Kuwano, Y.; Watanabe, K.; Fukuzumi, Y.; and 11 others: Molecular dynamics of MHC genesis unraveled by sequence analysis of the 1,796,938-bp HLA class I region. *Proc.Nat. Acad. Sci.* 96: 13282-13287, 1999.
- [43552] 17104.Gasparini, P.; Borgato, L.; Piperno, A.; Girelli, D.; Olivieri, O.; Gottardi, E.; Roetto, A.; Dianzani, I.; Fargion, S.; Schinaia, G.; Cappellini, M. D.; Gandini, G.; Pignatti, P. F.; Fiorelli, G.; De Sandre, G.; Camaschella, C.: Linkage analysis of 6p21 polymorphic markers and the hereditary hemochromatosis: localization of the gene centromeric to HLA-F. *Hum. Molec. Genet.* 2: 571-576, 1993.
- [43553] 17105.Engel, W.; Klemme, B.; Ebrecht, A.: Serological evidence for H-Y antigen in XO-female mice. *Hum. Genet.* 57: 68-70, 1981.
- [43554] 17106.Lau, Y.-F.: Personal Communication. San Francisco, Calif. 5/4/1987.

- [43555] 17107.Lau, Y.-F.; Chan, K.; Kan, Y. W.; Goldberg, E.: Isolation of amale-specific and conserved gene using an anti-H-Y antibody. (Abstract) Am.J. Hum. Genet. 39: A142 only, 1986.
- [43556] 17108.Lau, Y.-F.; Chan, K.; Kan, Y. W.; Goldberg, E.: Structure and expression of a gene isolated with an anti-H-Y antibody. (Abstract) Clin.Res. 35: 647A only, 1987.
- [43557] 17109.Muller, U.: Identification and function of serologically detectable H-Y antigen. Hum. Genet. 61: 91-94, 1982.
- [43558] 17110.Shapiro, M.; Erickson, R. P.: Evidence that the serological determinant of H-Y antigen is carbohydrate. Nature 290: 503-505, 1981.
- [43559] 17111.Wiberg, U.; Mayerova, A.; Muller, U.; Fredga, K.; Wolf, U.: X-linked genes of the H-Y antigen system in the wood lemming (*Myopus schisticolor*). Hum.Genet. 60: 163-166, 1982.
- [43560] 17112.Wolf, U.: Zum Mechanismus der Gonadendifferenzierung. Bull. Schweiz.Akad. Med. Wiss. 34: 357-368, 1978.
- [43561] 17113.van den Boogaard, M.-J. H.; Dorland, M.; Beemer, F. A.; Ploos van Amstel, H. K.: MSX1 mutation is associated with orofacial clefting and tooth agenesis in humans.

(Letter) Nature Genet. 24: 342–343,2000. Note: Erratum:
Nature Genet. 25: 125 only, 2000.

- [43562] 17114.Del Campo, M.; Jones, M. C.; Veraksa, A. N.; Curry, C. J.; Jones,K. L.; Mascarello, J. T.; Ali-Kahn-Catts, Z.; Drumheller, T.; McGinnis,W.: Monodactylous limbs and abnormal genitalia are associated withhemizyosity for the human 2q31 region that includes the HOXD cluster. Am.J. Hum. Genet. 65: 104–110, 1999.
- [43563] 17115.Zakany, J.; Kmita, M.; Alarcon, P.; de la Pompa, J.–L.; Duboule,D.: Localized and transient transcription of Hox genes suggests a link between patterning and the segmentation clock. Cell 106: 207–217,2001.
- [43564] 17116.Perez, O. D.; Kinoshita, S.; Hitoshi, Y.; Payan, D. G.; Kitamura,T.; Nolan, G. P.; Lorens, J. B.: Activation of the PKB/AKT pathwayby ICAM–2. Immunity 16: 51–65, 2002.
- [43565] 17117.Sansom, D.; Borrow, J.; Solomon, E.; Trowsdale, J.: The humanICAM2 gene maps to 17q23–25. Genomics 11: 462–464, 1991.
- [43566] 17118.Staunton, D. E.; Dustin, M. L.; Springer, T. A.: Functional cloningof ICAM–2, a cell adhesion ligand for LFA–1 homologous to ICAM–1. Nature 339:61–64, 1989.
- [43567] 17119.Rotem–Yehudar, R.; Galperin, E.; Horowitz, M.: Association ofinsulin–like growth factor 1 receptor with

EHD1 and SNAP29. *J. Biol.Chem.* 276: 33054–33060, 2001.

[43568] 17120.Scott, B. A.; Avidan, M. S.; Crowder, C. M.: Regulation of hypoxicdeath in *C. elegans* by the insulin/IGF receptor homolog DAF-2. *Science* 296:2388–2391, 2002.

[43569] 17121.Tatar, M.; Kopelman, A.; Epstein, D.; Tu, M.–P.; Yin, C.–M.; Garofalo,R. S.: A mutant *Drosophila* insulin receptor homolog that extendslife–span and impairs neuroendocrine function. *Science* 292: 107–110,2001.

[43570] 17122.Ullrich, A.; Gray, A.; Tam, A. W.; Yang–Feng, T.; Tsubokawa, M.;Collins, C.; Henzel, W.; Le Bon, T.; Kathuria, S.; Chen, E.; Jacobs,S.; Francke, U.; Ramachandran, J.; Fujita–Yamaguchi, Y.: Insulin–likegrowth factor I receptor primary structure: comparison with insulinreceptor suggests structural determinants that define functional specificity. *EMBOJ.* 5: 2503–2512, 1986.

[43571] 17123.Werner, H.; Karnieli, E.; Rauscher, F. J., III; LeRoth, D.: Wild–typeand mutant p53 differentially regulate transcription of the insulin–likegrowth factor I receptor gene. *Proc. Nat. Acad. Sci.* 93: 8318–8323,1996.

[43572] 17124.Barton, D. E.; Yang–Feng, T. L.; Mason, A. J.; Seeburg, P. H.;Francke, U.: INHA, INHBA and INHBB, the loci for the three subunitsof inhibin, mapped in mouse and

man. (Abstract) Cytogenet. Cell Genet. 46:578 only, 1987.

- [43573] 17125. Barton, D. E.; Yang-Feng, T. L.; Mason, A. J.; Seeburg, P. H.; Francke, U.: Mapping of genes for inhibin subunits alpha, beta(A) and beta(B) on human and mouse chromosomes and studies of jsd mice. Genomics 5:91-99, 1989.
- [43574] 17126. Bremner, W. J.: Inhibin: from hypothesis to clinical application. (Editorial) New Eng. J. Med. 321: 826-827, 1989.
- [43575] 17127. Lappohn, R. E.; Burger, H. G.; Bouma, J.; Bangah, M.; Krans, M.; de Bruijn, H. W. A.: Inhibin as a marker for granulosa-cell tumors. New Eng. J. Med. 321: 790-793, 1989.
- [43576] 17128. Mason, A. J.; Hayflick, J. S.; Ling, N.; Esch, F.; Ueno, N.; Ying, S.-Y.; Guillemin, R.; Niall, H.; Seeburg, P. H.: Complementary DNA sequences of ovarian follicular fluid inhibin show precursor structure and homology with transforming growth factor-beta. Nature 318: 659-663, 1985.
- [43577] 17129. Mason, A. J.; Niall, H. D.; Seeburg, P. H.: Structure of two human ovarian inhibins. Biochem. Biophys. Res. Commun. 135: 957-964, 1986.
- [43578] 17130. Matzuk, M. M.; Finegold, M. J.; Mather, J. P.; Krum-

men, L.; Lu, H.; Bradley, A.: Development of cancer cachexia-like syndrome and adrenal tumors in inhibin-deficient mice. *Proc. Nat. Acad. Sci.* 91:8817–8821, 1994.

[43579] 17131. Matzuk, M. M.; Finegold, M. J.; Su, J.-G. J.; Hsueh, A. J. W.; Bradley, A.: Alpha-inhibin is a tumour-suppressor gene with gonadal specificity in mice. *Nature* 360: 313–319, 1992.

[43580] 17132. Mellor, S. L.; Richards, M. G.; Pedersen, J. S.; Robertson, D. M.; Risbridger, G. P.: Loss of the expression and localization of inhibin alpha-subunit in high grade prostate cancer. *J. Clin. Endocr. Metab.* 83: 969–975, 1998.

[43581] 17133. Meunier, H.; Rivier, C.; Evans, R. M.; Vale, W.: Gonadal and extragonadal expression of inhibin alpha, beta-A and beta-B subunits in various tissues predicts diverse functions. *Proc. Nat. Acad. Sci.* 85:247–251, 1988.

[43582] 17134. Ramasharma, K.; Li, C. H.: Characteristics of binding of human seminal alpha-inhibin-92 to human pituitary membranes. *Proc. Nat. Acad. Sci.* 84: 3595–3598, 1987.

[43583] 17135. Schmitt, J. F.; Millar, D. S.; Pedersen, J. S.; Clark, S. L.; Venter, D. J.; Frydenberg, M.; Molloy, P. L.; Risbridger, G. P.: Hypermethylation of the inhibin alpha-subunit gene in prostate carcinoma. *Molec. Endocr.* 16: 213–220, 2002.

- [43584] 17136.Thirunavukarasu, P.; Stephenson, T.; Forray, J.; Stanton, P. G.;Groome, N.; Wallace, E.; Robertson, D. M.: Changes in molecular weightforms of inhibin A and pro-alpha-C in maternal serum during humanpregnancy. J. Clin. Endocr. Metab. 86: 5794–5804, 2001.
- [43585] 17137.Lafuse, W. P.; Zwillig, B. S.: Localization of the inhibin beta-Bgene on mouse chromosome 1. Mammalian Genome 4: 399–400, 1993.
- [43586] 17138.Salmenkivi, K.; Arola, J.; Voutilainen, R.; Ilvesmaki, V.; Haglund,C.; Kahri, A. I.; Heikkila, P.; Liu, J.: Inhibin/activin beta-B-subunitexpression in pheochromocytomas favors benign diagnosis. J. Clin.Endocr. Metab. 86: 2231–2235, 2001.
- [43587] 17139.Schrewe, H.; Gendron-Maguire, M.; Harbison, M. L.; Gridley, T.: Mice homozygous for a null mutation of activin beta B are viableand fertile. Mech. Dev. 47: 43–51, 1994.
- [43588] 17140.Nicoll, J. A. R.; Mrak, R. E.; Graham, D. I.; Stewart, J.; Wilcock,G.; MacGowan, S.; Esiri, M. M.; Murray, L. S.; Dewar, D.; Love, S.;Moss, T.; Griffin, W. S. T.: Association of interleukin-1 gene polymorphismswith Alzheimer's disease. Ann. Neurol. 47: 365–368, 2000.
- [43589] 17141.Rogers, J.: An IL-1-alpha susceptibility polymor-

phism in Alzheimer's disease: new fuel for the inflammation hypothesis. (Editorial) *Neurology* 55:464–465, 2000.

- [43590] 17142. Sabatino, M.; Boyce, B.; Aufdemorte, T.; Bonewald, L.; Mundy, G. R.: Infusions of recombinant human interleukins 1 alpha and 1 beta cause hypercalcemia in normal mice. *Proc. Nat. Acad. Sci.* 85:5235–5239, 1988.
- [43591] 17143. Silver, A. R. J.; Masson, W. K.; George, A. M.; Adam, J.; Cox, R.: The IL-1 alpha and beta genes are closely linked (less than 70kb) on mouse chromosome 2. *Somat. Cell Molec. Genet.* 16: 549–556, 1990.
- [43592] 17144. Dickensheets, H. L.; Venkataraman, C.; Schindler, U.; Donnelly, R. P.: Interferons inhibit activation of STAT6 by interleukin 4 in human monocytes by inducing SOCS-1 gene expression. *Proc. Nat. Acad. Sci.* 96: 10800–10805, 1999.
- [43593] 17145. Kotanides, H.; Reich, N. C.: Interleukin-4-induced STAT6 recognizes and activates a target site in the promoter of the interleukin-4 receptor gene. *J. Biol. Chem.* 271: 25555–25561, 1996.
- [43594] 17146. Takahashi, M.; Yoshida, M. C.; Satoh, H.; Hilgers, J.; Yaoita, Y.; Honjo, T.: Chromosomal mapping of the mouse IL-4 and human IL-5 genes. *Genomics* 4: 47–52, 1989.
- [43595] 17147. Caggana, M.; Walker, K.; Reilly, A. A.; Conroy, J. M.;

Duva, S.;Walsh, A. C.: Population-based studies reveal differences in the allelic frequencies of two functionally significant human interleukin-4 receptor polymorphisms in several ethnic groups. *Genet. Med.* 1:267-271, 1999.

[43596] 17148. Deichmann, K. A.; Heinzmann, A.; Forster, J.; Dischinger, S.; Mehl, C.; Brueggenolte, E.; Hildebrandt, F.; Moseler, M.; Kuehr, J.: Linkage and allelic association of atopy and markers flanking the IL4-receptor gene. *Clin. Exp. Allergy* 28: 151-155, 1998.

[43597] 17149. MacLeod, J. N.; Lee, A. K.; Liebhaber, S. A.; Cooke, N. E.: Developmental control and alternative splicing of the placentally expressed transcripts from the human growth hormone gene cluster. *J. Biol. Chem.* 267:14219-14226, 1992.

[43598] 17150. Wurzel, J. M.; Parks, J. S.; Herd, J. E.; Nielsen, P. V.: A gene deletion is responsible for absence of human chorionic somatomammotropin. *DNA* 1:251-257, 1982.

[43599] 17151. Gallin, J. I.: Personal Communication. Bethesda, Md. 10/12/1990.

[43600] 17152. McCombs, J. L.; Teng, C. T.; Pentecost, B. T.; Magnuson, V. L.; Moore, C. M.; McGill, J. R.: Chromosomal localization of human lactotransferringene (LTF) by in situ hybridization. *Cytogenet. Cell Genet.* 47:16-17, 1988.

- [43601] 17153.Moriuchi, M.; Moriuchi, H.: A milk protein lactoferrin enhances human T cell leukemia virus type I and suppresses HIV-1 infection. *J.Immun.* 166: 4231-4236, 2001.
- [43602] 17154.Naylor, S. L.; Marshall, A.; Solomon, A.; McGill, J. R.; McCombs, J.; Magnuson, V. L.; Moore, C. M.; Lalley, P. A.; Pentecost, B. T.; Teng, C.: Lactoferrin maps to human chromosome 3(q21-q23) and mouse chromosome 9. (Abstract) *Cytogenet. Cell Genet.* 46: 669 only, 1987.
- [43603] 17155.Powell, M. J.; Ogden, J. E.: Nucleotide sequence of human lactoferrin cDNA. *Nucleic Acids Res.* 18: 4013 only, 1990.
- [43604] 17156.Qiu, J.; Hendrixson, D. R.; Baker, E. N.; Murphy, T. F.; St. Geme, J. W., III; Plaut, A. G.: Human milk lactoferrin inactivates two putative colonization factors expressed by *Haemophilus influenzae*. *Proc.Nat. Acad. Sci.* 95: 12641-12646, 1998.
- [43605] 17157.Rado, T. A.; Wei, X.; Benz, E. J., Jr.: Isolation of lactoferrin cDNA from a human myeloid library and expression of mRNA during normal and leukemic myelopoiesis. *Blood* 70: 989-993, 1987.
- [43606] 17158.Singh, P. K.; Parsek, M. R.; Greenberg, E. P.; Welsh, M. J.: A component of innate immunity prevents bacterial biofilm development. *Nature* 417:552-555, 2002.

- [43607] 17159.Teng, C. T.; Pentecost, B. T.; Marshall, A.; Solomon, A.; Bowman,B. H.; Lalley, P. A.; Naylor, S. L.: Assignment of the lactotransferringene to human chromosome 3 and to mouse chromosome 9. *Somat. CellMolec. Genet.* 13: 689–693, 1987.
- [43608] 17160.Yang, F.; Lum, J.; Baldwin, W. D.; Brune, J. L.; van Bragt, P.;Bowman, B. H.: Genetic analysis of human iron binding glycoproteins.(Abstract) *Am. J. Hum. Genet.* 35: 184A only, 1983.
- [43609] 17161.Tanaka, A. R.; Ikeda, Y.; Abe–Dohmae, S.; Arakawa, R.; Sadanami,K.; Kidera, A.; Nakagawa, S.; Nagase, T.; Aoki, R.; Kioka, N.; Amachi,T.; Yokoyama, S.; Ueda, K.: Human ABCA1 contains a large amino terminalextracellular domain homologous to an epitope of Sjogren's syndrome. *Biochem.Biophys. Res. Commun.* 283: 1019–1025, 2001.
- [43610] 17162.Xu, W.; Gelber, S.; Orr–Urtreger, A.; Armstrong, D.; Lewis, R.A.; Ou, C.–N.; Patrick, J.; Role, L.; De Biasi, M.; Beaudet, A. L.: Megacystis, mydriasis, and ion channel defect in mice lacking thealpha–3 neuronal nicotinic acetylcholine receptor. *Proc. Nat. Acad.Sci.* 96: 5746–5751, 1999.
- [43611] 17163.Gallardo, M. E.; Lopez–Rios, J.; Fernaud–Espinosa,

I.; Granadino,B.; Sanz, R.; Ramos, C.; Ayuso, C.; Seller, M. J.; Brunner, H. G.;Bovolenta, P.; Rodriguez de Cordoba, S.: Genomic cloning and characterization of the human homeobox gene SIX6 reveals a cluster of SIX genes in chromosome 14 and associates SIX6 hemizyosity with bilateral anophthalmia and pituitary anomalies. Genomics 61: 82–91, 1999.

[43612] 17164.Sun, X.–H.: Constitutive expression of the Id1 gene impairs mouse B cell development. Cell 79: 893–900, 1994.

[43613] 17165.Nowell, P. C.; Hungerford, D. A.: Chromosome studies on normal and leukemic human leukocytes. J. Nat. Cancer Inst. 25: 85–109, 1960.

[43614] 17166.Dowbenko, D. J.; Diep, A.; Taylor, B. A.; Lysis, A. J.; Lasky, L. A.: Characterization of the murine homing receptor gene reveals correspondence between protein domains and coding exons. Genomics 9:270–277, 1991.

[43615] 17167.Couturier, J.; Sastre–Garau, X.; Schneider–Mau–noury, S.; Labib, A.; Orth, G.: Integration of papillomavirus DNA near myc genes in genital carcinomas and its consequences for proto–oncogene expression. J. Virol. 65: 4534–4538, 1991.

[43616] 17168.Habraken, Y.; Sung, P.; Prakash, S.; Prakash, L.:

Transcriptionfactor TFIIH and DNA endonuclease Rad2 constitute yeast nucleotideexcision repair factor 3: implications for nucleotide excision repairand Cockayne syndrome. Proc. Nat. Acad. Sci. 93: 10718–10722, 1996.

[43617] 17169.Abruzzo, L. V.; Jaffe, E. S.; Cotelingam, J. D.; Whang–Peng, J.;Del Duca, V., Jr.; Medeiros, L. J.: T–cell lymphoblastic lymphomawith eosinophilia associated with subsequent myeloid malignancy. Am.J. Surg. Path. 16: 236–245, 1992.

[43618] 17170.Chesi, M.; Nardini, E.; Brents, L. A.; Schrock, E.; Ried, T.; Kuehl,W. M.; Bergsagel, P. L.: Frequent translocation t(4;14)(p16.3;q32.3)in multiple myeloma is associated with increased expression and activatingmutations of fibroblast growth factor receptor 3. Nature Genet. 16:260–264, 1997.

[43619] 17171.Demiroglu, A.; Steer, E. J.; Heath, C.; Taylor, K.; Bentley, M.;Allen, S. L.; Koduru, P.; Brody, J. P.; Hawson, G.; Rodwell, R.; Doody,M.–L.; Carnicero, F.; Reiter, A.; Goldman, J. M.; Melo, J. V.; Cross,N. C. P.: The t(8;22) in chronic myeloid leukemia fuses BCR to FGFR1:transforming activity and specific inhibition of FGFR1 fusion proteins. Blood 98:3778–3783, 2001.

[43620] 17172.Guasch, G.; Mack, G. J.; Popovici, C.; Dastugue, N.;

Birnbaum,D.; Rattner, J. B.; Pebusque, M.-J.: FGFR1 is fused to the centrosome-associated protein CEP110 in the 8p12 stem cell myeloproliferative disorder with t(8;9)(p12;q33). *Blood* 95: 1788–1796, 2000.

[43621] 17173. Baroni, M. G.; Oelbaum, R. S.; Pozzilli, P.; Stocks, J.; Li, S.-R.; Fiore, V.; Galton, D. J.: Polymorphisms at the GLUT1 (HepG2) and GLUT4 (muscle/adipocyte) glucose transporter genes and non-insulin-dependent diabetes mellitus (NIDDM). *Hum. Genet.* 88: 557–561, 1992.

[43622] 17174. Kelley, M. J.; Pech, M.; Seuanez, H. N.; Rubin, J. S.; O'Brien, S. J.; Aaronson, S. A.: Emergence of the keratinocyte growth factor multigene family during the great ape radiation. *Proc. Nat. Acad. Sci.* 89: 9287–9291, 1992.

[43623] 17175. Mattei, M.-G.; deLapeyriere, O.; Bresnick, J.; Dickson, C.; Birnbaum, D.; Mason, I.: Mouse Fgf7 (fibroblast growth factor 7) and Fgf8 (fibroblast growth factor 8) genes map to chromosomes 2 and 19 respectively. *Mammalian Genome* 6: 196–197, 1995.

[43624] 17176. Rubin, J. S.; Osada, H.; Finch, P. W.; Taylor, W. G.; Rudikoff, S.; Aaronson, S. A.: Purification and characterization of a newly identified growth factor specific for epithelial cells. *Proc. Nat. Acad. Sci.* 86: 802–806, 1989.

[43625] 17177. Werner, S.; Smola, H.; Liao, X.; Longaker, M. T.;

Krieg, T.; Hofschneider, P. H.; Williams, L. T.: The function of KGF in morphogenesis of epithelium and reepithelialization of wounds. *Science* 266: 819–822, 1994.

[43626] 17178. Zimonjic, D. B.; Kelley, M. J.; Rubin, J. S.; Aaronson, S. A.; Popescu, N. C.: Fluorescence in situ hybridization analysis of keratinocyte growth factor gene amplification and dispersion in evolution of great apes and humans. *Proc. Nat. Acad. Sci.* 94: 11461–11465, 1997.

[43627] 17179. Edwards, Y. H.; Parkar, M.; Povey, S.; West, L. F.; Parrington, J. M.; Solomon, E.: Human myosin heavy chain genes assigned to chromosome 17 using a human cDNA clone as probe. *Ann. Hum. Genet.* 49: 101–109, 1985.

[43628] 17180. Rappold, G. A.; Vosberg, H.-P.: Chromosomal localization of a human myosin heavy-chain gene by in situ hybridization. *Hum. Genet.* 65: 195–197, 1983.

[43629] 17181. Sparkes, R. S.; Zollman, S.; Klisak, I.; Kirchgessner, T. G.; Komaromy, M. C.; Mohandas, T.; Schotz, M. C.; Lussis, A. J.: Human genes involved in lipolysis of plasma lipoproteins: mapping of loci for lipoprotein lipase to 8p22 and hepatic lipase to 15q21. *Genomics* 1: 138–144, 1987.

[43630] 17182. Baumgartner, M.; Holzfeind, P.; Redl, B.: Assignment of the gene for human tear prealbumin (LCN1), a member of the lipocalin superfamily, to chromosome

8q24. Cytogenet. Cell Genet. 65: 101–103, 1994.

- [43631] 17183. Blaker, M.; Kock, K.; Ahlers, C.; Buck, F.; Schmale, H.: Molecular cloning of human von Ebner's gland protein, a member of the lipocalin superfamily highly expressed in lingual salivary glands. Biochim. Biophys. Acta 1172: 131–137, 1993.
- [43632] 17184. Liu, Y.; Wu, Y.-P.; Wada, R.; Neufeld, E. B.; Mullin, K. A.; Howard, A. C.; Pentchev, P. G.; Vanier, M. T.; Suzuki, K.; Proia, R. L.: Alleviation of neuronal ganglioside storage does not improve the clinical course of the Niemann–Pick C disease mouse. Hum. Molec. Genet. 9:1087–1092, 2000.
- [43633] 17185. Ito, M.; Yuan, C.-X.; Malik, S.; Gu, W.; Fondell, J. D.; Yamamura, S.; Fu, Z.-Y.; Zhang, X.; Qin, J.; Roeder, R. G.: Identity between TRAP and SMCC complexes indicates novel pathways for the function of nuclear receptors and diverse mammalian activators. Molec. Cell 3:361–370, 1999.
- [43634] 17186. Nagase, T.; Seki, N.; Ishikawa, K.; Tanaka, A.; Nomura, N.: Prediction of the coding sequences of unidentified human genes. V. The coding sequences of 40 new genes (KIAA0161–KIAA0200) deduced by analysis of cDNA clones from human cell line KG-1. DNA Res. 3: 17–24,

1996.

- [43635] 17187. Makino, K.; Kuwahara, H.; Masuko, N.; Nishiyama, Y.; Morisaki, T.; Sasaki, J.; Nakao, M.; Kuwano, A.; Nakata, M.; Ushio, Y.; Saya, H.: Cloning and characterization of NE-dlg: a novel human homolog of the Drosophila discs large (dlg) tumor suppressor protein interacts with the APC protein. *Oncogene* 14: 2425–2433, 1997.
- [43636] 17188. Stathakis, D. G.; Lee, D.; Bryant, P. J.: DLG3, the gene encoding human neuroendocrine Dlg (NE-Dlg), is located within the 1.8-Mb dystonia-parkinsonism region at Xq13.1. *Genomics* 49: 310–313, 1998.
- [43637] 17189. Maeda, K.; Matsushashi, S.; Hori, K.; Xin, Z.; Mukai, T.; Tabuchi, K.; Egashira, M.; Niikawa, N.: Cloning and characterization of a novel human gene, TM4SF6, encoding a protein belonging to the transmembrane 4 superfamily, and mapped to Xq22. *Genomics* 52: 240–242, 1998.
- [43638] 17190. Todd, S. C.; Doctor, V. S.; Levy, S.: Sequences and expression of six new members of the tetraspanin/TM4SF family. *Biochim. Biophys. Acta* 1399: 101–104, 1998.
- [43639] 17191. Crew, A. J.; Clark, J.; Fisher, C.; Gill, S.; Grimer, R.; Chand, A.; Shipley, J.; Gusterson, B. A.; Cooper, C. S.: Fusion of SYT to two genes, SSX1 and SSX2, encoding proteins with homology to the Kruppel-associated box in hu-

man synovial sarcoma. EMBO J. 14: 2333–2340, 1995.

[43640] 17192.Vaccari, T.; Beltrame, M.; Ferrari, S.; Bianchi, M. E.: Hmg4,a new member of the Hmg1/2 gene family. Genomics 49: 247–252, 1998.

[43641] 17193.Wilke, K.; Wiemann, S.; Gaul, R.; Gong, W.; Poustka, A.: Isolationof human and mouse HMG2a cDNAs: evidence for an HMG2a-specific 3-primeuntranslated region. Gene 198: 269–274, 1997.

[43642] 17194.Anson, D. S.; Blake, D. J.; Winship, P. R.; Birnbaum, D.; Brownlee,G. G.: Nullisomic deletion of the mcf.2 transforming gene in twohaemophilia B patients. EMBO J. 7: 2795–2799, 1988.

[43643] 17195.Imbeaud, S.; Belville, C.; Messika-Zeitoun, L.; Rey, R.; di Clemente,N.; Josso, N.; Picard, J.-Y.: A 27 base-pair deletion of the anti-Mulleriantype II receptor gene is the most common cause of the persistent Mullerianduct syndrome. Hum. Molec. Genet. 5: 1269–1277, 1996.

[43644] 17196.Imbeaud, S.; Carre-Eusebe, D.; Rey, R.; Belville, C.; Josso, N.;Picard, J.-Y.: Molecular genetics of the persistent Mullerian ductsyndrome: a study of 19 families. Hum. Molec. Genet. 3: 125–131,1994.

[43645] 17197.Imbeaud, S.; Faure, E.; Lamarre, I.; Mattei, M.-G.; di Clemente,N.; Tizard, R.; Carre-Eusebe, D.; Belville, C.;

Tragethon, L.; Tonkin, C.; Nelson, J.; McAuliffe, M.; Bidart, J.-M.; Lababidi, A.; Josso, N.; Cate, R. L.; Picard, J.-V.: Insensitivity to anti-Mullerian hormone due to a mutation in the human anti-Mullerian hormone receptor. *Nature Genet.* 11: 382–388, 1995.

[43646] 17198. Lang-Muritano, M.; Biason-Lauber, A.; Gitzelmann, C.; Belville, C.; Picard, Y.; Schoenle, E. J.: A novel mutation in the anti-mullerian hormone gene as cause of persistent mullerian duct syndrome. *Europ. J. Pediat.* 160: 652–654, 2001.

[43647] 17199. Picard, J.-Y.; Benarous, R.; Guerrier, D.; Josso, N.; Kahn, A.: Cloning and expression of cDNA for anti-mullerian hormone. *Proc. Nat. Acad. Sci.* 83: 5464–5468, 1986.

[43648] 17200. Tezcan, I.; Xu, W.; Gurgey, A.; Tuncer, M.; Cetin, M.; Oner, C.; Yetgin, S.; Ersoy, F.; Aizencang, G.; Astrin, K. H.; Desnick, R. J.: Congenital erythropoietic porphyria successfully treated by allogeneic bone marrow transplantation. *Blood* 92: 4053–4058, 1998.

[43649] 17201. Xu, W.; Astrin, K. H.; Desnick, R. J.: Molecular basis of congenital erythropoietic porphyria: mutations in the human uroporphyrinogen III synthase gene. *Hum. Mutat.* 7: 187–192, 1996.

- [43650] 17202. Pennarun, G.; Escudier, E.; Chapelin, C.; Bridoux, A.-M.; Cacheux, V.; Roger, G.; Clement, A.; Goossens, M.; Amselem, S.; Duriez, B.: Loss-of-function mutations in a human gene related to *Chlamydomonas reinhardtii* dynein IC78 result in primary ciliary dyskinesia. *Am.J. Hum. Genet.* 65: 1508–1519, 1999.
- [43651] 17203. Weiss, P.; Tietze, F.; Gahl, W. A.; Seppala, R.; Ashwell, G.: Identification of the metabolic defect in sialuria. *J. Biol. Chem.* 264:17635–17636, 1989.
- [43652] 17204. Bassi, M. T.; Ramesar, R. S.; Caciotti, B.; Winship, I. M.; DeGrandi, A.; Riboni, M.; Townes, P. L.; Beighton, P.; Ballabio, A.; Borsani, G.: X-linked late-onset sensorineural deafness caused by a deletion involving OA1 and a novel gene containing WD-40 repeats. *Am.J. Hum. Genet.* 64: 1604–1616, 1999.
- [43653] 17205. Converse, P. J.: Personal Communication. Baltimore, Md. 8/24/2001.
- [43654] 17206. Disteche, C. M.; Dinulos, M. B.; Bassi, M. T.; Elliott, R. W.; Rugarli, E. I.: Mapping of the murine *Tbl1* gene reveals a new rearrangement between mouse and human X chromosomes. *Mammalian Genome* 9: 1062–1064, 1998.
- [43655] 17207. Dong, X.; Tsuda, L.; Zavitz, K. H.; Lin, M.; Li, S.; Carthew, R. W.; Zipursky, S. L.: *ebi* regulates epidermal

growth factor receptorsignaling pathways in Drosophila.
Genes Dev. 13: 954–965, 1999.

- [43656] 17208.Matsuzawa, S.; Reed, J. C.: Siah–1, SIP, and Ebi col-
laborate ina novel pathway for beta–catenin degradation
linked to p53 responses. Molec.Cell 7: 915–926, 2001.
- [43657] 17209.Zhang, J.; Kalkum, M.; Chait, B. T.; Roeder, R. G.:
The N–CoR–HDAC3nuclear receptor corepressor complex
inhibits the JNK pathway throughthe integral subunit
GPS2. Molec. Cell 9: 611–623, 2002.
- [43658] 17210.Mu, J.; Roach, P. J.: Characterization of human
glycogenin–2,a self–glucosylating initiator of liver glyco-
gen metabolism. J. Biol.Chem. 273: 34850–34856, 1998.
- [43659] 17211.Mu, J.; Skurat, A. V.; Roach, P. J.: Glycogenin–2, a
novel self–glucosylatingprotein involved in liver glycogen
biosynthesis. J. Biol. Chem. 272:27589–27597, 1997.
- [43660] 17212.Delbridge, M. L.; Lingenfelter, P. A.; Disteche, C.
M.; MarshallGraves, J. A.: The candidate spermatogenesis
gene RBMY has a homologueon the human X chromo-
some. (Letter) Nature Genet. 22: 223–224, 1999.
- [43661] 17213.Le Coniat, M.; Soulard, M.; Della Valle, V.; Larsen,
C.–J.; Berger,R.: Localization of the human gene encoding
heterogeneous nuclearRNA ribonucleoprotein G
(hnRNP–G) to chromosome 6p12. Hum. Genet.

88:593–595, 1992.

[43662] 17214.Mazeyrat, S.; Saut, N.; Mattei, M.–G.; Mitchell, M. J.: RBMY evolved on the Y chromosome from a ubiquitously transcribed X–Y identical gene. (Letter) *Nature Genet.* 22: 224–226, 1999.

[43663] 17215.Venables, J. P.; Elliott, D. J.; Makarova, O. V.; Makarov, E. M.; Cooke, H. J.; Eperon, I. C.: RBMY, a probable human spermatogenesis factor, and other hnRNP G proteins interact with Tra2–beta and affect splicing. *Hum. Molec. Genet.* 9: 685–694, 2000.

[43664] 17216.Vogt, P. H.; Affara, N.; Davey, P.; Hammer, M.; Jobling, M. A.; Lau, Y. F.; Mitchell, M.; Schempp, W.; Tyler-Smith, C.; Williams, G.; Yen, P.; Rappold, G. A: Report of the Third International Workshop on Y Chromosome Mapping 1997. Heidelberg, Germany, April 13–16, 1997. *Cytogenet. Cell Genet.* 79: 1–20, 1997.

[43665] 17217.Wilcken, B.; Don, N.; Greenaway, R.; Hammond, J.; Sosula, L.: Sialuria: a second case. *J. Inher. Metab. Dis.* 10: 97–102, 1987.

[43666] 17218.Sutherland, G. R.; Baker, E.; Callen, D. F.; Hyland, V. J.; Wong, G.; Clark, S.; Jones, S. S.; Eglinton, L. K.; Shannon, M. F.; Lopez, A. F.; Vadas, M. A.: Interleukin 4 is at 5q31 and interleukin 6 is at 7p15. *Hum. Genet.* 79: 335–337,

1988.

- [43667] 17219.Symmons, D. P.; Jones, M.; Osborne, J.; Sills, J.; Southwood,T. R.; Woo, P.: Pediatric rheumatology in the United Kingdom: datafrom the British Pediatric Rheumatology Group National DiagnosticRegister. J. Rheum. 23: 1975–1980, 1996.
- [43668] 17220.Tosato, G.; Seamon, K. B.; Goldman, N. D.; Sehgal, P. B.; May,L. T.; Washington, G. C.; Jones, K. D.; Pike, S. E.: Monocyte–derivedhuman B–cell growth factor identified as interferon–beta–2 (BSF–2,IL–6). Science 239: 502–504, 1988.
- [43669] 17221.Villuendas, G.; San Millan, J. L.; Sancho, J. and Escobar–Morreale,H. F.: The –597 G–A and –174 G–C polymorphisms in the promoter ofthe IL–6 gene are associated with hyperandrogenism. J. Clin. Endocr.Metab. 87: 1134–1141, 2002.
- [43670] 17222.Zilberstein, A.; Ruggieri, R.; Korn, J. H.; Revel, M.: Structureand expression of cDNA and genes for human interferon–beta–2, a distinctspecies inducible by growth–stimulatory cytokines. EMBO J. 5: 2529–2537,1986.
- [43671] 17223.Gaedigk, R.; Duncan, A. M. V.; Miyazaki, I.; Robinson, B. H.; Dosch,H.–M.: ICA1 encoding p69, a protein linked to the development oftype 1 diabetes, maps to hu–

man chromosome 7p22. Cytogenet. Cell Genet.
66:274–276, 1994.

- [43672] 17224. Gaedigk, R.; Karges, W.; Hui, M. F.; Scherer, S. W.; Dosch, H.–M.: Genomic organization and transcript analysis of ICAp69, a target antigen in diabetic autoimmunity. Genomics 38: 382–391, 1996.
- [43673] 17225. Pietropaolo, M.; Castano, L.; Babu, S.; Buelow, R.; Kuo, Y.–L.S.; Martin, S.; Martin, A.; Powers, A. C.; Prochazka, M.; Naggert, J.; Leiter, E. H.; Eisenbarth, G. S.: Islet cell autoantigen 69 kD (ICA69): molecular cloning and characterization of a novel diabetes-associated autoantigen. J. Clin. Invest. 92: 359–371, 1993.
- [43674] 17226. Cavalieri, R. L.; Havell, E. A.; Vilcek, J.; Pestka, S.: Synthesis of human interferon by *Xenopus laevis* oocytes: two structural genes for interferons in human cells. Proc. Nat. Acad. Sci. 74: 3287–3291, 1977.
- [43675] 17227. Chany, C.; Finaz, C.; Weil, D.; Vignal, M.; Van Cong, N.; de Grouchy, J.: Investigations on the chromosomal localizations of the human and chimpanzee interferon genes: possible role of chromosomes 9 and 13. Ann. Genet. 23: 201–207, 1980.
- [43676] 17228. Derynck, R.; Content, J.; DeClercq, E.; Volckaert, G.; Tavernier, J.; Devos, R.; Fiers, W.: Isolation and structure of

a human fibroblastinterferon gene. *Nature* 285: 542–547, 1980.

- [43677] 17229.Diaz, M. O.; Le Beau, M. M.; Pitha, P.; Rowley, J. D.: Interferon and c-ets-1 genes in the translocation (9;11)(p22;q23) in human acute monocytic leukemia. *Science* 231: 265–267, 1986.
- [43678] 17230.Diaz, M. O.; Ziemin, S.; Le Beau, M. M.; Pitha, P.; Smith, S. D.; Chilcote, R. R.; Rowley, J. D.: Homozygous deletion of the alpha- and beta(1)-interferon genes in human leukemia and derived cell lines. *Proc.Nat. Acad. Sci.* 85: 5259–5263, 1988.
- [43679] 17231.Erickson, B. W.; May, L. T.; Sehgal, P. B.: Internal duplication in human alpha-1 and beta-1 interferons. *Proc. Nat. Acad. Sci.* 81:7171–7175, 1984.
- [43680] 17232.Henry, L.; Sizun, J.; Turleau, C.; Boue, J.; Azoulay, M.; Junien, C.: The gene for human fibroblast interferon (IFB) maps to 9p21. *Hum.Genet.* 68: 67–69, 1984.
- [43681] 17233.Houghton, M.; Eaton, M. A. W.; Stewart, A. G.; Smith, J. C.; Doel, S. M.; Catlin, G. H.; Lewis, H. M.; Patel, T. P.; Emtage, J. S.; Carey, N. H.; Porter, A. G.: The complete amino acid sequence of human fibroblastinterferon as deduced using synthetic oligodeoxyribonucleotide primers of reverse transcriptase. *Nucleic Acids Res.* 8: 2885–2894,

1980.

- [43682] 17234.Houghton, M.; Jackson, I. J.; Porter, A. G.; Doel, S. M.; Catlin, G. H.; Barber, C.; Carey, N. H.: The absence of introns within a human fibroblast interferon gene. *Nucleic Acids Res.* 9: 247–266, 1981.
- [43683] 17235.Knight, E., Jr.: Human fibroblast interferon: amino acid analysis and amino terminal amino acid sequence. *Science* 207: 525–527, 1980.
- [43684] 17236.May, L. T.; Landsberger, F. R.; Inouye, M.; Sehgal, P. B.: Significance of similarities in patterns: an application to beta interferon-related DNA on human chromosome 2. *Proc. Nat. Acad. Sci.* 82: 4090–4094, 1985.
- [43685] 17237.Meager, A.; Graves, H.; Burke, D. C.; Swallow, D. M.: Involvement of a gene on chromosome 9 in human fibroblast interferon production. *Nature* 280:493–495, 1979.
- [43686] 17238.Meager, A.; Graves, H. E.; Walker, J. R.; Burke, D. C.; Swallow, D. M.; Westerveld, A.: Tentative assignment of the gene for human fibroblast interferon to chromosome 9. (Abstract) *Cytogenet. Cell Genet.* 25: 183–184, 1979.
- [43687] 17239.Ohlsson, M.; Feder, J.; Cavalli-Sforza, L. L.; von Gabain, A.: Close linkage of alpha and beta interferons and infrequent duplication of beta interferon in humans.

Proc. Nat. Acad. Sci. 82: 4473–4476,1985.

- [43688] 17240.Ohno, S.; Taniguchi, T.: Structure of a chromosomal gene for human interferon beta. Proc. Nat. Acad. Sci. 78: 5305–5309, 1981.
- [43689] 17241.Owerbach, D.; Rutter, W. J.; Shows, T. B.; Gray, P.; Goeddel, D. V.; Lawn, R. M.: Leukocyte and fibroblast interferon genes are located on human chromosome 9. Proc. Nat. Acad. Sci. 78: 3123–3127,1981.
- [43690] 17242.Pitha, P. M.; Slate, D. L.; Raj, N. B. K.; Ruddle, F. H.: Human beta interferon gene localization and expression in somatic cell hybrids. Molec.Cell. Biol. 2: 564–570, 1982.
- [43691] 17243.Sagar, A. D.; Sehgal, P. B.; May, L. T.; Inouye, M.; Slate, D.L.; Shulman, L.; Ruddle, F. H.: Interferon-beta-related DNA is dispersed in the human genome. Science 223: 1312–1315, 1984.
- [43692] 17244.Sehgal, P. B.; May, L. T.; Sagar, A. D.; LaForge, K. S.; Inouye, M.: Isolation of novel human genomic DNA clones related to human interferon-beta(1) cDNA. Proc. Nat. Acad. Sci. 80: 3632–3636, 1983.
- [43693] 17245.Shepard, H. M.; Leung, D.; Stebbing, N.; Goeddel, D. V.: A single amino acid change in IFN-beta(1) abolishes its antiviral activity. Nature 294:563–567, 1981.
- [43694] 17246.Siegal, F. P.; Kadowaki, N.; Shodell, M.; Fitzgerald–

Bocarsly, P. A.; Shah, K.; Ho, S.; Antonenko, S.; Liu, Y.-J.:
The nature of the principal type 1 interferon-producing
cells in human blood. *Science* 284:1835–1837, 1999.

[43695] 17247. Erikson, J.; Griffin, C.; ar-Rushdi, A.; Valtieri, M.;
Hoxie, J.; Finan, J.; Emanuel, B. S.; Rovera, G.; Nowell, P. C.;
Croce, C. M.: Heterogeneity of chromosome 22 breakpoint
in Philadelphia-positive (Ph⁺) acute lymphocytic leukemia.
Proc. Nat. Acad. Sci. 83: 1807–1811, 1986.

[43696] 17248. Fioretos, T.; Heisterkamp, N.; Groffen, J.: No evi-
dence for genomic imprinting of the human BCR gene.
Blood 83: 3441–3444, 1994.

[43697] 17249. Fitzgerald, P. H.: Evidence that chromosome band
22q12 is concerned with cell proliferation in chronic
myeloid leukemia. *Hum. Genet.* 33:269–274, 1976.

[43698] 17250. Ganesan, T. S.; Rassool, F.; Guo, A.-P.; Th'ng, K. H.;
Dowding, C.; Hibbin, J. A.; Young, B. D.; White, H.; Ku-
maran, T. O.; Galton, D. A. G.; Goldman, J. M.: Rearrange-
ment of the bcr gene in Philadelphia chromosome-nega-
tive chronic myeloid leukemia. *Blood* 68: 957–960, 1986.

[43699] 17251. Goldman, J. M.; Melo, J. V.: Targeting the BCR-ABL
tyrosine kinase in chronic myeloid leukemia. (Editorial)
New Eng. J. Med. 344: 1084–1086, 2001.

[43700] 17252. Gorre, M. E.; Mohammed, M.; Ellwood, K.; Hsu, N.;

Paquette, R.; Rao, P. N.; Sawyers, C. L.: Clinical resistance to STI-571 cancertherapy caused by BCR-ABL gene mutation or amplification. Science 293:876-883, 2001.

[43701] 17253. Groffen, J.; Stephenson, J. R.; Heisterkamp, N.; de Klein, A.; Bartram, C. R.; Grosveld, G.: Philadelphia chromosomal breakpoints are clustered within a limited region, bcr, on chromosome 22. Cell 36:93-99, 1984.

[43702] 17254. Grossman, A.; Silver, R. T.; Arlin, Z.; Coleman, M.; Camposano, E.; Gascon, P.; Benn, P. A.: Fine mapping of chromosome 22 breakpoints within the breakpoint cluster region (bcr) implies a role for bcr exon 3 in determining disease duration in chronic myeloid leukemia. Am. J. Hum. Genet. 45: 729-738, 1989.

[43703] 17255. Grosveld, G.; Verwoerd, T.; van Agthoven, T.; de Klein, A.; Ramachandran, K. L.; Heisterkamp, N.; Stam, K.; Groffen, J.: The chronic myelocytic cell line K562 contains a breakpoint in bcr and produces a chimeric bcr/c-abl transcript. Molec. Cell. Biol. 6: 607-616, 1986.

[43704] 17256. Haas, O. A.: Are ABL and BCR imprinted? No definitive answers, but more questions. Leukemia 9: 740-745, 1995.

[43705] 17257. Haas, O. A.; Argyriou-Tirita, A.; Lion, T.: Parental origin of chromosomes involved in the translocation

t(9;22). Nature 359: 414–416,1992.

- [43706] 17258.Hariharan, I. K.; Adams, J. M.: cDNA sequence for human bcr,the gene that translocates to the abl oncogene in chronic myeloidleukaemia. EMBO J. 6: 115–119, 1987.
- [43707] 17259.Heisterkamp, N.; Jenster, G.; ten Hoeve, J.; Zovich, D.; Pattengale,P. K.; Groffen, J.: Acute leukaemia in bcr/abl transgenic mice. Nature 344:251–253, 1990.
- [43708] 17260.Heisterkamp, N.; Stam, K.; Groffen, J.; de Klein, A.; Grosveld,G.: Structural organization of the bcr gene and its role in the Ph–1translocation. Nature 315: 758–761, 1985.
- [43709] 17261.Hermans, A.; Heisterkamp, N.; von Lindern, M.; van Baal, S.; Meijer,D.; van der Plas, D.; Wiedemann, L. M.; Groffen, J.; Bootsma, D.;Grosveld, G.: Unique fusion of bcr and c–abl genes in Philadelphiachromosome positive acute lymphoblastic leukemia. Cell 51: 33–40,1987.
- [43710] 17262.Huettner, C. S.; Zhang, P.; Van Etten, R. A.; Tenen, D. G.: Reversibilityof acute B–cell leukaemia induced by BCR–ABL1. Nature Genet. 24:57–60, 2000.
- [43711] 17263.Jacobs, A.: Benzene and leukemia. Brit. J. Haemat. 72: 119–121,1989.
- [43712] 17264.Klein, G.: Specific chromosomal translocations and the genesisof the B–cell–derived tumors in mice and men.

Cell 32: 311–315,1983.

- [43713] 17265.Koeffler, H. P.; Golde, D. W.: Chronic myelogenous leukemia--newconcepts. New Eng. J. Med. 304: 1201–1209 and 1269–1274, 1981.
- [43714] 17266.Kohno, S.–I.; Sandberg, A. A.: Chromosomes and causation of humancancer and leukemia. XXXIX. Usual and unusual findings in Ph(1)–positiveCML. Cancer 46: 2227–2237, 1980.
- [43715] 17267.Laurent, E.; Talpaz, M.; Kantarjian, H.; Kurzrock, R.: The BCRgene and Philadelphia chromosome–positive leukemogenesis. CancerRes. 61: 2343–2355, 2001.
- [43716] 17268.Lillicrap, D. A.; Sterndale, H.: Familial chronic myeloid leukaemia.(Letter) Lancet II: 699, 1984.
- [43717] 17269.Lim, Y.–M.; Wong, S.; Lau, G.; Witte, O. N.; Colicelli, J.: BCR/ABLinhibition by an escort/phosphatase fusion protein. Proc. Nat. Acad.Sci. 97: 12233–12238, 2000.
- [43718] 17270.Litz, C. E.; Copenhaver, C. M.: Paternal origin of the rearrangedmajor breakpoint cluster region in chronic myeloid leukemia. Blood 83:3445–3448, 1994.
- [43719] 17271.Maru, Y.; Witte, O. N.: The BCR gene encodes a novel serine/threoninekinase activity within a single exon. Cell 67: 459–468, 1991.
- [43720] 17272.Melo, J. V.; Yan, X.–H.; Diamond, J.; Goldman, J. M.:

Lack of imprinting of the ABL gene. (Letter) *Nature Genet.* 8: 318–319, 1994.

[43721] 17273. Melo, J. V.; Yan, X.-H.; Diamond, J.; Goldman, J. M.: Balanced parental contribution to the ABL component of the BCR-ABL gene in chronic myeloid leukemia. *Leukemia* 9: 734–745, 1995.

[43722] 17274. Mes-Masson, A.-M.; McLaughlin, J.; Daley, G. Q.; Paskind, M.; Witte, O. N.: Overlapping cDNA clones define the complete coding region for the P210(c-abl) gene product associated with chronic myelogenous leukemia cells containing the Philadelphia chromosome. *Proc. Nat. Acad. Sci.* 83: 9768–9772, 1986.

[43723] 17275. Mills, K. I.; MacKenzie, E. D.; Birnie, G. D.: The site of the breakpoint within the bcr is a prognostic factor in Philadelphia-positive CML patients. *Blood* 72: 1237–1241, 1988.

[43724] 17276. Mittelman, F.; Levan, G.: Clustering of aberrations to specific chromosomes in human neoplasms. III. Incidence and geographic distribution of chromosome aberrations in 856 cases. *Hereditas* 89: 207–232, 1978.

[43725] 17277. Nowell, P. C.; Hungerford, D. A.: A minute chromosome in human chronic granulocytic leukemia. *Science* 132: 1497, 1960.

- [43726] 17278.Kusari, J.; Takata, Y.; Hatada, E.; Freidenberg, G.; Kolterman,O.; Olefsky, J. M.: Insulin resistance and diabetes due to differentmutations in the tyrosine kinase domain of both insulin receptor genealleles. J. Biol. Chem. 266: 5260–5267, 1991.
- [43727] 17279.Lander, E. S.; Botstein, D.: Homozygosity mapping: a way to map human recessive traits with the DNA of inbred children. Science 236:1567–1570, 1987.
- [43728] 17280.Kramer, F.; White, K.; Pauleikhoff, D.; Gehrig, A.; Passmore,L.; Rivera, A.; Rudolph, G.; Kellner, U.; Andrassi, M.; Lorenz, B.;Rohrschneider, K.; Blankenagel, A.; Jurklies, B.; Schilling, H.; Schutt,F.; Holz, F. G.; Weber, B. H. F.: Mutations in the VMD2 gene areassociated with juvenile-onset vitelliform macular dystrophy (Bestdisease) and adult vitelliform macular dystrophy but not age-relatedmacular degeneration. Europ. J. Hum. Genet. 8: 286–292, 2000.
- [43729] 17281.Sagane, K.; Ohya, Y.; Hasegawa, Y.; Tanaka, I.: Metalloproteinase-like,disintegrin-like, cysteine-rich proteins MDC2 and MDC3: novel humancellular disintegrins highly expressed in the brain. Biochem. J. 334:93–98, 1998.
- [43730] 17282.Alsobrook, J. P., II; Zohar, A. H.; Leboyer, M.; Chabane, N.; Ebstein,R. P.; Pauls, D. L.: Association between

the COMT locus and obsessive-compulsive disorder in females but not males. *Am. J. Med. Genet.*

(*Neuropsychiat. Genet.*) 114: 116–120, 2002.

[43731] 17283. Karayiorgou, M.; Sobin, C.; Blundell, M. L.; Galke, B. L.; Malinova, L.; Goldberg, P.; Ott, J.; Gogos, J. A.: Family-based association studies support a sexually dimorphic effect of COMT and MAOA on genetic susceptibility to obsessive-compulsive disorder. *Biol. Psychiat.* 45:1178–1189, 1999.

[43732] 17284. Taira, M.; Yoshida, T.; Miyagawa, K.; Sakamoto, H.; Terada, M.; Sugimura, T.: cDNA sequence of human transforming gene hst and identification of the coding sequence required for transforming activity. *Proc. Nat. Acad. Sci.* 84: 2980–2984, 1987.

[43733] 17285. Krill, A. E.; Morse, P. A.; Potts, A. M.; Klien, B. A.: Hereditary vitelliruptive macular degeneration. *Am. J. Ophthalmol.* 61: 1405–1415, 1966.

[43734] 17286. Maloney, W. F.; Robertson, D. M.; Miller, S. A.: Hereditary vitelliform macular degeneration--variable fundus findings within a single pedigree. *Arch. Ophthalmol.* 95: 979–983, 1977.

[43735] 17287. Mansergh, F. C.; Kenna, P. F.; Rudolph, G.; Meitinger, T.; Farrar, G. J.; Kumar-Singh, R.; Scorer, J.;

Hally, A. M.; Mynett-Johnson, L.; Humphries, M. M.; Kiang, S.; Humphries, P.: Evidence for genetic heterogeneity in Best's vitelliform macular dystrophy. *J. Med. Genet.* 32:855–858, 1995.

[43736] 17288. Marmorstein, A. D.; Marmorstein, L. Y.; Rayborn, M.; Wang, X.; Hollyfield, J. G.; Petrukhin, K.: Bestrophin, the product of the Best vitelliform macular dystrophy gene (VMD2), localizes to the basolateral plasma membrane of the retinal pigment epithelium. *Proc. Nat. Acad. Sci.* 97: 12758–12763, 2000.

[43737] 17289. Marquardt, A.; Stohr, H.; Passmore, L. A.; Kramer, F.; Rivera, A.; Weber, B. H. F.: Mutations in a novel gene, VMD2, encoding a protein of unknown properties cause juvenile-onset vitelliform macular dystrophy (Best's disease). *Hum. Molec. Genet.* 7: 1517–1525, 1998.

[43738] 17290. Nichols, B. E.; Bascom, R.; Litt, M.; McInnes, R.; Sheffield, V. C.; Stone, E. M.: Refining the locus for Best vitelliform macular dystrophy and mutation analysis of the candidate gene ROM1. *Am. J. Hum. Genet.* 54: 95–103, 1994.

[43739] 17291. Nordstrom, S.: Epidemiological studies of hereditary macular degeneration (Best's disease) in Swedish and Swedish-American populations. In: Eriksson, A. W.; Forsius,

H. R.; Nevanlinna, H. R.; Workman, P. L.; Norio, R. K.: Population Structure and Genetic Disorders. New York:Academic Press (pub.) 1980. Pp. 431–443.

- [43740] 17292.Nordstrom, S.: Personal Communication. Umea, Sweden 1978.
- [43741] 17293.Nordstrom, S.; Thorburn, W.: Dominantly inherited macular degeneration(Best's disease) in a homozygous father with 11 children. Clin. Genet. 18:211–216, 1980.
- [43742] 17294.O'Gorman, S.; Flaherty, W. A.; Fishman, G. A.; Berson, E. L.:Histopathologic findings in Best's vitelliform macular dystrophy. Arch.Ophthal. 106: 1261–1268, 1988.
- [43743] 17295.Petrukhin, K.; Koisti, M. J.; Bakall, B.; Li, W.; Xie, G.; Marknell,T.; Sandgren, O.; Forsman, K.; Holmgren, G.; Andreasson, S.; Vujic,M.; Bergen, A. A. B.; McGarty–Dugan, V.; Figueroa, D.; Austin, C.P.; Metzker, M. L.; Caskey, C. T.; Wadelius, C.: Identification ofthe gene responsible for Best macular dystrophy. Nature Genet. 19:241–247, 1998.
- [43744] 17296.Rivas, F.; Ruiz, C.; Rivera, H.; Moller, M.; Serrano–Lucas, J.I.; Cantu, J. M.: De novo del(6)(q25) associated with macular degeneration. Ann.Genet. 29: 42–44, 1986.
- [43745] 17297.Rosas, F. E.: Maculopatía hereditaria viteliforme de Best. Ann.Soc. Mex. Oft. 50: 157–171, 1976.
- [43746] 17298.Sorsby, A.; Savory, M.; Davey, J. B.; Fraser, R. J. L.:

Macularcysts: a dominantly inherited affection with a progressive course. *Brit.J. Ophthal.* 40: 144–158, 1956.

- [43747] 17299.Stohr, H.; Marquardt, A.; Rivera, A.; Cooper, P. R.; Nowak, N.J.; Shows, T. B.; Gerhard, D. S.; Weber, B. H. F.: A gene map of the Best's vitelliform macular dystrophy region in chromosome 11q12–q13.1. *GenomeRes.* 8: 48–56, 1998.
- [43748] 17300.Stone, E. M.; Nichols, B. E.; Streb, L. M.; Kimura, A. E.; Sheffield, V. C.: Genetic linkage of vitelliform macular degeneration (Best's disease) to chromosome 11q13. *Nature Genet.* 1: 246–250, 1992.
- [43749] 17301.Sun, H.; Tsunenari, T.; Yau, K.–W.; Nathans, J.: The vitelliform macular dystrophy protein defines a new family of chloride channels. *Proc.Nat. Acad. Sci.* 99: 4008–4013, 2002.
- [43750] 17302.Vail, D.; Shoch, D.: Hereditary degeneration of the macula. II. Follow–up report and histopathologic study. *Trans. Am. Ophthal. Soc.* 63:51–63, 1965.
- [43751] 17303.Vossius, A.: Ueber die Bestsche familiaere Maculadegeneration. *Arch.Ophthal.* 105: 1050–1059, 1921.
- [43752] 17304.Weber, B. H. F.; Walker, D.; Muller, B.: Molecular evidence for non–penetrance in Best's disease. *J. Med. Genet.* 31: 388–392, 1994.

- [43753] 17305.Weber, B. H. F.; Walker, D.; Muller, B.; Mar, L.: Best's vitelliformdystrophy (VMD2) maps between D11S903 and PYGM: no evidence for locus heterogeneity. *Genomics* 20: 267–274, 1994.
- [43754] 17306.White, K.; Marquardt, A.; Weber, B. H. F.: VMD2 mutations in vitelliform macular dystrophy (Best disease) and other maculopathies. *Hum.Mutat.* 15: 301–308, 2000.
- [43755] 17307.Yoder, F. E.; Cross, H. E.; Chase, G. A.; Fine, S. L.; Freidhoff, L.; Machan, C. H.; Bias, W. B.: Linkage studies of Best's macular dystrophy. *Clin. Genet.* 34: 26–30, 1988.
- [43756] 17308.Farndon, J. R.; Leight, G. S.; Dilley, W. G.; Baylin, S. B.; Smallridge, R. C.; Harrison, T. S.; Wells, S. A., Jr.: Familial medullary thyroid carcinoma without associated endocrinopathies: a distinct clinical entity. *Brit. J. Surg.* 73: 278–281, 1986.
- [43757] 17309.Wallis, D. E.; Roessler, E.; Hehr, U.; Nanni, L.; Wiltshire, T.; Richieri-Costa, A.; Gillesen-Kaesbach, G.; Zackai, E. H.; Rommens, J.; Muenke, M.: Mutations in the homeodomain of the human SIX3 gene cause holoprosencephaly. *Nature Genet.* 22: 196–198, 1999.
- [43758] 17310.McMichael, A.; Makgoba, W.: Complexity in human histocompatibility loci. *Nature* 293: 701–702, 1981.
- [43759] 17311.Moen, T.; Albrechtsen, D.; Flatmark, A.; Jakobsen,

A.; Jervell, J.; Halvorsen, S.; Solheim, B. G.; Thorsby, E.: Importance of HLA-DR matching in cadaveric renal transplantation: a prospective one-center study of 170 transplants. *New Eng. J. Med.* 303: 850-854, 1980.

[43760] 17312. Olerup, O.; Troye-Blomberg, M.; Schreuder, G. M. T.; Riley, E. M.: HLA-DR and -DQ gene polymorphism in West Africans is twice as extensive as in North European Caucasians: evolutionary implications. *Proc. Nat. Acad. Sci.* 88: 8480-8484, 1991.

[43761] 17313. Park, M. S.; Terasaki, P. I.; Bernoco, D.: Relationship between MT and DR antigens. In: Terasaki, P. I.: *Histo-compatibility Testing* 1980. Los Angeles: UCLA Press (pub.) 1980.

[43762] 17314. Park, M. S.; Terasaki, P. I.; Bernoco, D.; Iwaki, Y.: Evidence for a second B-cell locus separate from the DR locus. *Transplant. Proc.* 10: 823-828, 1978.

[43763] 17315. Pisella, P.-J.; Brignole, F.; Debbasch, C.; Lozato, P.-A.; Creuzot-Garcher, C.; Bara, J.; Saiag, P.; Warnet, J.-M.; Baudouin, C.: Flow cytometric analysis of conjunctival epithelium in ocular rosacea and keratoconjunctivitis sicca. *Ophthalmology* 107: 1841-1849, 2000.

[43764] 17316. Rollini, P.; Mach, B.; Gorski, J.: Linkage map of three HLA-DR beta-chain genes: evidence for a recent du-

- plication event. Proc.Nat. Acad. Sci. 82: 7197–7201, 1985.
- [43765] 17317.Sachs, J. A.; Jaraquemada, D.; Festenstein, H.: Intra HLA–D regionrecombinant maps HLA–DR between HLA–B and HLA–D. Tissue Antigens 17:43–56, 1981.
- [43766] 17318.Schwartz, B. D.: Personal Communication. St. Louis, Mo. 2/22/1983.
- [43767] 17319.Shackelford, D. A.; Mann, D. L.; van Rood, J. J.; Ferrara, G.B.; Strominger, J. L.: Human B–cell alloantigens DC1, MT1, and LB12are identical to each other but distinct from the HLA–DR antigen. Proc.Nat. Acad. Sci. 78: 4566–4570, 1981.
- [43768] 17320.Strominger, J. L.: Biology of the human histocompatibility leukocyteantigen (HLA) system and a hypothesis regarding the generation ofautoimmune diseases. J. Clin. Invest. 77: 1411–1415, 1986.
- [43769] 17321.Suciu–Foca, N.; Weiner, J.; Rohowsky, C.; McKiernan, P.; Susinno,E.; Rubinstein, P.: Indications that Dw determinants are controlledby distinct (but closely linked) genes. Transplant. Proc. 10: 799–804,1978.
- [43770] 17322.Thursz, M. R.; Thomas, H. C.; Greenwood, B. M.; Hill, A. V. S.: Heterozygote advantage for HLA class–II type in hepatitis B virusinfection. (Letter) Nature Genet. 17: 11–12, 1997.

- [43771] 17323.Tiercy, J.-M.; Gorski, J.; Jeannet, M.; Mach, B.: Identification and distribution of three serologically undetected alleles of HLA-DR by oligonucleotide-DNA typing analysis. *Proc. Nat. Acad. Sci.* 85:198-202, 1988.
- [43772] 17324.Tiwari, J. L.; Terasaki, P. I.: HLA and Disease Associations. New York: Springer-Verlag (pub.) 1985.
- [43773] 17325.Trowsdale, J.: Genetics and polymorphism: class II antigens. *Brit.Med. Bull.* 43: 15-36, 1987.
- [43774] 17326.Tsubota, K.; Fukagawa, K.; Fujihara, T.; Shimmura, S.; Saito, I.; Saito, K.; Takeuchi, T.: Regulation of human leukocyte antigen expression in human conjunctival epithelium. *Invest. Ophthal. Vis.Sci.* 40: 28-34, 1999.
- [43775] 17327.Walker, L. E.; Hewick, R.; Hunkapiller, M. W.; Hood, L. E.; Dreyer, W. J.; Reisfeld, R. A.: N-terminal amino acid sequences of the alpha and beta chains of HLA-DR1 and HLA-DR2 antigens. *Biochemistry* 23:185-188, 1983.
- [43776] 17328.Geraghty, D. E.; Pei, J.; Lipsky, B.; Hansen, J. A.; Taillon-Miller, P.; Bronson, S. K.; Chaplin, D. D.: Cloning and physical mapping of the HLA class I region spanning the HLA-E-to-HLA-F interval by using yeast artificial chromosomes. *Proc. Nat. Acad. Sci.* 89: 2669-2673, 1992.
- [43777] 17329.McAlpine, P. J.: Personal Communication. Winnipeg, Manitoba, Canada 6/22/1988.

- [43778] 17330.Stewart, W. E., II: The Interferon System. Berlin: Springer(pub.) 1979.
- [43779] 17331.Farfel, Z.; Iiri, T.; Shapira, H.; Roitman, A.; Mouallem, M.; Bourne, H. R.: Pseudohypoparathyroidism, a novel mutation in the beta/gamma- contact region of Gs- alpha impairs receptor stimulation. J.Biol. Chem. 271: 19653-19655, 1996.
- [43780] 17332.Heisler, L. K.; Cowley, M. A.; Tecott, L. H.; Fan, W.; Low, M.J.; Smart, J. L.; Rubinstein, M.; Tatro, J. B.; Marcus, J. N.; Holstege, H.; Lee, C. E.; Cone, R. D.; Elmquist, J. K.: Activation of central melanocortin pathways by fenfluramine. Science 297: 609-611, 2002.
- [43781] 17333.Folster-Holst, R.; Moises, H. W.; Yang, L.; Fritsch, W.; Weissenbach, J.; Christophers, E.: Linkage between atopy and the IgE high-affinity receptor gene at 11q13 in atopic dermatitis families. Hum. Genet. 102:236-239, 1998.
- [43782] 17334.Hill, M. R.; Cookson, W. O. C. M.: A new variant of the beta subunit of the high-affinity receptor for immunoglobulin E (Fc-epsilon-RI-betaE237G): associations with measures of atopy and bronchial hyper-responsiveness. Hum.Molec. Genet. 5: 959-962, 1996.
- [43783] 17335.Hupp, K.; Siwarski, D.; Mock, B. A.; Kinet, J.-P.:

Gene mapping of the three subunits of the high affinity FcR for IgE to mouse chromosomes 1 and 19. *J. Immun.* 143: 3787–3791, 1989.

- [43784] 17336. Kuster, H.; Zhang, L.; Brini, A. T.; MacGlashan, D. W. J.; Kinet, J.-P.: The gene and cDNA for the human high affinity immunoglobulin E receptor beta chain and expression of the complete human receptor. *J. Biol. Chem.* 267: 12782–12787, 1992.
- [43785] 17337. Nagata, H.; Mutoh, H.; Kumahara, K.; Arimoto, Y.; Tomemori, T.; Sakurai, D.; Arase, K.; Ohno, K.; Yamakoshi, T.; Nakano, K.; Okawa, T.; Numata, T.; Konno, A.: Association between nasal allergy and a coding variant of the Fc-epsilon-R1-beta gene Glu237Gly in a Japanese population. *Hum. Genet.* 109: 262–266, 2001.
- [43786] 17338. Shirakawa, T.; Li, A.; Dubowitz, M.; Dekker, J. W.; Shaw, A. E.; Faux, J. A.; Ra, C.; Cookson, W. O. C. M.; Hopkin, J. M.: Association between atopy and variants of the beta subunit of the high-affinity immunoglobulin E receptor. *Nature Genet.* 7: 125–130, 1994.
- [43787] 17339. Shirakawa, T.; Mao, X.-Q.; Sasaki, S.; Enomoto, T.; Kawai, M.; Morimoto, K.; Hopkin, J.: Association between atopic asthma and a coding variant of Fc-epsilon-R1-beta in a Japanese population. *Hum. Molec. Genet.* 5:

1129–1130, 1996.

[43788] 17340.Szepietowski, P.; Gaudray, P.: FCER1B, a candidate gene for atopy, is located in 11q13 between CD20 and TCN1. *Genomics* 19: 399–400, 1994.

[43789] 17341.Kuster, H.; Thompson, H.; Kinet, J.–P.: Characterization and expression of the gene for the human Fc receptor gamma subunit: definition of a new gene family. *J. Biol. Chem.* 265: 6448–6452, 1990.

[43790] 17342.Takayanagi, H.; Kim, S.; Matsuo, K.; Suzuki, H.; Suzuki, T.; Sato, K.; Yokochi, T.; Oda, H.; Nakamura, K.; Ida, N.; Wagner, E. F.; Taniguchi, T.: RANKL maintains bone homeostasis through c–Fos–dependent induction of interferon–B. *Nature* 416: 744–749, 2002.

[43791] 17343.Taniguchi, T.; Fujii–Kuriyama, Y.; Muramatsu, M.: Molecular cloning of human interferon cDNA. *Proc. Nat. Acad. Sci.* 77: 4003–4006, 1980.

[43792] 17344.Taniguchi, T.; Mantei, N.; Schwarzstein, M.; Nagata, S.; Muramatsu, M.; Weissmann, C.: Human leukocyte and fibroblast interferons are structurally related. *Nature* 285: 547–549, 1980.

[43793] 17345.Taniguchi, T.; Ohno, S.; Fujii–Kuriyama, Y.; Muramatsu, M.: The nucleotide sequence of human fibroblast interferon cDNA. *Gene* 10:11–15, 1980.

- [43794] 17346.Tavernier, J.; Derynck, R.; Fiers, W.: Evidence for a unique human fibroblast interferon (IFN-beta1) chromosomal gene devoid of intervening sequences. *Nucleic Acids Res.* 9: 461-471, 1981.
- [43795] 17347.Tavernier, J.; Gheysen, D.; Duerinck, F.; Van der Heyden, J.; Fiers, W.: Deletion mapping of the inducible promoter of human IFN-beta gene. *Nature* 301: 634-636, 1983.
- [43796] 17348.Trent, J. M.; Olson, S.; Lawn, R.: Chromosomal localization of human leukocyte, fibroblast, and immune interferon genes by means of in situ hybridization. *Proc. Nat. Acad. Sci.* 79: 7809-7813, 1982.
- [43797] 17349.Weissenbach, J.; Chernajovsky, Y.; Zeevi, M.; Shulman, L.; Soreq, H.; Nir, U.; Wallach, D.; Perricaudet, M.; Tiollais, P.; Revel, M.: Two interferon mRNAs in human fibroblasts: in vitro translation and *Escherichia coli* cloning studies. *Proc. Nat. Acad. Sci.* 77: 7152-7156, 1980.
- [43798] 17350.Zinn, K.; DiMaio, D.; Maniatis, T.: Identification of two distinct regulatory regions adjacent to the human beta-interferon gene. *Cell* 34: 865-879, 1983.
- [43799] 17351.Bruns, G. A. P.; Eisenman, R. E.; Gerald, P. S.: Human mitochondrial NADP-dependent isocitrate dehydrogenase in man-mouse somatic cell hybrids. *Cytogenet. Cell*

Genet. 17: 200–211, 1976.

- [43800] 17352. Champion, M. J.; Brown, J. A.; Shows, T. B.: Assignment of cytoplasmic α -mannosidase (MAN-A) and confirmation of the mitochondrial isocitrate dehydrogenase (IDH-M) genes to the q11-qter region of chromosome 15 in man. Cytogenet. Cell Genet. 22: 498–502, 1978.
- [43801] 17353. Grzeschik, K.-H.: Assignment of a gene for human mitochondrial isocitrate dehydrogenase (ICD-M, EC 1.1.1.41.) to chromosome 15. Hum. Genet. 34: 23–28, 1976.
- [43802] 17354. Huh, T.-L.; Kim, Y.-O.; Oh, I.-U.; Song, B. J.; Inazawa, J.: Assignment of the human mitochondrial NAD(+)-specific isocitrate dehydrogenase α subunit (IDH3A) gene to 15q25.1–q25.2 by in situ hybridization. Genomics 32: 295–296, 1996.
- [43803] 17355. Oh, I.-U.; Inazawa, J.; Kim, Y.-O.; Song, B. J.; Huh, T.-L.: Assignment of the human mitochondrial NADP(+)-specific isocitrate dehydrogenase (IDH2) gene to 15q26.1 by in situ hybridization. Genomics 38: 104–106, 1996.
- [43804] 17356. Shimizu, N.; Giles, R. E.; Kucherlapati, R. S.; Shimizu, Y.; Ruddle, F. H.: Somatic cell genetic assignment

of the human gene for mitochondrial NADP-linked isocitrate dehydrogenase to the long arm of chromosome 15. *Somat. Cell Genet.* 3: 47–60, 1977.

- [43805] 17357. Appel, S.; Filter, M.; Reis, A.; Hennies, H. C.; Bergheim, A.; Ogilvie, E.; Arndt, S.; Simmons, A.; Lovett, M.; Hide, W.; Ramsay, M.; Reichwald, K.; Zimmermann, W.; Rosenthal, A.: Physical and transcriptional map of the critical region for keratolytic winter erythema (KWE) on chromosome 8p22–p23 between D8S550 and D8S1759. *Eur. J. Hum. Genet.* 10:17–25, 2002.
- [43806] 17358. Olavarria, E.; Craddock, C.; Dazzi, F.; Marin, D.; Markt, S.; Apperley, J. F.; Goldman, J. M.: Imatinib mesylate (STI571) in the treatment of relapse of chronic myeloid leukemia after allogeneic stem cell transplantation. *Blood* 99: 3861–3862, 2002.
- [43807] 17359. Pegoraro, L.; Matera, L.; Ritz, J.; Levis, A.; Palumbo, A.; Biagini, G.: Establishment of a Ph(1)–positive human cell line (BV173). *J. Nat. Cancer Inst.* 70: 447–451, 1983.
- [43808] 17360. Perrotti, D.; Cesi, V.; Trotta, R.; Guerzoni, C.; Santilli, G.; Campbell, K.; Iervolino, A.; Condorelli, F.; Gambacorti-Passerini, C.; Caligiuri, M. A.; Calabretta, B.: BCR–ABL suppresses C/EBP- α expression through inhibitory action of hnRNP E2. *Nature Genet.* 30:48–58, 2002.

- [43809] 17361.Prakash, O.; Yunis, J. J.: High resolution chromosomes of the t(9;22) positive leukemias. *Cancer Genet. Cytogenet.* 11: 361–367, 1984.
- [43810] 17362.Priest, J. R.; Robison, L. L.; McKenna, R. W.; Lindquist, L. L.; Warkentin, P. I.; LeBien, T. W.; Woods, W. G.; Kersey, J. H.; Coccia, P. F.; Nesbit, M. E., Jr.: Philadelphia chromosome positive childhood acute lymphoblastic leukemia. *Blood* 56: 15–22, 1980.
- [43811] 17363.Rowley, J. D.: A new consistent chromosomal abnormality in chronic myelogenous leukemia identified by quinacrine fluorescence and Giemsa staining. *Nature* 243: 290–293, 1973.
- [43812] 17364.Rubin, C. M.; Carrino, J. J.; Dickler, M. N.; Leibowitz, D.; Smith, S. D.; Westbrook, C. A.: Heterogeneity of genomic fusion of BCR and ABL in Philadelphia chromosome-positive acute lymphoblastic leukemia. *Proc. Nat. Acad. Sci.* 85: 2795–2799, 1988.
- [43813] 17365.Saglio, G.; Storlazzi, C. T.; Giugliano, E.; Surace, C.; Anelli, L.; Rege-Cambrin, G.; Zagaria, A.; Velasco, A. J.; Heiniger, A.; Scaravaglio, P.; Gomez, A. T.; Gomez, J. R.; Archidiacono, N.; Banfi, S.; Rocchi, M.: A 76-kb duplication maps close to the BCR gene on chromosome 22 and the ABL gene on chromosome 9: possible involvement in the

genesis of the Philadelphia chromosome translocation.

Proc. Nat. Acad. Sci. 99:9882–9887, 2002.

[43814] 17366. Savage, D. G.; Antman, K. H.: Imatinib mesylate—a new oral targeted therapy. New Eng. J. Med. 346: 683–693, 2002.

[43815] 17367. Sawyers, C. L.: Chronic myeloid leukemia. New Eng. J. Med. 340:1330–1340, 1999.

[43816] 17368. Schaefer-Rego, K.; Dudek, H.; Popenoe, D.; Arlin, Z.; Mears, J.G.; Bank, A.; Leibowitz, D.: CML patients in blast crisis have breakpoints localized to a specific region of the BCR. Blood 70: 448–455, 1987.

[43817] 17369. Shtivelman, E.; Gale, R. P.; Dreazen, O.; Berrebi, A.; Zaizov, R.; Kubonishi, I.; Miyoshi, I.; Canaani, E.: bcr-abl RNA in patients with chronic myelogenous leukemia. Blood 69: 971–973, 1987.

[43818] 17370. Shtivelman, E.; Lifshitz, B.; Gale, R. P.; Canaani, E.: Fused transcript of abl and bcr genes in chronic myelogenous leukaemia. Nature 315:550–554, 1985.

[43819] 17371. Skorski, T.; Nieborowska-Skorska, M.; Nicolaides, N. C.; Szczylik, C.; Iversen, P.; Iozzo, R. V.; Zon, G.; Calabretta, B.: Suppression of Philadelphia-1 leukemia cell growth in mice by BCR-ABL antisense oligodeoxynucleotide. Proc. Nat. Acad. Sci. 91: 4504–4508, 1994.

- [43820] 17372. Stam, K.; Heisterkamp, N.; Grosveld, G.; de Klein, A.; Verma, R. S.; Coleman, M.; Dosik, H.; Groffen, J.: Evidence of a new chimeric bcr/c-abl mRNA in patients with chronic myelocytic leukemia and the Philadelphia chromosome. *New Eng. J. Med.* 313: 1429–1433, 1985.
- [43821] 17373. Stam, K.; Heisterkamp, N.; Reynolds, F. H., Jr.; Groffen, J.: Evidence that the phl gene encodes a 160,000–dalton phosphoprotein with associated kinase activity. *Molec. Cell. Biol.* 7: 1955–1960, 1987.
- [43822] 17374. Swan, D. C.; McBride, O. W.; Robbins, K. C.; Keithley, D. A.; Reddy, E. P.; Aaronson, S. A.: Chromosomal mapping of the simian sarcoma virus onc gene analogue in human cells. *Proc. Nat. Acad. Sci.* 79: 4691–4695, 1982.
- [43823] 17375. Tanabe, T.; Kuwabara, T.; Warashina, M.; Tani, K.; Taira, K.; Asano, S.: Oncogene inactivation in a mouse model: tissue invasion by leukaemic cells is stalled by loading them with a designer ribozyme. *Nature* 406: 473–474, 2000.
- [43824] 17376. Teyssier, J. R.; Bartram, C. R.; Deville, J.; Potron, G.; Pigeon, F.: C-abl oncogene and chromosome 22 'bcr' juxtaposition in chronic myelogenous leukemia. *New Eng. J. Med.* 312: 1393–1394, 1985.
- [43825] 17377. Tkachuk, D. C.; Westbrook, C. A.; Andreeff, M.;

Donlon, T. A.;Cleary, M. L.; Suryanarayan, K.; Homge, M.; Redner, A.; Gray, J.;Pinkel, D.: Detection of bcr–abl fusion in chronic myelogeneous leukemia by in situ hybridization. Science 250: 559–562, 1990.

[43826] 17378.Verhest, A.; Monsieur, R.: Philadelphia chromosome–positive thrombocythemia with leukemic transformation. (Letter) New Eng. J. Med. 308: 1603,1983.

[43827] 17379.Verma, R. S.; Dosik, H.: Heteromorphisms of the Philadelphia(Ph–1) chromosome in patients with chronic myelogenous leukaemia (CML).I. Classification and clinical significance. Brit. J. Haemat. 45:215–222, 1980.

[43828] 17380.Cockerham, G. C.; Hidayat, A. A.; Bijwaard, K. E.; Sheng, Z.–M.: Re–evaluation of 'reactive lymphoid hyperplasia of the uvea': an immunohistochemical and molecular analysis of 10 cases. Ophthalmology 107:151–158, 2000.

[43829] 17381.Guru, S. C.; Agarwal, S. K.; Manickam, P.; Olufemi, S.–E.; Crabtree,J. S.; Weisemann, J. M.; Kester, M. B.; Kim, Y. S.; Wang, Y.; Emmert–Buck,M. R.; Liotta, L. A.; Spiegel, A. M.; Boguski, M. S.; Roe, B. A.;Collins, F. S.; Marx, S. J.; Burns, L.; Chandrasekharappa, S. C.:A transcript map for the 2.8–Mb region containing the multiple endocrineneoplasia type 1 locus. Genome Res. 7: 725–735, 1997.

[43830] 17382.Frossard, P. M.; Lestringant, G. G.: Association be–

tween a dimorphic site on chromosome 12 and clinical diagnosis of hypertension in three independent populations. Clin. Genet. 48: 284–287, 1995.

- [43831] 17383. Glenn, C. L.; Wang, W. Y. S.; Benjafield, A. V.; Morris, B. J.: Linkage and association of tumor necrosis factor receptor 2 locus with hypertension, hypercholesterolemia and plasma shed receptor. Hum. Molec. Genet. 9: 1943–1949, 2000.
- [43832] 17384. Lasky, L. A.; Singer, M. S.; Dowbenko, D.; Imai, Y.; Henzel, W. J.; Grimley, C.; Fennie, C.; Gillett, N.; Watson, S. R.; Rosen, S. D.: An endothelial ligand for L-selectin is a novel mucin-like molecule. Cell 69:927–938, 1992.
- [43833] 17385. Lasky, L. A.; Singer, M. S.; Yednock, T. A.; Dowbenko, D.; Fennie, C.; Rodriguez, H.; Nguyen, T.; Stachel, S.; Rosen, S. D.: Cloning of a lymphocyte homing receptor reveals a lectin domain. Cell 56:1045–1055, 1989.
- [43834] 17386. Ord, D. C.; Ernst, T. J.; Zhou, L.-J.; Rambaldi, A.; Spertini, O.; Griffin, J.; Tedder, T. F.: Structure of the gene encoding the human leukocyte adhesion molecule-1 (TQ1, Leu-8) of lymphocytes and neutrophils. J. Biol. Chem. 265: 7760–7767, 1990.
- [43835] 17387. Siegelman, M. H.; Weissman, I. L.: Human homologue of mouse lymph node homing receptor: evolutionary

conservation at tandem cell interaction domains. Proc. Nat. Acad. Sci. 86: 5562–5566, 1989.

- [43836] 17388. Sitrin, R. G.; Pan, P. M.; Blackwood, R. A.; Huang, J.; Petty, H. R.: Cutting edge: evidence for a signaling partnership between urokinase receptors (CD87) and L-selectin (CD62L) in human polymorphonuclear neutrophils. J. Immun. 166: 4822–4825, 2001.
- [43837] 17389. Tedder, T. F.; Isaacs, C. M.; Ernst, T. J.; Demetri, G. D.; Adler, D. A.; Distech, C. M.: Isolation and chromosomal localization of cDNAs encoding a novel human lymphocyte cell surface molecule, LAM-1: homology with the mouse lymphocyte homing receptor and other human adhesion proteins. J. Exp. Med. 170: 123–133, 1989.
- [43838] 17390. Aizawa, S.; Nakano, H.; Ishida, T.; Horie, R.; Nagai, M.; Ito, K.; Yagita, H.; Okumura, K.; Inoue, J.; Watanabe, T.: Tumor necrosis factor receptor-associated factor (TRAF) 5 and TRAF2 are involved in CD30-mediated NF- κ B activation. J. Biol. Chem. 272: 2042–2045, 1997.
- [43839] 17391. Durkop, H.; Latza, U.; Hummel, M.; Eitelbach, F.; Seed, B.; Stein, H.: Molecular cloning and expression of a new member of the nerve growth factor receptor family that is characteristic for Hodgkin's disease. Cell 68: 421–427, 1992.

- [43840] 17392.Kurts, C.; Carbone, F. R.; Krummel, M. F.; Koch, K. M.; Miller, J. F. A. P.; Heath, W. R.: Signalling through CD30 protects against autoimmune diabetes mediated by CD8 T cells. *Nature* 398: 341–344, 1999.
- [43841] 17393.McClive, P. J.; Morahan, G.: Assignment of the mouse homologues of 6 loci from HSA1p to chromosomes 3 and 4. *Genomics* 23: 243–246, 1994.
- [43842] 17394.Stein, H.; Gerdes, J.; Schwab, U.; Lemke, H.; Mason, D. Y.; Ziegler, A.; Schienle, W.; Diehl, V.: Identification of Hodgkin and Sternberg–Reed cells as a unique cell type derived from a newly detected small cell population. *Int. J. Cancer* 30: 445–449, 1982.
- [43843] 17395.de Lau, W.; Clevers, H.: LEF1 turns over a new leaf. *Nature Genet.* 28:3–5, 2001.

- [43844] 17396.Hovanes, K.; Li, T. W. H.; Munguia, J. E.; Truong, T.; Milovanovic,T.; Marsh, J. L.; Holcombe, R. F.; Waterman, M. L.: Beta-catenin-sensitive isoforms of lymphoid enhancer factor-1 are selectively expressed in colon cancer. *Nature Genet.* 28: 53-57, 2001.
- [43845] 17397.Love, J. J.; Li, X.; Case, D. A.; Giese, K.; Grosschedl, R.; Wright,P. E.: Structural basis for DNA bending by the architectural transcription factor LEF-1. *Nature* 376: 791-795, 1995.
- [43846] 17398.Milatovich, A.; Travis, A.; Grosschedl, R.; Francke, U.: LEF1,the gene for lymphoid enhancer-binding factor 1, mapped to human chromosome4 (q23-q25) and distal mouse chromosome 3 by Southern blot analysis and fluorescence in situ hybridization. (Abstract) *Cytogenet. Cell-Genet.* 58: 1888 only, 1991.
- [43847] 17399.Milatovich, A.; Travis, A.; Grosschedl, R.; Francke, U.: Gene for lymphoid enhancer-binding factor 1 (LEF1) mapped to human chromosome4 (q23-q25) and mouse chromosome 3 near Egf. *Genomics* 11: 1040-1048,1991.
- [43848] 17400.van Genderen, C.; Okamura, R. M.; Farinas, I.; Quo, R. G.; Parslow,T. G.; Bruhn, L.; Grosschedl, R.: Development of several organs that require inductive epithelial-mesenchymal interactions is impaired in LEF-1-deficient

mice. *Genes Dev.* 8: 2691–2703, 1994.

- [43849] 17401. Waterman, M. L.; Fischer, W. H.; Jones, K. A.: A thymus-specific member of the HMG protein family regulates the human T cell receptor C alpha enhancer. *Genes Dev.* 5: 656–669, 1991.
- [43850] 17402. Zhou, P.; Byrne, C.; Jacobs, J.; Fuchs, E.: Lymphoid enhancer factor 1 directs hair follicle patterning and epithelial cell fate. *Genes Dev.* 9: 700–713, 1995.
- [43851] 17403. White, P. S.; Jensen, S. J.; Rajalingam, V.; Stairs, D.; Sulman, E. P.; Maris, J. M.; Biegel, J. A.; Wooster, R.; Brodeur, G. M.: Physical mapping of the CA6, ENO1, and SLC2A5 (GLUT5) genes and reassignment of SLC2A5 to 1p36.2. *Cytogenet. Cell Genet.* 81: 60–64, 1998.
- [43852] 17404. Contractor, A.; Swanson, G.; Heinemann, S. F.: Kainate receptors are involved in short- and long-term plasticity at mossy fiber synapses in the hippocampus. *Neuron* 29: 209–216, 2001.
- [43853] 17405. Paschen, W.; Hedreen, J. C.; Ross, C. A.: RNA editing of the glutamate receptor subunits GluR2 and GluR6 in human brain tissue. *J. Neurochem.* 63: 1596–1602, 1994.
- [43854] 17406. Barbon, A.; Barlati, S.: Genomic organization, proposed alternative splicing mechanisms, and RNA editing structure of GRIK1. *Cytogenet. Cell Genet.* 88: 236–239,

2000.

- [43855] 17407.Eubanks, J. H.; Puranam, R. S.; Kleckner, N. W.; Bettler, B.; Heinemann, S. F.; McNamara, J. O.: The gene encoding the glutamate receptor subunit GluR5 is located on human chromosome 21q21.1–22.1 in the vicinity of the gene for familial amyotrophic lateral sclerosis. *Proc. Nat. Acad. Sci.* 90: 178–182, 1993.
- [43856] 17408.Gregor, P.; Gaston, S. M.; Yang, X.; O'Regan, J. P.; Rosen, D.R.; Tanzi, R. E.; Patterson, D.; Haines, J. L.; Horvitz, H. R.; Uhl, G. R.; Brown, R. H., Jr.: Genetic and physical mapping of the GLUR5 glutamate receptor gene on human chromosome 21. *Hum. Genet.* 94:565–570, 1994.
- [43857] 17409.Gregor, P.; Reeves, R. H.; Jabs, E. W.; Yang, X.; Dackowski, W.; Rochelle, J. M.; Brown, R. H., Jr.; Haines, J. L.; O'Hara, B. F.; Uhl, G. R.; Seldin, M. F.: Chromosomal localization of glutamate receptor genes: relationship to familial amyotrophic lateral sclerosis and other neurological disorders of mice and humans. *Proc. Nat. Acad. Sci.* 90: 3053–3057, 1993.
- [43858] 17410.Gregor, P.; Seldin, M. F.; Reeves, R.; Jabs, E.; Yang, X.; Rochelle, J. M.; O'Hara, B. F.; Uhl, G. R.: Chromosomal localization of glutamate receptor genes: candidates for

familial amyotrophic lateral sclerosis and other neurological disorders of mice and man. (Abstract) *Neurosci. Abstracts* 18: 395 only, 1992.

- [43859] 17411. Paschen, W.; Djuricic, B.: Extent of RNA editing of glutamate receptor subunit GluR5 in different brain regions of the rat. *Cell. Molec. Neurobiol.* 14: 259–270, 1994.
- [43860] 17412. Potier, M.-C.; Dutriaux, A.; Lambolez, B.; Bochet, P.; Rossier, J.: Assignment of the human glutamate receptor gene GLUR5 to 21q22 by screening a chromosome 21 YAC library. *Genomics* 15: 696–697, 1993.
- [43861] 17413. Sander, T.; Hildmann, T.; Kretz, R.; Furst, R.; Sailer, U.; Bauer, G.; Schmitz, B.; Beck-Mannagetta, G.; Wienker, T. F.; Janz, D.: Allelic association of juvenile absence epilepsy with a glutamate receptor gene (GRIK1) polymorphism. *Am. J. Med. Genet.* 74: 416–421, 1997.
- [43862] 17414. Smolders, I.; Bortolotto, Z. A.; Clarke, V. R. J.; Warre, R.; Khan, G. M.; O'Neill, M. J.; Ornstein, P. L.; Bleakman, D.; Ogden, A.; Weiss, B.; Stables, J. P.; Ho, K. H.; Ebinger, G.; Collingridge, G. L.; Lodge, D.; Michotte, Y.: Antagonists of GLU-K5-containing kainate receptors prevent pilocarpine-induced limbic seizures. *Nature Neurosci.* 5: 796–804, 2002.

- [43863] 17415.McNamara, J. O.; Eubanks, J. H.; McPherson, J. D.; Wasmuth, J.J.; Evans, G. A.; Heinemann, S. F.: Chromosomal localization of humanglutamate receptor genes. *J. Neurosci.* 12: 2555–2562, 1992.
- [43864] 17416.Armstrong, N.; Gouaux, E.: Mechanisms for activation and antagonism of an AMPA-sensitive glutamate receptor: crystal structures of the GluR2 ligand binding core. *Neuron* 28: 165–181, 2000.
- [43865] 17417.Brusa, R.; Zimmermann, F.; Koh, D.-S.; Feldmeyer, D.; Gass, P.; Seeburg, P. H.; Sprengel, R.: Early-onset epilepsy and postnatal lethality associated with an editing-deficient GluR-B allele in mice. *Science* 270:1677–1680, 1995.
- [43866] 17418.Hollmann, M.; Hartley, M.; Heinemann, S.: Ca^{2+} permeability of KA-AMPA-gated glutamate receptor channels depends on subunit composition. *Science* 252:851–853, 1991.
- [43867] 17419.Kohler, M.; Kornau, H. C.; Seeburg, P. H.: The organization of the gene for the functionally dominant α -amino-3-hydroxy-5-methylisoxazole-4-propionic acid receptor subunit Glu4-B. *J. Biol. Chem.* 269: 17367–17370, 1994.
- [43868] 17420.Sun, W.; Ferrer-Montiel, A. V.; Schinder, A. F.;

McPherson, J.P.; Evans, G. A.; Montal, M.: Molecular cloning, chromosomal mapping, and functional expression of human brain glutamate receptors. *Proc.Nat. Acad. Sci.* 89: 1443–1447, 1992.

[43869] 17421.Sun, Y.; Olson, R.; Horning, M.; Armstrong, N.; Mayer, M.; Gouaux,E.: Mechanism of glutamate receptor desensitization. *Nature* 417:245–253, 2002.

[43870] 17422.Chui, D.; Oh-Eda, M.; Liao, Y.-F.; Panneerselvam, K.; Lai, A.;Marek, K. W.; Freeze, H. H.; Moremen, K. W.; Fukuda, M. N.; Marth,J. D.: Alpha-mannosidase-II deficiency results in dyserythropoiesis and unveils an alternate pathway in oligosaccharide biosynthesis. *Cell* 90:157–167, 1997.

[43871] 17423.Chui, D.; Sellakumar, G.; Green, R. S.; Sutton-Smith, M.; McQuistan,T.; Marek, K. W.; Morris, H. R.; Dell, A.; Marth, J. D.: Genetic remodeling of protein glycosylation in vivo induces autoimmune disease. *Proc.Nat. Acad. Sci.* 98: 1142–1147, 2001.

[43872] 17424.Gasparini, P.; del Giudice, E. M.; Delaunay, J.; Totaro, A.; Granatiero,M.; Melchionda, S.; Zelante, L.; Iolascon, A.: Localization of the congenital dyserythropoietic anemia II locus to chromosome 20q11.2 by genomewide search. *Am. J. Hum. Genet.* 61: 1112–1116, 1997.

- [43873] 17425.Misago, M.; Liao, Y.-F.; Kudo, S.; Eto, S.; Mattei, M.-G.; Moremen,K. W.; Fukuda, M. N.: Molecular cloning and expression of cDNAs encoding human alpha-mannosidase II and a previously unrecognized alpha-mannosidasell(X) isozyme. *Proc. Nat. Acad. Sci.* 92: 11766-11770, 1995.
- [43874] 17426.Moremen, K. W.; Robbins, P. W.: Isolation, characterization, and expression of cDNAs encoding murine alpha-mannosidase II, a Golgi enzyme that controls conversion of high mannose to complex N-glycans. *J.Cell Biol.* 115: 1521-1534, 1991.
- [43875] 17427.Wada, A.; Sakamoto, H.; Katoh, O.; Yoshida, T.; Yokota, J.; Little,P. F. R.; Sugimura, T.; Terada, M.: Two homologous oncogenes, HST1 and INT2, are closely located in human genome. *Biochem. Biophys. Res. Commun.* 157: 828-835, 1988.
- [43876] 17428.Yoshida, T.; Tsutsumi, M.; Sakamoto, H.; Miya-gawa, K.; Teshima,S.; Sugimura, T.; Terada, M.: Expression of the HST1 oncogene in human germ cell tumors. *Biochem. Biophys. Res. Commun.* 155: 1324-1329, 1988.
- [43877] 17429.Nimmo, E.; Padua, R.-A.; Hughes, D.; Brook, J. D.; Williamson,R.; Johnson, K. J.: Confirmation and refinement of the localisation of the c-MEL locus on chromosome 19

by physical and genetic mapping. Hum.Genet. 81:
382–384, 1989.

- [43878] 17430.Nimmo, E.; Williamson, R.; Johnson, K.: Localization of the c-MELgene to 19(cen-p13.2). (Abstract) Cytogenet. Cell Genet. 51: 1053only, 1989.
- [43879] 17431.Nimmo, E. R.; Sanders, P. G.; Padua, R. A.; Hughes, D.; Williamson,R.; Johnson, K. J.: The MEL gene: a new member of the RAB/YPT classof RAS-related genes. Onco-gene 6: 1347–1351, 1991.
- [43880] 17432.Padua, R. A.; Barrass, N.; Currie, G. A.: A novel transforminggene in a human malignant melanoma cell line. Nature 311: 671–673,1984.
- [43881] 17433.Spurr, N. K.; Hughes, D.; Goodfellow, P. N.; Brook, J. D.; Padua,R. A.: Chromosomal assignment of c-MEL, a human transforming oncogene,to chromosome 19(p13.2-q13.2). Somat. Cell Molec. Genet. 12: 637–640,1986.
- [43882] 17434.Dabiri, G. A.; Young, C. L.; Rosenbloom, J.; South-wick, F. S.:Molecular cloning of human macrophage cap-ping protein cDNA: a uniquemember of the gelsolin/villin family expressed primarily in macrophages. J.Biol. Chem. 267: 16545–16552, 1992.
- [43883] 17435.Mishra, V. S.; Henske, E. P.; Kwiatkowski, D. J.;

Southwick, F.S.: The human actin-regulatory protein cap G: gene structure and chromosome location. *Genomics* 23: 560–565, 1994.

[43884] 17436. Southwick, F. S.; DiNubile, M. J.: Rabbit alveolar macrophages contain a Ca^{2+} -sensitive, 41,000-Dalton protein which reversibly blocks the 'barbed' ends of actin filaments but does not sever them. *J. Biol. Chem.* 261: 14191–14195, 1986.

[43885] 17437. Eichbaum, Q.; Clerc, P.; Bruns, G.; McKeon, F.; Ezekowitz, R. A.B.: Assignment of the human macrophage mannose receptor gene (MRC1) to 10p13 by in situ hybridization and PCR-based somatic cell hybrid mapping. *Genomics* 22: 656–658, 1994.

[43886] 17438. Harris, N.; Peters, L. L.; Eicher, E. M.; Rits, M.; Raspberry, D.; Eichbaum, Q. G.; Super, M.; Ezekowitz, R. A. B.: The exon-intron structure and chromosomal localization of the mouse macrophage mannose receptor gene, *Mcr1*: identification of a ricin-like domain at the N-terminus of the receptor. *Biochem. Biophys. Res. Commun.* 198:682–692, 1994.

[43887] 17439. Kim, S. J.; Ruiz, N.; Bezouska, K.; Drickamer, K.: Organization of the gene encoding the human macrophage mannose receptor (MRC1). *Genomics* 14:721–727, 1992.

- [43888] 17440.Lee, S. J.; Evers, S.; Roeder, D.; Parlow, A. F.; Risteli, J.; Risteli, L.; Lee, Y. C.; Feizi, T.; Langen, H.; Nussenzweig, M. C.: Mannose receptor-mediated regulation of serum glycoprotein homeostasis. *Science* 295:1898–1901, 2002.
- [43889] 17441.Bernhagen, J.; Calandra, T.; Mitchell, R. A.; Martin, S. B.; Tracey, K. J.; Voelter, W.; Manogue, K. R.; Cerami, A.; Bucala, R.: MIF is a pituitary-derived cytokine that potentiates lethal endotoxaemia. *Nature* 365:756–759, 1993.
- [43890] 17442.Bloom, B. R.; Bennett, B.: Mechanism of a reaction in vitro associated with delayed-type hypersensitivity. *Science* 153: 80–82, 1966.
- [43891] 17443.Bozza, M.; Kolakowski, L. F., Jr.; Jenkins, N. A.; Gilbert, D.J.; Copeland, N. G.; David, J. R.; Gerard, C.: Structural characterization and chromosomal location of the mouse macrophage migration inhibitory factor gene and pseudogenes. *Genomics* 27: 412–419, 1995.
- [43892] 17444.Bucala, R.: MIF rediscovered: cytokine, pituitary hormone, and glucocorticoid-induced regulator of the immune response. *FASEB J.* 10:1607–1613, 1996.
- [43893] 17445.Budarf, M.; McDonald, T.; Sellinger, B.; Kozak, C.; Graham, C.; Wistow, G.: Localization of the human gene for macrophage migration inhibitory factor (MIF) to chromosome 22q11.2. *Genomics* 39: 235–236, 1997.

- [43894] 17446.Cato, A. C. B.; Sillar, G. M.; Kioussis, J.; Burdon, R. H.: Molecularcloning of cDNA sequences coding for the major (beta-, gamma-, delta-,and epsilon) heat-shock polypeptides of HeLa cells. *Gene* 16: 27-34,1981.
- [43895] 17447.Cummings, C. J.; Sun, Y.; Opal, P.; Antalffy, B.; Mestril, R.;Orr, H. T.; Dillmann, W. H.; Zoghbi, H. Y.: Over-expression of inducibleHSP70 chaperone suppresses neuropathology and improves motor functionin SCA1 mice. *Hum. Molec. Genet.* 10: 1511-1518, 2001.
- [43896] 17448.Gaskins, H. R.; Prochazka, M.; Nadeau, J. H.; Henson, V. W.; Leiter,E. H.: Localization of a mouse heat shock Hsp70 gene within the H-2complex. *Immunogenetics* 32: 286-289, 1990.
- [43897] 17449.Goate, A. M.; Cooper, D. N.; Hall, C.; Leung, T. K. C.; Solomon,E.; Lim, L.: Localization of a human heat-shock HSP70 gene sequenceto chromosome 6 and detection of two other loci by somatic-cell hybridand restriction fragment length polymorphism analysis. *Hum. Genet.* 75:123-128, 1987.
- [43898] 17450.Fukai, K.; Oh, J.; Frenk, E.; Almodovar, C.; Spritz, R. A.: Linkagedisequilibrium mapping of the gene for Hermansky-Pudlak syndrome tochromosome 10q23.1-q23.3. *Hum. Molec. Genet.* 4: 1665-1669, 1995.

- [43899] 17451.Gwynn, B.; Eicher, E. M.; Peters, L. L.: Genetic localization of Cd63, a member of the transmembrane 4 superfamily, reveals two distinct loci in the mouse genome. *Genomics* 35: 389–391, 1996.
- [43900] 17452.Hotta, H.; Ross, A. H.; Huebner, K.; Isobe, M.; Wendeborn, S.; Chao, M. V.; Ricciardi, R. P.; Tsujimoto, Y.; Croce, C. M.; Koprowski, H.: Molecular cloning and characterization of an antigen associated with early stages of melanoma tumor progression. *Cancer Res.* 48:2955–2962, 1988.
- [43901] 17453.Metzelaar, M. J.; Wijngaard, P. L. J.; Peters, P. J.; Sixma, J.J.; Nieuwenhuis, H. K.; Clevers, H. C.: CD63 antigen: a novel lysosomal membrane glycoprotein, cloned by a screening procedure for intracellular antigens in eukaryotic cells. *J. Biol. Chem.* 266: 3239–3245, 1991.
- [43902] 17454.Nishibori, M.; Cham, B.; McNicol, A.; Shalev, A.; Jain, N.; Gerrard, J. M.: The protein CD63 is in platelet dense granules, is deficient in a patient with Hermansky–Pudlak syndrome, and appears identical to granulophysin. *J. Clin. Invest.* 91: 1775–1782, 1993.
- [43903] 17455.Azizi, E.; Friedman, J.; Pavlovsky, F.; Iscovich, J.; Bornstein, A.; Shafir, R.; Trau, H.; Brenner, H.; Nass, D.: Familial cutaneous malignant melanoma and tumors of the

nervous system. *Cancer* 76:1571–1578, 1995.

- [43904] 17456.Doege, K.; Rhodes, C.; Sasaki, M.; Hassell, J. R.; Yamada, Y.:Molecular biology of cartilage proteoglycan (aggrecan) and link protein.In:Sandel, L. J.; Boyd, C. D.: Extracellular Matrix Genes. New York:Academic Press (pub.) 1990. Pp. 137–152.
- [43905] 17457.Finkelstein, J. E.; Doege, K.; Yamada, Y.; Pyeritz, R. E.; Graham,J. M., Jr.; Moeschler, J. B.; Pauli, R. M.; Hecht, J. T.; Francomano,C. A.: Analysis of the chondroitin sulfate proteoglycan core protein(CSPGCP) gene in achondroplasia and pseudoachondroplasia. *Am. J.Hum. Genet.* 48: 97–102, 1991.
- [43906] 17458.Just, W.; Klett, C.; Vetter, U.; Vogel, W.: Assignment of thehuman aggrecan gene AGC1 to 15q25–q26.2 by in situ hybridization. *Hum.Genet.* 92: 516–518, 1993.
- [43907] 17459.Kimata, K.; Barrach, H. J.; Brown, K. S.; Penny-packer, J. P.:Absence of proteoglycan core protein in cartilage from the cmd/cmd(cartilage deficiency) mouse. *J. Biol. Chem.* 256: 6961–6968, 1981.
- [43908] 17460.Korenberg, J. R.; Chen, X. N.; Doege, K.; Grover, J.; Roughley,P. J.: Assignment of the human aggrecan gene (AGC1) to 15q26 usingfluorescence in situ hybridization analysis. *Genomics* 16: 546–548,1993.

- [43909] 17461.Lane, P. W.: New mutants and linkages: small with kinky tail. MouseNewsletter 80: 165, 1988.
- [43910] 17462.Watanabe, H.; Kimata, K.; Line, S.; Strong, D.; Gao, L.; Kozak,C. A.; Yamada, Y.: Mouse cartilage matrix deficiency (cmd) causedby a 7 bp deletion in the aggrecan gene. Nature Genet. 7: 154–157,1994.
- [43911] 17463.Watanabe, H.; Nakata, K.; Kimata, K.; Nakanishi, I.; Yamada, Y.: Dwarfism and age-associated spinal degeneration of heterozygotecmd mice defective in aggrecan. Proc. Nat. Acad. Sci. 94: 6943–6947,1997.
- [43912] 17464.Fong, L. Y. Y.; Fidanza, V.; Zanesi, N.; Lock, L. F.; Siracusa,L. D.; Mancini, R.; Siprashvili, Z.; Ottey, M.; Martin, S. E.; Druck,T.; McCue, P. A.; Croce, C. M.; Huebner, K.: Muir–Torre–like syndromein Fhit–deficient mice. Proc. Nat. Acad. Sci. 97: 4742–4747, 2000.
- [43913] 17465.Azen, E. A.; Carlson, D. M.; Clements, S.; Lalley, P. A.; Vanin,E.: Salivary proline–rich protein genes on chromosome 8 of mouse. Science 226:967–969, 1984.
- [43914] 17466.Azen, E. A.; Davisson, M. T.; Cherry, M.; Taylor, B. A.: Prp (proline–richprotein) genes linked to markers Es–12 (esterase–12), Ea–10 (erythrocytealloantigen), and loci on distal mouse chromosome 6. Genomics 5:415–422, 1989.

- [43915] 17467.Azen, E. A.; Denniston, C. L.: Genetic polymorphism of human salivary proline-rich proteins, further genetic analysis. *Biochem. Genet.* 12:109–120, 1974.
- [43916] 17468.Azen, E. A.; Oppenheim, F. G.: Genetic polymorphism of proline-rich human salivary proteins. *Science* 180: 1067–1069, 1973.
- [43917] 17469.Baird, M.; Neuweiler, J.; Balazs, I.: Linkage studies between proline rich-protein (SPC) genes and RFLPs from chromosome 12. (Abstract) *Cytogenet. Cell Genet.* 40: 573–574, 1985.
- [43918] 17470.Degand, P.; Aubert, J. P.; Boersma, A.; Richet, C.; Loucheux-Lefebvre, M. H.; Biserte, G.: Parotid alpha-amylase activity: a possible role for proline-rich proteins. *FEBS Lett.* 63: 137–140, 1976.
- [43919] 17471.Goodman, P. A.; Yu, P.-L.; Azen, E. A.; Karn, R. C.: The human salivary protein complex (SPC): a large block of related genes. *Am. J. Hum. Genet.* 37: 785–797, 1985.
- [43920] 17472.Oppenheim, F. G.; Hay, D. I.; Fraublau, C.: Proline-rich proteins from human parotid saliva. Isolation and partial characterization. *Biochemistry* 10:4233–4238, 1971.
- [43921] 17473.Adkins, S.; Gan, K. N.; Mody, M.; La Du, B. N.: Molecular basis for the polymorphic forms of human serum paraoxonase/arylesterase: glutamine or arginine at

position 191, for the respective A and Ballozymes. Am. J. Hum. Genet. 52: 598–608, 1993.

- [43922] 17474. Antikainen, M.; Murtomaki, S.; Syvanne, M.; Pahlman, R.; Tahvanainen, E.; Jauhiainen, M.; Frick, M. H.; Ehnholm, C.: The gln–arg191 polymorphism of the human paraoxonase gene (HUMPONA) is not associated with the risk of coronary artery disease in Finns. J. Clin. Invest. 98: 883–885, 1996.
- [43923] 17475. Augustinsson, K.–B.; Henricson, B.: A genetically controlled esterase in rat plasma. Biochim. Biophys. Acta 124: 323–331, 1966.
- [43924] 17476. Barbieri, M.; Bonafe, M.; Marfella, R.; Ragno, E.; Giugliano, D.; Franceschi, C.; Paolisso, G.: LL–paraoxonase genotype is associated with a more severe degree of homeostasis model assessment IR in healthy subjects. J. Clin. Endocr. Metab. 87: 222–225, 2002.
- [43925] 17477. Brophy, V. H.; Jampsa, R. L.; Clendenning, J. B.; McKinstry, L. A.; Jarvik, G. P.; Furlong, C. E.: Effects of 5–prime regulatory–region polymorphisms on paraoxonase–gene (PON1) expression. Am. J. Hum. Genet. 68: 1428–1436, 2001.
- [43926] 17478. Cherry, N.; Mackness, M.; Durrington, P.; Povey, A.; Dippnall, M.; Smith, T.; Mackness, B.: Paraoxonase (PON1)

polymorphisms in farmers attributing ill health to sheep dip. *Lancet* 359: 763–764, 2002.

- [43927] 17479. Clendenning, J. B.; Humbert, R.; Green, E. D.; Wood, C.; Traver, D.; Furlong, C. E.: Structural organization of the human PON1 gene. *Genomics* 35: 586–589, 1996.
- [43928] 17480. Coates, P. M.; Mestriner, M. A.; Hopkinson, D. A.: A preliminary interpretation of the esterase isozymes of human tissues. *Ann. Hum. Genet.* 39: 1–20, 1975.
- [43929] 17481. Davies, H. G.; Richter, R. J.; Keifer, M.; Broomfield, C. A.; Sowalla, J.; Furlong, C. E.: The effect of the human serum paraoxonase polymorphism is reversed with diazoxon, soman and sarin. *Nature Genet.* 14: 334–336, 1996.
- [43930] 17482. Deakin, S.; Leviev, I.; Nicaud, V.; Meynet, M.–C. B.; Tiret, L.; James, R. W.: Paraoxonase–1 L55M polymorphism is associated with an abnormal oral glucose tolerance test and differentiates high risk coronary disease families. *J. Clin. Endocr. Metab.* 87: 1268–1273, 2002.
- [43931] 17483. Eckerson, H. W.; Romson, J.; Wyte, C.; La Du, B. N.: The human serum paraoxonase polymorphism: identification of phenotypes by their response to salts. *Am. J. Hum. Genet.* 35: 214–227, 1983.
- [43932] 17484. Eckerson, H. W.; Wyte, C. M.; La Du, B. N.: The hu–

man serum paraoxonase/arylesterase polymorphism. *Am. J. Hum. Genet.* 35: 1126–1136, 1983.

[43933] 17485. Eiberg, H.; Mohr, J.: Linkage relations of the paraoxonase polymorphism with 43 marker systems. (Abstract) *Cytogenet. Cell Genet.* 25: 150, 1979.

[43934] 17486. Rey-Campos, J.; Chouard, T.; Yaniv, M.; Cereghini, S.: vHNF1 is a homeoprotein that activates transcription and forms heterodimers with HNF1. *EMBO J.* 10: 1445–1457, 1991.

[43935] 17487. Shih, D. Q.; Bussen, M.; Sehayek, E.; Ananthanarayanan, M.; Shneider, B. L.; Suchy, F. J.; Shefer, S.; Bollileni, J. S.; Gonzalez, F. J.; Breslow, J. L.; Stoffel, M.: Hepatocyte nuclear factor-1- α is an essential regulator of bile acid and plasma cholesterol metabolism. *Nature-Genet.* 27: 375–382, 2001.

[43936] 17488. Urhammer, S. A.; Hansen, T.; Ekstrom, C. T.; Eiberg, H.; Pederson, O.: The Ala/Val98 polymorphism of the hepatocyte nuclear factor-1- α gene contributes to the interindividual variation in serum C-peptide response during an oral glucose tolerance test: evidence from studies of 231 glucose-tolerant first degree relatives of type 2 diabetic probands. *J. Clin. Endocr. Metab.* 83: 4506–4509, 1998.

- [43937] 17489. Urhammer, S. A.; Moller, A. M.; Nyholm, B.; Ekstrom, C. T.; Eiberg, H.; Clausen, J. O.; Hansen, T.; Pedersen, O.; Schmitz, O.: The effect of two frequent amino acid variants of the hepatocyte nuclear factor-1 α gene on estimates of the pancreatic beta-cell function in Caucasian glucose-tolerant first-degree relatives of type 2 diabetic patients. *J. Clin. Endocr. Metab.* 83: 3992–3995, 1998.
- [43938] 17490. Urhammer, S. A.; Rasmussen, S. K.; Kaisaki, P. J.; Oda, N.; Yamagata, K.; Moller, A. M.; Fridberg, M.; Hansen, L.; Hansen, T.; Bell, G. I.; Pedersen, O.: Genetic variation in the hepatocyte nuclear factor-1 α gene in Danish Caucasians with late-onset NIDDM. *Diabetologia* 40:473–475, 1997.
- [43939] 17491. Vaxillaire, M.; Rouard, M.; Yamagata, K.; Oda, N.; Kaisaki, P. J.; Boriraj, V. V.; Chevre, J.-C.; Boccio, V.; Cox, R. D.; Lathrop, G. M.; Dussoix, P.; Philippe, J.; Timsit, J.; Charpentier, G.; Velho, G.; Bell, G. I.; Froguel, P.: Identification of nine novel mutations in the hepatocyte nuclear factor 1 α gene associated with maturity-onset diabetes of the young (MODY3). *Hum. Molec. Genet.* 6: 583–586, 1997.
- [43940] 17492. Yamada, S.; Nishigori, H.; Onda, H.; Utsugi, T.;

Yanagawa, T.;Maruyama, T.; Onigata, K.; Nagashima, K.; Nagai, R.; Morikawa, A.;Takeuchi, T.; Takeda, J.: Identification of mutations in the hepatocytenuclear factor (HNF)-1-alpha gene in Japanese subjects with IDDM. Diabetes 46:1643-1647, 1997.

[43941] 17493.Yamagata, K.; Oda, N.; Kalsaki, P. J.; Menzel, S.; Furuta, H.;Vaxillaire, M.; Southam, L.; Cox, R. D.; Lathrop, G. M.; Borhaj, V.V.; Chen, X.; Cox, N. J.; Oda, Y.; Yano, H.; Le Beau, M. M.; Yamada,S.; Nishigori, H.; Takeda, J.; Fajans, S. S.; Hattersley, A. T.; Iwasaki,N.; Hansen, T.; Pedersen, O.; Polonsky, K. S.; Turner, R. C.; Velho,G.; Chevre, J.-C.; Froguel, P.; Bell, G. I.: Mutations in the hepatocytenuclear factor-1-alpha gene in maturity-onset diabetes of the young(MODY3). Nature 384: 455-457, 1996.

[43942] 17494.Yoshiuchi, I.; Yamagata, K.; Yoshimoto, M.; Zhu, Q.; Yang, Q.;Nammo, T.; Uenaka, R.; Kinoshita, E.; Hanafusa, T.; Miyagawa, J.;Matsuzawa, Y.: Analysis of a non-functional HNF-1-alpha (TCF1) mutationin Japanese subjects with familial type 1 diabetes. Hum. Mutat. 18:345-351, 2001.

[43943] 17495.Dhanasekaran, S. M.; Barrette, T. R.; Ghosh, D.; Shah, R.; Varambally,S.; Kurachi, K.; Pienta, K. J.; Rubin, M. A.; Chinnaiyan, A. M.:Delineation of prognostic biomark-

ers in prostate cancer. *Nature* 412:822–826, 2001.

- [43944] 17496. Leytus, S. P.; Loeb, K. R.; Hagen, F. S.; Kurachi, K.; Davie, E.W.: A novel trypsin-like serine protease (hepsin) with a putative transmembrane domain expressed by human liver and hepatoma cells. *Biochemistry* 27:1067–1074, 1988.
- [43945] 17497. Tsuji, A.; Torres-Rosado, A.; Arai, T.; Le Beau, M. M.; Lemons, R. S.; Chou, S.-H.; Kurachi, K.: Hepsin, a cell membrane-associated protease: characterization, tissue distribution, and gene localization. *J. Biol. Chem.* 266: 16948–16953, 1991.
- [43946] 17498. Wu, Q.; Yu, D.; Post, J.; Halks-Miller, M.; Sadler, J. E.; Morser, J.: Generation and characterization of mice deficient in hepsin, a hepatic transmembrane serine protease. *J. Clin. Invest.* 101: 321–326, 1998.
- [43947] 17499. Cannella, B.; Hoban, C. J.; Gao, Y.-L.; Garcia-Arenas, R.; Lawson, D.; Marchionni, M.; Gwynne, D.; Raine, C. S.: The neuregulin, glial growth factor 2, diminishes autoimmune demyelination and enhances remyelination in a chronic relapsing model for multiple sclerosis. *Proc. Nat. Acad. Sci.* 95: 10100–10105, 1998.
- [43948] 17500. Falls, D. L.; Rosen, K. M.; Corfas, G.; Lane, W. S.; Fischbach, G. D.: ARIA, a protein that stimulates acetyl-

choline receptor synthesis, is a member of the Neu ligand family. *Cell* 72: 801–815, 1993.

- [43949] 17501. Fernandez, P.-A.; Tang, D. G.; Cheng, L.; Prochiantz, A.; Mudge, A. W.; Raff, M. C.: Evidence that axon-derived neuregulin promotes oligodendrocyte survival in the developing rat optic nerve. *Neuron* 28:81–90, 2000.
- [43950] 17502. Holmes, W. E.; Sliwkowski, M. X.; Akita, R. W.; Henzel, W. J.; Lee, J.; Park, J. W.; Yansura, D.; Abadi, N.; Raab, H.; Lewis, G. D.; Shepard, H. M.; Kuang, W.-J.; Wood, W. I.; Goeddel, D. V.; Vandlen, R. L.: Identification of heregulin, a specific activator of p185(erbB2). *Science* 256:1205–1210, 1992.
- [43951] 17503. Huang, Y. Z.; Won, S.; Ali, D. W.; Wang, Q.; Tanowitz, M.; Du, Q. S.; Pelkey, K. A.; Yang, D. J.; Xiong, W. C.; Salter, M. W.; Mei, L.: Regulation of neuregulin signaling by PSD-95 interacting with ErbB4 at CNS synapses. *Neuron* 26: 443–455, 2000.
- [43952] 17504. Lee, J.; Wood, W. I.: Assignment of heregulin (HGL) to human chromosome 8p22–p11 by PCR analysis of somatic cell hybrid DNA. *Genomics* 16:790–791, 1993.
- [43953] 17505. Martinou, J.-C.; Falls, D. L.; Fischbach, G. D.; Merlie, J. P.: Acetylcholine receptor-inducing activity stimulates expression of the epsilon-subunit gene of the muscle

acetylcholine receptor. Proc.Nat. Acad. Sci. 88:
7669–7673, 1991.

- [43954] 17506.Ni, C.-Y.; Murphy, M. P.; Golde, T. E.; Carpenter, G.: Gamma-secretase cleavage and nuclear localization of ErbB-4 receptor tyrosine kinase. Science 294:2179–2181, 2001.
- [43955] 17507.Orr-Urtreger, A.; Trakhtenbrot, L.; Ben-Levy, R.; Wen, D.; Rechavi, G.; Lonai, P.; Yarden, Y.: Neural expression and chromosomal mapping of NEU differentiation factor to 8p12–p21. Proc. Nat. Acad. Sci. 90:1867–1871, 1993.
- [43956] 17508.Boyd, D.; Jain, S. K.; Crampton, J.; Barrett, K. J.; Drysdale, J.: Isolation and characterization of a cDNA clone for human ferritin heavy chain. Proc. Nat. Acad. Sci. 81: 4751–4755, 1984.
- [43957] 17509.Caskey, J. H.; Jones, C.; Miller, Y. E.; Seligman, P. A.: Human ferritin gene is assigned to chromosome 19. Proc. Nat. Acad. Sci. 80:482–486, 1983.
- [43958] 17510.Costanzo, F.; Colombo, M.; Staempfli, S.; Santoro, C.; Marone, M.; Frank, R.; Delius, H.; Cortese, R.: Structure of gene and pseudogenes of human apoferritin H. Nucleic Acids Res. 14: 721–736, 1986.
- [43959] 17511.Cragg, S. J.; Drysdale, J.; Worwood, M.: Genes for

the 'H' subunit of human ferritin are present on a number of human chromosomes. *Hum.Genet.* 71: 108–112, 1985.

[43960] 17512.Eisenstein, R. S.: Iron regulatory proteins and the molecular control of mammalian iron metabolism. *Annu. Rev. Nutr.* 20: 627–662, 2000.

[43961] 17513.Ferreira, C.; Bucchini, D.; Martin, M.–E.; Levi, S.; Arosio, P.; Grandchamp, B.; Beaumont, C.: Early embryonic lethality of H ferritin gene deletion in mice. *J. Biol. Chem.* 275: 3021–3024, 2000.

[43962] 17514.Ferreira, C.; Santambrogio, P.; Martin, M.–E.; Andrieu, V.; Feldmann, G.; Henin, D.; Beaumont, C.: H ferritin knockout mice: a model of hyperferritinemia in the absence of iron overload. *Blood* 98: 525–532, 2001.

[43963] 17515.Gailani, M. R.; Petty, E. M.; Horsthemke, B.; Arnold, A.; Marx, S. J.; Bale, A. E.: Physical mapping of chromosome 11q12–13 by pulsed field gel electrophoresis (PFGE). (Abstract) *Cytogenet. Cell Genet.* 58:1959 only, 1991.

[43964] 17516.Gatti, R. A.; Shaked, R.; Mohandas, T. K.; Salser, W.: Human ferritin genes: chromosomal assignments and polymorphisms. *Am. J. Hum. Genet.* 41: 654–667, 1987.

[43965] 17517.Harrison, P. M.; Arosio, P.: The ferritins: molecular properties, iron storage function and cellular regulation. *Biochim. Biophys. Acta* 1275: 161–203, 1996.

- [43966] 17518.Hentze, M. W.; Keim, S.; Papadopoulos, P.; O'Brien, S.; Modi,W.; Drysdale, J.; Leonard, W. J.; Harford, J. B.; Klausner, R. D.: Cloning, characterization, expression, and chromosomal localization of a human ferritin heavy-chain gene. *Proc. Nat. Acad. Sci.* 83:7226–7230, 1986.
- [43967] 17519.Kato, J.; Fujikawa, K.; Kanda, M.; Fukuda, N.; Sasaki, K.; Takayama,T.; Kobune, M.; Takada, K.; Takimoto, R.; Hamada, H.; Ikeda, T.; Niitsu,Y.: A mutation, in the iron-responsive element of H ferritin mRNA,causing autosomal dominant iron overload. *Am. J. Hum. Genet.* 69:191–197, 2001.
- [43968] 17520.Leibold, E. A.; Munro, H. N.: Cytoplasmic protein binds in vitro to a highly conserved sequence in the 5-prime untranslated region of ferritin heavy- and light-subunit mRNAs. *Proc. Nat. Acad. Sci.* 85:2171–2175, 1988.
- [43969] 17521.Anderson, L. A.; Friedman, L.; Osborne-Lawrence, S.; Lynch, E.;Weissenbach, J.; Bowcock, A.; King, M.-C.: High-density genetic map of the BRCA1 region of chromosome 17q12–q21. *Genomics* 17: 618–623,1993.
- [43970] 17522.Fryns, J. P.; Kleczkowska, A.; Moerman, P.; Vandenberghe, K.:Hereditary hydronephrosis and the short arm of chromosome 6. (Letter) *Hum.Genet.* 91: 514–515,

1993.

- [43971] 17523. Groenen, P. M. A.; Garcia, E.; Debeer, P.; Devriendt, K.; Fryns, J. P.; Van de Ven, W. J. M.: Structure, sequence, and chromosome 19 localization of human USF2 and its rearrangement in a patient with multicystic renal dysplasia. *Genomics* 38: 141–148, 1996.
- [43972] 17524. Groenen, P. M. A.; Vanderlinden, G.; Devriendt, K.; Fryns, J.-P.; Van de Ven, W. J. M.: Rearrangement of the human CDC5L gene by at(6;19)(p21;q13.1) in a patient with multicystic renal dysplasia. *Genomics* 49: 218–229, 1998.
- [43973] 17525. Galizzi, J.-P.; Zuber, C. E.; Harada, N.; Gorman, D. M.; Kastelein, R.; Banchereau, J.; Howard, M.; Miyajima, A.: Molecular cloning of a cDNA encoding the human interleukin 4 receptor. *Int. Immun.* 2: 669–675, 1990.
- [43974] 17526. Grimbacher, B.; Holland, S. M.; Puck, J. M.: The interleukin-4 receptor variant Q576R in hyper-IgE syndrome. (Letter) *New Eng. J. Med.* 338: 1073–1074, 1998.
- [43975] 17527. Idzerda, R. L.; March, C. J.; Mosley, B.; Lyman, S. D.; VandenBos, T.; Gimpel, S. D.; Din, W. S.; Grabstein, K. H.; Widmer, M. B.; Park, L. S.; Cosman, D.; Beckmann, M. P.: Human interleukin-4 receptor confers biological responsiveness and defines a novel receptor superfamily. *J. Exp. Med.* 171: 861–873, 1990.

- [43976] 17528.Khurana Hershey, G. K.; Friedrich, M. F.; Esswein, L. A.; Thomas,M. L.; Chatila, T. A.: The association of atopy with a gain-of-functionmutation in the alpha subunit of the interleukin-4 receptor. *NewEng. J. Med.* 337: 1720-1725, 1997.
- [43977] 17529.Kruse, S.; Japha, T.; Tedner, M.; Sparholt, S. H.; Forster, J.;Kuehr, J.; Deichmann, K. A.; Abello, A.: The polymorphisms S503P and Q576R in the interleukin-4 receptor alpha gene are associatedwith atopy and influence the signal transduction. *Immunology* 96:365-371, 1999.
- [43978] 17530.Mitsuyasu, H.; Izuhara, K.; Mao, X.-Q.; Gao, P.-S.; Arinobu, Y.;Enomoto, T.; Kawai, M.; Sasaki, S.; Dake, Y.; Hamasaki, N.; Shirakawa,T.; Hopkin, J. M.: Ile50val variant of IL4R-alpha upregulates IgEsynthesis and associates with atopic asthma. (Letter) *Nature Genet.* 19:119-120, 1998.
- [43979] 17531.Olson, M.; Hood, L.; Cantor, C.; Botstein, D.: A common languagefor physical mapping of the human genome. *Science* 245: 1434-1435,1989.
- [43980] 17532.Sutherland, G. R.; Baker, E.; Fernandez, K. E. W.; Callen, D. F.;Goodwin, R. G.; Lupton, S.; Namen, A. E.; Shannon, M. F.; Vadas, M.A.: The gene for human interleukin 7 (IL7) is at 8q12-13. *Hum. Genet.* 82:371-372,

1989.

- [43981] 17533.Watanabe, M.; Ueno, Y.; Yajima, T.; Iwao, Y.; Tsuchiya, M.; Ishikawa,H.; Aiso, S.; Hibi, T.; Ishii, H.: Interleukin 7 is produced by humanintestinal epithelial cells and regulates the proliferation of intestinalmucosal lymphocytes. J. Clin. Invest. 95: 2945–2953, 1995.
- [43982] 17534.Corcoran, A. E.; Riddell, A.; Krooshoop, D.; Venkittaraman, A. R.: Impaired immunoglobulin gene rearrangement in mice lacking the IL–7receptor. Nature 391: 904–907, 1998.
- [43983] 17535.Ober, C.; Leavitt, S. A.; Tsalenko, A.; Howard, T. D.; Hoki, D.M.; Daniel, R.; Newman, D. L.; Wu, X.; Parry, R.; Lester, L. A.; Solway,J.; Blumenthal, M.; King, R. A.; Xu, J.; Meyers, D. A.; Bleecker,E. R.; Cox, N. J.: Variation in the interleukin 4–receptor alphagene confers susceptibility to asthma and atopy in ethnically diversepopulations. Am. J. Hum. Genet. 66: 517–526, 2000.
- [43984] 17536.Ober, C.; Tsalenko, A.; Willadsen, S.; Newman, D.; Daniel, R.;Wu, X.; Andal, J.; Hoki, D.; Schneider, D.; True, K.; Schou, C.; Parry,R.; Cox, N.: Genome–wide screen for atopy susceptibility allelesin the Hutterites. Clin. Exp. Allergy 29 (suppl. 4): 11–15, 1999.
- [43985] 17537.Patuzzo, C.; Trabetti, E.; Malerba, G.; Martinati, L.

C.; Boner, A. L.; Pescolliderung, L.; Zanoni, G.; Pignatti, P. F.: No linkage or association of the IL-4R-alpha gene Q576R mutation with atopic asthma in Italian families. (Letter) J. Med. Genet. 37: 382-384, 2000.

[43986] 17538. Pritchard, M. A.; Baker, E.; Whitmore, S. A.; Sutherland, G. R.; Idzerda, R. L.; Park, L. S.; Cosman, D.; Jenkins, N. A.; Gilbert, D. J.; Copeland, N. G.; Beckmann, M. P.: The interleukin-4 receptor gene (IL4R) maps to 16p11.2-16p12.1 in human and to the distal region of mouse chromosome 7. Genomics 10: 801-806, 1991.

[43987] 17539. Suzuki, H.; Chung, F.; Palmer, E.; Sasaki, T.; Ohara, N.; Taylor, B. A.; Ohara, J.-I.: Gene mapping of mouse IL-4 receptor: the locus of interleukin 4 (IL-4) receptor gene and lymphocyte function associated antigen 1 (LFA-1) gene are closely linked on chromosome 7. Immunogenetics 34: 252-256, 1991.

[43988] 17540. Zurawski, S. M.; Vega, F., Jr.; Huyghe, B.; Zurawski, G.: Receptors for interleukin-13 and interleukin-4 are complex and share a novel component that functions in signal transduction. EMBO J. 12: 2663-2670, 1993.

[43989] 17541. Koshland, M. E.: The coming of age of the immunoglobulin J chain. Annu. Rev. Immun. 3: 425-453, 1985.

- [43990] 17542. Max, E. E.; McBride, O. W.; Morton, C. C.; Robinson, M. A.: Human β chain gene: chromosomal localization and associated restriction fragment length polymorphisms. *Proc. Nat. Acad. Sci.* 83: 5592–5596, 1986.
- [43991] 17543. Jones, C.; Penny, L.; Mattina, T.; Yu, S.; Baker, E.; Voullaire, L.; Langdon, W. Y.; Sutherland, G. R.; Richards, R. I.; Tunnacliffe, A.: Association of a chromosome deletion syndrome with a fragile site within the proto-oncogene CBL2. *Nature* 376: 145–149, 1995.
- [43992] 17544. Brunton, L. L.; Lupton, S. D.: An STS in the human IL7 gene located at 18q12–13. *Nucleic Acids Res.* 18: 1315 only, 1990.
- [43993] 17545. Carvalho, T. L.; Mota-Santos, T.; Cumano, A.; Demengeot, J.; Vieira, P.: Arrested B lymphopoiesis and persistence of activated B cells in adult interleukin 7 $^{-/-}$ mice. *J. Exp. Med.* 194: 1141–1150, 2001.
- [43994] 17546. Goodwin, R. G.; Lupton, S.; Schmierer, A.; Hjerrild, K. J.; Jerzy, R.; Clevenger, W.; Gillis, S.; Cosman, D.; Namen, A. E.: Human interleukin 7: molecular cloning and growth factor activity on human and murine B-lineage cells. *Proc. Nat. Acad. Sci.* 86: 302–306, 1989.
- [43995] 17547. Namen, A. E.; Lupton, S.; Hjerrild, K.; Wignall, J.; Mochizuki, D. Y.; Schmierer, A.; Mosley, B.; March, C. J.;

Urdal, D.; Gillis, S.; Cosman, D.; Goodwin, R. G.: Stimulation of B-cell progenitors by cloned murine interleukin-7. *Nature* 333: 571-573, 1988.

[43996] 17548. Namen, A. E.; Schmierer, A. E.; March, C. J.; Overell, R. W.; Park, L. S.; Urdal, D. L.; Mochizuki, D. Y.: B-cell precursor growth-promoting activity: purification and characterization of a growth factor active on lymphocyte precursors. *J. Exp. Med.* 167: 988-1002, 1988.

[43997] 17549. Goodwin, R. G.; Friend, D.; Ziegler, S. F.; Jerzy, R.; Falk, B. A.; Gimpel, S.; Cosman, D.; Dower, S. K.; March, C. J.; Namen, A. E.; Park, L. S.: Cloning of the human and murine interleukin-7 receptors: demonstration of a soluble form and homology to a new receptor superfamily. *Cell* 60: 941-951, 1990.

[43998] 17550. Kondo, M.; Takeshita, T.; Higuchi, M.; Nakamura, M.; Sudo, T.; Nishikawa, S.-I.; Sugamura, K.: Functional participation of the IL-2 receptor gamma chain in IL-7 receptor complexes. *Science* 263: 1453-1454, 1994.

[43999] 17551. Lai, S. Y.; Molden, J.; Goldsmith, M. A.: Shared gamma(c) subunit within the human interleukin-7 receptor complex: a molecular basis for the pathogenesis of X-linked severe combined immunodeficiency. *J. Clin. Invest.* 99: 169-177, 1997.

- [44000] 17552.Lynch, M.; Baker, E.; Park, L. S.; Sutherland, G. R.; Goodwin,R. G.: The interleukin-7 receptor gene is at 5p13. Hum. Genet. 89:566-568, 1992.
- [44001] 17553.Maki, K.; Sunaga, S.; Komagata, Y.; Kodaira, Y.; Mabuchi, A.; Karasuyama,H.; Yokomuro, K.; Miyazaki, J.; Ikuta, K.: Interleukin 7 receptor-deficientmice lack gamma/delta T cells. Proc. Nat. Acad. Sci. 93: 7172-7177,1996.
- [44002] 17554.Noguchi, M.; Nakamura, Y.; Russell, S. M.; Ziegler, S. F.; Tsang,M.; Cao, X.; Leonard, W. J.: Interleukin-2 receptor gamma chain:a functional component of the interleukin-7 receptor. Science 262:1877-1880, 1993.
- [44003] 17555.Peschon, J. J.; Morrissey, P. J.; Grabstein, K. H. et al.: Earlylymphocyte expansion is severely impaired in interleukin 7 receptor-deficientmice. J. Exp. Med. 180: 1955-1960, 1994.
- [44004] 17556.Puel, A.; Ziegler, S. F.; Buckley, R. H.; Leonard, W. J.: DefectiveIL7R expression in T-B+NK+ severe combined immunodeficiency. NatureGenet. 20: 394-397, 1998.
- [44005] 17557.Mezei, M. M.; Mankodi, A.; Brais, B.; Marineau, C.; Thornton, C.A.; Rouleau, G. A.; Karpati, G.: Minimal expansion of the GCG repeatin the PABP2 gene does not predispose to sporadic inclusion body myositis. Neurology

52:669–670, 1999.

- [44006] 17558.Aleman, A.; Verhaar, H. J. J.; De Haan, E. H. F.; De Vries, W.R.; Samson, M. M.; Drent, M. L.; Van Der Veen, E. A.; Koppeschaar, H. P. F.: Insulin-like growth factor-I and cognitive function in healthy older men. *J. Clin. Endocr. Metab.* 84: 471–475, 1999.
- [44007] 17559.Baker, J.; Liu, J.-P.; Robertson, E. J.; Efstratiadis, A.: Role of insulin-like growth factors in embryonic and post-natal growth. *Cell* 75:73–82, 1993.
- [44008] 17560.Bowcock, A.; Sartorelli, V.: Polymorphism and mapping of the IGF1 gene, and absence of association with stature among African Pygmies. *Hum.Genet.* 85: 349–354, 1990.
- [44009] 17561.Camacho-Hubner, C.; Woods, K. A.; Miraki-Moud, F.; Hindmarsh, P.C.; Clark, A. J.; Hansson, Y.; Johnston, A.; Baxter, R. C.; Savage, M. O.: Effects of recombinant human insulin-like growth factor I (IGF-I) therapy on the growth hormone-IGF system of a patient with a partial IGF-I gene deletion. *J. Clin. Endocr. Metab.* 84: 1611–1616, 1999.
- [44010] 17562.Goddard, A. D.; Covello, R.; Luoh, S.-M.; Clackson, T.; Attie, K. M.; Gesundheit, N.; Rundle, A. C.; Wells, J. A.; Carlsson, L. M.S.: Mutations of the growth hormone receptor in children with idiopathic short stature. *New Eng. J.*

Med. 333: 1093–1098, 1995.

- [44011] 17563.Guler, H.–P.; Binz, K.; Eigenmann, E.; Jaggi, S.; Zimmermann, D.; Zapf, J.; Froesch, E. R.: Small stature and insulin–like growth factors: prolonged treatment of minipoodles with recombinant human insulin–like growth factor I. *Acta Endocr.* 121: 456–464, 1989.
- [44012] 17564.Fajans, S. S.: Maturity–onset diabetes of the young (MODY). *Diabetes Metab. Rev.* 5: 579–606, 1989.
- [44013] 17565.Grosz, M. D.; Womack, J. E.; Skow, L. C.: Syntenic conservation of HSP70 genes in cattle and humans. *Genomics* 14: 863–868, 1992.
- [44014] 17566.Harrison, G. S.; Drabkin, H. A.; Kao, F.–T.; Hartz, J.; Hart, I.M.; Chu, E. H. Y.; Wu, B. J.; Morimoto, R. I.: Chromosomal location of human genes encoding major heat–shock protein HSP70. *Somat. Cell Molec. Genet.* 13: 119–130, 1987.
- [44015] 17567.Harrison, G. S.; Morimoto, R.; Kao, F.–T.; Chu, E. H. Y.; Wu, B.J.; Drabkin, H.: Chromosomal location of human genes encoding the major heat shock protein HSP70. (Abstract) *Am. J. Hum. Genet.* 39:A157 only, 1986.
- [44016] 17568.Hickey, E.; Brandon, S. E.; Sadis, S.; Smale, G.; Weber, L. A.: Molecular cloning of sequences encoding the human heat–shock proteins and their expression during

hyperthermia. *Gene* 43: 147–154, 1986.

- [44017] 17569. Imai, Y.; Soda, M.; Hatakeyama, S.; Akagi, T.; Hashikawa, T.; Nakayama, K.; Takahashi, R.: CHIP is associated with Parkin, a gene responsible for familial Parkinson's disease, and enhances its ubiquitin ligase activity. *Molec. Cell* 10: 55–67, 2002.
- [44018] 17570. Laroia, G.; Cuesta, R.; Brewer, G.; Schneider, R. J.: Control of mRNA decay by heat shock–ubiquitin–proteasome pathway. *Science* 284:499–502, 1999.
- [44019] 17571. Leung, T. K. C.; Hall, C.; Rajendran, M.; Spurr, N. K.; Lim, L.: The human heat–shock genes HSPA6 and HSPA7 are both expressed and localize to chromosome 1. *Genomics* 12: 74–79, 1992.
- [44020] 17572. Milner, C. M.; Campbell, R. D.: Polymorphic analysis of the three MHC–linked HSP70 genes. *Immunogenetics* 36: 357–362, 1992.
- [44021] 17573. Milner, C. M.; Campbell, R. D.: Structure and expression of the three MHC–linked HSP70 genes. *Immunogenetics* 32: 242–251, 1990.
- [44022] 17574. Pelham, H. R. B.: Speculations on the functions of the major heat shock and glucose–regulated proteins. *Cell* 46: 959–961, 1986.
- [44023] 17575. Sargent, C. A.; Dunham, I.; Trowsdale, J.; Campbell,

R. D.: Human major histocompatibility complex contains genes for the major heatshock protein HSP70. *Proc. Nat. Acad. Sci.* 86: 1968–1972, 1989.

[44024] 17576. Slater, A.; Cato, A. C. B.; Sillar, G. M.; Kioussis, J.; Burdon, R. H.: The pattern of protein synthesis induced by heat shock of HeLa cells. *Europ. J. Biochem.* 117: 341–346, 1981.

[44025] 17577. Leung, T. K. C.; Rajendran, M. Y.; Monfries, C.; Hall, C.; Lim, L.: The human heat-shock protein family: expression of a novel heat-inducible HSP70 (HSP70B-prime) and isolation of its cDNA and genomic DNA. *Biochem. J.* 267: 125–132, 1990.

[44026] 17578. Schiller, P.; Amin, J.; Ananthan, J.; Brown, M. E.; Scott, W. A.; Voellmy, R.: Cis-acting elements involved in the regulated expression of a human HSP70 gene. *J. Molec. Biol.* 203: 97–105, 1988.

[44027] 17579. Voellmy, R.; Ahmed, A.; Schiller, P.; Bromley, P.; Rungger, D.: Isolation and functional analysis of a human 70,000-dalton heatshock protein gene segment. *Proc. Nat. Acad. Sci.* 82: 4949–4953, 1985.

[44028] 17580. Rebbe, N. F.; Hickman, W. S.; Ley, T. J.; Stafford, D. W.; Hickman, S.: Nucleotide sequence and regulation of a human 90-kDa heat shock protein gene. *J. Biol. Chem.*

264: 15006–15011, 1989.

- [44029] 17581.Wolpowitz, D.; Mason, T. B. A.; Dietrich, P.; Mendelsohn, M.;Talmage, D. A.; Role, L. W.: Cysteine-rich domain isoforms of the neuregulin-1 gene are required for maintenance of peripheral synapses. *Neuron* 25:79–91, 2000.
- [44030] 17582.Bolton, P.; Powell, J.; Rutter, M.; Buckle, V.; Yates, J. R. W.;Ishikawa-Brush, Y.; Monaco, A. P.: Autism, mental retardation, multiple exostoses and short stature in a female with 46,X,t(X;8)(p22.13;q22.1). *Psychiat.Genet.* 5: 51–55, 1995.
- [44031] 17583.David, G.; van der Schueren, B.; Marynen, P.; Cassiman, J.-J.;van den Berghe, H.: Molecular cloning of amphiglycan, a novel integral membrane heparan sulfate proteoglycan expressed by epithelial and fibroblastic cells. *J. Cell Biol.* 118: 961–969, 1992.
- [44032] 17584.Ishikawa-Brush, Y.; Powell, J. F.; Bolton, P.; Miller, A. P.; Francis, F.; Willard, H. F.; Lehrach, H.; Monaco, A. P.: Autism and multiple exostoses associated with an X;8 translocation occurring within the GRPR gene and 3-prime to the SDC2 gene. *Hum. Molec. Genet.* 6: 1241–1250, 1997.
- [44033] 17585.Spring, J.; Goldberger, O. A.; Jenkins, N. A.; Gilbert,

D. J.; Copeland, N. G.; Bernfield, M.: Mapping of the synde-
can genes in the mouse: linkage with members of the Myc
gene family. *Genomics* 21:597–601, 1994.

[44034] 17586. Grenningloh, G.; Pribilla, I.; Prior, P.; Multhaup, G.;
Beyreuther, K.; Taleb, O.; Betz, H.: Cloning and expression
of the 58 kd beta subunit of the inhibitory glycine recep-
tor. *Neuron* 4: 963–970, 1990.

[44035] 17587. Handford, C. A.; Lynch, J. W.; Baker, E.; Webb, G.
C.; Ford, J. H.; Sutherland, G. R.; Schofield, P. R.: The hu-
man glycine receptor beta subunit: primary structure,
functional characterisation and chromosomal localisation
of the human and murine genes. *Molec. Brain Res.*
35:211–219, 1996.

[44036] 17588. Milani, N.; Mulhardt, C.; Weber, R. G.; Lichter, P.;
Kioschis, P.; Poustka, A.; Becker, C.-M.: The human glycine
receptor beta subunit gene (GLRB): structure, refined chro-
mosomal localization, and population polymorphism. *Ge-
nomics* 50: 341–345, 1998.

[44037] 17589. Mulhardt, C.; Fischer, M.; Gass, P.; Simon-Cha-
zottes, D.; Guenet, J.-L.; Kuhse, J.; Betz, H.; Becker, C.-M.:
The spastic mouse: aberrant splicing of glycine receptor
beta subunit mRNA caused by intronic insertion of L1 ele-
ment. *Neuron* 13: 1003–1015, 1994.

- [44038] 17590. Browner, M. F.; Nakano, K.; Bang, A. G.; Fletterick, R. J.: Human muscle glycogen synthase cDNA sequence: a negatively charged protein with an asymmetric charge distribution. *Proc. Nat. Acad. Sci.* 86:1443–1447, 1989.
- [44039] 17591. Groop, L. C.; Kankuri, M.; Schalin-Jantti, C.; Ekstrand, A.; Nikula-Ijas, P.; Widen, E.; Kuusmanen, E.; Eriksson, J.; Franssila-Kallunki, A.; Saloranta, C.; Koskimies, S.: Association between polymorphism of the glycogen synthase gene and non-insulin-dependent diabetes mellitus. *New Eng. J. Med.* 328: 10–14, 1993.
- [44040] 17592. Huang, X.; Vaag, A.; Hansson, M.; Weng, J.; Laurila, E.; Groop, L.: Impaired insulin-stimulated expression of the glycogen synthase gene in skeletal muscle of type 2 diabetic patients is acquired rather than inherited. *J. Clin. Endocr. Metab.* 85: 1584–1590, 2000.
- [44041] 17593. Kadowaki, T.; Kadowaki, H.; Yazaki, Y.: Polymorphism of the glycogen synthase gene and non-insulin-dependent diabetes mellitus. (Letter) *New Eng. J. Med.* 328: 1569, 1993.
- [44042] 17594. Lehto, M.; Stoffel, M.; Groop, L.; Espinosa, R., III; Le Beau, M. M.; Bell, G. I.: Assignment of the gene encoding glycogen synthase (GYS) to human chromosome 19, band q13.3. *Genomics* 15: 460–461, 1993.

- [44043] 17595.Seldin, M. F.; Mott, D.; Bhat, D.; Petro, A.; Kuhn, C. M.; Kingsmore,S. F.; Bogardus, C.; Opara, E.; Feinglos, M. N.; Surwit, R. S.: Glycogensynthase: a putative locus for diet-induced hyperglycemia. J. Clin.Invest. 94: 269–276, 1994.
- [44044] 17596.Zouali, H.; Velho, G.; Froguel, P.: Polymorphism of the glycogensynthase gene and non-insulin-dependent diabetes mellitus. (Letter) NewEng. J. Med. 328: 1568, 1993.
- [44045] 17597.David, J. R.: Delayed hypersensitivity in vitro: its mediationby cell-free substances formed by lymphoid cell-antigen interaction. Proc.Nat. Acad. Sci. 56: 72–77, 1966.
- [44046] 17598.Reche, P. A.; Soumelis, V.; Gorman, D. M.; Clifford, T.; Liu,M.; Travis, M.; Zurawski, S. M.; Johnston, J.; Liu, Y.-J.; Spits,H.; de Waal Malefyt, R.; Kastelein, R. A.; Bazan, J. F.: Human thymicstromal lymphopoietin preferentially stimulates myeloid cells. J.Immun. 167: 336–343, 2001.
- [44047] 17599.Kalmar, G. B.; Kay, R. J.; LaChance, A. C.; Cornell, R. B.: Primarystructure and expression of a human CTP:phosphocholine cytidyltransferase. Biochim.Biophys. Acta 1219: 328–334, 1994.
- [44048] 17600.Rutherford, M. S.; Rock, C. O.; Jenkins, N. A.;

Gilbert, D. J.; Tessner, T. G.; Copeland, N. G.; Jackowski, S.: The gene for murine CTP:phosphocholine cytidyltransferase (Ctpct) is located on mouse chromosome 16. *Genomics* 18: 698–701, 1993.

[44049] 17601. Tabas, J. A.; Zasloff, M.; Wasmuth, J. J.; Emanuel, B. S.; Altherr, M. R.; McPherson, J. D.; Wozney, J. M.; Kaplan, F. S.: Bone morphogenetic protein: chromosomal localization of human genes for BMP1, BMP2A, and BMP3. *Genomics* 9: 283–289, 1991.

[44050] 17602. Gopal Rao, V. V. N.; Loffler, C.; Wozney, J. M.; Hansmann, I.: The gene for bone morphogenetic protein 2A (BMP2A) is localized to human chromosome 20p12 by radioactive and nonradioactive in situ hybridization. *Hum. Genet.* 90: 299–302, 1992.

[44051] 17603. Wang, E. A.; Rosen, V.; D'Alessandro, J. S.; Bauduy, M.; Cordes, P.; Harada, T.; Israel, D. I.; Hewick, R. M.; Kerns, K. M.; LaPan, P.; Luxenberg, D. P.; McQuaid, D.; Moutsatsos, I. K.; Nove, J.; Wozney, J. M.: Recombinant human bone morphogenetic protein induces bone formation. *Proc. Nat. Acad. Sci.* 87: 2220–2224, 1990.

[44052] 17604. White, P. M.; Morrison, S. J.; Orimoto, K.; Kubu, C. J.; Verdi, J. M.; Anderson, D. J.: Neural crest stem cells undergo cell-intrinsic developmental changes in sensitivity to

instructive differentiation signals. Neuron 29: 57–71, 2001.

- [44053] 17605.Wozney, J. M.; Rosen, V.; Celeste, A. J.; Mitsock, L. M.; Whitters, M. J.; Kriz, R. W.; Hewick, R. M.; Wang, E. A.: Novel regulators of bone formation: molecular clones and activities. Science 242:1528–1534, 1988.
- [44054] 17606.Connor, J. M.: Fibrodysplasia ossificans progressiva: lessons from rare maladies. (Editorial) New Eng. J. Med. 335: 591–593, 1996.
- [44055] 17607.Dooley, C. A.; Attia, G. R.; Rainey, W. E.; Moore, D. R.; Carr, B. R.: Bone morphogenetic protein inhibits ovarian androgen production. J.Clin. Endocr. Metab. 85: 3331–3337, 2000.
- [44056] 17608.McAlpine, P. J.: Personal Communication. Winnipeg, Manitoba, Canada 7/15/1992.
- [44057] 17609.Monsoro-Burq, A.–H.; le Douarin, N. M.: BMP4 plays a key role in left–right patterning in chick embryos by maintaining Sonic Hedgehog asymmetry. Molec. Cell 7: 789–799, 2001.
- [44058] 17610.Shafritz, A. B.; Shore, E. M.; Gannon, F. H.; Zasloff, M. A.; Taub, R.; Muenke, M.; Kaplan, F. S.: Overexpression of an osteogenic morphogen in fibrodysplasia ossificans progressiva. New Eng. J. Med. 335:555–561, 1996.

- [44059] 17611.Tabas, J. A.; Hahn, G. V.; Cohen, R. B.; Seaunez, H. N.; Modi,W. S.; Wozney, J. M.; Zasloff, M.; Kaplan, F. S.: Chromosomal assignmentof the human gene for bone morphogenetic protein 4. Clin. Orthop.Rel. Res. 293: 310–316, 1993.
- [44060] 17612.Tucker, A. S.; Matthews, K. L.; Sharpe, P. T.: Trans–formationof tooth type induced by inhibition of BMP signaling. Science 282:1136–1138, 1998.
- [44061] 17613.van den Wijngaard, A.; Olde Weghuis, D.; Boersma, C. J. C.; vanZoelen, E. J. J.; Geurts van Kessel, A.; Olijve, W.: Fine mappingof the human bone morphogenetic protein–4 gene (BMP4) to chromosome14q22–q23 by in situ hybridization. Genomics 27: 559–560, 1995.
- [44062] 17614.van den Wijngaard, A.; van Kraay, M.; van Zoelen, E. J. J.; Olijve,W.; Boersma, C. J. C.: Genomic organization of the human bone morphogeneticprotein–4 gene: molecular basis for multiple transcripts. Biochem.Biophys. Res. Commun. 219: 789–794, 1996.
- [44063] 17615.Amano, S.; Scott, I. C.; Takahara, K.; Koch, M.; Champlaud, M.–F.;Gerecke, D. R.; Keene, D. R.; Hudson, D. L.; Nishiyama, T.; Lee, S.;Greenspan, D. S.; Burgeson, R. E.: Bone morphogenetic protein 1 isan extracellular processing enzyme of the laminin 5 gamma–2 chain. J.Biol.

Chem. 275: 22728–22735, 2000.

- [44064] 17616.Bond, J. S.; Beynon, R. J.: The astacin family of metalloendopeptidases. *ProteinSci.* 4: 1247–1261, 1995.
- [44065] 17617.Ceci, J. D.; Kingsley, D. M.; Silan, C. M.; Copeland, N. G.; Jenkins, N. A.: An interspecific backcross linkage map of the proximal half of mouse chromosome 14. *Genomics* 6: 673–678, 1990.
- [44066] 17618.Kessler, E.; Takahara, K.; Biniaminov, L.; Brusel, M.; Greenspan, D.: Bone morphogenic protein–1: the type I procollagen C–proteinase. *Science* 271:360–362, 1996.
- [44067] 17619.Donalies, M.; Cramer, M.; Ringwald, M.; Starzinski-Powitz, A.: Expression of M–cadherin, a member of the cadherin multigene family, correlates with differentiation of skeletal muscle cells. *Proc. Nat. Acad. Sci.* 88: 8024–8028, 1991.
- [44068] 17620.Kaupmann, K.; Becker–Follmann, J.; Scherer, G.; Jockusch, H.; Starzinski–Powitz, A.: The gene for the cell adhesion molecule M–cadherin maps to mouse chromosome 8 and human chromosome 16q24.1–qter and is near the E–cadherin (uvomorulin) locus in both species. *Genomics* 14: 488–490, 1992.
- [44069] 17621.Kremmidiotis, G.; Baker, E.; Crawford, J.; Eyre, H. J.; Nahmias, J.; Callen, D. F.: Localization of human cadherin

genes to chromosomeregions exhibiting cancer-related loss of heterozygosity. *Genomics* 49:467–471, 1998.

[44070] 17622.Garcia–Castro, M. I.; Vielmetter, E.; Bronner–Fraser, M.: N–cadherin,a cell adhesion molecule involved in establishment of embryonic left–rightasymmetry. *Science* 288: 1047–1051, 2000.

[44071] 17623.Hermiston, M. L.; Gordon, J. I.: Inflammatory bowel disease andadenomas in mice expressing a dominant negative N–cadherin. *Science* 270:1203–1206, 1995.

[44072] 17624.Miyatani, S.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.;Takeichi, M.: Genomic structure and chromosomal mapping of the mouseN–cadherin gene. *Proc. Nat. Acad. Sci.* 89: 8443–8447, 1992.

[44073] 17625.Takeichi, M.: Cadherins: a molecular family essential for selectivecell–cell adhesion and animal morphogenesis. *Trends Genet.* 3: 213–217,1987.

[44074] 17626.Tanaka, H.; Shan, W.; Phillips, G. R.; Arndt, K.; Bozdagi, O.;Shapiro, L.; Huntley, G. W.; Benson, D. L.; Colman, D. R.: Molecularmodification of N–cadherin in response to synaptic activity. *Neuron* 25:93–107, 2000.

[44075] 17627.Wallis, J.; Fox, M. F.; Walsh, F. S.: Structure of the human N–cadheringene: YAC analysis and fine chromosomal mapping to 18q11.2. *Genomics* 22:172–179, 1994.

- [44076] 17628. Walsh, F. S.; Barton, C. H.; Putt, W.; Moore, S. E.; Kelsell, D.; Spurr, N.; Goodfellow, P. N.: N-cadherin gene maps to human chromosome 18 and is not linked to the E-cadherin gene. *J. Neurochem.* 55: 805–812, 1990.
- [44077] 17629. Claverie, J.-M.; Hardelin, J.-P.; Legouis, R.; Leveillé, J.; Bougueleret, L.; Mattei, M.-G.; Petit, C.: Characterization and chromosomal assignment of a human cDNA encoding a protein related to the murine 102-kDa cadherin-associated protein (alpha-catenin). *Genomics* 15: 13–20, 1993.
- [44078] 17630. Cook, S. A.; Bronson, R. T.; Donahue, L. R.; Ben-Arie, N.; Davisson, M. T.: Cerebellar deficient folia (cdf): a new mutation on mouse chromosome 6. *Mammalian Genome* 8: 108–112, 1997.
- [44079] 17631. Herrenknecht, K.; Ozawa, M.; Eckerskorn, C.; Lottspeich, F.; Lenter, M.; Kemler, R.: The uvomorulin-anchorage protein alpha-catenin is a vinculin homologue. *Proc. Nat. Acad. Sci.* 88: 9156–9160, 1991.
- [44080] 17632. Park, C.; Falls, W.; Finger, J. H.; Longo-Guess, C. M.; Ackerman, S. L.: Deletion in *Catna2*, encoding alpha-N-catenin, causes cerebellar and hippocampal lamination defects and impaired startle modulation. *Nature Genet.* 31: 279–284, 2002.

- [44081] 17633. Magnuson, V. L.; McCombs, J. L.; Lee, C.-C.; Yang, F.; Bowman, B. H.; McGill, J. R.: Human alpha-2-HS-glycoprotein localized to 3q27-q29 by in situ hybridization. *Cytogenet. Cell Genet.* 47: 72-74, 1988.
- [44082] 17634. Seroussi, E.; Kedra, D.; Kost-Alimova, M.; Sandberg-Nordqvist, A.-C.; Fransson, I.; Jacobs, J. F. M.; Fu, Y.; Pan, H.-Q.; Roe, B. A.; Imreh, S.; Dumanski, J. P.: TOM1 genes map to human chromosome 22q13.1 and mouse chromosome 8C1 and encode proteins similar to the endosomal proteins HGS and STAM. *Genomics* 57: 380-388, 1999.
- [44083] 17635. Hanson, I.; Churchill, A.; Love, J.; Axton, R.; Moore, T.; Clarke, M.; Meire, F.; van Heyningen, V.: Missense mutations in the most ancient residues of the PAX6 paired domain underlie a spectrum of human congenital eye malformations. *Hum. Molec. Genet.* 8: 165-172, 1999.
- [44084] 17636. O'Farrell, P. H.: High resolution two-dimensional electrophoresis of proteins. *J. Biol. Chem.* 250: 4007-4021, 1975.
- [44085] 17637. Osawa, M.; Umetsu, K.; Ohki, T.; Nagasawa, T.; Suzuki, T.; Takeichi, S.: Molecular evidence for human alpha2-HS glycoprotein (AHSG) polymorphism. *Hum. Genet.* 99: 18-21, 1997.

- [44086] 17638.Pedersen, K. O.: Fetuin, a new globulin isolated from serum.(Letter) *Nature* 154: 575, 1944.
- [44087] 17639.Rizzu, P.; Baldini, A.: Three members of the human cystatin genesuperfamily, AHSG, HRG, and KNG, map within one megabase of genomicDNA at 3q27. *Cytogenet. Cell Genet.* 70: 26–28, 1995.
- [44088] 17640.Terkelsen, O. B. F.; Jahnen–Dechent, W.; Nielsen, H.; Moos, T.;Fink, E.; Nawratil, P.; Muller–Esterl, W.; Mollgard, K.: Rat fetuin:distribution of protein and mRNA in embryonic and neonatal rat tissues. *Anat.Embryol.* 197: 125–133, 1998.
- [44089] 17641.Umetsu, K.; Kashimura, S.; Ikeda, N.; Suzuki, T.: A new alpha-2-HS-glycoproteintyping by isoelectric focusing. *Hum. Genet.* 67: 70–71, 1984.
- [44090] 17642.Umetsu, K.; Kashimura, S.; Ikeda, N.; Suzuki, T.: A new alpha-2-HS-glycoproteinallele (AHS*5*) in two Japanese families. *Hum. Genet.* 68: 264–265,1984.
- [44091] 17643.Umetsu, K.; Yuasa, I.; Ikuta, M.; Suzuki, T.: Alpha-2-HS-glycoprotein(A2HS) polymorphism in a Japanese population: existence of two newvariants. *Jpn. J. Hum. Genet.* 32: 263–266, 1987.
- [44092] 17644.Umetsu, K.; Yuasa, I.; Suzuki, T.: The polymorphism of desialyzedalpha-2-HS-glycoprotein (AHS): iso-

electric focusing in 2.5M urea as a method for identification of genetic variants. Hum. Genet. 73:372–373, 1986.

[44093] 17645. Yoshioka, Y.; Gejyo, F.; Marti, T.; Rickli, E. E.; Burgi, W.; Offner, G. D.; Troxler, R. F.; Schmid, K.: The complete amino acid sequence of the A-chain of human plasma alpha-2HS-glycoprotein. J. Biol. Chem. 261: 1665–1676, 1986.

[44094] 17646. Zelinski, T.; Kaita, H.; Lewis, M.; Coghlan, G.; Craig, D.: The sequence of chromosome 3 loci AHSB:TF:CHE1. Hum. Hered. 37: 1–6, 1987.

[44095] 17647. Elpeleg, O. N.; Shaag, A.; Glustein, J. Z.; Anikster, Y.; Joseph, A.; Saada, A.: Lipoamide dehydrogenase deficiency in Ashkenazi Jews: an insertion mutation in the mitochondrial leader sequence. Hum. Mutat. 10: 256–257, 1997.

[44096] 17648. Feigenbaum, A. S.; Robinson, B. H.: The structure of the human dihydrolipoamide dehydrogenase gene (DLD) and its upstream elements. Genomics 17:376–381, 1993.

[44097] 17649. Hong, Y. S.; Kerr, D. S.; Craigen, W. J.; Tan, J.; Pan, Y.; Lusk, M.; Patel, M. S.: Identification of two mutations in a compound heterozygous child with dihydrolipoamide dehydrogenase deficiency. Hum. Molec. Genet. 5: 1925–1930, 1996.

- [44098] 17650. Johnson, M.; Yang, H.-S.; Johanning, G. L.; Patel, M. S.: Characterization of the mouse dihydrolipoamide dehydrogenase (Dld) gene: genomic structure, promoter sequence, and chromosomal localization. *Genomics* 41: 320–326, 1997.
- [44099] 17651. Liu, T.-C.; Kim, H.; Arizmendi, C.; Kitano, A.; Patel, M. S.: Identification of two missense mutations in a dihydrolipoamide dehydrogenase-deficient patient. *Proc. Nat. Acad. Sci.* 90: 5186–5190, 1993.
- [44100] 17652. Matalon, R.; Michals, K.; Stumpf, D.; Goodman, S.; Parks, J.: Lactic acidosis due to lipoamide dehydrogenase (LAD) deficiency: improvement after oral lipoic acid. (Abstract) *Am. J. Hum. Genet.* 33: 48A, 1981.
- [44101] 17653. Munnich, A.; Saudubray, J. M.; Taylor, J.; Charpentier, C.; Marsac, C.; Rocchiccioli, F.; Amedee-Manesme, O.; Coude, F. X.; Frezal, J.; Robinson, B. H.: Congenital lactic acidosis, alpha-ketoglutaric aciduria and variant form of maple syrup urine disease due to a single enzyme defect: dihydrolipoyl dehydrogenase deficiency. *Acta Paediat. Scand.* 71: 167–171, 1982.
- [44102] 17654. Olson, S.; Song, B. J.; Huh, T.-L.; Chi, Y.-T.; Veech, R. L.; McBride, O. W.: Three genes for enzymes of the pyruvate dehydrogenase complex map to human chromo-

somes 3, 7, and X. Am. J. Hum. Genet. 46:340–349, 1990.

[44103] 17655.Otulakowski, G.; Robinson, B. H.: Isolation and sequence determination of cDNA clones for porcine and human lipoamide dehydrogenase. J.Biol. Chem. 262: 17313–17318, 1987.

[44104] 17656.Otulakowski, G.; Robinson, B. H.; Willard, H. F.: Gene for lipoamide dehydrogenase maps to human chromosome 7. Somat. Cell Molec. Genet. 14:411–414, 1988.

[44105] 17657.Pons, G.; Raefsky–Estrin, C.; Carothers, D. J.; Pepin, R. A.; Javed, A. A.; Jesse, B. W.; Ganapathi, M. K.; Samols, D.; Patel, M.S.: Cloning and cDNA sequence of the dihydrolipoamide dehydrogenase component of human alpha-ketoacid dehydrogenase complexes. Proc.Nat. Acad. Sci. 85: 1422–1426, 1988.

[44106] 17658.Robinson, B. H.; Taylor, J.; Kahler, S. G.; Kirkman, H. N.: Lacticacidemia, neurologic deterioration and carbohydrate dependence in a girl with dihydrolipoyl dehydrogenase deficiency. Europ. J. Pediat. 136:35–39, 1981.

[44107] 17659.Robinson, B. H.; Taylor, J.; Sherwood, W. G.: Deficiency of dihydrolipoyl dehydrogenase (a component of the pyruvate and alpha-ketoglutarate dehydrogenase complexes): a cause of congenital lactic acidosis in infancy. Pediat. Res. 11: 1198–1202, 1977.

- [44108] 17660.Sakaguchi, Y.; Yoshino, M.; Aramaki, S.; Yoshida, I.; Yamashita,F.; Kuhara, T.; Matsumoto, I.; Hayashi, T.: Dihydrolipoyl dehydrogenasedeficiency: a therapeutic trial with branched-chain amino acid restriction. *Europ.J. Pediat.* 145: 271-274, 1986.
- [44109] 17661.Scherer, S. W.; Otulakowski, G.; Robinson, B. H.; Tsui, L.-C.: Localization of the human dihydrolipoamide dehydrogenase gene (DLD)to 7q31-q32. *Cytogenet. Cell Genet.* 56: 176-177, 1991.
- [44110] 17662.Shaag, A.; Saada, A.; Berger, I.; Mandel, H.; Joseph, A.; Feigenbaum,A.; Elpeleg, O. N.: Molecular basis of lipoamide dehydrogenase deficiencyin Ashkenazi Jews. *Am. J. Med. Genet.* 82: 177-182, 1999.
- [44111] 17663.Martin-Subero, J. I.; Gesk, S.; Harder, L.; Sonoki, T.; Tucker,P. W.; Schlegelberger, B.; Grote, W.; Novo, F. J.; Calasanz, M. J.;Hansmann, M. L.; Dyer, M. J. S.; Siebert, R.: Recurrent involvementof the REL and BCL11A loci in classical Hodgkin lymphoma. *Blood* 99:1474-1477, 2002.
- [44112] 17664.Hankinson, S. E.; Willett, W. C.; Colditz, G. A.; Hunter, D. J.;Michaud, D. S.; Deroo, B.; Rosner, B.; Speizer, F. E.; Pollak, M.: Circulating concentrations of insulin-like growth factor-I and riskof breast cancer. *Lancet* 351: 1393-1396, 1998.

- [44113] 17665. Jones, C.; Slijepcevic, P.; Marsh, S.; Baker, E.; Langdon, W. Y.; Richards, R. I.; Tunnacliffe, A.: Physical linkage of the fragile site FRA11B and a Jacobsen syndrome chromosome deletion breakpoint in 11q23.3. *Hum. Molec. Genet.* 3: 2123–2130, 1994.
- [44114] 17666. Esumi, N.; Budarf, M.; Ciccarelli, L.; Sellinger, B.; Kozak, C.A.; Wistow, G.: Conserved gene structure and genomic linkage for D-dopachrome tautomerase (DDT) and MIF. *Mammalian Genome* 9: 753–757, 1998.
- [44115] 17667. Kleemann, R.; Hausser, A.; Geiger, G.; Mischke, R.; Burger-Kentischer, A.; Flieger, O.; Johannes, F.-J.; Roger, T.; Calandra, T.; Kapurniotu, A.; Grell, M.; Finkelmeier, D.; Brunner, H.; Bernhagen, J.: Intracellular action of the cytokine MIF to modulate AP-1 activity and the cell cycle through Jab1. *Nature* 408: 211–216, 2000.
- [44116] 17668. Kozak, C. A.; Adamson, M. C.; Buckler, C. E.; Segovia, L.; Paralkar, V.; Wistow, G.: Genomic cloning of mouse MIF (macrophage inhibitory factor) and genetic mapping of the human and mouse expressed gene and nine mouse pseudogenes. *Genomics* 27: 405–411, 1995.
- [44117] 17669. Paralkar, V.; Wistow, G.: Cloning the human gene for macrophage migration inhibitory factor (MIF). *Genomics* 19: 48–51, 1994.

- [44118] 17670. Pastrana, D. V.; Raghavan, N.; Fitzgerald, P.; Eisinger, S. W.; Metz, C.; Bucala, R.; Schleimer, R. P.; Bickel, C.; Scott, A. L.: Filarial nematode parasites secrete a homologue of the human cytokine macrophage migration inhibitory factor. *Infect. Immun.* 66: 5955–5963, 1998.
- [44119] 17671. Roger, T.; David, J.; Glauser, M. P.; Calandra, T.: MIF regulates innate immune response through modulation of Toll-like receptor 4. *Nature* 414: 920–924, 2001.
- [44120] 17672. Weiser, W. Y.; Temple, P. A.; Witek-Giannotti, J. S.; Remold, H. G.; Clark, S. C.; David, J. R.: Molecular cloning of a cDNA encoding a human macrophage migration inhibitory factor. *Proc. Nat. Acad. Sci.* 86: 7522–7526, 1989.
- [44121] 17673. Emi, M.; Asaoka, H.; Matsumoto, A.; Itakura, H.; Kurihara, Y.; Wada, Y.; Kanamori, H.; Yazaki, Y.; Takahashi, E.; Lepert, M.; Lalouel, J.-M.; Kodama, T.; Mukai, T.: Structure, organization, and chromosomal mapping of the human macrophage scavenger receptor gene. *J. Biol. Chem.* 268: 2120–2125, 1993.
- [44122] 17674. Latil, A.; Lidereau, R.: Genetic aspects of prostate cancer. *Virchows Arch.* 432: 389–406, 1998.
- [44123] 17675. Matsumoto, A.; Naito, M.; Itakura, H.; Ikemoto, S.; Asaoka, H.; Hayakawa, I.; Kanamori, H.; Aburatani, H.; Takaku, F.; Suzuki, H.; Kobari, Y.; Miyai, T.; Takahashi, K.;

Cohen, E. H.; Wydro, R.; Housman, D. E.; Kodama, T.: Human macrophage scavenger receptors: primary structure, expression, and localization in atherosclerotic lesions. *Proc. Nat. Acad. Sci.* 87: 9133–9137, 1990.

- [44124] 17676. Xu, J.; Zheng, S. L.; Hawkins, G. A.; Faith, D. A.; Kelly, B.; Isaacs, S. D.; Wiley, K. E.; Chang, B.; Ewing, C. M.; Bujnovszky, P.; Carpten, J. D.; Bleecker, E. R.; Walsh, P. C.; Trent, J. M.; Meyers, D. A.; Isaacs, W. B.: Linkage and association studies of prostate cancer susceptibility: evidence for linkage at 8p22–23. *Am. J. Hum. Genet.* 69: 341–350, 2001.
- [44125] 17677. Xu, J.; Zheng, S. L.; Komiya, A.; Mychaleckyj, J. C.; Isaacs, S. D.; Hu, J. J.; Sterling, D.; Lange, E. M.; Hawkins, G. A.; Turner, A.; Ewing, C. M.; Faith, D. A.; and 19 others: Germline mutations and sequence variants of the macrophage scavenger receptor 1 gene are associated with prostate cancer risk. *Nature Genet.* 32: 321–325, 2002.
- [44126] 17678. Holness, C. L.; Simmons, D. L.: Molecular cloning of CD68, a human macrophage marker related to lysosomal glycoproteins. *Blood* 81:1607–1613, 1993.
- [44127] 17679. Jones, E.; Quinn, C. M.; See, C. G.; Montgomery, D. S.; Ford, M. J.; Kolbe, K.; Gordon, S.; Greaves, D. R.: The linked human elongation initiation factor 4A1 (EIF4A1) and

CD68 genes map to chromosome 17p13. *Genomics* 53:248–250, 1998.

- [44128] 17680. Chavrier, P.; Janssen–Timmen, U.; Mattei, M.–G.; Zerial, M.; Bravo, R.; Charnay, P.: Structure, chromosome location, and expression of the mouse zinc finger gene Krox–20: multiple gene products and coregulation with the proto–oncogene c–fos. *Molec. Cell. Biol.* 9: 787–797, 1989.
- [44129] 17681. Le Beau, M. M.; Espinosa, R., III; Neuman, W. L.; Stock, W.; Roulston, D.; Larson, R. A.; Keinänen, M.; Westbrook, C. A.: Cytogenetic and molecular delineation of the smallest commonly deleted region of chromosome 5 in malignant myeloid diseases. *Proc. Nat. Acad. Sci.* 90: 5484–5488, 1993.
- [44130] 17682. Barrow, L. L.; Simin, K.; Jones, J. M.; Lee, D. C.; Meisler, M. H.: Conserved linkage of early growth response 4, annexin 4, and transforming growth factor alpha on mouse chromosome 6. *Genomics* 19:388–390, 1994.
- [44131] 17683. Crosby, S. D.; Veile, R. A.; Donis–Keller, H.; Baraban, J. M.; Bhat, R. V.; Simburger, K. S.; Milbrandt, J.: Neural–specific expression, genomic structure, and chromosomal localization of the gene encoding the zinc–finger transcription factor NGFI–C. *Proc. Nat. Acad. Sci.*

89:4739–4743, 1992.

- [44132] 17684.Boerkoel, C. F.; Takashima, H.; Bacino, C. A.; Daentl, D.; Lupski, J. R.: EGR2 mutation R359W causes a spectrum of Dejerine–Sottas neuropathy. *Neurogenetics* 3:153–157, 2001.
- [44133] 17685.Willcutt, E. G.; Pennington, B. F.; Smith, S. D.; Cardon, L. R.; Gayan, J.; Knopik, V. S.; Olson, R. K.; DeFries, J. C.: Quantitative trait locus for reading disability on chromosome 6p is pleiotropic for attention–deficit/hyperactivity disorder. *Am. J. Med. Genet.* 114:260–268, 2002.
- [44134] 17686.Inazu, A.; Brown, M. L.; Hesler, C. B.; Agellon, L. B.; Koizumi, J.; Takata, K.; Maruhama, Y.; Mabuchi, H.; Tall, A. R.: Increased high–density lipoprotein levels caused by a common cholesteryl–ester transfer protein gene mutation. *New Eng. J. Med.* 323: 1234–1238, 1990.
- [44135] 17687.Saito, F.: A pedigree of homozygous familial hyperalphalipoproteinemia. *Metabolism* 33:629–633, 1984.
- [44136] 17688.Andres, D. A.; Milatovich, A.; Ozcelik, T.; Wenzlau, J. M.; Brown, M. S.; Goldstein, J. L.; Francke, U.: cDNA cloning of the two subunits of human CAAX farnesyl transferase and chromosomal mapping of FNTA and FNTB loci and related sequences. *Genomics* 18: 105–112, 1993.

- [44137] 17689.Epstein, W. W.; Lever, D.; Leining, L. M.; Bruenger, E.; Rilling, H. C.: Quantitation of prenylcysteines by a selective cleavage reaction. *Proc.Nat. Acad. Sci.* 88: 9668–9670, 1991.
- [44138] 17690.Long, S. B.; Casey, P. J.; Beese, L. S.: Reaction path of proteinfarnesyltransferase at atomic resolution. *Nature* 419: 645–650, 2002.
- [44139] 17691.Porter, J. C.; Messer, A.: Genetic mapping of farnesyltransferasealpha(Fnta) to mouse chromosome 8. *Mammalian Genome* 7: 622–623, 1996.
- [44140] 17692.Schafer, W. R.; Rine, J.: Protein prenylation: genes, enzymes, targets, and functions. *Annu. Rev. Genet.* 26: 209–237, 1992.
- [44141] 17693.Wang, T.; Danielson, P. D.; Li, B.; Shah, P. C.; Kim, S. D.; Donahoe, P. K.: The p21ras farnesyltransferase alpha subunit in TGF-beta andactivin signaling. *Science* 271: 1120–1122, 1996.
- [44142] 17694.Zhang, F. L.; Diehl, R. E.; Kohl, N. E.; Gibbs, J. B.; Giros, B.; Casey, P. J.; Omer, C. A.: cDNA cloning and expression of rat andhuman protein geranylgeranyltransferase type-I. *J. Biol. Chem.* 269:3175–3180, 1994.
- [44143] 17695.Roifman, C. M.; Zhang, J.; Chitayat, D.; Sharfe, N.: A partialdeficiency of interleukin-7R-alpha is sufficient to

abrogate T-cell development and cause severe combined immunodeficiency. *Blood* 96:2803–2807, 2000.

[44144] 17696. Abraham, R.; Myers, A.; Wavrant-DeVrieze, F.; Hamshire, M. L.; Thomas, H. V.; Marshall, H.; Compton, D.; Spurlock, G.; Turic, D.; Hoogendoorn, B.; Kwon, J. M.; Petersen, R. C.; and 12 others: Substantial linkage disequilibrium across the insulin-degrading enzyme locus but no association with late-onset Alzheimer's disease. *Hum. Genet.* 109:646–652, 2001.

[44145] 17697. Affholter, J. A.; Fried, V. A.; Roth, R. A.: Human insulin-degrading enzyme shares structural and functional homologies with *E. coli* protease III. *Science* 242: 1415–1418, 1988.

[44146] 17698. Affholter, J. A.; Hsieh, C.-L.; Francke, U.; Roth, R. A.: Insulin-degrading enzyme: stable expression of the human complementary DNA, characterization of its protein product, and chromosomal mapping of the human and mouse genes. *Molec. Endocr.* 4: 1125–1135, 1990.

[44147] 17699. Edbauer, D.; Willem, M.; Lammich, S.; Steiner, H.; Haass, C.: Insulin-degrading enzyme rapidly removes the beta-amyloid precursor protein intracellular domain (AICD). *J. Biol. Chem.* 277: 13389–13393, 2002.

[44148] 17700. Espinosa, R., III; Lemons, R. S.; Perlman, R. K.; Kuo,

W.-L.; Rosner, M. R.; Le Beau, M. M.: Localization of the gene encoding insulin-degrading enzyme to human chromosome 10, bands q23-q25. *Cytogenet. Cell Genet.* 57:184-186, 1991.

[44149] 17701. Qiu, W. Q.; Walsh, D. M.; Ye, Z.; Vekrellis, K.; Zhang, J.; Podlisny, M. B.; Rosner, M. R.; Safavi, A.; Hersh, L. B.; Selkoe, D. J.: Insulin-degrading enzyme regulates extracellular levels of amyloid beta-protein by degradation. *J. Biol. Chem.* 273: 32730-32738, 1998.

[44150] 17702. Qiu, W. Q.; Ye, Z.; Kholodenko, D.; Seubert, P.; Selkoe, D. J.: Degradation of amyloid beta-protein by a metalloprotease secreted by microglia and other neural and non-neural cells. *J. Biol. Chem.* 272:6641-6646, 1997.

[44151] 17703. Glasgow, B. J.; Heinzmann, C.; Kojis, T.; Sparkes, R. S.; Mohandas, T.; Bateman, J. B.: Assignment of tear lipocalin gene to human chromosome 9q34-9qter. *Curr. Eye Res.* 12: 1019-1023, 1993.

[44152] 17704. Herzog, H.; Baumgartner, M.; Holzfeind, P.; Redl, B.: Confirmation of 9q34 as the chromosomal site of the human lipocalin LCN1 gene. *Cytogenet. Cell Genet.* 69: 108-109, 1995.

[44153] 17705. Holzfeind, P.; Merschak, P.; Rogatsch, H.; Culig, Z.;

Feichtinger,H.; Klocker, H.; Redl, B.: Expression of the gene for tear lipocalin/vonEbner's gland protein in human prostate. FEBS Lett. 395: 95–98,1996.

[44154] 17706.Lacazette, E.; Pitiot, G.; Jobert, S.; Mallet, J.; Gachon, A. M.F.: Fine genetic mapping of LCN1/D9S1826 within 9q34. Ann. Hum.Genet. 61: 449–455, 1997.

[44155] 17707.Lassagne, H.; Gachon, A. M. F.: Cloning of a human lacrimal lipocalinsecreted in tears. Exp. Eye Res. 56: 605–609, 1993.

[44156] 17708.Lassagne, H.; Nguyen, V. C.; Mattei, M. G.; Gachon, A. M. F.:Assignment of LCN1 to human chromosome 9 is confirmed. Cytogent.Cell Genet. 71: 104, 1995.

[44157] 17709.Lassagne, H.; Ressot, C.; Mattei, M. G.; Gachon, A. M. F.: Assignmentof the human tear lipocalin gene (LCN1) to 9q34 by in situ hybridization. Genomics 18:160–161, 1993.

[44158] 17710.Nguyen Van Cong; Weil, D.; Finaz, C.; Cohen–Haguenauer, O.; Gross,M.–S.; Jegou–Foubert, C.; de Tand, M.–F.; Cochet, C.; de Grouchy,J.; Frezal, J.: Panel of twenty–five independent man–rodent hybridsfor human genetic marker mapping. Ann. Genet. 29: 20–26, 1986.

[44159] 17711.Pervaiz, S.; Brew, K.: Homology and structure–function correlationsbetween alpha–1–acid glycoprotein and

serum retinol-binding protein and its relatives. *FASEB J.* 1: 209–214, 1987.

[44160] 17712.Redl, B.; Holzfeind, P.; Lottspeich, F.: cDNA cloning and sequencing reveals human tear prealbumin to be a member of the lipophilic-ligand carrier protein superfamily. *J. Biol.Chem.* 28: 20282–20287, 1992.

[44161] 17713.Redl, B.; Wojnar, P.; Ellemunter, H.; Feichtinger, H.: Identification of a lipocalin in mucosal glands of the human tracheobronchial tree and its enhanced secretion in cystic fibrosis. *Lab. Invest.* 78:1121–1129, 1998.

[44162] 17714.van't Hof, W.; Blankenvoorde, M. F. J.; Veerman, E. C. I.; NieuwAmerongen, A. V.: The salivary lipocalin Von Ebner's Gland protein is a cysteine proteinase inhibitor. *J. Biol. Chem.* 272: 1837–1841, 1997.

[44163] 17715.Huebner, K.; Cannizzaro, L. A.; Croce, C. M.; Frey, A. Z.; Wallner, B. P.; Hecht, B. K.; Hecht, F.: Chromosome localization of the human genes for lipocortin I and the lipocortin II family. (Abstract) *Cytogenet.Cell Genet.* 46: 631 only, 1987.

[44164] 17716.Huebner, K.; Cannizzaro, L. A.; Frey, A. Z.; Hecht, B. K.; Hecht, F.; Croce, C. M.; Wallner, B. P.: Chromosomal localization of the human genes for lipocortin I and lipocortin II. *Oncogene Res.* 2:299–310, 1988.

- [44165] 17717.Spano, F.; Raugei, G.; Palla, E.; Colella, C.; Melli, M.: Characterization of the human lipocortin-2-encoding multigene family: its structure suggests the existence of a short amino acid unit undergoing duplication. *Gene* 95:243-251, 1990.
- [44166] 17718.Takahashi, S.; Reddy, S. V.; Chirgwin, J. M.; Devlin, R.; Haipek, C.; Anderson, J.; Roodman, G. D.: Cloning and identification of annexin II as an autocrine/paracrine factor that increases osteoclast formation and bone resorption. *J. Biol. Chem.* 269: 28696-28701, 1994.
- [44167] 17719.Harman, S. M.; Metter, E. J.; Blackman, M. R.; Landis, P. K.; Carter, H. B.: Serum levels of insulin-like growth factor I (IGF-I), IGF-II, IGF-binding protein-3, and prostate-specific antigen as predictors of clinical prostate cancer. *J. Clin. Endocr. Metab.* 85: 4258-4265, 2000.
- [44168] 17720.Holly, J.: Insulin-like growth factor-I and new opportunities for cancer prevention. *Lancet* 351: 1373-1375, 1998.
- [44169] 17721.Hoppener, J. W. M.; de Pagter-Holthuisen, P.; Geurts van Kessel, A. H. M.; Jansen, M.; Kittur, S. D.; Antonarakis, S. E.; Lips, C. J. M.; Sussenbach, J. S.: The human gene encoding insulin-like growth factor I is located on chromosome 12. *Hum. Genet.* 69: 157-160, 1985.

- [44170] 17722. Le Bouc, Y.; Dreyer, D.; Jaeger, F.; Binoux, M.; Sondermeyer, P.: Complete characterization of the human IGF-I nucleotide sequence isolated from a newly constructed adult liver cDNA library. *FEBS Lett.* 196: 108–112, 1986.
- [44171] 17723. Hellstrom, A.; Perruzzi, C.; Ju, M.; Engstrom, E.; Hard, A.-L.; Liu, J.-L.; Albertsson-Wikland, K.; Carlsson, B.; Niklasson, A.; Sjodell, L.; LeRoith, D.; Senger, D. R.; Smith, L. E. H.: Low IGF-I suppresses VEGF-survival signaling in retinal endothelial cells: direct correlation with clinical retinopathy of prematurity. *Proc. Nat. Acad. Sci.* 98: 5804–5808, 2001.
- [44172] 17724. Lembo, G.; Rockman, H. A.; Hunter, J. J.; Steinmetz, H.; Koch, W. J.; Ma, L.; Printz, M. P.; Ross, J., Jr.; Chien, K. R.; Powell-Braxton, L.: Elevated blood pressure and enhanced myocardial contractility in mice with severe IGF-1 deficiency. *J. Clin. Invest.* 98: 2648–2655, 1996.
- [44173] 17725. Li, C. H.; Yamashiro, D.; Gospodarowicz, D.; Kaplan, S. L.; Van Vliet, G.: Total synthesis of insulin-like growth factor I (somatomedin C). *Proc. Nat. Acad. Sci.* 80: 2216–2220, 1983.
- [44174] 17726. Liu, J.-P.; Baker, J.; Perkins, A. S.; Robertson, E. J.; Efstratiadis, A.: Mice carrying null mutations of the genes encoding insulin-like growth factor I (Igf-1) and type 1 IGF

receptor (Igf1r). Cell 75:59–72, 1993.

- [44175] 17727.Mathews, L. S.; Norstedt, G.; Palmiter, R. D.: Regulation of insulin-like growth factor I gene expression by growth hormone. Proc.Nat. Acad. Sci. 83: 9343–9347, 1986.
- [44176] 17728.Morton, C.; Rall, L.; Bell, G.; Shows, T.: Human insulin-like growth factor–1 (IGF1) is encoded at 12q22–q24.1, and insulin-like growth factor–2 (IGF2) is at 11p15. (Abstract) Cytogenet. Cell Genet. 40:703 only, 1985.
- [44177] 17729.Musaro, A.; McCullagh, K.; Paul, A.; Houghton, L.; Dobrowolny, G.; Molinaro, M.; Barton, E. R.; Sweeney, H. L.; Rosenthal, N.: Localized Igf–1 transgene expression sustains hypertrophy and regeneration in senescent skeletal muscle. Nature Genet. 27: 195–200, 2001.
- [44178] 17730.Musaro, A.; McCullagh, K. J. A.; Naya, F. J.; Olson, E. N.; Rosenthal, N.: IGF–1 induces skeletal myocyte hypertrophy through calcineurin association with GATA–2 and NF–ATc1. Nature 400: 581–585, 1999.
- [44179] 17731.Playford, M. P.; Bicknell, D.; Bodmer, W. F.; Macaulay, V. M.: Insulin-like growth factor 1 regulates the location, stability, and transcriptional activity of beta-catenin. Proc. Nat. Acad. Sci. 97:12103–12108, 2000.

- [44180] 17732.Powell–Braxton, L.; Hollingshead, P.; Warburton, C.; Dowd, M.;Pitts–Meek, S.; Dalton, D.; Gillett, N.; Stewart, T. A.: IGF–I isrequired for normal embryonic growth in mice. *Genes Dev.* 7: 2609–2617,1993.
- [44181] 17733.Rapp, R.; Deger, A.; Blum, W.; Koch, R.; Weber, U.: Characterizationof the protein which binds insulin–like growth factor in human serum. *Europ.J. Biochem.* 172: 421–425, 1988.
- [44182] 17734.Rotwein, P.: Two insulin–like growth factor I mes–senger RNAsare expressed in human liver. *Proc. Nat. Acad. Sci.* 83: 77–81, 1986.
- [44183] 17735.Schoenle, E. J.; Zenobi, P. D.; Torresani, T.; Werder, E. A.;Zachmann, M.; Froesch, E. R.: Recombinant human insulin–like growthfactor I (rhIGF I) reduces hypergly–caemia in patients with extremeinsulin resistance. *Dia–betologia* 34: 675–679, 1991.
- [44184] 17736.Semsarian, C.; Wu, M.–J.; Ju, Y.–K.; Marciniak, T.; Yeoh, T.;Allen, D. G.; Harvey, R. P.; Graham, R. M.: Skeletal muscle hypertrophyis mediated by a Ca^{2+} –dependent calcineurin signalling pathway. *Nature* 400:576–581, 1999.
- [44185] 17737.Svoboda, M. E.; Van Wyk, J. J.; Klapper, D. G.; Fel–lows, R. E.;Grissom, F. E.; Schleuter, R. J.: Purification of

somatomedin-C from human plasma: chemical and biological properties, partial sequence analysis, and relationship to other somatomedins. *Biochemistry* 19:790–797, 1980.

[44186] 17738. Taylor, B. A.; Grieco, D.: Localization of the gene encoding insulin-like growth factor I on mouse chromosome 10. *Cytogenet. Cell Genet.* 56: 57–58, 1991.

[44187] 17739. Tricoli, J. V.; Rall, L. B.; Scott, J.; Bell, G. I.; Shows, T. B.: Insulin-like growth factor genes: chromosome organization and association with disease. (Abstract) *Am. J. Hum. Genet.* 36: 121S only, 1984.

[44188] 17740. Tricoli, J. V.; Rall, L. B.; Scott, J.; Bell, G. I.; Shows, T. B.: Localization of insulin-like growth factor genes to human chromosomes 11 and 12. *Nature* 310: 784–786, 1984.

[44189] 17741. Ullrich, A.; Berman, C. H.; Dull, T. J.; Gray, A.; Lee, J. M.: Isolation of the human insulin-like growth factor I gene using a single synthetic DNA probe. *EMBO J.* 3: 361–364, 1984.

[44190] 17742. Gough, N. M.; Rakar, S.; Harpur, A.; Wilks, A. F.: Localization of genes for two members of the JAK family of protein tyrosine kinases to murine chromosomes 4 and 19. *Mammalian Genome* 6: 247–248, 1995.

- [44191] 17743.Ihle, J. N.: Cytokine receptor signalling. *Nature* 377: 591–594,1995.
- [44192] 17744.Pritchard, M. A.; Baker, E.; Callen, D. F.; Sutherland, G. R.;Wilks, A. F.: Two members of the JAK family of protein tyrosine kinasesmap to chromosomes 1p31.3 and 9p24. *Mammalian Genome* 3: 36–38, 1992.
- [44193] 17745.Campbell, G. S.; Argetsinger, L. S.; Ihle, J. N.; Kelly, P. A.;Rillema, J. A.; Carter–Su, C.: Activation of JAK2 tyrosine kinaseby prolactin receptors in Nb2 cells and mouse mammary gland explants. *Proc.Nat. Acad. Sci.* 91: 5232–5236, 1994.
- [44194] 17746.Huang, L. J.; Constantinescu, S. N.; Lodish, H. F.: The N-terminaldomain of Janus kinase 2 is required for Golgi processing and cellsurface expression of erythropoietin receptor. *Molec. Cell* 8: 1327–1338,2001.
- [44195] 17747.Neubauer, H.; Cumano, A.; Muller, M.; Wu, H.; Huffstadt, U.; Pfeffer,K.: Jak2 deficiency defines an essential developmental checkpointin definitive hematopoiesis. *Cell* 93: 397–409, 1998.
- [44196] 17748.Parganas, E.; Wang, D.; Stravopodis, D.; Topham, D. J.; Marine,J.–C.; Teglund, S.; Vanin, E. F.; Bodner, S.; Colamonici, O. R.; vanDeursen, J. M.; Grosveld, G.; Ihle, J. N.: Jak2 is essential forsignaling through a variety of cy–

tokine receptors. Cell 93: 385–395,1998.

- [44197] 17749.Saltzman, A.; Stone, M.; Franks, C.; Searfoss, G.; Munro, R.; Jaye,M.; Ivashchenko, Y.: Cloning and characterization of human Jak–2kinase: high mRNA expression in immune cells and muscle tissue. Biochem.Biophys. Res. Commun. 246: 627–633, 1998.
- [44198] 17750.Schwaller, J.; Parganas, E.; Wang, D.; Cain, D.; Aster, J. C.;Williams, I. R.; Lee, C.–K.; Gerthner, R.; Kitamura, T.; Frantsve,J.; Anastasiadou, E.; Loh, M. L.; Levy, D. E.; Ihle, J. N.; Gilliland,D. G.: Stat5 is essential for the myelo– and lymphoproliferativedisease induced by TEL/ JAK2. Molec. Cell 6: 693–704, 2000.
- [44199] 17751.Watling, D.; Guschin, D.; Muller, M.; Silvennoinen, O.; Witthuhn,B. A.; Quelle, F. W.; Rogers, N. C.; Schindler, C.; Stark, G. R.;Ihle, J. N.; Kerr, I. M.: Complementation by the protein tyrosinekinase JAK2 of a mutant cell line defective in the interferon–gammasignal transduction pathway. Nature 366: 166–170, 1993.
- [44200] 17752.Cihakova, D.; Trebusak, K.; Heino, M.; Fadeyev, V.; Tiulpakov,A.; Battelino, T.; Tar, A.; Halasz, Z.; Blumel, P.; Tawfik, S.; Krohn,K.; Lebl, J.; Peterson, P.; MEWPE–APECED Study Group: Novel AIREmutations and P450 cytochrome autoantibodies in Central and EasternEuropean patients

with APECED. Hum. Mutat. 18: 225–232, 2001.

[44201] 17753.Clemente, M. G.; Obermayer–Straub, P.; Meloni, A.; Strassburg, C. P.; Arangino, V.; Tukey, R. H.; de Virgiliis, S.; Manns, M. P.: Cytochrome P450 1A2 is a hepatic autoantigen in autoimmune polyglandular syndrome type 1. J. Clin. Endocr. Metab. 82: 1353–1361, 1997.

[44202] 17754.Craig, J. M.; Schiff, L. H.; Boone, J. E.: Chronic moniliasis associated with Addison's disease. Am. J. Dis. Child. 89: 669–684, 1955.

[44203] 17755.Ekwall, O.; Hedstrand, H.; Haavik, J.; Perheentupa, J.; Betterle, C.; Gustafsson, J.; Husebye, E.; Rorsman, F.; Kampe, O.: Pteridin–dependent hydroxylases as autoantigens in autoimmune polyendocrine syndrome type I. J. Clin. Endocr. Metab. 85: 2944–2950, 2000.

[44204] 17756.Cetani, F.; Barbesino, G.; Borsari, S.; Pardi, E.; Cianferotti, L.; Pinchera, A.; Marcocci, C.: A novel mutation of the autoimmune regulator gene in an Italian kindred with autoimmune polyendocrinopathy–candidiasis–ectodermal dystrophy, acting in a dominant fashion and strongly cosegregating with hypothyroid autoimmune thyroiditis. J. Clin. Endocr. Metab. 86: 4747–4752, 2001.

[44205] 17757.Finnish–German APECED Consortium: An autoimmune disease, APECED, caused by mutations in a novel

gene featuring two PHD-type zinc-fingerdomains. *Nature Genet.* 17: 399–403, 1997.

[44206] 17758.Foz, M.; Mirada, A.; Guardia, J.: Endocrine disorders in a family.(Letter) *Lancet* II: 269 only, 1970.

[44207] 17759.Gass, J. D. M.: The syndrome of keratoconjunctivitis, superficialmoniliasis, idiopathic hypoparathyroidism and Addison's disease. *Am.J. Ophthal.* 54: 660–674, 1962.

[44208] 17760.Gylling, M.; Tuomi, T.; Bjorses, P.; Kontiainen, S.; Partanen,J.; Christie, M. R.; Knip, M.; Perheentupa, J.; Miettinen, A.: Beta-cellautoantibodies, human leukocyte antigen II alleles, and type 1 diabetesin autoimmune polyendocrinopathy–candidiasis–ectodermal dystrophy. *J.Clin. Endocr. Metab.* 85: 4434–4440, 2000.

[44209] 17761.Hedstrand, H.; Ekwall, O.; Haavik, J.; Landgren, E.; Betterle,C.; Perheentupa, J.; Gustafsson, J.; Husebye, E.; Rorsman, F.; Kampe,O.: Identification of tyrosine hydroxylase as an autoantigen in autoimmune polyendocrine syndrome type I. *Biochem. Biophys. Res. Commun.* 267:456–461, 2000.

[44210] 17762.Heino, M.; Peterson, P.; Kudoh, J.; Shimizu, N.; Antonarakis,S. E.; Scott, H. S.; Krohn, K.: APECED mutations in the autoimmuneregulator (AIRE) gene. *Hum. Mutat.* 18: 205–211, 2001.

- [44211] 17763.Heino, M.; Scott, H. S.; Chen, Q.; Peterson, P.; Maenpaa, U.;Papasavvas, M.-P.; Mittaz, L.; Barras, C.; Rossier, C.; Chrousos,G. P.; Stratakis, C. A.; Nagamine, K.; Kudoh, J.; Shimizu, N.; Maclaren,N.; Antonarakis, S. E.; Krohn, K.: Mutation analyses of North AmericanAPS-1 patients. Hum. Mutat. 13: 69-74, 1999.
- [44212] 17764.Hendrix, T. R.: Personal Communication. Baltimore, Md. 1/14/1985.
- [44213] 17765.Hiekkala, H.: Idiopathic hypoparathyroidism, adrenal insufficiencyand moniliasis in children. Ann. Paediat. Fenn. 10: 213-222, 1964.
- [44214] 17766.Hung, W.; Migeon, C. J.; Parrott, R. H.: A possible autoimmunebasis for Addison's disease in three siblings, one with idiopathichypoparathyroidism, pernicious anemia and superficial moniliasis. NewEng. J. Med. 269: 658-663, 1963.
- [44215] 17767.Husebye, E. S.; Gebre-Medhin, G.; Tuomi, T.; Perheentupa, J.;Landin-Olsson, M.; Gustafsson, J.; Rorsman, F.; Kampe, O.: Autoantibodiesagainst aromatic L-amino acid decarboxylase in autoimmune polyendocrinesyndrome type I. J. Clin. Endocr. Metab. 82: 147-150, 1997.
- [44216] 17768.Ishii, T.; Suzuki, Y.; Ando, N.; Matsuo, N.; Ogata, T.: Novelmutations of the autoimmune regulator gene in

two siblings with autoimmune polyendocrinopathy–candidiasis–ectodermal dystrophy. *J. Clin. Endocrinol. Metab.* 85: 2922–2926, 2000.

- [44217] 17769. Kenny, F. M.; Holliday, M. D.: Hypoparathyroidism, moniliasis, Addison's and Hashimoto's disease. Hypercalcemia treated with intravenously administered sodium sulfate. *New Eng. J. Med.* 271: 708–713, 1964.
- [44218] 17770. Krohn, K.; Uibo, R.; Aavik, E.; Peterson, P.; Savilahti, K.: Identification by molecular cloning of an autoantigen associated with Addison's disease as steroid 17- α -hydroxylase. *Lancet* 339: 770–773, 1992.
- [44219] 17771. Kunin, A. S.; MacKay, B. R.; Burns, S. L.; Halberstam, M. J.: The syndrome of hypoparathyroidism and adrenocortical insufficiency, a possible sequel of hepatitis: case report and review of the literature. *Am. J. Med.* 34: 856–866, 1963.
- [44220] 17772. Louria, D. B.; Shannon, D. C.; Johnson, G.; Caroline, L.; Okas, A.; Taschdjian, C.: The susceptibility to moniliasis in children with endocrine hypofunction. *Trans. Assoc. Am. Phys.* 80: 236–249, 1967.
- [44221] 17773. Maghnie, M.; Cosi, G.; Genovese, E.; Manca-Bitti, M. L.; Cohen, A.; Zecca, S.; Tinelli, C.; Gallucci, M.; Bernasconi, S.; Boscherini, B.; Severi, F.; Arico, M.: Central

diabetes insipidus in children and young adults. *New Eng. J. Med.* 343: 998–1007, 2000.

[44222] 17774. Marieb, N. J.; Melby, J. C.; Lyall, S. S.: Isolated hypoadosteronism associated with idiopathic hypoparathyroidism. *Arch. Intern. Med.* 134:424–429, 1974.

[44223] 17775. McKusick, V. A.: Personal Communication. Baltimore, Md. 1985.

[44224] 17776. Meloni, A.; Perniola, R.; Faa, V.; Corvaglia, E.; Cao, A.; Rosatelli, M. C.: Delineation of the molecular defects in the AIRE gene in autoimmune polyendocrinopathy–candidiasis–ectodermal dystrophy patients from Southern Italy. *J. Clin. Endocr. Metab.* 87: 841–846, 2002.

[44225] 17777. Nagamine, K.; Peterson, P.; Scott, H. S.; Kudoh, J.; Minoshima, S.; Heino, M.; Krohn, K. J. E.; Lalioti, M. D.; Mullis, P. E.; Antonarakis, S. E.; Kawasaki, K.; Asakawa, S.; Ito, F.; Shimizu, N.: Positional cloning of the APECED gene. *Nature Genet.* 17: 393–398, 1997.

[44226] 17778. Neufeld, M.; Maclaren, N.; Blizzard, R.: Autoimmune polyglandular syndrome. *Pediat. Ann.* 9: 154–162, 1980.

[44227] 17779. Neufeld, M.; Maclaren, N. K.; Blizzard, R. M.: Two types of autoimmune Addison's disease associated with different polyglandular autoimmune (PGA) syndromes.

Medicine 60: 355–362, 1981.

- [44228] 17780. Nithiyananthan, R.; Heward, J. M.; Allahabadia, A.; Barnett, A.H.; Franklyn, J. A.; Gough, S. C. L.: A heterozygous deletion of the autoimmune regulator (AIRE1) gene, autoimmune thyroid disease, and type 1 diabetes: no evidence for association. *J. Clin. Endocr. Metab.* 85: 1320–1322, 2000.
- [44229] 17781. Okano, M.; Bell, D. W.; Haber, D. A.; Li, E.: DNA methyltransferases Dnmt3a and Dnmt3b are essential for de novo methylation and mammalian development. *Cell* 99: 247–257, 1999.
- [44230] 17782. Wijmenga, C.; van den Heuvel, L. P. W. J.; Strengman, E.; Luyten, J. A. F. M.; van der Burgt, I. J. A. M.; de Groot, R.; Smeets, D. F. C. M.; Draaisma, J. M. T.; van Dongen, J. J.; De Abreu, R. A.; Pearson, P. L.; Sandkuijl, L. A.; Weemaes, C. M. R.: Localization of the ICF syndrome to chromosome 20 by homozygosity mapping. *Am. J. Hum. Genet.* 63: 803–809, 1998.
- [44231] 17783. Xu, G.-L.; Bestor, T. H.; Bourc'his, D.; Hsieh, C.-L.; Tommerup, N.; Bugge, M.; Hulten, M.; Qu, X.; Russo, J. J.; Viegas-Pequignot, E.: Chromosome instability and immunodeficiency syndrome caused by mutations in a DNA methyltransferase gene. *Nature* 402: 187–191, 1999.

- [44232] 17784. Chuang, D. T.; Shih, V. E.: Disorders of branched chain amino acid and keto acid metabolism. In: Scriver, C. R.; Beaudet, A. L.; Sly, W. S.; Valle, D.: *Metabolic and Molecular Bases of Inherited Disease*. New York: McGraw-Hill (pub.) (7th ed.): 1995. Pp. 1239–1277.
- [44233] 17785. Smeitink, J.; van den Heuvel, L.: Human mitochondrial complex I in health and disease. *Am. J. Hum. Genet.* 64: 1505–1510, 1999.
- [44234] 17786. Pizarro, T. T.; Michie, M. H.; Bentz, M.; Woraratanadham, J.; Smith, M. F., Jr.; Foley, E.; Moskaluk, C. A.; Bickston, S. J.; Cominelli, F.: IL-18, a novel immunoregulatory cytokine, is up-regulated in Crohn's disease: expression and localization in intestinal mucosal cells. *J. Immun.* 162: 6829–6835, 1999.
- [44235] 17787. Le Coniat, M.; Kinet, J. P.; Berger, R.: The human genes for the α and γ subunits of the mast cell receptor for immunoglobulin E are located on human chromosome band 1q23. *Immunogenetics* 32: 183–186, 1990.
- [44236] 17788. 't Hart, L. M.; Stolk, R. P.; Heine, R. J.; Grobbee, D. E.; van der Does, F. E. E.; Maassen, J. A.: Association of the insulin-receptor variant met-985 with hyperglycemia and non-insulin-dependent diabetes mellitus in the Nether-

lands: a population-based study. *Am. J. Hum. Genet.* 59: 1119–1125, 1996.

[44237] 17789. Accili, D.; Drago, J.; Lee, E. J.; Johnson, M. D.; Cool, M. H.; Salvatore, P.; Asico, L. D.; Jose, P. A.; Taylor, S. I.; Westphal, H.: Early neonatal death in mice homozygous for a null allele of the insulin receptor gene. *Nature Genet.* 12: 106–109, 1996.

[44238] 17790. Accili, D.; Frapier, C.; Mosthaf, L.; McKeon, C.; El-bein, S. C.; Permutt, M. A.; Ramos, E.; Lander, E.; Ullrich, A.; Taylor, S. I.: A mutation in the insulin receptor gene that impairs transport of the receptor to the plasma membrane and causes insulin-resistant diabetes. *EMBO J.* 8: 2509–2517, 1989.

[44239] 17791. Al-Gazali, L. I.; Khalil, M.; Devadas, K.: A syndrome of insulin resistance resembling leprechaunism in five sibs of consanguineous parents. *J. Med. Genet.* 30: 470–475, 1993.

[44240] 17792. Bar, R. S.; Muggeo, M.; Roth, J.; Kahn, C. R.; Havrankova, J.; Imperato-McGinley, J.: Insulin resistance, acanthosis nigricans, and normal insulin receptors in a young woman: evidence for a postreceptor defect. *J. Clin. Endocr. Metab.* 47: 620–625, 1978.

[44241] 17793. Barbetti, F.; Gejman, P. V.; Taylor, S. I.; Raben, N.;

Cama, A.; Bonora, E.; Pizzo, P.; Moghetti, P.; Muggeo, M.; Roth, J.: Detection of mutations in insulin receptor gene by denaturing gradient gel electrophoresis. *Diabetes* 41:408–415, 1992.

- [44242] 17794. Barnes, N. D.; Palumbo, P. J.; Hayles, A. B.; Folgar, H.: Insulin resistance, skin changes, and virilization: a recessively inherited syndrome possibly due to pineal gland dysfunction. *Diabetologia* 10:285–289, 1974.
- [44243] 17795. Belke, D. D.; Betuing, S.; Tuttle, M. J.; Graveleau, C.; Young, M. E.; Pham, M.; Zhang, D.; Cooksey, R. C.; McClain, D. A.; Litwin, S. E.; Taegtmeyer, H.; Severson, D.; Kahn, C. R.; Abel, E. D.: Insulin signaling coordinately regulates cardiac size, metabolism, and contractile protein isoform expression. *J. Clin. Invest.* 109: 629–639, 2002.
- [44244] 17796. Benecke, H.; Flier, J. S.; Moller, D. E.: Alternatively spliced variants of the insulin receptor protein: expression in normal and diabetic human tissues. *J. Clin. Invest.* 89: 2066–2070, 1992.
- [44245] 17797. Bruning, J. C.; Gautam, D.; Burks, D. J.; Gillette, J.; Schubert, M.; Orban, P. C.; Klein, R.; Krone, W.; Muller-Wieland, D.; Kahn, C. R.: Role of brain insulin receptor in control of body weight and reproduction. *Science* 289: 2122–2125, 2000.

- [44246] 17798. Bruning, J. C.; Michael, M. D.; Winnay, J. N.; Hayashi, T.; Horsch, D.; Accili, D.; Goodyear, L. J.; Kahn, C. R.: A muscle-specific insulin receptor knockout exhibits features of the metabolic syndrome of NIDDM without altering glucose tolerance. *Molec. Cell* 2: 559–569, 1998.
- [44247] 17799. Cama, A.; de la Luz Sierra, M.; Ottini, L.; Kadowaki, T.; Gorden, P.; Imperato-McGinley, J.; Taylor, S. I.: A mutation in the tyrosine kinase domain of the insulin receptor associated with insulin resistance in an obese woman. *J. Clin. Endocr. Metab.* 73: 894–901, 1991.
- [44248] 17800. Henry, I.; Humphries, S. E.; Tata, F.; Barichard, F.; Holm, M.; Williamson, R.; Junien, C.: The gene for HMG CoA reductase (HMGCR) is on human chromosome 5. (Abstract) *Cytogenet. Cell Genet.* 40:649–650, 1985.
- [44249] 17801. Humphries, S. E.; Tata, F.; Henry, I.; Barichard, F.; Holm, M.; Junien, C.; Williamson, R.: The isolation, characterisation, and chromosomal assignment of the gene for human 3-hydroxy-3-methylglutaryl coenzyme A reductase, (HMG-CoA reductase). *Hum. Genet.* 71: 254–258, 1985.
- [44250] 17802. Julier, C.; Delepine, M.; Keavney, B.; Terwilliger, J.; Davis, S.; Weeks, D. E.; Bui, T.; Jeunemaitre, X.; Velho, G.; Froguel, P.; Ratcliffe, P.; Corvol, P.; Soubrier, F.; Lathrop, G.

M.: Geneticsusceptibility for human familial essential hypertension in a regionof homology with blood pressure linkage on rat chromosome 10. Hum.Molec. Genet. 6: 2077–2085, 1997.

- [44251] 17803.Rutherford, S.; Johnson, M. P.; Curtain, R. P.; Griffiths, L.R.: Chromosome 17 and the inducible nitric oxide synthase gene inhuman essential hypertension. Hum. Genet. 109: 408–415, 2001.
- [44252] 17804.Aprelikova, O.; Pajusola, K.; Partanen, J.; Armstrong, E.; Alitalo,R.; Bailey, S. K.; McMahon, J.; Wasmuth, J.; Huebner, K.; Alitalo,K.: FLT4, a novel class III receptor tyrosine kinase in chromosome5q33–qter. Cancer Res. 52: 746–748, 1992.
- [44253] 17805.Dumont, D. J.; Jussila, L.; Taipale, J.; Lymboussaki, A.; Mustonen,T.; Pajusola, K.; Breitman, M.; Alitalo, K.: Cardiovascular failurein mouse embryos deficient in VEGF receptor–3. Science 282: 946–949,1998.
- [44254] 17806.Evans, A. L.; Brice, G.; Sotirova, V.; Mortimer, P.; Beninson,J.; Burnand, K.; Rosbotham, J.; Child, A.; Sarfarazi, M.: Mappingof primary congenital lymphedema to the 5q35.3 region. Am. J. Hum.Genet. 64: 547–555, 1999.
- [44255] 17807.Ferrell, R. E.; Levinson, K. L.; Esman, J. H.; Kimak, M. A.; Lawrence,E. C.; Barmada, M. M.; Finegold, D. N.:

Hereditary lymphedema: evidence for linkage and genetic heterogeneity. *Hum. Molec. Genet.* 7: 2073–2078, 1998.

- [44256] 17808. Galland, F.; Karamysheva, A.; Mattei, M.-G.; Rosnet, O.; Marchetto, S.; Birnbaum, D.: Chromosomal localization of FLT4, a novel receptor-tyrosine kinase gene. *Genomics* 13: 475–478, 1992.
- [44257] 17809. Irrthum, A.; Karkkainen, M. J.; Devriendt, K.; Alitalo, K.; Vikkula, M.: Congenital hereditary lymphedema caused by a mutation that inactivates VEGFR3 tyrosine kinase. *Am. J. Hum. Genet.* 67: 295–301, 2000.
- [44258] 17810. Kaipainen, A.; Korhonen, J.; Mustonen, T.; van Hinsbergh, V. W. M.; Fang, G.-H.; Dumont, D.; Breitman, M.; Alitalo, K.: Expression of the fms-like tyrosine kinase 4 gene becomes restricted to lymphatic endothelium during development. *Proc. Nat. Acad. Sci.* 92: 3566–3570, 1995.
- [44259] 17811. Karkkainen, M. J.; Ferrell, R. E.; Lawrence, E. C.; Kimak, M. A.; Levinson, K. L.; McTigue, M. A.; Alitalo, K.; Finegold, D. N.: Missense mutations interfere with VEGFR-3 signalling in primary lymphoedema. *Nature Genet.* 25: 153–159, 2000.
- [44260] 17812. Karkkainen, M. J.; Saaristo, A.; Jussila, L.; Karila, K. A.; Lawrence, E. C.; Pajusola, K.; Bueler, H.; Eichmann, A.; Kauppinen, R.; Kettunen, M. I.; Yla-Herttuala, S.; Finegold,

D. N.; Ferrell, R. E.; Alitalo, K.: A model for gene therapy of human hereditary lymphedema. *Proc. Nat. Acad. Sci.* 98: 12677–12682, 2001.

[44261] 17813. Lee, J.; Gray, A.; Yuan, J.; Luoh, S.-M.; Avraham, H.; Wood, W. I.: Vascular endothelial growth factor-related protein: a ligand and specific activator of the tyrosine kinase receptor Flt4. *Proc. Nat. Acad. Sci.* 93: 1988–1992, 1996.

[44262] 17814. Lyon, M. F.; Glenister, P. H.: New Mutants. *Mouse Newsletter* 71:26 only, 1984.

[44263] 17815. Lyon, M. F.; Glenister, P. H.: Gene order of Chyt-Re on chromosome 11. *Mouse Newsletter* 74: 96 only, 1986.

[44264] 17816. Milroy, W. F.: An undescribed variety of hereditary oedema. *New York Med. J.* 56: 505–508, 1892.

[44265] 17817. Offori, T. W.; Platt, C. C.; Stephens, M.; Hopkinson, G. B.: Angiosarcoma in congenital hereditary lymphoedema (Milroy's disease): diagnostic beacons and a review of the literature. *Clin. Exp. Derm.* 18:174–177, 1993.

[44266] 17818. Pajusola, K.; Aprelikova, O.; Korhonen, J.; Kaipainen, A.; Pertovaara, L.; Alitalo, R.; Alitalo, K.: FLT4 receptor tyrosine kinase contains seven immunoglobulin-like loops and is expressed in multiple human tissues and

cell lines. *Cancer Res.* 52: 5738–5743, 1992.

[44267] 17819. Pajusola, K.; Aprelikova, O.; Pelicci, G.; Weich, H.; Claesson-Welsh, L.; Alitalo, K.: Signalling properties of FLT4, a proteolytically processed receptor tyrosine kinase related to two VEGF receptors. *Oncogene* 9:3545–3555, 1994.

[44268] 17820. Walter, J. W.; North, P. E.; Waner, M.; Mizeracki, A.; Blei, F.; Walker, J. W. T.; Reinisch, J. F.; Marchuk, D. A.: Somatic mutation of vascular endothelial growth factor receptors in juvenile hemangioma. *Genes Chromosomes Cancer* 33: 295–303, 2002.

[44269] 17821. Ragoussis, J.; Senger, G.; Trowsdale, J.; Campbell, I. G.: Genomic organization of the human folate receptor genes on chromosome 11q13. *Genomics* 14:423–430, 1992.

[44270] 17822. Barber, R. C.; Shaw, G. M.; Lammer, E. J.; Greer, K. A.; Biela, T. A.; Lacey, S. W.; Wasserman, C. R.; Finnell, R. H.: Lack of association between mutations in the folate receptor- α gene and spina bifida. *Am. J. Med. Genet.* 76: 310–317, 1998.

[44271] 17823. Bowcock, A. M.; Lacey, S.; Saltman, D.; Mohandas, T. K.; Kamen, B. A.; Taggart, R. T.: Localization of the folate receptor gene to chromosome 11q13. (Abstract) *Cyto-*

genet. Cell Genet. 58: 1955 only,1991.

- [44272] 17824.Campbell, I. G.; Jones, T. A.; Foulkes, W. D.; Trowsdale, J.:Folate-binding protein is a marker for ovarian cancer. Cancer Res. 51:5329-5338, 1991.
- [44273] 17825.Kranes, A.; Balogh, K., Jr.: Liver disease in a patient with von Hippel-Lindau disease. New Eng. J. Med. 275: 950-959, 1966.
- [44274] 17826.Lamiell, J. M.: Personal Communication. San Francisco, Calif. 9/1987.
- [44275] 17827.Lamiell, J. M.; Salazar, F. G.; Hsia, Y. E.: Von Hippel-Lindau disease affecting 43 members of a single kindred. Medicine 68: 1-29,1989.
- [44276] 17828.Latif, F.; Tory, K.; Gnarr, J.; Yao, M.; Duh, F.-M.; Orcutt, M. L.; Stackhouse, T.; Kuzmin, I.; Modi, W.; Geil, L.; Schmidt, L.; Zhou, F.; Li, H.; Wei, M. H.; Chen, F.; Glenn, G.; Choyke, P.; Walther, M. M.; Weng, Y.; Duan, D.-S. R.; Dean, M.; Glavac, D.; Richards, F.M.; Crossey, P. A.; Ferguson-Smith, M. A.; Le Paslier, D.; Chumakov, I.; Cohen, D.; Chinnault, A. C.; Maher, E. R.; Linehan, W. M.; Zbar, B.; Lerman, M. I.: Identification of the von Hippel-Lindau disease tumor suppressor gene. Science 260: 1317-1320, 1993.
- [44277] 17829.Lee, S.; Chen, D. Y. T.; Humphrey, J. S.; Gnarr, J. R.; Linehan, W. M.; Klausner, R. D.: Nuclear/cytoplasmic lo-

calization of the vonHippel–Lindau tumor suppressor gene product is determined by celldensity. Proc. Nat. Acad. Sci. 93: 1770–1775, 1996.

[44278] 17830.Lenz, T.; Thiede, H. M.; Nussberger, J.; Atlas, S. A.; Distler,A.; Schulte, K. L.: Hyperreninemia and secondary hyperaldosteronism in a patient with pheochromocytoma and von Hippel–Lindau disease. Nephron 62:345–350, 1992.

[44279] 17831.Lindau, A.: Zur Frage der Angiomatosis Retinae und Ihrer Hirncomplication. ActaOphthal. 4: 193–226, 1927.

[44280] 17832.Loeb, D. B.; Pericak–Vance, M. A.; Stajich, J. M.; Vance, J. M.: A novel mutation in the von Hippel–Lindau gene. Hum. Molec. Genet. 3:1423–1424, 1994.

[44281] 17833.Lui, W. O.; Chen, J.; Glasker, S.; Bender, B. U.; Madura, C.;Khoo, S. K.; Kort, E.; Larsson, C.; Neumann, H. P. H.; Teh, B. T.: Selective loss of chromosome 11 in pheochromocytomas associatedwith the VHL syndrome. Oncogene 21: 1117–1122, 2002.

[44282] 17834.Maddock, I. R.; Moran, A.; Maher, E. R.; Teare, M. D.; Norman,A.; Payne, S. J.; Whitehouse, R.; Dodd, C.; Lavin, M.; Hartley, N.;Super, M.; Evans, D. G. R.: A genetic register for von Hippel–Lindaudisease. J. Med. Genet. 33:

120–127, 1996.

- [44283] 17835. Maher, E. R.; Bentley, E.; Yates, J. R. W.; Barton, D.; Jennings, A.; Fellows, I. W.; Ponder, M. A.; Ponder, B. A. J.; Benjamin, C.; Harris, R.; Ferguson-Smith, M. A.: Mapping of von Hippel–Lindau disease to chromosome 3p confirmed by genetic linkage analysis. *J. Neurol. Sci.* 100: 27–30, 1990.
- [44284] 17836. Maher, E. R.; Bentley, E.; Yates, J. R. W.; Latif, F.; Lerman, M.; Zbar, B.; Affara, N. A.; Ferguson-Smith, M. A.: Mapping of the von Hippel–Lindau disease locus to a small region of chromosome 3p by genetic linkage analysis. *Genomics* 10: 957–960, 1991.
- [44285] 17837. Maher, E. R.; Iselius, L.; Yates, J. R. W.; Littler, M.; Benjamin, C.; Harris, R.; Sampson, J.; Williams, A.; Ferguson-Smith, M. A.; Morton, N.: Von Hippel–Lindau disease: a genetic study. *J. Med. Genet.* 28: 443–447, 1991.
- [44286] 17838. Maher, E. R.; Yates, J. R. W.; Ferguson-Smith, M. A.: Statistical analysis of the two stage mutation model in von Hippel–Lindau disease, and in sporadic cerebellar haemangioblastoma and renal cell carcinoma. *J. Med. Genet.* 27: 311–314, 1990.
- [44287] 17839. Mahon, P. C.; Hirota, K.; Semenza, G. L.: FIH-1: a novel protein that interacts with HIF-1- α and VHL to

mediate repression of HIF-1 transcriptional activity. *Genes Dev.* 15: 2675–2686, 2001.100. Manski, T. J.; Heffner, D. K.; Glenn, G. M.; Patronas, N. J.; Pikus, A. T.; Katz, D.; Lebovics, R.; Sledjeski, K.; Choyke, P. L.; Zbar, B.; Linehan, W. M.; Oldfield, E. H.: Endolymphatic sac tumors: a source of morbid hearing loss in von Hippel–Lindau disease. *J.A.M.A.* 277:1461–1466, 1997.101. Maxwell, P. H.; Wiesener, M. S.; Chang, G.–W.; Clifford, S. C.; Vaux, E. C.; Cockman, M. E.; Wykoff, C. C.; Pugh, C. W.; Maher, E.R.; Ratcliffe, P. J.: The tumour suppressor protein VHL targets hypoxia-inducible factors for oxygen-dependent proteolysis. *Nature* 399: 271–275, 1999.102. McCabe, C. M.; Flynn, H. W., Jr.; Shields, C. L.; Shields, J.A.; Regillo, C. D.; McDonald, H. R.; Berrocal, M. H.; Gass, J. D.M.; Mieler, W. F.: Juxtapapillary capillary hemangiomas: clinical features and visual acuity outcomes. *Ophthalmology* 107: 2240–2249, 2000.103. McKusick, V. A.; Abbey, H.; Bettersby, E. J.; Borhani, N. O.; Boyer, S. H., IV; Cohen, B. H.; Ferguson–Smith, M. A.; Franke, F.R.; Gordon, H.; Handmaker, S. D.; Harris, W. S.; Hawkins, M. R.; and 13 others: Medical genetics 1960. *J. Chronic Dis.* 14: 1–198, 1961. Fig. 71.104. Melmon, K. L.; Rosen, S. W.: Lindau's disease: review of the literature and study of a large kindred. *Am. J.*

Med. 36: 595–617, 1964.105. Min, J.-H.; Yang, H.; Ivan, M.; Gertler, F.; Kaelin, W. G., Jr.; Pavletich, N. P.: Structure of an HIF-1-alpha-pVHL complex: hydroxyproline recognition in signaling. Science 296: 1886–1889, 2002.106. Mukhopadhyay, D.; Knebelmann, B.; Cohen, H. T.; Ananth, S.; Sukhatme, V. P.: The von Hippel-Lindau tumor suppressor gene product interacts with Sp1 to repress vascular endothelial growth factor promoter activity. Molec. Cell. Biol. 17: 5629–5639, 1997.107. Neumann, H. P. H.; Berger, D. P.; Sigmund, G.; Blum, U.; Schmidt, D.; Parmer, R. J.; Volk, B.; Kirste, G.: Pheochromocytomas, multiple endocrine neoplasia type 2, and von Hippel-Lindau disease. New Eng. J. Med. 329: 1531–1538, 1993.108. Neumann, H. P. H.; Eng, C.; Mulligan, L. M.; Glavac, D.; Zauner, I.; Ponder, B. A. J.; Crossey, P. A.; Maher, E. R.; Brauch, H.: Consequences of direct genetic testing for germline mutations in the clinical management of families with multiple endocrine neoplasia, type II. J.A.M.A. 274:1149–1151, 1995.109. Neumann, H. P. H.; Wiestler, O. D.: Clustering of features of von Hippel-Lindau syndrome: evidence for a complex genetic locus. Lancet 337:1052–1054, 1991.110. Nibbelink, D. W.; Peters, B. H.; McCormick, W. F.: On the association of pheochromocytoma and cerebellar hemang-

gioblastoma. Neurology 19:455–460, 1969.111. Oberstrass, J.; Reifenberger, G.; Reifenberger, J.; Wechsler, W.; Collins, V. P.: Mutation of the von Hippel–Lindau tumour suppressorgene in capillary haemangioblastomas of the central nervous system. J.Path. 179: 151–156, 1996.112. Ohh, M.; Yauch, R. L.; Lonergan, K. M.; Whaley, J. M.; Stemmer–Rachamimov, A. O.; Louis, D. N.; Gavin, B. J.; Kley, N.; Kaelin, W. G., Jr.; Iliopoulos, O.: The von Hippel–Lindau tumor suppressor protein is required for proper assembly of an extracellular fibronectin matrix. Molec. Cell 1:959–968, 1998.113. Olschwang, S.; Richard, S.; Boisson, C.; Giraud, S.; Laurent–Puig, P.; Resche, F.; Thomas, G.: Germline mutation profile of the VHL gene in von Hippel–Lindau disease and in sporadic hemangioblastoma. Hum.Mutat. 12: 424–430, 1998.114. Otenasek, F. J.; Silver, M. L.: Spinal hemangioma (hemangioblastoma) in Lindau's disease: report of six cases in a single family. J. Neurosurg. 18:295–300, 1961.115. Pack, S. D.; Zbar, B.; Pak, E.; Ault, D. O.; Humphrey, J. S.; Pham, T.; Hurley, K.; Weil, R. J.; Park, W.–S.; Kuzmin, I.; Stolle, C.; Glenn, G.; Liotta, L. A.; Lerman, M. I.; Klausner, R. D.; Linehan, W. M.; Zhuang, Z.: Constitutional von Hippel–Lindau (VHL) gene deletions detected in VHL families by fluorescence in situ

hybridization. *Cancer Res.* 59: 5560–5564, 1999.116.

Pause, A.; Lee, S.; Lonergan, K. M.; Klausner, R. D.: The vonHippel–Lindau tumor suppressor gene is required for cell cycle exit upon serum withdrawal. *Proc. Nat. Acad. Sci.* 95: 993–998, 1998.117. Price, E. B., Jr.: Papillary cystadenoma of the epididymis: a clinicopathologic analysis of 20 cases. *Arch. Path.* 91: 456–470, 1971.118. Probst, A.; Lotz, M.; Heitz, P.: Von Hippel–Lindau's disease, syringomyelia and multiple endocrine tumors: a complex neuroendocrinopathy. *Virchows Arch. Path. Anat. Histol.* 378: 265–272, 1978.119. Prowse, A. H.; Webster, A. R.; Richards, F. M.; Richard, S.; Olschwang, S.; Resche, F.; Affara, N. A.; Maher, E. R.: Somatic inactivation of the VHL gene in Von Hippel–Lindau disease tumors. *Am. J. Hum. Genet.* 60: 765–771, 1997.120. Rho, Y. M.: Von Hippel–Lindau's disease: a report of 5 cases. *Canad. Med. Assoc. J.* 101: 135–142, 1969.121. Richard, S.; Croisille, L.; Yvert, J.; Casadevall, N.; Eschwege, P.; Aghakhani, N.; David, P.; Gaudric, A.; Scigalla, P.; Hermine, O.: Paradoxical secondary polycythemia in von Hippel–Lindau patient treated with anti-vascular endothelial growth factor receptor therapy. *Blood* 99: 3851–3853, 2002.122.

Richards, F. M.; Crossey, P. A.; Phipps, M. E.; Foster, K.;

Latif, F.; Evans, G.; Sampson, J.; Lerman, M. I.; Zbar, B.; Affara, N. A.; Ferguson-Smith, M. A.; Maher, E. R.: Detailed mapping of germline deletions of the von Hippel-Lindau disease tumour suppressor gene. *Hum. Molec. Genet.* 3: 595-598, 1994.123. Richards, F. M.; Maher, E. R.; Latif, F.; Phipps, M. E.; Tory, K.; Lush, M.; Crossey, P. A.; Oostra, B.; Gustavson, K. H.; Green, J.; Turner, G.; Yates, J. R. W.; Linehan, W. M.; Affara, N. A.; Lerman, M.; Zbar, B.; Ferguson-Smith, M. A.: Detailed genetic mapping of the von Hippel-Lindau disease tumour suppressor gene. *J. Med. Genet.* 30:104-107, 1993.124. Richards, F. M.; Phipps, M. E.; Latif, F.; Yao, M.; Crossey, P. A.; Foster, K.; Linehan, W. M.; Affara, N. A.; Lerman, M. I.; Zbar, B.; Ferguson-Smith, M. A.; Maher, E. R.: Mapping the von Hippel-Lindau disease tumour suppressor gene: identification of germline deletions by pulsed field gel electrophoresis. *Hum. Molec. Genet.* 2: 879-882, 1993.125. Richards, F. M.; Schofield, P. N.; Fleming, S.; Maher, E. R.: Expression of the von Hippel-Lindau disease tumour suppressor gene during human embryogenesis. *Hum. Molec. Genet.* 5: 639-644, 1996.126. Rubenstein, J. L.; Yaari, H.: von Hippel-Lindau and the genetics of astrocytoma. *J. Nat. Cancer Inst.* 86: 142-143, 1994.127. Sander, S.; Normann, T.; Mathisen,

W.: Pheochromocytoma associated with von-Hippel-Lindau's disease in a family. *Scand. J. Urol. Nephrol.* 4:259-263, 1970.128. Schechterman, L.: Lindau's disease: report of an unusual case and two additional cases in a Negro family. *Med. Ann. D.C.* 30: 64-76, 1961.129.

Schimke, R. N.; Collins, D. L.; Rothberg, P. G.: Functioning carotid paraganglioma in the von Hippel-Lindau syndrome. (Letter) *Am. J. Med. Genet.* 80: 533-534, 1998.130.

Schoenfeld, A.; Davidowitz, E. J.; Burk, R. D.: A second major native von Hippel-Lindau gene product, initiated from an internal translation start site, functions as a tumor suppressor. *Proc. Nat. Acad. Sci.* 95: 8817-8822, 1998.131.

Seizinger, B. R.; Farmer, G.; Haines, J.; Anderson, K.; Whaley, J.; Hettlich, C.; Decker, J.; Rouleau, G.; Smith, D.; Drabkin, H.; Filling-Katz, M.; Neumann, H.; Collins, D.; Hsia, E.; Green, J.; Waziri, M.; Gusella, J.; Li, F.: Isolating the gene(s) for von Hippel-Lindau disease and renal cell carcinoma. (Abstract) *Am. J. Hum. Genet.* 45(suppl.): A32, 1989.132.

Seizinger, B. R.; Rouleau, G. A.; Ozelius, L. J.; Lane, A. H.; Farmer, G. E.; Lamiell, J. M.; Haines, J.; Yuen, J. W.; Collins, D.; Majoor-Krakauer, D.; et al.: Von Hippel-Lindau disease maps to the region of chromosome 3 associated with renal cell carcinoma. *Nature*

332: 268–269, 1988.133. Seizinger, B. R.; Smith, D. I.; Filling–Katz, M. R.; Neumann, H.; Green, J. S.; Choyke, P. L.; Anderson, K. M.; Freiman, R. N.; Klauck, S. M.; Whaley, J.; Decker, H.–J. H.; Hsia, Y. E.; Collins, D.; Halperin, J.; Lamiell, J. M.; Oostra, B.; Waziri, M. H.; Gorin, M. B.; Scherer, G.; Drabkin, H. A.; Aronin, N.; Schinzel, A.; Martuza, R. L.; Gusella, J. F.; Haines, J. L.: Genetic flanking markers refined diagnostic criteria and provide insights into the genetics of Von Hippel Lindau disease. *Proc. Nat. Acad. Sci.* 88: 2864–2868, 1991.134. Sgambati, M. T.; Stolle, C.; Choyke, P. L.; Walther, M. M.; Zbar, B.; Linehan, W. M.; Glenn, G. M.: Mosaicism in von Hippel–Lindau disease: lessons from kindreds with germline mutations identified in offspring with mosaic parents. *Am. J. Hum. Genet.* 66: 84–91, 2000.135. Sharp, W. V.; Platt, R. L.: Familial pheochromocytoma: association with von–Hippel–Lindau's disease. *Angiology* 22: 141–146, 1971.136. Shokeir, M. H. K.: Von Hippel–Lindau syndrome: a report on three kindreds. *J. Med. Genet.* 7: 155–157, 1970.137. Silver, M. L.: Hereditary vascular tumors of the nervous system. *J.A.M.A.* 156: 1053–1056, 1954.138. Stolle, C.; Glenn, G.; Zbar, B.; Humphrey, J. S.; Choyke, P.; Walther, M.; Pack, S.; Hurley, K.; Audrey, C.; Klausner, R.; Linehan, W. M.: Im–

proved detection of germline mutations in the von Hippel-Lindau disease tumor suppressor gene. *Hum. Mutat.* 12: 417–423, 1998.

139. Thomas, M.; Burnside, R. M.: Von Hippel-Lindau disease. *Am.J. Ophthal.* 51: 140–146, 1961.

140. Tisherman, S. E.; Gregg, F. J.; Danowski, T. S.: Familial pheochromocytoma. *J.A.M.A.* 182: 152–156, 1962.

141. Tisherman, S. E.; Tisherman, B. G.; Tisherman, S. A.; Dunmire, S.; Levey, G. S.; Mulvihill, J. J.: Three-decade investigation of familial pheochromocytoma: an allele of von Hippel-Lindau disease? *Arch.Int. Med.* 153: 2550–2556, 1993.

142. Tory, K.; Brauch, H.; Linehan, M.; Barba, D.; Oldfield, E.; Filling-Katz, M.; Seizinger, B.; Nakamura, Y.; White, R.; Marshall, F. F.; Lerman, M. I.; Zbar, B.: Specific genetic change in tumors associated with von Hippel-Lindau disease. *J. Nat. Cancer Inst.* 81: 1097–1101, 1989.

143. Tsuda, H.; Fukushima, S.; Takahashi, M.; Hikosaka, Y.; Hayashi, K.: Familial bilateral papillary cystadenoma of the epididymis: report of three cases in siblings. *Cancer* 37: 1831–1839, 1976.

144. Tyers, M.; Willems, A. R.: One ring to rule a superfamily of E3 ubiquitin ligases. *Science* 284: 602–604, 1999.

145. Vance, J. M.; Small, K. W.; Jones, M. A.; Stajich, J. M.; Yamaoka, L. H.; Roses, A. D.; Hung, W.-Y.; Pericak-Vance, M. A.: Confir-

mation of linkage in von Hippel–Lindau disease. *Genomics* 6: 565–567, 1990.146. van der Harst, E.; de Krijger, R. R.; Dinjens, W. N. M.; Weeks, L. E.; Bonjer, H. J.; Bruining, H. A.; Lamberts, S. W. J.; Koper, J. W.: Germline mutations in the *vhl* gene in patients presenting with pheochromocytomas. *Int. J. Cancer*. 77: 337–340, 1998.147. Versteeg, R.: Aberrant methylation in cancer. (Editorial) *Am. J. Hum. Genet.* 60: 751–754, 1997.148. Vogelstein, B.: Personal Communication. Baltimore, Md. 1/6/1995.149. von Hippel, E.: Ueber eine sehr seltene Erkrankung der Netzhaut. *Albrecht von Graefes Arch. Ophthalm.* 59: 83–106, 1904.150. Webster, A. R.; Maher, E. R.; Bird, A. C.; Moore, A. T.: Risk of multisystem disease in isolated ocular angioma (haemangioblastoma). *J. Med. Genet.* 37: 62–63, 2000.151. Webster, A. R.; Richards, F. M.; MacDonald, F. E.; Moore, A. T.; Maher, E. R.: An analysis of phenotypic variation in the familial cancer syndrome von Hippel–Lindau disease: evidence for modifier effects. *Am. J. Hum. Genet.* 63: 1025–1035, 1998.152. Wells, R. A.; Reeders, S. T.; Green, J.; Johnson, G. J.; Robson, K. J. H.: Linkage studies of von Hippel–Lindau syndrome using multiple-locus hypervariable DNA probes. (Abstract) *Cytogenet. Cell Genet.* 46: 714, 1987.153. Wesolowski, D. P.; Ellwood, R.

A.; Schwab, R. E.; Farah, J.:Hippel–Lindau syndrome in identical twins. *Brit. J. Radiol.* 54:982–986, 1981.154.

Wiesener, M. S.; Seyfarth, M.; Warnecke, C.; Jurgensen, J. S.;Rosenberger, C.; Morgan, N. V.; Maher, E. R.; Frei, U.; Eckardt, K.–U.: Paraneoplastic erythrocytosis associated with an inactivating pointmutation of the von Hippel–Lindau gene in a renal cell carcinoma. *Blood* 99:3562–3565, 2002.155.

Wise, K. S.; Gibson, J. A.: Von Hippel–Lindau's disease andphaeochromocytoma. *Brit. Med. J.* 1: 441, 1971.156.

Wyburn–Mason, R.: Arteriovenous aneurysm of mid–brain and retina, facial naevi and mental changes. *Brain* 66: 163–203, 1943.157.

Zatyka, M.; da Silva, N. F.; Clifford, S. C.; Morris, M. R.;Wiesener, M. S.; Eckardt, K.–U.; Houlston, R. S.; Richards, F. M.;Latif, F.; Maher, E. R.: Identification of cyclin D1 and other noveltargets for the von Hippel–Lindau tumor suppressor gene by expressionarray analysis and investigation of cyclin D1 genotype as a modifierin von Hippel–Lindau disease. *Cancer Res.* 62: 3803–3811, 2002.158.

Zbar, B.; Kishida, T.; Chen, F.; Schmidt, L.; Maher, E. R.; Richards,F. M.; Crossey, P. A.; Webster, A. R.; Affara, N. A.; Ferguson–Smith,M. A.; Brauch, H.; Glavac, D.; and 14 others: Germline mutation–sin the Von Hippel–Lindau disease (VHL) gene in families

from North America, Europe, and Japan. *Hum. Mutat.* 8: 348–357, 1996.159. Zhuang, Z.; Emmert–Buck, M. R.; Roth, M. J.; Gnarr, J.; Linehan, W. M.; Liotta, L. A.; Luben–sky, I. A.: Von Hippel–Lindau disease gene deletion detected in microdissected sporadic human colon carcinomaspecimens. *Hum. Path.* 27: 152–156, 1996.

[44288] 17840. Thomas, J. T.; Kilpatrick, M. W.; Lin, K.; Erlacher, L.; Lembessis, P.; Costa, T.; Tsipouras, P.; Luyten, F. P.: Dis–ruption of human limb morphogenesis by a dominant neg–ative mutation in CDMP1. *Nature Genet.* 58–64, 1997.

[44289] 17841. Devlin, R. H.; Deeb, S.; Brunzell, J.; Hayden, M. R.: Partial gene duplication involving exon–Alu interchange results in lipoprotein lipase deficiency. *Am. J. Hum. Genet.* 46: 112–119, 1990.

[44290] 17842. Dichek, H. L.; Fojo, S. S.; Beg, O. U.; Skarlatos, S. I.; Brunzell, J. D.; Cutler, G. B., Jr.; Brewer, H. B., Jr.: Identifi–cation of 2 separate allelic mutations in the lipoprotein li–pase gene of a patient with the familial hyperchylomi–cronemia syndrome. *J. Biol. Chem.* 266:473–477, 1991.

[44291] 17843. Eckel, R. H.: Lipoprotein lipase: a multifunctional enzyme relevant to common metabolic diseases. *New Eng. J. Med.* 320: 1060–1068, 1989.

[44292] 17844. Emi, M.; Hata, A.; Robertson, M.; Iverius, P.–H.;

Hegele, R.;Lalouel, J.-M.: Lipoprotein lipase deficiency resulting from a nonsensemutation in exon 3 of the lipoprotein lipase gene. *Am. J. Hum. Genet.* 47:107–111, 1990.

[44293] 17845.Emi, M.; Wilson, D. E.; Iverius, P.-H.; Wu, L.; Hata, A.; Hegele,R.; Williams, R. R.; Lalouel, J.-M.: Missense mutation (gly-to-glu188)of human lipoprotein lipase imparting functional deficiency. *J. Biol.Chem.* 265: 5910–5916, 1990.

[44294] 17846.Faustinella, F.; Chang, A.; Van Biervliet, J. P.; Rosseneu, M.;Vinaimont, N.; Smith, L. C.; Chen, S.-H.; Chan, L.: Catalytic triadresidue mutation (asp156-to-gly) causing familial lipoprotein lipasedeficiency: co-inheritance with a nonsense mutation (ser447-to-ter)in a Turkish family. *J. Biol. Chem.* 266: 14418–14424, 1991.

[44295] 17847.Feoli-Fonseca, J. C.; Levy, E.; Godard, M.; Lambert, M.: Familiallipoprotein lipase deficiency in infancy: clinical, biochemical, andmolecular study. *J. Pediat.* 133: 417–423, 1998.

[44296] 17848.Fisher, R. M.; Humphries, S. E.; Talmud, P. J.: Common variationin the lipoprotein lipase gene: effects on plasma lipids and riskof atherosclerosis. *Atherosclerosis* 135: 145–159, 1997.

- [44297] 17849. Franklin, S. M.: Splenomegaly with lipaemia. Proc. Roy. Soc. Med. 30: 711, 1937.
- [44298] 17850. Funke, H.; Klug, J.; Assmann, G.: Hind III RFLP in the lipoprotein lipase gene, (LPL). Nucleic Acids Res. 15: 9102, 1987.
- [44299] 17851. Funke, H.; Reckwerth, A.; Stapenhorst, D.; Schulze Beiering, M.; Jansen, M.; Assmann, G.: Bst NI (Eco RII) RFLP in the lipoprotein lipase gene (LPL). Nucleic Acids Res. 16: 2741, 1988.
- [44300] 17852. Gagne, C.; Brun, L. D.; Julien, P.; Moorjani, S.; Lupien, P. J.: Primary lipoprotein lipase activity deficiency: clinical investigation of a French Canadian population. Canad. Med. Assoc. J. 140: 405–411, 1989.
- [44301] 17853. Gilbert, B.; Rouis, M.; Griglio, S.; de Lumley, L.; Laplaud, P.–M.: Lipoprotein lipase (LPL) deficiency: a new patient homozygote for the preponderant mutation gly88–to–glu in the human LPL gene and review of reported mutations: 75% are clustered in exons 5 and 6. Ann. Genet. 44:25–32, 2001.
- [44302] 17854. Ginzinger, D. G.; Lewis, M. E. S.; Ma, Y.; Jones, B. R.; Liu, G.; Jones, S. D.; Hayden, M. R.: A mutation in the lipoprotein lipase gene is the molecular basis of chylomicronemia in a colony of domestic cats. J. Clin. Invest. 97:

1257–1266, 1996.

[44303] 17855. Gotoda, T.; Yamada, N.; Kawamura, M.; Kozaki, K.; Mori, N.; Ishibashi, S.; Shimano, H.; Takaku, F.; Yazaki, Y.; Furuichi, Y.; Murase, T.: Heterogeneous mutations in the human lipoprotein lipase gene in patients with familial lipoprotein lipase deficiency. *J. Clin. Invest.*

88:1856–1864, 1991.

[44304] 17856. Gotoda, T.; Yamada, N.; Murase, T.; Miyake, S.; Murakami, R.; Kawamura, M.; Kozaki, K.; Mori, N.; Shimano, H.; Shimada, M.; Yazaki, Y.: A newly identified null allelic mutation in the human lipoprotein lipase (LPL) gene of a compound heterozygote with familial LPL deficiency. *Biochim. Biophys. Acta* 1138: 353–356, 1992.

[44305] 17857. Hata, A.; Emi, M.; Luc, G.; Basdevant, A.; Gamber, P.; Iversen, P.-H.; Lalouel, J.-M.: Compound heterozygote for lipoprotein lipase deficiency: ser-to-thr(244) and transition in 3-prime splice site of intron 2 (AG-to-AA) in the lipoprotein lipase gene. *Am. J. Hum. Genet.* 47: 721–726, 1990.

[44306] 17858. Hata, A.; Ridinger, D. N.; Sutherland, S. D.; Emi, M.; Kwong, L. K.; Shuhua, J.; Lubbers, A.; Guy-Grand, B.; Basdevant, A.; Iversen, P. H.; Wilson, D. E.; Lalouel, J.-M.: Missense mutations in exon 5 of the human lipoprotein lipase

gene: inactivation correlates with loss of dimerization. J. Biol. Chem. 267: 20132–20139, 1992.

- [44307] 17859. Haubenwallner, S.; Horl, G.; Shachter, N. S.; Presta, E.; Fried, S. K.; Hofler, G.; Kostner, G. M.; Breslow, J. L.; Zechner, R.: A novel missense mutation in the gene for lipoprotein lipase resulting in a highly conservative amino acid substitution (asp180-to-glu) causes familial chylomicronemia (type I hyperlipoproteinemia). Genomics 18:392–396, 1993.
- [44308] 17860. Havel, R. J.; Gordon, R. S.: Idiopathic hyperlipemia: metabolic studies in an affected family. J. Clin. Invest. 39: 1777–1790, 1960.
- [44309] 17861. Heaney, A. P.; Sharer, N.; Rameh, B.; Braganza, J. M.; Durrington, P. N.: Prevention of recurrent pancreatitis in familial lipoprotein lipase deficiency with high-dose antioxidant therapy. J. Clin. Endocr. Metab. 84: 1203–1205, 1999.
- [44310] 17862. Heinzmann, C.; Ladas, J.; Antonarakis, S.; Kirchgessner, T.; Schotz, M.; Lusi, A. J.: RFLP for the human lipoprotein lipase (LPL) gene: HindIII. Nucleic Acids Res. 15: 6763, 1987.
- [44311] 17863. Heinzmann, C.; Kirchgessner, T.; Kwiterovich, P. O.; Ladas, J. A.; Derby, C.; Antonarakis, S. E.; Lusi, A. J.: DNA

polymorphismhaplotypes of the human lipoprotein lipase gene: possible associationwith high density lipoprotein levels. Hum. Genet. 86: 578–584, 1991.

- [44312] 17864.Henderson, H.; Ma, Y.; Kastelein, J.; Roederer, G.; Julien, P.;Brunzell, J.; Hayden, M. R.: Identification of the molecular defectsunderlying chylomicronemia in the majority of 75 separate probandswith LPL deficiency.(Abstract) Clin. Res. 39: 336A, 1991.
- [44313] 17865.Henderson, H. E.; Bijvoet, S. M.; Mannens, M. A. M. M.; Bruin,T.; Erkelens, D. W.; Hayden, M. R.; Kastelein, J. J. P.: Ile225–to–thrloop mutation in the lipoprotein lipase (LPL) gene is a de novo event. Am.J. Med. Genet. 78: 313–316, 1998.
- [44314] 17866.Sjalander, A.; Birgander, R.; Rannug, A.; Alexandrie, A.–K.; Tornling,G.; Beckman, G.: Association between the p21 codon 31A1 (arg) alleleand lung cancer. Hum. Hered. 46: 221–225, 1996.
- [44315] 17867.Fryns, J. P.; van den Berghe, K.; van Assche, A.; van den Berghe,H.: Prenatal diagnosis of campomelic dwarfism. Clin. Genet. 19:199–201, 1981.
- [44316] 17868.Mellows, H. J.; Pryse–Davies, J.; Bennett, M. J.; Carter, C. O.: The camptomelic syndrome in two female siblings. Clin. Genet. 18:137–141, 1980.

- [44317] 17869. Millauer, B.; Shawver, L. K.; Plate, K. H.; Risau, W.; Ullrich, A.: Glioblastoma growth inhibited in vivo by a dominant-negative Flk-1 mutant. *Nature* 367: 576–579, 1994.
- [44318] 17870. Cho, K. R.; Oliner, J. D.; Simons, J. W.; Hedrick, L.; Fearon, E. R.; Preisinger, A. C.; Hedge, P.; Silverman, G. A.; Vogelstein, B.: The DCC gene: structural analysis and mutations in colorectal carcinomas. *Genomics* 19: 525–531, 1994.
- [44319] 17871. Fazeli, A.; Dickinson, S. L.; Hermiston, M. L.; Tighe, R. V.; Steen, R. G.; Small, C. G.; Stoeckli, E. T.; Keino-Masu, K.; Masu, M.; Rayburn, H.; Simons, J.; Bronson, R. T.; Gordon, J. I.; Tessier-Lavigne, M.; Weinberg, R. A.: Phenotype of mice lacking functional Deleted in colorectal cancer (Dcc) gene. *Nature* 386: 796–804, 1997.
- [44320] 17872. Fearon, E. R.; Cho, K. R.; Nigro, J. M.; Kern, S. E.; Simons, J. W.; Ruppert, J. M.; Hamilton, S. R.; Preisinger, A. C.; Thomas, G.; Kinzler, K. W.; Vogelstein, B.: Identification of a chromosome 18q gene that is altered in colorectal cancers. *Science* 247: 49–56, 1990.
- [44321] 17873. Gotley, D. C.; Reeder, J. A.; Fawcett, J.; Walsh, M. D.; Bates, P.; Simmons, D. L.; Antalis, T. M.: The deleted in colon cancer (DCC) gene is consistently expressed in col-

orectal cancers and metastases. *Oncogene* 13:787–795, 1996.

[44322] 17874.Hohne, M. W.; Halatsch, M.–E.; Kahl, G. F.; Weinel, R. J.: Frequent loss of expression of the potential tumor suppressor gene DCC in ductal pancreatic adenocarcinoma. *Cancer Res.* 52: 2616–2619, 1992.

[44323] 17875.Jen, J.; Kim, H.; Piantadosi, S.; Liu, Z.–F.; Levitt, R. C.; Sistonen, P.; Kinzler, K. W.; Vogelstein, B.; Hamilton, S. R.: Allelic loss of chromosome 18q and prognosis in colorectal cancer. *New Eng. J. Med.* 331: 213–221, 1994.

[44324] 17876.Keino–Masu, K.; Masu, M.; Hinck, L.; Leonardo, E. D.; Chan, S.S.–Y.; Culotti, J. G.; Tessier–Lavigne, M.: Deleted in colorectal cancer (DCC) encodes a netrin receptor. *Cell* 87: 175–185, 1996.

[44325] 17877.Keynes, R.; Cook, G. M. W.: Axon guidance molecules. *Cell* 83:161–169, 1995.

[44326] 17878.Kolodziej, P. A.; Timpe, L. C.; Mitchell, K. J.; Fried, S. R.; Goodman, C. S.; Jan, L. Y.; Jan, Y. N.: Frazzled encodes a Drosophila member of the DCC immunoglobulin subfamily and is required for CNS and motor axon guidance. *Cell* 87: 197–204, 1996.

[44327] 17879.Maesawa, C.; Tamura, G.; Ogasawara, S.; Suzuki, Y.; Sakata, K.; Sugimura, J.; Nishizuka, S.; Sato, N.; Ishida,

K.; Saito, K.; Satodate, R.: Loss of heterozygosity at the DCC gene locus is not crucial for the acquisition of metastatic potential in oesophageal squamous cell carcinoma. *Europ. J. Cancer*. 32A: 896–898, 1996.

[44328] 17880. Mehlen, P.; Rabizadeh, S.; Snipas, S. J.; Assa-Munt, N.; Salvesen, G. S.; Bredesen, D. E.: The DCC gene product induces apoptosis by a mechanism requiring receptor proteolysis. *Nature* 395: 801–804, 1998.

[44329] 17881. Miyake, S.; Nagai, K.; Yoshino, K.; Oto, M.; Endo, M.; Yuasa, Y.: Point mutations and allelic deletion of tumor suppressor gene DCC in human esophageal squamous cell carcinomas and their relation to metastasis. *Cancer Res.* 54: 3007–3010, 1994.

[44330] 17882. Nigro, J. M.; Cho, K. R.; Fearon, E. R.; Kern, S. E.; Ruppert, J. M.; Oliner, J. D.; Kinzler, K. W.; Vogelstein, B.: Scrambled exons. *Cell* 64:607–613, 1991.

[44331] 17883. Shibata, D.; Reale, M. A.; Lavin, P.; Silverman, M.; Fearon, E. R.; Steele, G., Jr.; Jessup, J. M.; Loda, M.; Summerhayes, I. C.: The DCC protein and prognosis in colorectal cancer. *New Eng. J. Med.* 335:1727–1732, 1996.

[44332] 17884. Stein, E.; Tessier-Lavigne, M.: Hierarchical organization of guidance receptors: silencing of netrin attraction by Slit through a Robo/DCC receptor complex. *Science*

291: 1928–1938, 2001.

- [44333] 17885.Tanaka, K.; Oshimura, M.; Kikuchi, R.; Seki, M.; Hayashi, T.;Miyaki, M.: Suppression of tumorigenicity in human colon carcinomacells by introduction of normal chromosome 5 or 18. *Nature* 349:340–342, 1991.
- [44334] 17886.Uchino, S.; Tsuda, H.; Noguchi, M.; Yokota, J.; Terada, M.; Saito,T.; Kobayashi, M.; Sugimura, T.; Hirohashi, S.: Frequent loss ofheterozygosity at the DCC locus in gastric cancer. *Cancer Res.* 52:3099–3102, 1992.
- [44335] 17887.Vogelstein, B.: Personal Communication. Baltimore, Md. 11/30/1995.
- [44336] 17888.Zetter, B. R.: Adhesion molecules in tumor metastasis. *Semin.Cancer Biol.* 4: 219–229, 1993.
- [44337] 17889.Devor, E. J.; Grandy, D. K.; Civelli, O.; Litt, M.; Burgess, A.K.; Isenberg, K. E.; van de Wetering, B. J. M.; Oostra, B.: Geneticlinkage is excluded for the D2–dopamine receptor lambda–HD2G1 andflanking loci on chromosome 11q22–q23 in Tourette syndrome. *Hum.Hered.* 40: 105–108, 1990.
- [44338] 17890.Freemantle, S. J.; Taylor, S. M.; Krystal, G.; Moran, R. G.: Upstreamorganization of and multiple transcripts from the human folylpoly–gamma–glutamatesynthetase gene. *J. Biol. Chem.* 270: 9579–9584, 1995.

- [44339] 17891. Garrow, T. A.; Admon, A.; Shane, B.: Expression cloning of a human cDNA encoding folylpoly(γ -glutamate) synthetase and determination of its primary structure. *Proc. Nat. Acad. Sci.* 89: 9151–9155, 1992.
- [44340] 17892. Jones, C.; Kao, F.-T.: Regional mapping of the folylpolyglutamate synthetase gene (FPGS) to 9cen–q34. (Abstract) *Cytogenet. Cell Genet.* 37:499–500, 1984.
- [44341] 17893. Jones, C.; Kao, F.-T.; Taylor, R. T.: Chromosomal assignment of the gene for folylpolyglutamate synthetase to human chromosome 9. *Cytogenet. Cell Genet.* 28: 181–194, 1980.
- [44342] 17894. Jones, C.; Kao, F. T.: Assignment of the human gene complementing the auxotrophic marker GAT⁻ to chromosome 9. (Abstract) *Cytogenet. Cell Genet.* 25: 168, 1979.
- [44343] 17895. Kao, F. T.; Puck, T. T.: Genetics of somatic mammalian cells. VII. Induction and isolation of nutritional mutants in Chinese hamster cells. *Proc. Nat. Acad. Sci.* 60: 1275–1281, 1968.
- [44344] 17896. Sussman, D. J.; Milman, G.; Shane, B.: Characterization of human folylpolyglutamate synthetase expressed in Chinese hamster ovary cells. *Somat. Cell Molec. Genet.*

12: 531–540, 1986.

- [44345] 17897. Taylor, R. T.; Hanna, M. L.: Folate-dependent enzymes in cultured Chinese hamster cells: folylpolyglutamate synthetase and its absence in mutants auxotrophic for glycine, adenosine and thymidine. *Arch. Biochem. Biophys.* 181: 331–344, 1977.
- [44346] 17898. Taylor, S. M.; Freemantle, S. J.; Moran, R. G.: Structural organization of the human folylpolygamma-glutamate synthetase gene: evidence for a single genomic locus. *Cancer Res.* 55: 6030–6034, 1995.
- [44347] 17899. Matsuo, K.; Owens, J. M.; Tonko, M.; Elliott, C.; Chambers, T. J.; Wagner, E. F.: Fos1 is a transcriptional target of c-Fos during osteoclast differentiation. *Nature Genet.* 24: 184–187, 2000.
- [44348] 17900. Buters, J. T. M.; Tang, B.-K.; Pineau, T.; Gelboin, H. V.; Kimura, S.; Gonzalez, F. J.: Role of CYP1A2 in caffeine pharmacokinetics and metabolism: studies using mice deficient in CYP1A2. *Pharmacogenetics* 6: 291–296, 1996.
- [44349] 17901. Butler, M. A.; Iwasaki, M.; Guengerich, F. P.; Kadlubar, F. F.: Human cytochrome P-450(PA) (P-450IA2), the phenacetin O-deethylase, is primarily responsible for the hepatic 3-demethylation of caffeine and N-oxidation of carcinogenic arylamines. *Proc. Nat. Acad. Sci.*

86:7696–7700, 1989.

- [44350] 17902.Christiansen, L.; Bygum, A.; Jensen, A.; Thomsen, K.; Brandrup,F.; Horder, M.; Petersen, N. E.: Association between CYP1A2 polymorphismand susceptibility to porphyria cutanea tarda. Hum. Genet. 107:612–614, 2000.
- [44351] 17903.Devonshire, H. W.; Kong, I.; Cooper, M.; Sloan, T. P.; Idle, J.R.; Smith, R. L.: The contribution of genetically determined oxidationstatus to inter–individual variation in phenacetin disposition. Brit.J. Clin. Pharm. 16: 157–166, 1983.
- [44352] 17904.Ikeya, K.; Jaiswal, A. K.; Owens, R. A.; Jones, J. E.; Nebert,D. W.; Kimura, S.: Human CYP1A2: sequence, gene structure, comparisonwith the mouse and rat orthologous gene, and differences in liver1A2 mRNA expression. Molec. Endocr. 3: 1399–1408, 1989.
- [44353] 17905.Jaiswal, A. K.; Nebert, D. W.; McBride, O. W.; Gonzalez, F. J.: Human P(3)450: cDNA and complete protein sequence, repetitive Alusequences in the 3–prime non–translated region, and localization ofgene to chromosome 15. J. Exp. Path. 3: 1–17, 1987.
- [44354] 17906.Liang, H.–C. L.; Li, H.; McKinnon, R. A.; Duffy, J. J.; Potter,S. S.; Puga, A.; Nebert, D. W.: Cyp1a2(–/–) null mutant mice developnormally but show deficient drug

metabolism. Proc. Nat. Acad. Sci. 93:1671–1676, 1996.

[44355] 17907.Nebert, D. W.: Personal Communication. Bethesda, Md. 2/3/1988.

[44356] 17908.Sesardic, D.; Boobis, A. R.; Edwards, R. J.; Davies, D. S.: A form of cytochrome P450 in man, orthologous to form d in the rat, catalyses the O-deethylation of phenacetin and is inducible by cigarette smoking. Brit. J. Clin. Pharm. 26: 363–372, 1988.

[44357] 17909.Tantcheva-Poor, I.; Zaigler, M.; Rietbrock, S.; Fuhr, U.: Estimation of cytochrome P-450 CYP1A2 activity in 863 healthy Caucasians using a saliva-based caffeine test. Pharmacogenetics 9: 131–144, 1999.

[44358] 17910.Wooding, S. P.; Watkins, W. S.; Bamshad, M. J.; Dunn, D. M.; Weiss, R. B.; Jorde, L. B.: DNA sequence variation in a 3.7-kb noncoding sequence 5-prime of the CYP1A2 gene: implications for human population history and natural selection. Am. J. Hum. Genet. 71: 528–542, 2002.

[44359] 17911.Bale, A. E.; Mitchell, A. L.; Gonzalez, F. J.; McBride, O. W.: Localization of CYP2F1 by multipoint linkage analysis and pulsed-field gel electrophoresis. Genomics 10: 284–286, 1991.

[44360] 17912.Mitchell, A. L.; Bale, A. E.; Gonzalez, F.; McBride, O.

W.: Mapping of cytochrome p450 IIA, IIB, and IIF subfamilies on chromosome 19 by linkage analysis. (Abstract) Cytogenet. Cell Genet. 51: 1045, 1989.

- [44361] 17913. Namburo, P. T.; Kimura, S.; McBride, O. W.; Kozak, C. A.; Gelboin, H. V.; Gonzalez, F. J.: The human CYP2F gene subfamily: identification of a cDNA encoding a new cytochrome P450, cDNA-directed expression, and chromosome mapping. Biochemistry 29: 5491–5499, 1990.
- [44362] 17914. Imaoka, S.; Yoneda, Y.; Sugimoto, T.; Hiroi, T.; Yamamoto, K.; Nakatani, T.; Funae, Y.: CYP4B1 is a possible risk factor for bladder cancer in humans. Biochem. Biophys. Res. Commun. 277: 776–780, 2000.
- [44363] 17915. Namburo, P. T.; Gonzalez, F. J.; McBride, O. W.; Gelboin, H. V.; Kimura, S.: Identification of a new P450 expressed in human lung: complete cDNA sequence, cDNA-directed expression, and chromosome mapping. Biochemistry 28: 8060–8066, 1989.
- [44364] 17916. Thum, T.; Borlak, J.: Gene expression in distinct regions of the heart. Lancet 355: 979–983, 2000.
- [44365] 17917. Xu, L.; Xia, J.; Jiang, H.; Zhou, J.; Li, H.; Wang, D.; Pan, Q.; Long, Z.; Fan, C.; Deng, H.-X.: Mutation analysis of hereditary multiple exostoses in the Chinese. Hum. Genet.

105: 45–50, 1999.

- [44366] 17918.Joutel, A.; Favrole, P.; Labauge, P.; Chabriat, H.; Le-scoat, C.;Andreux, F.; Domenga, V.; Cecillon, M.; Vahedi, K.; Ducros, A.; Cave-Riant,F.; Bousser, M. G.; Tournier-Lasserre, E.: Skin biopsy immunostainingwith a Notch3 monoclonal antibody for CADASIL diagnosis. *Lancet* 358:2049–2051, 2001.
- [44367] 17919.Barinaga, M.: An intriguing new lead on Hunting-ton's disease. *Science* 271:1233–1234, 1996.
- [44368] 17920.Burke, J. R.; Enghild, J. J.; Martin, M. E.; Jou, Y.-S.; Myers,R. M.; Roses, A. D.; Vance, J. M.; Strittmatter, W. J.: Huntingtinand DRPLA proteins selectively interact with the enzyme GAPDH. *NatureMed.* 2: 347–350, 1996.
- [44369] 17921.Eubanks, J. H.; Djabali, M.; Selleri, L.; Grandy, D. K.; Civelli,O.; McElligott, D. L.; Evans, G. A.: Structure and linkage of theD2 dopamine receptor and neural cell adhe-sion molecule genes on humanchromosome 11q23. *Ge-nomics* 14: 1010–1018, 1992.
- [44370] 17922.Gejman, P. V.; Ram, A.; Gelernter, J.; Friedman, E.; Cao, Q.;Pickar, D.; Blum, K.; Noble, E. P.; Kranzler, H. R.; O'Malley, S.;Hamer, D. H.; Whitsitt, F.; Rao, P.; DeLisi, L. E.; Virkkunen, M.;Linnoila, M.; Goldman, D.; Gershon, E. S.: No structural mutationin the dopamine D2 receptor gene

in alcoholism or schizophrenia: analysis using denaturing gradient gel electrophoresis. J.A.M.A. 271: 204–208, 1994.

[44371] 17923. Gelernter, J.; Grandy, D. K.; Bunzow, J.; Pakstis, A.

J.; Civelli, O.; Retief, A. E.; Litt, M.; Kidd, K. K.: D(2)

dopamine receptor locus (probe hD2G1) maps close to

D11S29 (probe L7) and is also linked to PBGD (probe

PBGD0.0) and D11S84 (probe p-2-7-ID6) on 11q.

(Abstract) Cytogenet. Cell Genet. 51: 1002 only, 1989.

[44372] 17924. Gelernter, J.; Pakstis, A. J.; Grandy, D.; Litt, M.;

Retief, A. E.; Kennedy, J. L.; Hing-Loh, A.; Schoolfield, G.;

Civelli, O.; Kidd, K. K.: Linkage map of eight human chro-

mosome 11q markers, including DRD2, spanning 60 cM.

Cytogenet. Cell Genet. 60: 26–28, 1992.

[44373] 17925. Gelernter, J.; Pakstis, A. J.; Pauls, D. L.; Kurlan, R.;

Gancher, S. T.; Civelli, O.; Grandy, D.; Kidd, K. K.: Gilles de

la Tourette syndrome is not linked to D2-dopamine recep-

tor. Arch. Gen. Psychiat. 47: 1073–1077, 1990.

[44374] 17926. Giros, B.; Sokoloff, P.; Martres, M.-P.; Riou, J.-F.;

Emorine, L. J.; Schwartz, J.-C.: Alternative splicing directs

the expression of two D(2) dopamine receptor isoforms.

Nature 342: 923–926, 1989.

[44375] 17927. Grandy, D. K.; Litt, M.; Allen, L.; Bunzow, J. R.; Mar-

chionni, M.; Makam, H.; Reed, L.; Magenis, R. E.; Civelli, O.:

The human dopamine D(2) receptor gene is located on chromosome 11 at q22–q23 and identifies a TaqI RFLP. *Am. J. Hum. Genet.* 45: 778–785, 1989.

- [44376] 17928. Grandy, D. K.; Marchionni, M. A.; Makam, H.; Stofko, R. E.; Alfano, M.; Frothingham, L.; Fischer, J. B.; Burke–Howie, K. J.; Bunzow, J. R.; Server, A. C.; Civelli, O.: Cloning of the cDNA and gene for a human D2 dopamine receptor. *Proc. Nat. Acad. Sci.* 86: 9762–9766, 1989.
- [44377] 17929. Klein, C.; Brin, M. F.; Kramer, P.; Sena–Esteves, M.; de Leon, D.; Doheny, D.; Bressman, S.; Fahn, S.; Breakefield, X. O.; Ozelius, L. J.: Association of a missense change in the D2 dopamine receptor with myoclonus dystonia. *Proc. Nat. Acad. Sci.* 96: 5173–5176, 1999.
- [44378] 17930. Maldonado, R.; Salardi, A.; Valverde, O.; Samad, T. A.; Roques, B. P.; Borrelli, E.: Absence of opiate rewarding effects in mice lacking dopamine D2 receptors. *Nature* 388: 586–589, 1997.
- [44379] 17931. Milligan, G.: Receptors as kissing cousins. *Science* 288: 65–67, 2000.
- [44380] 17932. Monsma, F. J., Jr.; McVittie, L. D.; Gerfen, C. R.; Mahan, L. C.; Sibley, D. R.: Multiple D(2) dopamine receptors produced by alternative RNA splicing. *Nature* 342: 926–929, 1989.

- [44381] 17933.Oliveri, R. L.; Annesi, G.; Zappia, M.; Civitelli, D.; Montesanti,R.; Branca, D.; Nicoletti, G.; Spadafora, P.; Pasqua, A. A.; Cittadella,R.; Andreoli, V.; Gambardella, A.; Aguglia, U.; Quattrone, A.: DopamineD2 receptor gene polymorphism and the risk of levodopa-induced dyskinesias in PD. *Neurology* 53: 1425–1430, 1999.
- [44382] 17934.Rocheville, M.; Lange, D. C.; Kumar, U.; Patel, S. C.; Patel,R. C.; Patel, Y. C.: Receptors for dopamine and somatostatin: formationof hetero-oligomers with enhanced functional activity. *Science* 288:154–157, 2000.
- [44383] 17935.Smith, M.; Wasmuth, J.; McPherson, J. D.; Wagner, C.; Grandy,D.; Civelli, O.; Potkin, S.; Litt, M.: Cosegregation of an 11q22.3–9p22translocation with affective disorder: proximity of the dopamine D2receptor gene relative to the translocation breakpoint. (Abstract) *Am.J. Hum. Genet.* 45 (suppl.): A220 only, 1989.
- [44384] 17936.St Clair, D.; Blackwood, D.; Muir, W.; Carothers, A.; Walker,M.; Spowart, G.; Gosden, C.; Evans, H. J.: Association within a familyof a balanced autosomal translocation with major mental illness. *Lancet* 336:13–16, 1990.
- [44385] 17937.Suarez, B. K.; Parsian, A.; Hampe, C. L.; Todd, R. D.; Reich,T.; Cloninger, C. R.: Linkage disequilibria at the D2 dopamine receptorlocus (DRD2) in alcoholics and con-

trols. Genomics 19: 12–20, 1994.

- [44386] 17938.Usiello, A.; Baik, J.–H.; Rouge–Pont, F.; Picetti, R.; Dierich,A.; LeMeur, M.; Piazza, P. V.; Borrelli, E.: Distinct functions ofthe two isoforms of dopamine D2 receptors. Nature 408: 199–203,2000.
- [44387] 17939.Wang, J.; Liu, Z.–L.; Chen, B.: Association study of dopamineD2, D3 receptor gene polymorphisms with motor fluctuations in PD. Neurology 56:1757–1759, 2001.
- [44388] 17940.Accili, D.; Fishburn, C. S.; Drago, J.; Steiner, H.; Lachowicz,J. E.; Park, B.–H.; Gauda, E. B.; Lee, E. J.; Cool, M. H.; Sibley,D. R.; Gerfen, C. R.; Westphal, H.; Fuchs, S.: A targeted mutationof the D3 dopamine receptor gene is associated with hyperactivityin mice. Proc. Nat. Acad. Sci. 93: 1945–1949, 1996.
- [44389] 17941.Asico, L. D.; Ladines, C.; Fuchs, S.; Accili, D.; Carey, R. M.;Semeraro, C.; Pocchiari, F.; Felder, R. A.; Eisner, G. M.; Jose, P.A.: Disruption of the dopamine D3 receptor gene produces renin–dependenthypertension. J. Clin. Invest. 102: 493–498, 1998.
- [44390] 17942.Crocq, M.–A.; Mant, R.; Asherson, P.; Williams, J.; Hode, Y.; Mayerova,A.; Collier, D.; Lannfelt, L.; Sokoloff, P.; Schwartz, J.–C.; Gill,M.; Macher, J.–P.; McGuffin, P.; Owen, M. J.: Association between schizophrenia and ho–

mozygosity at the dopamine D3 receptor gene. J.Med. Genet. 29: 858–860, 1992.

- [44391] 17943.Guillin, O.; Diaz, J.; Carroll, P.; Griffon, N.; Schwartz, J.–C.;Sokoloff, P.: BDNF controls dopamine D3 receptor expression and triggersbehavioural sensitization. Nature 411: 86–89, 2001.
- [44392] 17944.Ilani, T.; Ben–Shachar, D.; Strous, R. D.; Mazor, M.; Sheinkman,A.; Kotler, M.; Fuchs, S.: A peripheral marker for schizophrenia:increased levels of D3 dopamine receptor mRNA in blood lymphocytes. Proc.Nat. Acad. Sci. 98: 625–628, 2001.
- [44393] 17945.Le Coniat, M.; Sokoloff, P.; Hillion, J.; Martres, M.–P.; Giros,B.; Pilon, C.; Schwartz, J.–C.; Berger, R.: Chromosomal localizationof the human D–3 dopamine receptor gene. Hum. Genet. 87: 618–620,1991.
- [44394] 17946.Nanko, S.; Sasaki, T.; Fukuda, R.; Hattori, M.; Dai, X. Y.; Kazamatsuri,H.; Kuwata, S.; Juji, T.; Gill, M.: A study of the association betweenschizophrenia and the dopamine D3 receptor gene. Hum. Genet. 92:336–338, 1993.
- [44395] 17947.Nothen, M. M.; Cichon, S.; Propping, P.; Fimmers, R.; Schwab, S.G.; Wildenauer, D. B.: Excess of homozygosity at the dopamine D3receptor gene in schizophrenia not

confirmed. (Letter) J. Med. Genet. 30:708–712, 1993.

- [44396] 17948. Bevilacqua, M.; Butcher, E.; Furie, B.; Furie, B.; Gal-
latin, M.; Gimbrone, M.; Harlan, J.; Kishimoto, K.; Lasky, L.;
McEver, R.; Paulson, J.; Rosen, S.; Seed, B.; Siegelman, M.;
Springer, T.; Stoolman, L.; Tedder, T.; Varki, A.; Wagner,
D.; Weissman, I.; Zimmerman, G.: Selectins: a family of ad-
hesion receptors. (Letter) Cell 67: 233 only, 1991.
- [44397] 17949. Bevilacqua, M. P.; Stengelin, S.; Gimbrone, M. A.,
Jr.; Seed, B.: Endothelial leukocyte adhesion molecule 1:
an inducible receptor for neutrophils related to comple-
ment regulatory proteins and lectins. Science
243:1160–1165, 1989.
- [44398] 17950. Collins, T.; Williams, A.; Johnston, G. I.; Kim, J.;
Eddy, R.; Shows, T.; Gimbrone, M. A., Jr.; Bevilacqua, M. P.:
Structure and chromosomal location of the gene for en-
dothelial-leukocyte adhesion molecule 1. J. Biol. Chem.
266: 2466–2473, 1991.
- [44399] 17951. DeLisser, H. M.; Christofidou-Solomidou, M.; Sun,
J.; Nakada, M. T.; Sullivan, K. E.: Loss of endothelial surface
expression of E-selectin in a patient with recurrent infec-
tions. Blood 94: 884–894, 1999.
- [44400] 17952. Wang, N.; Chintala, S. K.; Fini, M. E.; Schuman, J. S.:
Activation of a tissue-specific stress response in the aque-

ous outflow pathway of the eye defines the glaucoma disease phenotype. *Nature Med.* 7:304–309, 2001.

- [44401] 17953. Watson, M. L.; Kingsmore, S. F.; Johnston, G. I.; Siegelman, M. H.; Le Beau, M. M.; Lemons, R. S.; Bora, N. S.; Howard, T. A.; Weissman, I. L.; McEver, R. P.; Seldin, M. F.: Genomic organization of the selectin family of leukocyte adhesion molecules on human and mouse chromosome 1. *J. Exp. Med.* 172: 263–272, 1990.
- [44402] 17954. Wenzel, K.; Felix, S.; Kleber, F. X.; Brachold, R.; Menke, T.; Schattke, S.; Schulte, K. L.; Glaser, C.; Rohde, K.; Baumann, G.; Speer, A.: E-selectin polymorphism and atherosclerosis: an association study. *Hum. Molec. Genet.* 3: 1935–1937, 1994.
- [44403] 17955. Zheng, F.; Chevalier, J. A.; Zhang, L. Q.; Virgil, D.; Ye, S. Q.; Kwiterovich, P. O.: An HphI polymorphism in the E-selectin gene is associated with premature coronary artery disease. *Clin. Genet.* 59:58–64, 2001.
- [44404] 17956. Burgess, W. H.; Mehlman, T.; Marshak, D. R.; Fraser, B. A.; Maciag, T.: Structural evidence that endothelial cell growth factor beta is the precursor of both endothelial cell growth factor alpha and acidic fibroblast growth factor. *Proc. Nat. Acad. Sci.* 83: 7216–7220, 1986.
- [44405] 17957. Eckenstein, F. P.: Fibroblast growth factors in the

nervous system. *J. Neurobiol.* 25: 1467–1480, 1994.

- [44406] 17958. Gautschi-Sova, P.; Muller, T.; Bohlen, P.: Amino acid sequence of human acidic fibroblast growth factor. *Biochem. Biophys. Res. Commun.* 140: 874–880, 1986.
- [44407] 17959. Jaye, M.; Howk, R.; Burgess, W.; Ricca, G. A.; Chiu, I.-M.; Ravera, M. W.; O'Brien, S. J.; Modi, W. S.; Maciag, T.; Drohan, W. N.: Human endothelial cell growth factor: cloning, nucleotide sequence, and chromosome localization. *Science* 233: 541–545, 1986.
- [44408] 17960. Jung, J.; Zheng, M.; Goldfarb, M.; Zaret, K. S.: Initiation of mammalian liver development from endoderm by fibroblast growth factors. *Science* 284:1998–2003, 1999.
- [44409] 17961. Mergia, A.; Eddy, R.; Abraham, J. A.; Fiddes, J. C.; Shows, T. B.: The genes for basic and acidic fibroblast growth factors are on different human chromosomes. *Biochem. Biophys. Res. Commun.* 138:644–651, 1986.
- [44410] 17962. Pellegrini, L.; Burke, D. F.; von Delft, F.; Mulloy, B.; Blundell, T. L.: Crystal structure of fibroblast growth factor receptor ectodomain bound to ligand and heparin. *Nature* 407: 1029–1034, 2000.
- [44411] 17963. Plotnikov, A. N.; Hubbard, S. R.; Schlessinger, J.; Mohammadi, M.: Crystal structures of two FGF–FGFR complexes reveal the determinants of ligand–receptor speci-

ficity. Cell 101: 413–424, 2000.

- [44412] 17964.Wang, W.–P.; Lehtoma, K.; Varban, M. L.; Krishnan, I.; Chiu, I.–M.: Cloning of the gene coding for human class 1 heparin–binding growthfactor and its expression in fetal tissues. Molec. Cell. Biol. 9:2387–2395, 1989.
- [44413] 17965.Gamez, J.; Ferreiro, C.; Accarino, M. L.; Guarner, L.; Tadesse,S.; Marti, R. A.; Andreu, A. L.; Raguer, N.; Cervera, C.; Hirano,M.: Phenotypic variability in a Spanish family with MNGIE. Neurology 59:455–457, 2002.
- [44414] 17966.Hagiwara, K.; Stenman, G.; Honda, H.; Sahlin, P.; Andersson, A.;Miyazono, K.; Heldin, C. H.; Ishikawa, F.; Takaku, F.: Organizationand chromosomal localization of the human platelet–derived endothelialcell growth factor gene. Molec. Cell. Biol. 11: 2125–2132, 1991.
- [44415] 17967.Nishino, I.; Spinazzola, A.; Hirano, M.: Thymidine phosphorylasegene mutations in MNGIE, a human mitochondrial disorder. Science 283:689–692, 1999.
- [44416] 17968.Stenman, G.; Sahlin, P.; Dumanski, J. P.; Hagiwara, K.; Ishikawa,F.; Miyazono, K.; Collins, V. P.; Heldin, C.–H.: Regional localizationof the human platelet–derived endothelial cell growth factor (ECGF1)gene to chromosome 22q13. Cytogenet. Cell Genet. 59: 22–23, 1992.
- [44417] 17969.Stenman, G.; Sahlin, P.; Hagiwara, K.; Dumanski, J.;

Collins, V.;Heldin, C.-H.: Mapping of the human platelet-derived endothelialcell growth factor (PD-ECGF) gene to chromosome 22q13. (Abstract) Cytogenet.Cell Genet. 58: 2051 only, 1991.

[44418] 17970.Kayes-Wandover, K.; White, P. C.: Steroidogenic enzyme gene expressionin the human heart. J. Clin. Endocr. Metab. 85: 2519-2525, 2000.

[44419] 17971.Sinke, R. J.; Tanigami, A.; Nakamura, Y.; Geurts van Kessel, A.: Reverse mapping of the gene encoding the human fos-related antigen-1(fra-1) within chromosome band 11q13. Genomics 18: 165 only, 1993.

[44420] 17972.Cox, D. R.; Epstein, L. B.; Epstein, C. J.: Genes coding for sensitivityto interferon (IfRec) and soluble superoxide dismutase (SOD-1) arelinked in mouse and man and map to mouse chromosome 16. Proc. Nat.Acad. Sci. 77: 2168-2172, 1980.

[44421] 17973.De Clercq, E.; Edy, V. G.; Cassiman, J.-J.: Chromosome 21 doesnot code for an interferon receptor. Nature 264: 249-251, 1976.

[44422] 17974.Epstein, L. B.; Epstein, C. J.: Localization of the gene AVG forthe antiviral expression of immune and classical interferon to the distal portion of the long arm of chromosome 21. J. Infect. Dis. 133(suppl.): A56-A62,

1976.

- [44423] 17975.Faltynek, C. R.; Branca, A. A.; McCandless, S.; Baglioni, C.:Characterization of an interferon receptor on human lymphoblastoidcells. Proc. Nat. Acad. Sci. 80: 3269–3273, 1983.
- [44424] 17976.Fournier, A.; Zhang, Z. Q.; Tan, Y. H.: Human beta:alpha but notgamma interferon binding site is a product of the chromosome 21 interferonaction gene. Somat. Cell Molec. Genet. 11: 291–295, 1985.
- [44425] 17977.Lutfalla, G.; Gardiner, K.; Proudhon, D.; Vielh, E.; Uze, G.:The structure of the human interferon alpha/beta receptor gene. J.Biol. Chem. 267: 2802–2809, 1992.
- [44426] 17978.Lutfalla, G.; Roeckel, N.; Mogensen, K. E.; Mattei, M. G.; Uze,G.: Assignment of the human interferon–alpha receptor gene to chromosome21q22.1 by in situ hybridization. J. Interferon Res. 10: 515–517,1990.
- [44427] 17979.Maroun, L. E.: Interferon action and chromosome 21 trisomy. (Letter) J.Theor. Biol. 86: 603–606, 1980.
- [44428] 17980.Novick, D.; Cohen, B.; Rubinstein, M.: The human interferon alpha/betareceptor: characterization and molecular cloning. Cell 77: 391–400,1994.
- [44429] 17981.Raziuddin, A.; Sarkar, F. H.; Dutkowski, R.; Shulman, L.; Ruddle,F. H.; Gupta, S. L.: Receptors for human

alpha and beta interferon but not for gamma interferon are specified by human chromosome 21. Proc.Nat. Acad. Sci. 81: 5504–5508, 1984.

- [44430] 1982.Revel, M.; Bash, D.; Ruddle, F. H.: Antibodies to a cell–surface component coded by human chromosome 21 inhibit action of interferon. Nature 260:139–141, 1976.
- [44431] 1983.Topham, M. K.; Prescott, S. M.: Mammalian diacylglycerol kinases, a family of lipid kinases with signaling functions. J. Biol. Chem. 274:11447–11450, 1999.
- [44432] 1984.Calabrese, G.; Crescenzi, C.; Morizio, E.; Palka, G.; Guerra, E.;Alberti, S.: Assignment of TACSTD1 (alias TROP1, M4S1) to human chromosome 2p21 and refinement of mapping of TACSTD2 (alias TROP2, M1S1) to human chromosome 1p32 by in situ hybridization. Cytogenet. Cell Genet. 92:164–165, 2001.
- [44433] 1985.Fornaro, M.; Dell'Arciprete, R.; Stella, M.; Bucci, C.; Nutini, M.; Capri, M. G.; Alberti, S.: Cloning of the gene encoding Trop–2, a cell–surface glycoprotein expressed by human carcinomas. Int. J.Cancer 62: 610–618, 1995.
- [44434] 1986.Linnenbach, A. J.; Seng, B. A.; Wu, S.; Robbins, S.; Scollon, M.;Pyrc, J. J.; Druck, T.; Huebner, K.: Retroposition in a family of carcinoma–associated antigen genes. Molec. Cell Biol. 13: 1507–1515, 1993.

- [44435] 17987.Linnenbach, A. J.; Wojcierowski, J.; Wu, S.; Pyrc, J. J.; Ross,A. H.; Dietzschold, B.; Speicher, D.; Koprowski, H.: Sequence investigationof the major gastrointestinal tu-
mor-associated antigen gene family,GA733. Proc. Nat.
Acad. Sci. 86: 27-31, 1989.
- [44436] 17988.Ren, Z.; Lin, P.-Y.; Klintworth, G. K.; Iwata, F.; Mu-
nier, F. L.;Schorderet, D. F.; El Matri, L.; Theendakara, V.;
Basti, S.; Reddy,M.; Hejtmancik, J. F.: Allelic and locus het-
erogeneity in autosomalrecessive gelatinous drop-like
corneal dystrophy. Hum. Genet. 110:568-577, 2002.
- [44437] 17989.Tsujikawa, M.; Kurahashi, H.; Tanaka, T.; Nishida,
K.; Shimomura,Y.; Tano, Y.; Nakamura, Y.: Identification of
the gene responsiblefor gelatinous drop-like corneal dys-
trophy. Nature Genet. 21: 420-423,1999.
- [44438] 17990.Willecke, K.; Jungbluth, S.; Dahl, E.; Hennemann,
H.; Heynkes,R.; Grzeschik, K.-H.: Six genes of the human
connexin gene familycoding for gap junctional proteins
are assigned to four differenthuman chromosomes. Europ.
J. Cell Biol. 53: 275-280, 1990.
- [44439] 17991.Reed, K. E.; Westphale, E. M.; Larson, D. M.; Wang,
H.-Z.; Veenstra,R. D.; Beyer, E. C.: Molecular cloning and
functional expressionof human connexin37, an endothe-
lial cell gap junction protein. J.Clin. Invest. 91: 997-1004,

1993.

- [44440] 17992. Van Camp, G.; Coucke, P.; Speleman, F.; Van Roy, N.; Beyer, E.C.; Oostra, B. A.; Willems, P. J.: The gene for human gap junction protein connexin37 (GJA4) maps to chromosome 1p35.1, in the vicinity of D1S195. *Genomics* 30: 402–403, 1995.
- [44441] 17993. Gelb, B. D.; Zhang, J.; Cotter, P. D.; Gershin, I. F.; Desnick, R. J.: Physical mapping of the human connexin 40 (GJA5) flavin-containing monooxygenase 5, and natriuretic peptide receptor A genes on 1q21. *Genomics* 39:409–411, 1997.
- [44442] 17994. Kanter, H. L.; Saffitz, J. E.; Beyer, E. C.: Cardiac myocytes express multiple gap junction proteins. *Circ. Res.* 70: 438–444, 1992.
- [44443] 17995. Kanter, H. L.; Saffitz, J. E.; Beyer, E. C.: Molecular cloning of two human cardiac gap junction proteins, connexin40 and connexin45. *J. Molec. Cell. Cardiol.* 26: 861–868, 1994.
- [44444] 17996. Oviedo-Orta, E.; Hoy, T.; Evans, W. H.: Intercellular communication in the immune system: differential expression of connexin40 and 43, and perturbation of gap junction channel functions in peripheral blood and tonsil human lymphocyte subpopulations. *Immunology* 99:

578–590,2000.

- [44445] 17997.Seul, K. H.; Tadros, P. N.; Beyer, E. C.: Mouse connexin40: gene structure and promoter analysis. *Genomics* 46: 120–126, 1997.
- [44446] 17998.Britz–Cunningham, S. H.; Shah, M. M.; Zuppan, C. W.; Fletcher, W. H.: Mutations of the connexin43 gap-junction gene in patients with heart malformations and defects of laterality. *New Eng. J. Med.* 332:1323–1329, 1995.
- [44447] 17999.Brueckner, M.; D'Eustachio, P.; Horwich, A. L.: Linkage mapping of a mouse gene, *iv*, that controls left–right asymmetry of the heart and viscera. *Proc. Nat. Acad. Sci.* 86: 5035–5038, 1989.
- [44448] 18000.Burdine, R. D.; Schier, A. F.: Conserved and divergent mechanisms in left–right axis formation. *Genes Dev.* 14: 763–776, 2000.
- [44449] 18001.Casey, B.; Ballabio, A.: Connexin43 mutations in sporadic and familial defects of laterality. (Letter) *New Eng. J. Med.* 333: 941, 1995.
- [44450] 18002.Corcus, I. A.; Meese, E. U.; Loch–Caruso, R.: Human connexin 43 gene locus, *GJA1*, sublocalized to band 6q21–q23.2. *Cytogenet. Cell Genet.* 64: 31–32, 1993.
- [44451] 18003.Debrus, S.; Tuffery, S.; Matsuoka, R.; Galal, O.; Sarda, P.; Sauer, U.; Bozio, A.; Tanman, B.; Toutain, A.;

Claustres, M.; Le Paslier, D.; Bouvagnet, P.: Lack of evidence for connexin 43 gene mutations in human autosomal recessive lateralization defects. *J. Molec. Cell. Cardiol.* 29: 1423–1431, 1997.

[44452] 18004. Wijnen, J. T.; Oldenburg, M.; Bloemendal, H.; Meera Khan, P.: GS(gamma-S)-crystallin (CRYGS) assignment to chromosome 3. (Abstract) *Cytogenet. Cell Genet.* 51: 1108 only, 1989.

[44453] 18005. Hai, T.; Liu, F.; Coukos, W. J.; Green, M. R.: Transcription factor ATF cDNA clones: an extensive family of leucine zipper proteins able to selectively form DNA-binding heterodimers. *Genes Dev.* 3: 2083–2090, 1989.

[44454] 18006. Zucman, J.; Delattre, O.; Desmaze, C.; Epstein, A. L.; Stenman, G.; Speleman, F.; Fletcher, C. D. M.; Aurias, A.; Thomas, G.: EWS and ATF-1 gene fusion induced by t(12;22) translocation in malignant melanoma of soft parts. *Nature Genet.* 4: 341–345, 1993.

[44455] 18007. Barco, A.; Alarcon, J. M.; Kandel, E. R.: Expression of constitutively active CREB protein facilitates the late phase of long-term potentiation by enhancing synaptic capture. *Cell* 108: 689–703, 2002.

[44456] 18008. Barton, C. H.; Ajioka, J. W.; Roach, T. I. A.; Blackwell, J. M.: Mapping Creb-1 to chromosome 1 in the

mouse. Genomics 14: 790–792,1992.

[44457] 18009.Barton, K.; Muthusamy, N.; Chanyangam, M.; Fischer, C.; Clendenin,C.; Leiden, J. M.: Defective thymocyte proliferation and IL-2 production in transgenic mice expressing a dominant-negative form of CREB. Nature 379:81–85, 1996.

[44458] 18010.Carlezon, W. A., Jr.; Thome, J; Olson, V. G.; Lane-Ladd, S. B.;Brodkin, E. S.; Hiroi, N.; Duman, R. S.; Neve, R. L.; Nestler, E.J.: Regulation of cocaine reward by CREB. Science 282: 2272–2275,1998.

[44459] 18011.Cole, T. J.; Copeland, N. G.; Gilbert, D. J.; Jenkins, N. A.; Schutz,G.; Ruppert, S.: The mouse CREB (cAMP responsive element bindingprotein) gene: structure, promoter analysis, and chromosomal localization. Genomics 13:974–982, 1992.

[44460] 18012.Fentzke, R. C.; Korcarz, C. E.; Lang, R. M.; Lin, H.; Leiden, J.M.: Dilated cardiomyopathy in transgenic mice expressing a dominant-negativeCREB transcription factor in the heart. J. Clin. Invest. 101: 2415–2426,1998.

[44461] 18013.Herzig, S.; Long, F.; Jhala, U. S.; Hedrick, S.; Quinn, R.; Bauer,A.; Rudolph, D.; Schutz, G.; Yoon, C.; Puigserver, P.; Spiegelman,B.; Montminy, M.: CREB regulates hepatic gluconeogenesis throughthe coactivator PGC-1. Nature

413: 179–183, 2001.

- [44462] 18014. Benson, A. M.; Hunkeler, M. J.; Talalay, P.: Increase of NAD(P)H:quinone reductase by dietary antioxidants: possible role in protection against carcinogenesis and toxicity. *Proc. Nat. Acad. Sci.* 77: 5216–5220, 1980.
- [44463] 18015. Chen, L. Z.; Harris, P. C.; Apostolou, S.; Baker, E.; Holman, K.; Lane, S. A.; Nancarrow, J. K.; Whitmore, S. A.; Stallings, R. L.; Hildebrand, C. E.; Richards, R. I.; Sutherland, G. R.; Callen, D. F.: A refined physical map of the long arm of human chromosome 16. *Genomics* 10:308–312, 1991.
- [44464] 18016. Edwards, Y. H.; Hopkinson, D. A.; Carritt, B.: A genetic characterization of the human diaphorase-4 deficiency. *Ann. Hum. Genet.* 47: 97–105, 1983.
- [44465] 18017. Grzeschik, K.-H.: Assignment of a structural gene for a fourth human diaphorase (DIA-4) to chromosome 16 in man-mouse somatic cell hybrids. *Hum. Genet.* 53: 189–193, 1980.
- [44466] 18018. Jaiswal, A. K.; McBride, O. W.; Adesnik, M.; Nebert, D. W.: Human dioxygen-inducible cytosolic NAD(P)H:menadione oxidoreductase: cDNA sequence and localization of gene to chromosome 16. *J. Biol. Chem.* 263:13572–13578, 1988.

- [44467] 18019.Kelsey, K. T.; Ross, D.; Traver, R. D.; Christiani, D. C.; Zuo,Z. F.; Spitz, M. R.; Wang, M.; Xu, X.; Lee, B. K.; Schwartz, B. S.;Wiencke, J. K.: Ethnic variation in the prevalence of a common NAD(P)Hquinone oxidoreductase polymorphism and its implications for anti-cancerchemotherapy. *Brit. J. Cancer* 76: 852–854, 1997.
- [44468] 18020.Lavinha, J.; Morrison, N.; Glasgow, L.; Ferguson-Smith, M. A.:Further evidence for the regional localization of human APRT and DIA4on chromosome 16. (Abstract) *Cytogenet. Cell Genet.* 37: 517 only,1984.
- [44469] 18021.Moran, J. L.; Siegel, D.; Ross, D.: A potential mechanism underlyingthe increased susceptibility of individuals with a polymorphism inNAD(P)H:quinone oxidoreductase 1 (NQO1) to benzene toxicity. *Proc.Nat. Acad. Sci.* 96: 8150–8155, 1999.
- [44470] 18022.Povey, S.; Wilson, D.; Edwards, Y. H.: Assignment of a human diaphorase(DIA-4) to chromosome 16. *Ann. Hum. Genet.* 43: 349–353, 1980.
- [44471] 18023.Radjendirane, V.; Joseph, P.; Lee, Y.–H.; Kimura, S.; Klein-Szanto,A. J. P.; Gonzalez, F. J.; Jaiswal, A. K.: Disruption of the DT diaphorase(NQO1) gene in mice leads to increased menadione toxicity. *J. Biol.Chem.* 273: 7382–7389, 1998.

- [44472] 18024.Rothman, N.; Smith, M. T.; Hayes, R. B.; Traver, R. D.; Hoener,B.; Campleman, S.; Li, G. L.; Dosemeci, M.; Linet, M.; Zhang, L.;Xi, L.; Wacholder, S.; Lu, W.; Meyer, K. B.; Titenko–Holland, N.;Stewart, J. T.; Yin, S.; Ross, D.: Benzene poisoning, a risk factorfor hematological malignancy, is associated with the NQO1 609C–T mutationand rapid fractional excretion of chlorzoxazone. *Cancer Res.* 57:2839–2842, 1997.
- [44473] 18025.Smith, M. T.: Benzene, NQO1, and genetic susceptibility to cancer.(Commentary) *Proc. Nat. Acad. Sci.* 96: 7624–7626, 1999.
- [44474] 18026.Traver, R. D.; Horikoshi, T.; Danenberg, K. D.; Stadlbauer, T.H.; Danenberg, P. V.; Ross, D.; Gibson, N. W.: NAD(P)H:quinone oxidoreductasegene expression in human colon carcinoma cells: characterization ofa mutation which modulates DT–diaphorase activity and mitomycin sensitivity. *CancerRes.* 52: 797–802, 1992.
- [44475] 18027.Traver, R. D.; Siegel, D.; Beall, H. D.; Phillips, R. M.; Gibson,N. W.; Franklin, W. A.; Ross, D.: Characterization of a polymorphism in NAD(P)H: quinone oxidoreductase (DT–diaphorase). *Brit. J. Cancer* 75:69–75, 1997.
- [44476] 18028.Zhang, L.; Rothman, N.; Wang, Y.; Hayes, R. B.; Li, G.; Dosemeci,M.; Yin, S.; Kolachana, P.; Titenko–Holland,

N.; Smith, M. T.: Increasedaneusomy and long arm deletion of chromosomes 5 and 7 in the lymphocytesof Chinese workers exposed to benzene. Carcinogenesis 19: 1955–1961,1998.

[44477] 18029.Zhang, L.; Wang, Y.; Shang, N.; Smith, M. T.: Benzene metabolitesinduce the loss of long arm deletion of chromosomes 5 and 7 in humanlymphocytes. Leukemia Res. 22: 105–113, 1998.

[44478] 18030.Roychoudhury, A. K.; Nei, M.: Human Polymorphic Genes: World Distribution. New York: Oxford Univ. Press (pub.) 1988.

[44479] 18031.Anholt, R. R. H.; Pederson, P. L.; Desouza, E. B.; Synder, S. H.: The peripheral-type benzodiazepine receptor: localization to themitochondrial outer membrane. J. Biol. Chem. 261: 576–583, 1986.

[44480] 18032.DeBernardi, M. A.; Crowe, R. R.; Mocchetti, I.; Shows, T. B.; Eddy,R. L.; Costa, E.: Chromosomal localization of the human diazepambinding inhibitor gene. Proc. Nat. Acad. Sci. 85: 6561–6565, 1988.

[44481] 18033.Campanelli, J. T.; Hoch, W.; Rupp, F.; Kreiner, T.; Scheller, R.H.: Agrin mediates cell contact-induced acetylcholine receptor clustering. Cell 67:909–916, 1991.

[44482] 18034.DeChiara, T. M.; Bowen, D. C.; Valenzuela, D. M.;

Simmons, M. V.; Poueymirou, W. T.; Thomas, S.; Kinetz, E.; Compton, D. L.; Rojas, E.; Park, J. S.; Smith, C.; DiStefano, P. S.; Glass, D. J.; Burden, S. J.; Yancopoulos, G. D.: The receptor tyrosine kinase MuSK is required for neuromuscular junction formation in vivo. *Cell* 85: 501–512, 1996.

[44483] 18035. Anonymous: Freely associating. (Editorial) *Nature Genet.* 22:1–2, 1999.

[44484] 18036. Bell, G. I.; Rall, L. B.; Sanchez-Pescador, R.; Merryweather, J. P.; Scott, J.; Eddy, R. L.; Shows, T. B.: Human alpha-2-macroglobulin gene is located on chromosome 12. *Somat. Cell Molec. Genet.* 11:285–289, 1985.

[44485] 18037. Bergqvist, D.; Nilsson, I. M.: Hereditary alpha-2-macroglobulin deficiency. *Scand. J. Haemat.* 23: 433–436, 1979.

[44486] 18038. Blacker, D.; Crystal, A. S.; Wilcox, M. A.; Laird, N. M.; Tanzi, R. E.: Reply to Rudrasingham et al. *Nature Genet.* 22: 21–22, 1999.

[44487] 18039. Blacker, D.; Wilcox, M. A.; Laird, N. M.; Rodes, L.; Horvath, S. M.; Go, R. C. P.; Perry, R.; Watson, B., Jr.; Bassett, S. S.; McInnis, M. G.; Albert, M. S.; Hyman, B. T.; Tanzi, R. E.: Alpha-2 macroglobulin is genetically associated with Alzheimer disease. *Nature Genet.* 19:357–360, 1998.

- [44488] 18040.David, F.; Kan, C. C.; Lucotte, G.: Two Taq I RFLPs for human alpha-2 macroglobulin (alpha-2M) using a full length cDNA probe. *Nucleic Acids Res.* 15: 374 only, 1987.
- [44489] 18041.Chan, S. Y.; Empig, C. J.; Welte, F. J.; Speck, R. F.; Schmaljohn, A.; Kreisberg, J. F.; Goldsmith, M. A.: Folate receptor-alpha is a cofactor for cellular entry by Marburg and Ebola viruses. *Cell* 106:117-126, 2001.
- [44490] 18042.Czeizel, A. E.; Dudas, I.: Prevention of the first occurrence of neural-tube defects by periconceptional vitamin supplementation. *New Eng. J. Med.* 327: 1832-1835, 1992.
- [44491] 18043.De Marco, P.; Moroni, A.; Merello, E.; de Franchis, R.; Andreussi, L.; Finnell, R. H.; Barber, R. C.; Cama, A.; Capra, V.: Folate pathway gene alterations in patients with neural tube defects. *Am. J. Med. Genet.* 95: 216-223, 2000.
- [44492] 18044.Elwood, P. C.: Molecular cloning and characterization of the human folate-binding protein cDNA from placenta and malignant tissue culture (KB) cells. *J. Biol. Chem.* 264: 14893-14901, 1989.
- [44493] 18045.Elwood, P. C.; Nachmanoff, K.; Saikawa, Y.; Page, S. T.; Pacheco, P.; Roberts, S.; Chung, K.-N.: The divergent 5-prime termini of the alpha human folate receptor (hFR)

mRNAs originate from two tissue-specific promoters and alternative splicing: characterization of the alpha hFR gene structure. *Biochemistry* 36: 1467–1478, 1997.

- [44494] 18046. Boman, B. M.; Wildrick, D. M.; Alfaro, S. R.: Chromosome 18 allele loss at the D18S6 locus in human colorectal carcinomas. *Biochem. Biophys. Res. Commun.* 155: 463–469, 1988.
- [44495] 18047. Chan, S. S.-Y.; Zheng, H.; Su, M.-W.; Wilk, R.; Killeen, M. T.; Hedgecock, E. M.; Culotti, J. G.: UNC-40, a *C. elegans* homolog of the DCC (deleted in colorectal cancer), is required in motile cells responding to UNC-6 netrin cues. *Cell* 87: 187–195, 1996.
- [44496] 18048. Sarmanova, J.; Benesova, K.; Gut, I.; Nedelcheva-Kristensen, V.; Tynkova, L.; Soucek, P.: Genetic polymorphisms of biotransformation enzymes in patients with Hodgkin's and non-Hodgkin's lymphomas. *Hum. Molec. Genet.* 10: 1265–1273, 2001.
- [44497] 18049. Nadeau, J. H.; Eicher, E. M.: Conserved linkage of soluble aconitase and galactose-1-phosphate uridyl transferase in mouse and man: assignment of these genes to mouse chromosome 4. *Cytogenet. Cell Genet.* 34: 271–281, 1982.
- [44498] 18050. Albrecht, F. E.; Drago, J.; Felder, R. A.; Printz, M. P.;

Eisner, G. M.; Robillard, J. E.; Sibley, D. R.; Westphal, H. J.; Jose, P. A.: Role of the D-1A dopamine receptor in the pathogenesis of genetic hypertension. *J. Clin. Invest.* 97: 2283–2288, 1996.

[44499] 18051. Bermak, J. C.; Li, M.; Bullock, C.; Zhou, Q.-Y.: Regulation of transport of the dopamine D1 receptor by a new membrane-associated ER protein. *Nature Cell Biol.* 3: 492–498, 2001.

[44500] 18052. Boulton, J.; Lewis, M. S.; Wainscoat, J. S.: Physical linkage of glucocorticoid receptor (GRL) and dopamine D1 receptor (DRD1) on the long arm of chromosome 5. (Abstract) *Cytogenet. Cell Genet.* 58:1894 only, 1991.

[44501] 18053. Castner, S. A.; Williams, G. V.; Goldman-Rakic, P. S.: Reversal of antipsychotic-induced working memory deficits by short-term dopamine D1 receptor stimulation. *Science* 287: 2020–2022, 2000.

[44502] 18054. Deary, A.; Gingrich, J. A.; Falardeau, P.; Fremeau, R. T., Jr.; Bates, M. D.; Caron, M. G.: Molecular cloning and expression of the gene for a human D(1) dopamine receptor. *Nature* 347: 72–76, 1990.

[44503] 18055. Grandy, D. K.; Zhou, Q.-Y.; Allen, L.; Litt, R.; Magenis, R. E.; Civelli, O.; Litt, M.: A human D(1) dopamine receptor gene is located on chromosome 5 at q35.1 and

identifies an EcoRI RFLP. *Am. J. Hum. Genet.* 47: 828–834, 1990.

[44504] 18056. Krushkal, J.; Xiong, M.; Ferrell, R.; Sing, C. F.; Turner, S. T.; Boerwinkle, E.: Linkage and association of adrenergic and dopaminereceptor genes in the distal portion of the long arm of chromosome 5 with systolic blood pressure variation. *Hum. Molec. Genet.* 7:1379–1383, 1998.

[44505] 18057. Mayerhofer, A.; Hemmings, H. C., Jr.; Snyder, G. L.; Greengard, P.; Boddien, S.; Berg, U.; Brucker, C.: Functional dopamine-1 receptors and DARPP-32 are expressed in human ovary and granulosa luteal cells in vitro. *J. Clin. Endocr. Metab.* 84: 257–264, 1999.

[44506] 18058. Sunahara, R. K.; Niznik, H. B.; Weiner, D. M.; Stormann, T. M.; Brann, M. R.; Kennedy, J. L.; Gelernter, J. E.; Rozmahel, R.; Yang, Y.; Israel, Y.; Seeman, P.; O'Dowd, B. F.: Human dopamine D1 receptor encoded by an intronless gene on chromosome 5. *Nature* 347: 80–83, 1990.

[44507] 18059. Wilkie, T. M.; Chen, Y.; Gilbert, D. J.; Moore, K. J.; Yu, L.; Simon, M. I.; Copeland, N. G.; Jenkins, N. A.: Identification, chromosomal location, and genome organization of mammalian G-protein-coupled receptors. *Genomics* 18:175–184, 1993.

- [44508] 18060.Xu, M.; Moratalla, R.; Gold, L. H.; Hiroi, N.; Koob, G. F.; Graybiel, A. M.; Tonegawa, S.: Dopamine D1 receptor mutant mice are deficient in striatal expression of dynorphin and in dopamine-mediated behavioral responses. *Cell* 79: 729–742, 1994.
- [44509] 18061.Zhou, Q.-Y.; Grandy, D. K.; Thambi, L.; Kushner, J. A.; Van Tol, H. H. M.; Cone, R.; Pribnow, D.; Salon, J.; Bunzow, J. R.; Civelli, O.: Cloning and expression of human and rat D(1) dopamine receptors. *Nature* 347:76–80, 1990.
- [44510] 18062.Balk, J.-H.; Picetti, R.; Salardi, A.; Thirlet, G.; Dierich, A.; Depaulis, A.; Le Meur, M.; Borrelli, E.: Parkinsonian-like locomotor impairment in mice lacking dopamine D2 receptors. *Nature* 377: 424–428, 1995.
- [44511] 18063.Basu, S.; Nagy, J. A.; Pal, S.; Vasile, E.; Eckelhoefer, I. A.; Bliss, V. S.; Manseau, E. J.; Dasgupta, P. S.; Dvorak, H. F.; Mukhopadhyay, D.: The neurotransmitter dopamine inhibits angiogenesis induced by vascular permeability factor/vascular endothelial growth factor. *Nature Med.* 7: 569–574, 2001.
- [44512] 18064.Blum, K.; Noble, E. P.; Sheridan, P. J.; Montgomery, A.; Ritchie, T.; Jagadeeswaran, P.; Nogami, H.; Briggs, A. H.; Cohn, J. B.: Allelic association of human dopamine D(2)

receptor gene in alcoholism. J.A.M.A. 263:2055–2060, 1990.

[44513] 18065.Bolos, A. M.; Dean, M.; Lucas–Derse, S.; Ramsburg, M.; Brown, G.L.; Goldman, D.: Population and pedigree studies reveal a lack of association between the dopamine D(2) receptor gene and alcoholism. J.A.M.A.

264:3156–3160, 1990.

[44514] 18066.Bunzow, J. R.; Van Tol, H. H.; Grandy, D. K.; Albert, P.; Salon, J.; Christie, M.; Machida, C. A.; Neve, K. A.; Civelli, O.: Cloning and expression of a rat D2 dopamine receptor cDNA. Nature 336: 783–787, 1988.

[44515] 18067.Dal Toso, R.; Sommer, B.; Ewert, M.; Herb, A.; Pritchett, D. B.; Bach, A.; Shivers, B. D.; Seeburg, P. H.: The dopamine D(2) receptor: two molecular forms generated by alternative splicing. EMBO J. 8:4025–4034, 1989.

[44516] 18068.Seeman, P.; Guan, H.–C.; Van Tol, H. H. M.: Dopamine D4 receptors elevated in schizophrenia. Nature 365: 441–445, 1993.

[44517] 18069.Seeman, P.; Ulpian, C.; Chouinard, G.; Van Tol, H. H. M.; Dwosh, H.; Lieberman, J. A.; Siminovitch, K.; Liu, I. S. C.; Wayne, J.; Voruganti, P.; Hudson, C.; Serjeant, G. R.; Masibay, A. S.; Seeman, M. V.: Dopamine D4 receptor variant, D4GLYCINE194, in Africans, but not in Caucasians: no as–

sociation with schizophrenia. *Am. J. Med. Genet.* 54: 384–390,1994.

- [44518] 18070.Smalley, S. L.; Bailey, J. N.; Palmer, C. G.; Cantwell, D. P.; McGough, J. J.; Del'Homme, M. A.; Asarnow, J. R.; Woodward, J. A.; Ramsey, C.; Nelson, S. F.: Evidence that the dopamine D4 receptor is a susceptibility gene in attention deficit hyperactivity disorder. *Molec.Psychiat.* 3: 427–430, 1998.
- [44519] 18071.Meera Khan, P.: Personal Communication. Leiden, The Netherlands 1977.
- [44520] 18072.Takai, T.; Li, M.; Sylvestre, D.; Clynes, R.; Ravetch, J. V.: FcR gamma chain deletion results in pleiotropic effector cell defects. *Cell* 76:519–529, 1994.
- [44521] 18073.Garman, S. C.; Kinet, J.-P.; Jardetzky, T. S.: Crystal structure of the human high-affinity IgE receptor. *Cell* 95: 951–961, 1998.
- [44522] 18074.Garman, S. C.; Wurzburg, B. A.; Tarchevskaya, S. S.; Kinet, J.-P.; Jardetzky, T. S.: Structure of the Fc fragment of human IgE bound to its high-affinity receptor Fc-epsilon-RI-alpha. *Nature* 406: 259–266,2000.
- [44523] 18075.Liu, F.-T.; Albrandt, K.; Robertson, M. W.: cDNA heterogeneity suggests structural variants related to the high-affinity IgE receptor. *Proc.Nat. Acad. Sci.* 85:

5639–5643, 1988.

- [44524] 18076. Shimizu, A.; Tepler, I.; Benfey, P. N.; Berenstein, E. H.; Siraganian, R. P.; Leder, P.: Human and rat mast cell high-affinity immunoglobulin E receptors: characterization of putative alpha-chain gene products. *Proc. Nat. Acad. Sci.* 85: 1907–1911, 1988.
- [44525] 18077. Miyajima, I.; Dombrowicz, D.; Martin, T. R.; Ravetch, J. V.; Kinet, J.-P.; Galli, S. J.: Systemic anaphylaxis in the mouse can be mediated largely through IgG1 and Fc-gamma-RIII: assessment of the cardiopulmonary changes, mast cell degranulation, and death associated with active or IgE- or IgG1-dependent passive anaphylaxis. *J. Clin. Invest.* 99:901–914, 1997.
- [44526] 18078. Tepler, I.; Morton, C. C.; Shimizu, A.; Holcombe, R. F.; Eddy, R.; Shows, T. B.; Leder, P.: The gene for the human mast cell high-affinity IgE receptor alpha chain: chromosomal localization to 1q21–q23 and RFLP analysis. *Am. J. Hum. Genet.* 45: 761–765, 1989.
- [44527] 18079. Zhu, D.; Kepley, C. L.; Zhang, M.; Zhang, K.; Saxon, A.: A novel human immunoglobulin Fc-gamma-Fc-epsilon bifunctional fusion protein inhibits Fc-epsilon-RI-mediated degranulation. *Nature Med.* 8: 518–521, 2002.

- [44528] 18080.Bain, G.; Robanus Maandag, E. C.; Izon, D. J.; Am-
sen, D.; Kruisbeek, A. M.; Weintraub, B. C.; Krop, I.; Schlis-
sel, M. S.; Feeney, A. J.; van Roon, M.; van der Valk, M.; te
Riele, H. P. J.; Berns, A.; Murre, C.: E2A proteins are re-
quired for proper B cell development and initiation of im-
munoglobulin gene rearrangements. *Cell* 79: 885–892,
1994.
- [44529] 18081.El Ghouzzi, V.; Legeai-Mallet, L.; Aresta, S.;
Benoist, C.; Munnich, A.; de Gunzburg, J.; Bonaventure, J.:
Saethre-Chotzen mutations cause TWIST protein degrada-
tion or impaired nuclear location. *Hum. Molec. Genet.* 9:
813–819, 2000.
- [44530] 18082.Henthorn, P.; Kiledjian, M.; Kadesch, T.: Two dis-
tinct transcription factors that bind the immunoglobulin
enhancer mu-E5/kappa-E2 motif. *Science* 247:467–470,
1990.
- [44531] 18083.Kamps, M. P.; Murre, C.; Sun, X.; Baltimore, D.: A
new homeobox gene contributes the DNA binding domain
of the t(1;19) translocation protein in pre-B ALL. *Cell* 60:
547–555, 1990.
- [44532] 18084.Mellentin, J. D.; Murre, C.; Donlon, T. A.; McCaw, P.
S.; Smith, S. D.; Carroll, A. J.; McDonald, M. E.; Baltimore,
D.; Cleary, M. L.: The gene for enhancer binding proteins

E12/E47 lies at the t(1;19)breakpoint in acute leukemias.
Science 246: 379–382, 1989.

- [44533] 18085.Murre, C.; McCaw, P. S.; Baltimore, D. :Cell 56: 777–783, 1989.
- [44534] 18086.Nourse, J.; Mellentin, J. D.; Galili, N.; Wilkinson, J.; Stanbridge,E.; Smith, S. D.; Cleary, M. L.: Chromosomal translocation t(1;19)results in synthesis of a homeobox fusion mRNA that codes for a potentialchimeric transcrip-
tion factor. Cell 60: 535–545, 1990.
- [44535] 18087.Fan, Y.–S.; Eddy, R. L.; Byers, M. G.; Haley, L. L.; Henry, W.M.; Kayano, T.; Shows, T. B.; Bell, G. I.: Assign-
ment of genes encodingthree human glucose transporter/
transporter–like proteins (GLUT4,GLUT5 and GLUT6) to
chromosomes 17, 1 and 5, respectively. (Abstract) Cyto-
genet.Cell Genet. 51: 997 only, 1989.
- [44536] 18088.Gould, G. W.; Holman, G. D.: The glucose trans-
porter family: structure,function and tissue–specific ex-
pression. Biochem. J. 295: 329–341,1993.
- [44537] 18089.Hauguel–de Mouzon, S.; Challier, J. C.; Kacemi, A.; Cauzac, M.;Malek, A.; Girard, J.: The GLUT3 glucose trans-
porter isoform is differentiallyexpressed within human
placental cell types. J. Clin. Endocr. Metab. 82:2689–2694,
1997.

- [44538] 18090. Kayano, T.; Burant, C. F.; Fukumoto, H.; Gould, G. W.; Fan, Y.S.; Eddy, R. L.; Byers, M. G.; Shows, T. B.; Seino, S.; Bell, G. I.: Human facilitative glucose transporters: isolation, functional characterization, and gene localization of cDNAs encoding an isoform (GLUT5) expressed in small intestine, kidney, muscle, and adipose tissue and an unusual glucose transporter pseudogene-like sequence (GLUT6). *J. Biol. Chem.* 265:13276–13282, 1990.
- [44539] 18091. Kayano, T.; Fukumoto, H.; Eddy, R. L.; Fan, Y.-S.; Byers, M. G.; Shows, T. B.; Bell, G. I.: Evidence for a family of human glucose transporter-like proteins: sequence and gene localization of a protein expressed in fetal skeletal muscle and other tissues. *J. Biol. Chem.* 263:15245–15248, 1988.
- [44540] 18092. Aitken, D. A.; Ferguson-Smith, M. A.: Gene dosage evidence for the regional assignment of the GOT-S structural gene locus to 10q24–10q25. *Cytogenet. Cell Genet.* 22: 468–471, 1978.
- [44541] 18093. Creagan, R.; Tischfield, J.; McMorris, F. A.; Chen, S.-H.; Hirschi, M.; Chen, T.-T.; Ricciuti, F.; Ruddle, F. H.: Assignment of the genes for human peptidase A to chromosome 18 and cytoplasmic glutamic oxaloacetate-transaminase to chromosome 10 using somatic-cell hy-

brids. Cytogenet.Cell Genet. 12: 187–198, 1973.

- [44542] 18094.Ford, G. C.; Eichele, G.; Jansonius, J. N.: Three-dimensional structure of a pyridoxal-phosphate-dependent enzyme, mitochondrial aspartate aminotransferase. Proc. Nat. Acad. Sci. 77: 2559–2563, 1980.
- [44543] 18095.Gitelman, B. J.; Tomkins, D. J.; Partington, M. W.; Roberts, M.H.; Simpson, N. E.: Gene dosage studies of glutamic oxaloacetic transaminase (GOT) and hexokinase (HK) in two patients with possible partial trisomy 10q. (Abstract) Am. J. Hum. Genet. 32: 41A only, 1980.
- [44544] 18096.Junien, C.; Despoisse, S.; Turleau, C.; de Grouchy, J.; Bucher, T.; Fundele, R.: Assignment of phosphoglycerate mutase (PGAM) to human chromosome 10: regional mapping of GOT1 and PGAM to subbands 10q26.1 (or q25.3). Ann. Genet. 25: 25–27, 1982.
- [44545] 18097.Overhauser, J.; Mewar, R.; Rojas, K.; Lia, K.; Kline, A. D.; Silverman, G. A.: STS map of genes and anonymous DNA fragments on human chromosome 18 using a panel of somatic cell hybrids. Genomics 15: 387–391, 1993.
- [44546] 18098.Abdalla, S. A.; Pece-Barbara, N.; Vera, S.; Tapia, E.; Paez, E.; Bernabeu, C.; Letarte, M.: Analysis of ALK-1 and endoglin in newborns from families with hereditary hemorrhagic telangiectasia type 2. Hum.Molec. Genet. 9:

1227–1237, 2000.

- [44547] 18099. Bode-Lesniewska, B.; Dours-Zimmermann, M. T.; Odermatt, B. F.; Briner, J.; Heitz, P. U.; Zimmermann, D. R.: Distribution of the large aggregating proteoglycan versican in adult human tissues. *J. Histochem. Cytochem.* 44: 303–312, 1996.
- [44548] 18100. Dours-Zimmermann, M. T.; Zimmermann, D. R.: A novel glycosaminoglycan attachment domain identified in two alternative splice variants of human versican. *J. Biol. Chem.* 269: 32992–32998, 1994.
- [44549] 18101. Iozzo, R. V.; Naso, M. F.; Cannizzaro, L. A.; Wasmuth, J. J.; McPherson, J. D.: Mapping of the versican proteoglycan gene (CSPG2) to the long arm of human chromosome 5 (5q12–5q14). *Genomics* 14: 845–851, 1992.
- [44550] 18102. Kjellen, L.; Lindahl, U.: Proteoglycans: structures and interactions. *Annu. Rev. Biochem.* 60: 443–475, 1991.
- [44551] 18103. Naso, M. F.; Morgan, J. L.; Buchberg, A. M.; Siracusa, L. D.; Iozzo, R. V.: Expression pattern and mapping of the murine versican gene (Cspg2) to chromosome 13. *Genomics* 29: 297–300, 1995.
- [44552] 18104. Naso, M. F.; Zimmermann, D. R.; Iozzo, R. V.: Characterization of the complete genomic structure of the human versican gene and functional analysis of its pro-

moter. J. Biol. Chem. 269: 32999–33008, 1994.

- [44553] 18105.Zako, M.; Shinomura, T.; Ujita, M.; Ito, K.; Kimata, K.: Expression of PG-M(V3), an alternatively spliced form of PG-M without a chondroitin sulfate attachment region in mouse and human tissues. J. Biol. Chem. 270:3914–3918, 1995.
- [44554] 18106.Zimmermann, D. R.; Ruoslahti, E.: Multiple domains of the large fibroblast proteoglycan, versican. EMBO J. 8: 2975–2981, 1989.
- [44555] 18107.Byrne, E.; White, O.; Cook, M.: Familial dystonic choreoathetosis with myokymia; a sleep responsive disorder. J. Neurol. Neurosurg. Psychiat. 54: 1090–1092, 1991.
- [44556] 18108.Demirkiran, M.; Jankovic, J.: Paroxysmal dyskinesias: clinical features and classification. Ann. Neurol. 38: 571–579, 1995.
- [44557] 18109.Fink, J. K.; Rainier, S.; Wilkowski, J.; Jones, S. M.; Kume, A.; Hedera, P.; Albin, R.; Mathay, J.; Girbach, L.; Varvil, T.; Otterud, B.; Leppert, M.: Paroxysmal dystonic choreoathetosis: tight linkage to chromosome 2q. Am. J. Hum. Genet. 59: 140–145, 1996.
- [44558] 18110.Fouad, G. T.; Servidei, S.; Durcan, S.; Bertini, E.; Ptacek, L.J.: A gene for familial paroxysmal dyskinesia (FPD1) maps to chromosome 2q. Am. J. Hum. Genet. 59:

135–139, 1996.

- [44559] 18111.Grunder, S.; Geisler, H.–S.; Rainer, S.; Fink, J. K.: Acid-sensing ion channel (ASIC) 4 gene: physical mapping, genomic organisation, and evaluation as a candidate for paroxysmal dystonia. *Europ. J. Hum. Genet.* 9: 672–676, 2001.
- [44560] 18112.Hudgins, R. L.; Corbin, K. B.: An uncommon seizure disorder: familial paroxysmal choreoathetosis. *Brain* 89: 199–204, 1966.
- [44561] 18113.Koch, G.; Lalley, P. A.; McAvoy, M.; Shows, T. B.: Assignment of LIPA, associated with human acid lipase deficiency, to human chromosome 10 and comparative assignment to mouse chromosome 19. *Somat. Cell Genet.* 7: 345–358, 1981.
- [44562] 18114.Kadotani, H.; Hirano, T.; Masugi, M.; Nakamura, K.; Nakao, K.; Katsuki, M.; Nakanishi, S.: Motor discoordination results from combined gene disruption of the NMDA receptor NR2A and NR2C subunits, but not from single disruption of the NR2A or NR2C subunit. *J. Neurosci.* 16:7859–7867, 1996.
- [44563] 18115.Lin, Y. J.; Bovetto, S.; Carver, J. M.; Giordano, T.: Cloning of the cDNA for the human NMDA receptor NR2C subunit and its expression in the central nervous system

and periphery. *Molec. Brain Res.* 43:57–64, 1996.

[44564] 18116. Islam, M. Q.; Platz, A.; Szpirer, J.; Szpirer, C.; Levan, G.; Mannervik, B.: Chromosomal localization of human glutathione transferase genes of classes alpha, mu and pi. *Hum. Genet.* 82: 338–342, 1989.

[44565] 18117. Albert, L. J.; Inman, R. D.: Molecular mimicry and autoimmunity. *New Eng. J. Med.* 341: 2068–2074, 1999.

[44566] 18118. Asada, H.; Kawamura, Y.; Maruyama, K.; Kume, H.; Ding, R.-G.; Ji, F. Y.; Kanbara, N.; Kuzume, H.; Sanbo, M.; Yagi, T.; Obata, K.: Mice lacking the 65 kDa isoform of glutamic acid decarboxylase (GAD65) maintain normal levels of GAD67 and GABA in their brains but are susceptible to seizures. *Biochem. Biophys. Res. Commun.* 229: 891–895, 1996.

[44567] 18119. Baekkeskov, S.; Aanstoot, H.-J.; Christgau, S.; Reetz, A.; Solimena, M.; Cascalho, M.; Folli, F.; Richter-Olesen, H.; Camilli, P.-D.: Identification of the 64K autoantigen in insulin-dependent diabetes as the GABA-synthesizing enzyme glutamic acid decarboxylase. *Nature* 347:151–156, 1990.

[44568] 18120. Bu, D.-F.; Erlander, M. G.; Hitz, B. C.; Tillakaratne, N. J. K.; Kaufman, D. L.; Wagner-McPherson, C. B.; Evans, G. A.; Tobin, A. J.: Two human glutamate decarboxylases,

65-kDa GAD and 67-kDa GAD, are each encoded by a single gene. Proc. Nat. Acad. Sci. 89: 2115–2119, 1992.

[44569] 18121. Cram, D. S.; Barnett, L. D.; Joseph, J. L.; Harrison, L. C.: Cloning and partial nucleotide sequence of human glutamic acid decarboxylase cDNA from brain and pancreatic islets. Biochem. Biophys. Res. Commun. 176:1239–1244, 1991.

[44570] 18122. De Aizpurua, H. J.; Wilson, Y. M.; Harrison, L. C.: Glutamic acid decarboxylase autoantibodies in preclinical insulin-dependent diabetes. Proc. Nat. Acad. Sci. 89: 9841–9845, 1992.

[44571] 18123. Edelhoff, S.; Grubin, C. E.; Karlsen, A. E.; Adler, D. A.; Foster, D.; Disteché, C. M.; Lernmark, A.: Mapping of glutamic acid decarboxylase (GAD) genes. Genomics 17: 93–97, 1993.

[44572] 18124. Jones, D. B.; Armstrong, N. W.: Coxsackie virus and diabetes revisited. (Letter) Nature Med. 1: 284 only, 1995.

[44573] 18125. Karlsen, A. E.; Hagopian, W. A.; Grubin, C. E.; Dube, S.; Disteché, C. M.; Adler, D. A.; Barmeier, H.; Mathewes, S.; Grant, F. J.; Foster, D.; Lernmark, A.: Cloning and primary structure of a human islet isoform of glutamic acid decarboxylase from chromosome 10. Proc. Nat. Acad. Sci. 88: 8337–8341, 1991.

- [44574] 18126.Kash, S. F.; Johnson, R. S.; Tecott, L. H.; Noebels, J. L.; Mayfield, R. D.; Hanahan, D.; Baekkeskov, S.: Epilepsy in mice deficient in the 65-kDa isoform of glutamic acid decarboxylase. *Proc. Nat. Acad. Sci.* 94: 14060–14065, 1997.
- [44575] 18127.Kaufman, D. L.; Erlander, M. G.; Clare-Salzler, M.; Atkinson, M. A.; Maclaren, N. K.; Tobin, A. J.: Autoimmunity to two forms of glutamate decarboxylase in insulin-dependent diabetes mellitus. *J. Clin. Invest.* 89: 283–292, 1992.
- [44576] 18128.Lohmann, T.; Hawa, M.; Leslie, R. D. G.; Lane, R.; Picard, J.; Londei, M.: Immune reactivity to glutamic acid decarboxylase 65 in stiff-man syndrome and type 1 diabetes mellitus. *Lancet* 356: 31–35, 2000.
- [44577] 18129.Richter, W.; Endl, J.; Eiermann, T. H.; Brandt, M.; Kientsch-Engel, R.; Thivolet, C.; Jungfer, H.; Scherbaum, W. A.: Human monoclonal islet cell antibodies from a patient with insulin-dependent diabetes mellitus reveal glutamate decarboxylase as the target antigen. *Proc. Nat. Acad. Sci.* 89: 8467–8471, 1992.
- [44578] 18130.Tian, J.; Lehmann, P. V.; Kaufman, D. L.: T cell cross-reactivity between coxsackievirus and glutamate decarboxylase is associated with a murine diabetes susceptibility allele. *J. Exp. Med.* 180: 1979–1984, 1994.

- [44579] 18131.McKusick, V. A.: The morbid anatomy of the human genome: a review of gene mapping in clinical medicine (part 1). *Medicine* 65: 1–33, 1986.
- [44580] 18132.Panteghini, M.: Aspartate aminotransferase isoenzymes. *Clin.Biochem.* 23: 311–319, 1990.
- [44581] 18133.Pol, S.; Bousquet–Lemerrier, B.; Pave–Preux, M.; Bulle, F.; Passage, E.; Hanoune, J.; Mattei, M. G.; Barouki, R.: Chromosomal localization of human aspartate aminotransferase genes by in situ hybridization. *Hum.Genet.* 83: 159–164, 1989.
- [44582] 18134.Scott, E. M.; Wright, R. C.: An alternate method for demonstration of erythrocytic aminotransferases on starch gels. *Am. J. Hum. Genet.* 33:561–563, 1981.
- [44583] 18135.Spritz, R. A.; Emanuel, B. S.; Chern, C. J.; Mellman, W. J.: Gene dosage effect: intraband mapping of human soluble glutamic oxaloacetic transaminase. *Cytogenet. Cell Genet.* 23: 149–156, 1979.
- [44584] 18136.Tomkins, D. J.; Gitelman, B. J.; Roberts, M. H.: Confirmation of a de novo duplication, dup(10)(q24–q26), by GOT1 gene dosage studies. *Hum.Genet.* 63: 369–373, 1983.
- [44585] 18137.Wang, C.–Y.; Huang, Y.–Q.; Shi, J.–O.; Marron, M. P.; Ruan, Q.–G.; Hawkins–Lee, B.; Ochoa, B.; She, J.–X.: Ge–

netic homogeneity, high-resolution mapping, and mutation analysis of the urofacial (Ochoa) syndrome and exclusion of the glutamate oxaloacetate transaminase gene (GOT1) in the critical region as the disease gene. *Am. J. Med. Genet.* 84:454–459, 1999.

[44586] 18138. Wurzinger, K. H.; Mohrenweiser, H. W.: Studies on the genetic and non-genetic (physiological) variation of human erythrocyte glutamic oxaloacetic transaminase. *Ann. Hum. Genet.* 46: 191–201, 1982.

[44587] 18139. Abel, E. D.; Kaulbach, H. C.; Tian, R.; Hopkins, J. C. A.; Duffy, J.; Doetschman, T.; Minnemann, T.; Boers, M.-E.; Hadro, E.; Oberste-Berghaus, C.; Quist, W.; Lowell, B. B.; Ingwall, J. S.; Kahn, B. B.: Cardiac hypertrophy with preserved contractile function after selective deletion of GLUT4 from the heart. *J. Clin. Invest.* 104: 1703–1714, 1999.

[44588] 18140. Abel, E. D.; Peroni, O.; Kim, J. K.; Kim, Y.-B.; Boss, O.; Hadro, E.; Minnemann, T.; Shulman, G. I.; Kahn, B. B.: Adipose-selective targeting of the GLUT4 gene impairs insulin action in muscle and liver. *Nature* 409:729–733, 2001.

[44589] 18141. Bell, G. I.; Kayano, T.; Buse, J. B.; Burant, C. F.; Takeda, J.; Lin, D.; Fukumoto, H.; Seino, S.: Molecular biol-

ogy of mammalian glucose transporters. *Diabetes Care* 13: 198–208, 1990.

[44590] 18142. Bell, G. I.; Murray, J. C.; Nakamura, Y.; Kayano, T.; Eddy, R. L.; Fan, Y.-S.; Byers, M. G.; Shows, T. B.: Polymorphic human insulin-responsive glucose-transporter gene on chromosome 17p13. *Diabetes* 38: 1072–1075, 1989.

[44591] 18143. Birnbaum, M. J.: Identification of a novel gene encoding an insulin-responsive glucose transporter protein. *Cell* 57: 305–315, 1989.

[44592] 18144. Chiang, S.-H.; Baumann, C. A.; Kanzaki, M.; Thurnmond, D. C.; Watson, R. T.; Neudauer, C. L.; Macara, I. G.; Pessin, J. E.; Saltiel, A. R.: Insulin-stimulated GLUT4 translocation requires the CAP-dependent activation of TC10. *Nature* 410: 944–948, 2001.

[44593] 18145. Garvey, W. T.; Maianu, L.; Zhu, J.-H.; Brechtel-Hook, G.; Wallace, P.; Baron, A. D.: Evidence for defects in the trafficking and translocation of GLUT4 glucose transporters in skeletal muscle as a cause of human insulin resistance. *J. Clin. Invest.* 101: 2377–2386, 1998.

[44594] 18146. Ikemoto, S.; Thompson, K. S.; Itakura, H.; Lane, M. D.; Ezaki, O.: Expression of an insulin-responsive glucose transporter (GLUT4) minigene in transgenic mice: effect of exercise and role in glucose homeostasis. *Proc. Nat. Acad.*

Sci. 92: 865–869, 1995.

- [44595] 18147. Katz, E. B.; Stenbit, A. E.; Hatton, K.; DePinho, R.; Charron, M. J.: Cardiac and adipose tissue abnormalities but not diabetes in mice deficient in GLUT4. *Nature* 377: 151–155, 1995.
- [44596] 18148. Kusari, J.; Verma, U. S.; Buse, J. B.; Henry, R. R.; Olefsky, J. M.: Analysis of the gene sequences of the insulin receptor and the insulin-sensitive glucose transporter (GLUT-4) in patients with common-type non-insulin-dependent diabetes mellitus. *J. Clin. Invest.* 88: 1323–1330, 1991.
- [44597] 18149. Muraoka, A.; Sakura, H.; Kim, K.; Kishimoto, M.; Akanuma, Y.; Buse, J. B.; Yasuda, K.; Seino, S.; Bell, G. I.; Yazaki, Y.; Kasuga, M.; Kadowaki, T.: Polymorphism in exon 4a of the human GLUT4/muscle-fat facilitative glucose transporter gene detected by SSCP. *Nucleic Acids Res.* 19: 4313 only, 1991.
- [44598] 18150. Ribon, V.; Printen, J. A.; Hoffman, N. G.; Kay, B. K.; Saltiel, A. R.: A novel, multifunctional c-Cbl binding protein in insulin receptor signaling in 3T3-L1 adipocytes. *Molec. Cell. Biol.* 18: 872–879, 1998.
- [44599] 18151. Zisman, A.; Peroni, O. D.; Abel, E. D.; Michael, M. D.; Mauvais-Jarvis, F.; Lowell, B. B.; Wojtaszewski, J. F. P.;

Hirshman, M. F.; Virkamaki, A.; Goodyear, L. J.; Kahn, C. R.; Kahn, B. B.: Targeted disruption of the glucose transporter 4 selectively in muscle causes insulin resistance and glucose intolerance. *Nature Med.* 6: 924–928, 2000.

[44600] 18152. Astrin, K. H.; Arredondo-Vega, F. X.; Desnick, R. J.; Smith, M.: Assignment of the gene for cytosolic alanine aminotransferase (AAT1) to human chromosome 8. *Ann. Hum. Genet.* 46: 125–133, 1982.

[44601] 18153. Chen, S.-H.; Giblett, E. R.: Polymorphism of soluble glutamic-pyruvic transaminase: a new genetic marker in man. *Science* 173: 148–149, 1971.

[44602] 18154. Cook, P. J. L.; Jeremiah, S. J.; Buckton, K. E.: Exclusion mapping of GPT. (Abstract) *Cytogenet. Cell Genet.* 32: 258 only, 1982.

[44603] 18155. Mombaerts, P.; Wang, F.; Dulac, C.; Vassar, R.; Chao, S. K.; Nemes, A.; Mendelsohn, M.; Edmondson, J.; Axel, R.: The molecular biology of olfactory perception. *Cold Spring Harbor Symp. Quant. Biol.* 61: 135–145, 1996.

[44604] 18156. Reed, R. R.: Genetic approaches to mammalian olfaction. *Cold Spring Harbor Symp. Quant. Biol.* 61: 165–172, 1996.

[44605] 18157. Ronnett, G. V.; Snyder, S. H.: Molecular messengers of olfaction. *Trends Neurosci.* 15: 508–513, 1992.

- [44606] 18158.Schwab, S. G.; Hallmayer, J.; Lerer, B.; Albus, M.; Borrmann, M.;Honig, S.; Strauss, M.; Segman, R.; Lichtermann, D.; Knapp, M.; Trixler,M.; Maier, W.; Wildenauer, D. B.: Support for a chromosome 18p locusconferring susceptibility to functional psychoses in families with-schizophrenia, by association and linkage analysis. *Am. J. Hum. Genet.* 63:1139–1152, 1998.
- [44607] 18159.Davignon, I.; Barnard, M.; Gavrilova, O.; Sweet, K.; Wilkie, T.M.: Gene structure of murine Gna11 and Gna15: tandemly duplicatedGq class G protein alpha subunit genes. *Genomics* 31: 359–366, 1996.
- [44608] 18160.Jiang, M.; Pandey, S.; Tran, V. T.; Fong, H. K.: Guanine nucleotide-bindingregulatory proteins in retinal pigment epithelial cells. *Proc. Nat.Acad. Sci.* 88: 3907–3911, 1991.
- [44609] 18161.Offermanns, S.; Zhao, L.-P.; Gohla, A.; Sarosi, I.; Simon, M. I.;Wilkie, T. M.: Embryonic cardiomyocyte hypoplasia and craniofacialdefects in G-alpha-pha-q/G-alpha-11-mutant mice. *EMBO J.* 17: 4304–4312,1998.
- [44610] 18162.Strathmann, M. P.; Simon, M. I.: G-alpha-12 and G-alpha-13 subunitsdefine a fourth class of G protein alpha subunits. *Proc. Nat. Acad.Sci.* 88: 5582–5586, 1991.

- [44611] 18163.Ahmed, S. F.; Barr, D. G. D.; Bonthron, D. T.: GNAS1 mutations and progressive osseous heteroplasia. (Letter) New Eng. J. Med. 346:1669–1670, 2002.
- [44612] 18164.Ahmed, S. F.; Dixon, P. H.; Bonthron, D. T.; Stirling, H. F.; Barr, D. G. D.; Kelnar, C. J. H.; Thakker, R. V.: GNAS1 mutational analysis in pseudohypoparathyroidism. Clin. Endocr. 49: 525–531, 1998.
- [44613] 18165.Ahrens, W.; Hiort, O.; Staedt, P.; Kirschner, T.; Marschke, C.; Kruse, K.: Analysis of the GNAS1 gene in Albright's hereditary osteodystrophy. J.Clin. Endocr. Metab. 86: 4630–4634, 2001.
- [44614] 18166.Aldred, M. A.; Trembath, R. C.: Activating and inactivating mutations in the human GNAS1 gene. Hum. Mutat. 16: 183–189, 2000.
- [44615] 18167.Ashley, P. L.; Ellison, J.; Sullivan, K. A.; Bourne, H. R.; Cox, D. R.: Chromosomal assignment of the murine Gi and Gs genes. (Abstract) Am.J. Hum. Genet. 41: A155 only, 1987.
- [44616] 18168.Ballare, E.; Mantovani, S.; Lania, A.; Di Blasio, A. M.; Vallar, L.; Spada, A.: Activating mutations of the GS-alpha gene are associated with low levels of GS-alpha protein in growth hormone-secreting tumors. J.Clin. Endocr. Metab. 83: 4386–4390, 1998.

- [44617] 18169. Bastepe, M.; Pincus, J. E.; Sugimoto, T.; Tojo, K. Kanatani, M.; Azuma, Y.; Kruse, K.; Rosenbloom, A. L.; Koshiyama, H.; Juppner, H.: Positional dissociation between the genetic mutation responsible for pseudohypoparathyroidism type 1b and the associated methylation-defect at exon A/B: evidence for a long-range regulatory element within the imprinted GNAS1 locus. *Hum. Molec. Genet.* 10: 1231–1241, 2001.
- [44618] 18170. Bastepe, M.; Juppner, H.: GNAS1 mutations and progressive osseous heteroplasia. (Letter) *New Eng. J. Med.* 346: 1671 only, 2002.
- [44619] 18171. Bastepe, M.; Lane, A. H.; Juppner, H.: Parental uniparental isodisomy of chromosome 20q--and the resulting changes in GNAS1 methylation--as a plausible cause of pseudohypoparathyroidism. *Am. J. Hum. Genet.* 68:1283–1289, 2001.
- [44620] 18172. Bianco, P.; Riminucci, M.; Majolagbe, A.; Kuznetsov, S. A.; Collins, M. T.; Mankani, M. H.; Corsi, A.; Bone, H. G.; Wientroub, S.; Spiegel, A. M.; Fisher, L. W.; Robey, P. G.: Mutations of the GNAS1 gene, stromal cell dysfunction, and osteomalacic changes in non-McCune-Albright fibrous dysplasia of bone. *J. Bone Miner. Res.* 15: 120–128, 2000.
- [44621] 18173. Billestrup, N.; Swanson, L. W.; Vale, W.: Growth

hormone-releasing factor stimulates proliferation of somatotrophs in vitro. *Proc. Nat. Acad. Sci.* 83: 6854–6857, 1986.

[44622] 18174. Candelieri, G. A.; Glorieux, F. H.; Prud'Homme, J.; St.-Arnaud, R.: Increased expression of the c-fos proto-oncogene in bone from patients with fibrous dysplasia. *New Eng. J. Med.* 332: 1546–1551, 1995.

[44623] 18175. Candelieri, G. A.; Roughley, P. J.; Glorieux, F. H.: Polymerase chain reaction-based technique for the selective enrichment and analysis of mosaic *arg201* mutations in *G alpha s* from patients with fibrous dysplasia of bone. *Bone* 21: 201–206, 1997.

[44624] 18176. Carel, J. C.; Le Stunff, C.; Condamine, L.; Mallet, E.; Chaussain, J. L.; Adnot, P.; Garabedian, M.; Bougneres, P.: Resistance to the lipolytic action of epinephrine: a new feature of protein GS deficiency. *J. Clin. Endocr. Metab.* 84: 4127–4131, 1999.

[44625] 18177. Carter, A.; Bardin, C.; Collins, R.; Simons, C.; Bray, P.; Spiegel, A.: Reduced expression of multiple forms of the alpha subunit of the stimulatory GTP-binding protein in pseudohypoparathyroidism type Ia. *Proc. Nat. Acad. Sci.* 84: 7266–7269, 1987.

[44626] 18178. Cattanach, B. M.; Kirk, M.: Differential activity of

maternally and paternally derived chromosome regions in mice. *Nature* 315: 496–498, 1985.

[44627] 18179. Coutant, R.; Lumbroso, S.; Rey, R.; Lahlou, N.; Venara, M.; Rouleau, S.; Sultan, C.; Limal, J.-M.:

Macroorchidism due to autonomous hyperfunction of Sertoli cells and GS- α gene mutation: an unusual expression of McCune-Albright syndrome in a prepubertal boy. *J. Clin. Endocr. Metab.* 86: 1778–1781, 2001.

[44628] 18180. Kim, H.-S.; Nagalla, S. R.; Oh, Y.; Wilson, E.; Roberts, C. T. Jr.; Rosenfeld, R. G.: Identification of a family of low-affinity insulin-like growth factor binding proteins (IGFBPs): characterization of connective tissue growth factor as a member of the IGFBP superfamily. *Proc. Nat. Acad. Sci.* 94: 12981–12986, 1997.

[44629] 18181. Martinerie, C.; Viegas-Pequignot, E.; Guenard, I.; Dutrillaux, B.; Nguyen, V. C.; Bernheim, A.; Perbal, B.: Physical mapping of human loci homologous to the chicken *nov* proto-oncogene. *Oncogene* 7: 2529–2534, 1992.

[44630] 18182. Nakanishi, T.; Yamaai, T.; Asano, M.; Nawachi, K.; Suzuki, M.; Sugimoto, T.; Takigawa, M.: Overexpression of connective tissue growth factor/hypertrophic chondrocyte-specific gene product 24 decreases bone density in adult mice

and induces dwarfism. *Biochem. Biophys. Res. Commun.* 281: 678–681, 2001.

- [44631] 18183.O'Donovan, N.; Galvin, M.; Morgan, J. G.: Physical mapping of the CXC chemokine locus on human chromosome 4. *Cytogenet. Cell Genet.* 84:39–42, 1999.
- [44632] 18184.McGill, J. R.; Boyd, D.; Barrett, K. J.; Drysdale, J. W.; Moore, C. M.: Localization of human ferritin H (heavy) and L (light) subunits by in situ hybridization. (Abstract) *Am. J. Hum. Genet.* 36: 146S only, 1984.
- [44633] 18185.Murray, M. T.; White, K.; Munro, H. N.: Conservation of ferritin heavy subunit gene structure: implications for the regulation of ferritin gene expression. *Proc. Nat. Acad. Sci.* 84: 7438–7442, 1987.
- [44634] 18186.Papadopoulos, P.; Bhavsar, D.; Zappone, E.; David, V.; Jones, C.; Worwood, M.; Drysdale, J.: A second human ferritin H locus on chromosome 11. *Cytogenet. Cell Genet.* 61: 107–108, 1992.
- [44635] 18187.Richard, C. W.; Withers, D. A.; Meeker, T. C.; Myers, R. M.: A radiation hybrid map of the proximal long arm of human chromosome 11 containing the MEN-1 and bcl-1 disease locus. (Abstract) *Cytogenet. Cell Genet.* 58: 1970 only, 1991.
- [44636] 18188.Worwood, M.; Brook, J. D.; Cragg, S. J.; Hellkuhl, B.;

Jones,B. M.; Perera, P.; Roberts, S. H.; Shaw, D. J.: Assign-
ment of humanferritin genes to chromosomes 11 and
19q13.3–19qter. Hum. Genet. 69:371–374, 1985.

[44637] 18189.Wu, K.–J.; Polack, A.; Dalla–Favera, R.: Coordinated
regulationof iron–controlling genes, H–ferritin and IRP2,
by c–MYC. Science 283:676–679, 1999.

[44638] 18190.Yachou, A.; Mattei, M. G.; Roeckel, N.; Grand–
champ, B.; Beaumont,C.: Mouse ferritin H sequences map
to chromosomes 3, 6, and 19. Genomics 9:204–206,
1991.

[44639] 18191.Yachou, A.–K.; Renaudie, F.; Guenet, J.–L.; Simon–
Chazottes, D.;Jones, R.; Grandchamp, B.; Beaumont, C.:
Mouse ferritin H multigenefamily is polymorphic and con–
tains a single multiallelic functionalgene located on chro–
mosome 19. Genomics 10: 531–538, 1991.

[44640] 18192.Youssoufian, H.; Chance, P.; Tuck–Muller, C. M.;
Jabs, E. W.:Association of a new chromosomal deletion
[del(1)(q32q42)] with diaphragmatic hernia: assignment of
a human ferritin gene. Hum. Genet. 78: 267–270,1988.

[44641] 18193.Aguilar–Martinez, P.; Biron, C.; Masmajan, C.;
Jeanjean, P.; Schved,J.–F.: A novel mutation in the iron re–
sponsive element of ferritinL–subunit gene as a cause for
hereditary hyperferritinemia–cataractsyndrome. (Letter)

Blood 88: 1895–1903, 1996.

- [44642] 18194. Beaumont, C.; Leneuve, P.; Devaux, I.; Scoazec, J.-Y.; Berthier, M.; Loiseau, M.-N.; Grandchamp, B.; Bonneau, D.: Mutation in the iron responsive element of the L ferritin mRNA in a family with dominant hyperferritinaemia and cataract. *Nature Genet.* 11: 444–446, 1995.
- [44643] 18195. Brown, A. J. P.; Leibold, E. A.; Munro, H. N.: Isolation of cDNA clones for the light subunit of rat liver ferritin: evidence that the light subunit is encoded by a multi-gene family. *Proc. Nat. Acad. Sci.* 80: 1265–1269, 1983.
- [44644] 18196. Camaschella, C.; Zecchina, G.; Lockitch, G.; Roetto, A.; Campanella, A.; Arosio, P.; Levi, S.: A new mutation (G51C) in the iron-responsive element (IRE) of L-ferritin associated with hyperferritinaemia-cataract syndrome decreases the binding affinity of the mutated IRE for iron-regulatory proteins. *Brit. J. Haemat.* 108: 480–482, 2000.
- [44645] 18197. Cazzola, M.; Bergamaschi, G.; Tonon, L.; Arbustini, E.; Grasso, M.; Vercesi, E.; Barosi, G.; Bianchi, P. E.; Cairo, G.; Arosio, P.: Hereditary hyperferritinemia-cataract syndrome: relationship between phenotypes and specific mutations in the iron-responsive element of ferritin light-chain mRNA. *Blood* 90: 814–821, 1997.
- [44646] 18198. Cazzola, M.; Foglieni, B.; Bergamaschi, G.; Levi, S.;

Lazzarino, M.; Arosio, P.: A novel deletion of the L-ferritin iron-responsive element responsible for severe hereditary hyperferritinaemia-cataract syndrome. *Brit. J. Haemat.* 116: 667–670, 2002.

[44647] 18199. Xu, W.; Gorman, P. A.; Rider, S. H.; Hedge, P. J.; Moore, G.; Prichard, C.; Sheer, D.; Solomon, E.: Construction of a genetic map of human chromosome 17 by use of chromosome-mediated gene transfer. *Proc. Nat. Acad. Sci.* 85: 8563–8567, 1988.

[44648] 18200. Fishman, G. I.; Eddy, R. L.; Shows, T. B.; Rosenthal, L.; Levin, L. A.: The human connexin gene family of gap junction proteins: distinct chromosomal locations but similar structures. *Genomics* 10: 250–256, 1991.

[44649] 18201. Campbell, H. D.; Webb, G. C.; Kono, T.; Taniguchi, T.; Ford, J. H.; Young, I. G.: Assignment of the interleukin-2 receptor beta chain gene (IL-2rb) to band E on mouse chromosome 15. *Genomics* 12: 179–180, 1992.

[44650] 18202. Bonneau, D.; Huret, J. L.; Godeau, G.; Couet, D.; Putterman, M.; Tanzer, J.; Babin, P.; Larregue, M.: Recurrent ctg(7)(q31.3) and possible laminin involvement in a neonatal cutis laxa with a Marfan phenotype. *Hum. Genet.* 87: 317–319, 1991.

[44651] 18203. Buchanan, R.; Wyatt, G. P.: Marfan's syndrome pre-

senting as anintrapartum death. Arch. Dis. Child. 60: 1074–1076, 1985.

- [44652] 18204.Burgeson, R. E.; Chiquet, M.; Deutzmann, R.; Ekblom, P.; Engel,J.; Kleinman, H.; Martin, G. R.; Meneguzzi, G.; Paulsson, M.; Sanes,J.; Timpl, R.; Tryggvason, K.; Yamada, Y.; Yurchenco, P. D.: A new nomenclature for the laminins. Matrix Biol. 14: 209–211, 1994.
- [44653] 18205.Day, D. L.; Burke, B. A.: Pulmonary emphysema in a neonate with Marfan syndrome. Pediat. Radiol. 16: 518–521, 1986.
- [44654] 18206.Elliott, R. W.; Barlow, D.; Hogan, B. L. M.: Linkage of genes for laminin B1 and B2 subunits on chromosome 1 in mouse. In Vitro Cell Dev. Biol. 21: 477–484, 1985.
- [44655] 18207.Gross, D. M.; Robinson, L. K.; Smith, L. T.; Glass, N.; Rosenberg,H.; Duvic, M.: Severe perinatal Marfan syndrome. Pediatrics 84:83–89, 1989.
- [44656] 18208.Hohn, A. R.; Webb, H. M.: Cardiac studies of infant twins with Marfan's syndrome. Am. J. Dis. Child. 122: 526–528, 1971.
- [44657] 18209.Hopker, V. H.; Shewan, D.; Tessier-Lavigne, M.; Poo, M.; Holt,C.: Growth-cone attraction to netrin-1 is converted to repulsion by laminin-1. Nature 401: 69–73, 1999.

- [44658] 18210.Huret, J.; Bonneau, D.; Godeau, G.; Leheup, B.; Larregue, M.:Neonatal cutis laxa with Marfan habitus: this syndrome maps with B1laminin at the junction 7q31.3–q32. (Abstract) Cytogenet. Cell Genet. 58:1922 only, 1991.
- [44659] 18211.Iwamoto, Y.; Robey, F. A.; Graf, J.; Sasaki, M.; Kleinman, H.K.; Yamada, Y.; Martin, G. R.: YIGSR, a synthetic laminin pentapeptide,inhibits experimental metastasis formation. Science 238: 1132–1134,1987.
- [44660] 18212.Jaye, M.; Modi, W. S.; Ricca, G. A.; Mudd, R.; Chiu, I.–M.; O'Brien,S. J.; Drohan, W. N.: Isolation of a cDNA clone for the human laminin–B1chain and its gene localization. Am. J. Hum. Genet. 41: 605–615,1987.
- [44661] 18213.Kleinman, H. K.: Personal Communication. Bethesda, Md. 1/7/1982.
- [44662] 18214.Lababidi, Z.; Monzon, C.: Early cardiac manifestations of Marfan'ssyndrome in the newborn. Am. Heart J. 102: 943–945, 1981.
- [44663] 18215.Modi, W. S.; Jaye, M.; O'Brien, S. J.: Chromosomal localizationof a cDNA clone for the human B1 laminin chain. (Abstract) Cytogenet.Cell Genet. 46: 663 only, 1987.
- [44664] 18216.Neimann, N.; Rauber, G.; Marchal, C.; Vidailhet, M.;

Fall, M.: Maladie de Marfan chez un nouveau-né avec atteintes polyviscérales: étude anatomo-clinique. *Ann. Paediat.* 15: 619–624, 1968.

[44665] 18217. Pikkarainen, T.; Eddy, R.; Fukushima, Y.; Byers, M.; Shows, T.; Pihlajaniemi, T.; Saraste, M.; Tryggvason, K.: Human laminin B1 chain: a multidomain protein with gene (LAMB1) locus in the q22 region of chromosome 7. *J. Biol. Chem.* 262: 10454–10462, 1987.

[44666] 18218. Pikkarainen, T.; Savolainen, E.-R.; Tryggvason, K.: Nhe I and Hinc II polymorphisms in the human laminin B1 chain gene on 7q22. *Nucleic Acids Res.* 17: 4424 only, 1989.

[44667] 18219. Sasaki, M.; Kato, S.; Kohno, K.; Martin, G. R.; Yamada, Y.: Sequence of the cDNA encoding the laminin B1 chain reveals a multidomain protein containing cysteine-rich repeats. *Proc. Nat. Acad. Sci.* 84: 935–939, 1987.

[44668] 18220. Vuolteenaho, R.; Chow, L. T.; Tryggvason, K.: Structure of the human laminin B1 chain gene. *J. Biol. Chem.* 265: 15611–15616, 1990.

[44669] 18221. Gremlich, S.; Porret, A.; Hani, E. H.; Cherif, D.; Vionnet, N.; Froguel, P.; Thorens, B.: Cloning, functional expression, and chromosomal localization of the human pancreatic islet glucose-dependent insulinotrop-

ic polypeptide receptor. *Diabetes* 44: 1202–1208, 1995.

- [44670] 18222. Miyawaki, K.; Yamada, Y.; Ban, N.; Ihara, Y.; Tsukiyama, K.; Zhou, H.; Fujimoto, S.; Oku, A.; Tsuda, K.; Toyokuni, S.; Hiai, H.; Mizunoya, W.; and 9 others: Inhibition of gastric inhibitory polypeptide signaling prevents obesity. *Nature Med.* 8: 738–742, 2002.
- [44671] 18223. Stoffel, M.; Fernald, A. A.; Le Beau, M. M.; Bell, G. I.: Assignment of the gastric inhibitory polypeptide receptor gene (GIPR) to chromosome bands 19q13.2–q13.3 by fluorescence in situ hybridization. *Genomics* 28: 607–609, 1995.
- [44672] 18224. Usdin, T. B.; Mezey, E.; Button, D. C.; Brownstein, M. J.; Bonner, T. I.: Gastric inhibitory polypeptide receptor, a member of the secretin–vasoactive intestinal peptide receptor family, is widely distributed in peripheral organs and the brain. *Endocrinology* 133: 2861–2870, 1993.
- [44673] 18225. Volz, A.; Groke, R.; Lankat-Buttgereit, B.; Fehmann, H.-C.; Bode, H. P.; Goke, B.: Molecular cloning, functional expression, and signal transduction of the GIP-receptor cloned from a human insulinoma. *FEBS Lett.* 373: 23–29, 1995.
- [44674] 18226. Yamada, Y.; Hayami, T.; Nakamura, K.; Kaisaki, P. J.; Someya, Y.; Wang, C.-Z.; Seino, S.; Seino, Y.: Human

gastric inhibitory polypeptidereceptor: cloning of the gene (GIPR) and cDNA. *Genomics* 29: 773–776,1995.

[44675] 18227.Miyawaki, K.; Yamada, Y.; Yano, H.; Niwa, H.; Ban, N.; Ihara,Y.;Kubota, A.; Fujimoto, S.; Kajikawa, M.; Kuroe, A.; Tsuda, K.; Hashimoto,H.; Yamashita, T.; Jomori, T.; Tashiro, F.; Miyazaki, J.; Seino, Y.: Glucose intolerance caused by a defect in the entero–insular axis:a study in gastric inhibitory polypeptide receptor knockout mice. *Proc.Nat. Acad. Sci.* 96: 14843–14847, 1999.

[44676] 18228.Flejter, W. L.; Barcroft, C. L.; Guo, S.–W.; Lynch, E. D.; Boehnke,M.; Chandrasekharappa, S.; Hayes, S.; Collins, F. S.; Weber, B. L.;Glover, T. W.: Multicolor FISH mapping with Alu–PCR–amplified YACclone DNA determines the order of markers in the BRCA1 region on chromosome17q12–q21. *Genomics* 17: 624–631, 1993.

[44677] 18229.Fukushige, S.; Murotsu, T.; Matsubara, K.: Chromosomal assignmentof human genes for gastrin, thyrotropin (TSH)–beta subunit and C–erb–2by chromosome sorting combined with velocity sedimentation and southernhybridization. *Biochem. Biophys. Res. Commun.* 134: 477–483, 1986.

[44678] 18230.Justice, M. J.; Gilbert, D. J.; Kinzler, K. W.; Vogelstein, B.;Buchberg, A. M.; Ceci, J. D.; Matsuda, Y.; Chap–

man, V. M.; Patriotis, C.; Makris, A.; Tsiichlis, P. N.; Jenkins, N. A.; Copeland, N. G.: A molecular genetic linkage map of mouse chromosome 18 reveals extensive linkage conservation with human chromosomes 5 and 18. *Genomics* 13:1281–1288, 1992.

[44679] 18231. Lebacqz-Verheyden, A.-M.; Bertness, V.; Kirsch, I.; Hollis, G. F.; McBride, O. W.; Battey, J.: Human gastrin-releasing peptide gene maps to chromosome band 18q21. *Somat. Cell Molec. Genet.* 13: 81–86, 1987.

[44680] 18232. Naylor, S. L.; Sakaguchi, A. Y.; Spindel, E.; Chin, W. W.: Human gastrin-releasing peptide gene is located on chromosome 18. *Somat. Cell Molec. Genet.* 13: 87–91, 1987.

[44681] 18233. Naylor, S. L.; Spindel, E.; Chin, W. W.; Sakaguchi, A. Y.: Gastrin-releasing peptide gene is located on human chromosome 18. (Abstract) *Cytogenet. Cell Genet.* 40: 711 only, 1985.

[44682] 18234. Sausville, E. A.; Lebacqz-Verheyden, A.-M.; Spindel, E. R.; Cuttitta, F.; Gazdar, A. F.; Battey, J. F.: Expression of the gastrin-releasing peptide gene in human small cell lung cancer: evidence for alternative processing resulting in three distinct mRNAs. *J. Biol. Chem.* 261:2451–2457, 1986.

- [44683] 18235.Spindel, E. R.; Chin, W. W.; Price, J.; Rees, L. H.; Besser, G.M.; Habener, J. F.: Cloning and characterization of cDNAs encoding human gastrin-releasing peptide. Proc. Nat. Acad. Sci. 81: 5699–5703, 1984.
- [44684] 18236.Spindel, E. R.; Zilberberg, M. D.; Habener, J. F.; Chin, W. W.: Two prohormones for gastrin-releasing peptide are encoded by two mRNAs differing by 19 nucleotides. Proc. Nat. Acad. Sci. 83: 19–23, 1986.
- [44685] 18237.Kudo, S.; Chagnovich, D.; Rearden, A.; Mattei, M. G.; Fukuda, M.: Molecular analysis of a hybrid gene encoding human glycophorin variant Miltenberger V-like molecule. J. Biol. Chem. 265: 13825–13829, 1990.
- [44686] 18238.Kudo, S.; Fukuda, M.: Identification of a novel human glycophorin, glycophorin E, by isolation of genomic clones and complementary DNA clones utilizing polymerase chain reaction. J. Biol. Chem. 265: 1102–1110, 1990.
- [44687] 18239.Onda, M.; Fukuda, M.: Detailed physical mapping of the genes encoding glycophorins A, B, and E, as revealed by P1 plasmids containing human genomic DNA. Gene 159: 225–230, 1995.
- [44688] 18240.Onda, M.; Kudo, S.; Rearden, A.; Mattei, M.-G.; Fukuda, M.: Identification of a precursor genomic segment

that provided a sequence unique to glycophorin B and E genes. Proc. Nat. Acad. Sci. 90: 7220–7224, 1993.

[44689] 18241. Vignal, A.; Rahuel, C.; London, J.; Cherif Zahar, B.; Schaff, S.; Hattab, C.; Okubo, Y.; Cartron, J.-P.: A novel gene member of the human glycophorin A and B gene family: molecular cloning and expression. Europ. J.

Biochem. 191: 619–625, 1990.

[44690] 18242. Board, P. G.; Jones, I. M.; Bentley, A. K.: Molecular cloning and nucleotide sequence of human alpha-1 acid glycoprotein cDNA. Gene 44: 127–131, 1986.

[44691] 18243. Cox, D. W.; Francke, U.: Direct assignment of orosomucoid to human chromosome 9 and alpha-2-HS-glycoprotein to chromosome 3 using human fetal liver x rat hepatoma hybrids. Hum. Genet. 70: 109–115, 1985.

[44692] 18244. Dayhoff, M. O.: Atlas of Protein Sequence and Structure. Orosomucoid. Washington: National Biomedical Research Foundation (pub.) 5: 1972. Pp. D310–D316.

[44693] 18245. Dente, L.; Ciliberto, G.; Cortese, R.: Structure of the human alpha-1-acid glycoprotein gene: sequence homology with other human acute phase protein genes. Nucleic Acids Res. 13: 3941–3952, 1985.

[44694] 18246. Dente, L.; Pizza, M. G.; Metspalu, A.; Cortese, R.:

Structure and expression of the genes coding for human alpha-1-acid glycoprotein. EMBOJ. 6: 2289–2296, 1987.

[44695] 18247. Dente, L.; Rutherford, U.; Tripodi, M.; Wagner, E. F.; Cortese, R.: Expression of human alpha-1-acid glycoprotein genes in cultured cells and in transgenic mice. Genes Dev. 2: 259–266, 1988.

[44696] 18248. Eap, C. B.; Cuendet, C.; Baumann, P.: Orosomucoid (alpha-1 acid glycoprotein) phenotyping by use of immobilized pH gradients with 8 M urea and immunoblotting: a new variant encountered in a population study. Hum. Genet. 80: 183–185, 1988.

[44697] 18249. Eiberg, H.; Mohr, J.; Nielsen, L. S.: Linkage of orosomucoid (ORM) to ABO and AK1. (Abstract) Cytogenet. Cell Genet. 32: 272 only, 1982.

[44698] 18250. Johnson, A. M.; Schmid, K.; Alper, C. A.: Inheritance of human alpha(1)-acid glycoprotein (orosomucoid) variants. J. Clin. Invest. 48: 2293–2299, 1969.

[44699] 18251. Luckenbach, C.; Kompf, J.; Ritter, H.: Orosomucoid (ORM1) subtyping and formal genetics. Hum. Genet. 87: 429–432, 1991.

[44700] 18252. Rocchi, M.; Roncuzzi, L.; Santamaria, R.; Archidiacono, N.; Dente, L.; Romeo, G.: Mapping through somatic cell hybrids and cDNA probes of protein C to chromosome

2, factor X to chromosome 13, and alpha-1-acidglycoprotein to chromosome 9. Hum. Genet. 74: 30-33, 1986.

- [44701] 18253.Schmid, K.; Tokita, K.; Yoshizaki, H.: The alpha-1-acid glycoproteinvariants of normal Caucasian and Japanese individuals. J. Clin. Invest. 44:1394-1401, 1965.
- [44702] 18254.Tokita, K.; Schmid, K.: Variants of alpha-1-acid glycoprotein. Nature 200:266 only, 1963.
- [44703] 18255.Tomei, L.; Eap, C. B.; Baumann, P.; Dente, L.: Use of transgenicmice for the characterization of human alpha-1-acid glycoprotein (orosomucoid)variants. Hum. Genet. 84: 89-91, 1989.
- [44704] 18256.Umetsu, K.; Ikeda, N.; Kashimura, S.; Suzuki, T.: Orosomucoid(ORM) typing by print lectinofixation: a new technique for isoelectricfocusing--two common alleles in Japan. Hum. Genet. 71: 223-224,1985.
- [44705] 18257.Umetsu, K.; Yuasa, I.; Nishimura, H.; Sasaki, H.; Suzuki, T.:Genetic polymorphisms of orosomucoid and alpha-2-HS-glycoprotein ina Philippine population. Hum. Hered. 38: 287-290, 1988.
- [44706] 18258.Webb, G. C.; Earle, M. E.; Merritt, C.; Board, P. G.: Localizationof human alpha-1 glycoprotein genes to 9q31-q34.1. Cytogenet. CellGenet. 47: 18-21, 1988.

- [44707] 18259. Weidinger, S.; Muller, T.; Schwarzfischer, F.; Cleve, H.: Threenew orosomucoid (ORM) variants revealed by isoelectric focusing and print immunofixation. *Hum. Genet.* 77: 286–288, 1987.
- [44708] 18260. Yuasa, I.; Umetsu, K.; Vogt, U.; Nakamura, H.; Nanba, E.; Tamaki, N.; Irizawa, Y.: Human orosomucoid polymorphism: molecular basis of the three common ORM1 alleles, ORM1*F1, ORM1*F2, and ORM1*S. *Hum. Genet.* 99: 393–398, 1997.
- [44709] 18261. Yuasa, I.; Weidinger, S.; Umetsu, K.; Suenaga, K.; Ishimoto, G.; Eap, B. C.; Duche, J.-C.; Baumann, P.: Orosomucoid system: 17 additional orosomucoid variants and proposal for a new nomenclature. *Vox Sang.* 64:47–55, 1993.
- [44710] 18262. Triantafilou, K.; Triantafilou, M.; Dedrick, R. L.: A CD14-independent LPS receptor cluster. *Nature Immun.* 2: 338–345, 2001.
- [44711] 18263. Saito, S.; Okui, K.; Tokino, T.; Oshimura, M.; Nakamura, Y.: Isolation and mapping of 68 RFLP markers on human chromosome 6. *Am. J. Hum. Genet.* 50: 65–70, 1992.
- [44712] 18264. Takahashi, I.; Tanuma, R.; Hirata, M.; Hashimoto, K.: A cosmid clone at the D6S182 locus on human chro–

mosome 6p12 contains the 90-kDa heat shock protein beta-gene (HSP90-beta). Mammalian Genome 5: 121-122, 1994.

[44713] 18265. Durkin, A. S.; Maglott, D. R.; Vamvakopoulos, N. C.; Zoghbi, H. Y.; Nierman, W. C.: Assignment of an intron-containing human heat-shock protein gene (hsp90-beta, HSPCB) to chromosome 6 near TCTE1 (6p21) and two intronless pseudogenes to chromosomes 4 and 15 by polymerase chain reaction amplification from a panel of hybrid cell lines. Genomics 18:452-454, 1993.

[44714] 18266. Klinger, H. P.: Suppression of tumorigenicity in somatic cell hybrids. I. Suppression and reexpression of tumorigenicity in diploid human x D98/AH2 hybrids and independent segregation of tumorigenicity from other cell phenotypes. Cytogenet. Cell Genet. 27: 254-266, 1980.

[44715] 18267. Lichy, J. H.; Modi, W. S.; Seunanez, H. N.; Howley, P. M.: Identification of a human chromosome 11 gene which is differentially regulated in tumorigenic and nontumorigenic somatic cell hybrids of HeLa cells. Cell Growth Differ. 3: 541-548, 1992.

[44716] 18268. Stanbridge, E. J.: Suppression of malignancy in human cells. Nature 260:17-20, 1976.

[44717] 18269. Corrigall, V. M.; Arastu, M.; Khan, S.; Shah, C.; Fife,

M.; Smeets, T.; Tak, P.-P.; Panayi, G. S.: Functional IL-2 receptor beta (CD122) and gamma (CD132) chains are expressed by fibroblast-like synoviocytes: activation by IL-2 stimulates monocyte chemoattractant protein-1 production. *J. Immun.* 166: 4141-4147, 2001.

[44718] 18270. Gnarr, J. R.; Otani, H.; Wang, M. G.; McBride, O. W.; Sharon, M.; Leonard, W. J.: Human interleukin 2 receptor beta-chain gene: chromosomal localization and identification of 5-prime regulatory sequences. *Proc. Nat. Acad. Sci.* 87: 3440-3444, 1990.

[44719] 18271. Hatakeyama, M.; Tsudo, M.; Minamoto, S.; Kono, T.; Doi, T.; Miyata, T.; Miyasaka, M.; Taniguchi, T.: Interleukin-2 receptor beta chain gene: generation of three receptor forms by cloned human alpha and beta chain cDNA's. *Science* 244: 551-556, 1989.

[44720] 18272. Lamaze, C.; Dujancourt, A.; Baba, T.; Lo, C. G.; Benmerah, A.; Dautry-Varsat, A.: Interleukin 2 receptors and detergent-resistant membrane domains define a clathrin-independent endocytic pathway. *Molec. Cell* 7: 661-671, 2001.

[44721] 18273. Shibuya, H.; Yoneyama, M.; Nakamura, Y.; Harada, H.; Hatakeyama, M.; Minamoto, S.; Kono, T.; Doi, T.; White, R.; Taniguchi, T.: The human interleukin-2 receptor beta-

chain gene: genomic organization, promoter analysis and chromosomal assignment. *Nucleic Acids Res.* 18:3697–3703, 1990.

[44722] 18274. Suzuki, H.; Kundig, T. M.; Furlonger, C.; Wakeham, A.; Timms, E.; Matsuyama, T.; Schmits, R.; Simard, J. J. L.; Ohashi, P. S.; Griesser, H.; Taniguchi, T.; Paige, C. J.; Mak, T. W.: Deregulated T cell activation and autoimmunity in mice lacking interleukin-2 receptor beta. *Science* 268:1472–1476, 1995.

[44723] 18275. Tsudo, M.; Kitamura, F.; Miyasaka, M.: Characterization of the interleukin 2 receptor beta chain using three distinct monoclonal antibodies. *Proc. Nat. Acad. Sci.* 86: 1982–1986, 1989.

[44724] 18276. Alitalo, T.; Koistinen, R.; Aalto-Setälä, K.; Kontula, K.; Julkunen, M.; Janne, O.; de la Chapelle, A.: Gene for the human insulin-like growth factor-binding protein/placental protein 12: mapping to 7p12–p13 and demonstration of a BglII RFLP. (Abstract) *Cytogenet. Cell Genet.* 51:950 only, 1989.

[44725] 18277. Alitalo, T.; Kontula, K.; Koistinen, R.; Aalto-Setälä, K.; Julkunen, M.; Janne, O. A.; Seppälä, M.; de la Chapelle, A.: The gene encoding human low-molecular weight insulin-like growth-factor binding protein (IGF-BP25): re-

gional localization to 7p12–p13 and description of aDNA polymorphism. Hum. Genet. 83: 335–338, 1989.

- [44726] 18278. Ballard, F. J.; Baxter, R. C.; Binoux, M.; Clemmons, D. R.; Drop, S. L. S.; Hall, K.; Hintz, R. L.; Rechler, M. M.; Rutanen, E. M.; Schwander, J. C.: Report on the nomenclature of the IGF binding proteins. J. Clin. Endocr. Metab. 70: 817–818, 1990.
- [44727] 18279. Brewer, M. T.; Stetler, G. L.; Squires, C. H.; Thompson, R. C.; Busby, W. H.; Clemmons, D. R.: Cloning, characterization, and expression of a human insulin-like growth factor binding protein. Biochem. Biophys. Res. Commun. 152: 1289–1297, 1988.
- [44728] 18280. Brinkman, A.; Groffen, C.; Kortleve, D. J.; Geurts van Kessel, A.; Drop, S. L. S.: Isolation and characterization of a cDNA encoding the low molecular weight insulin-like growth factor binding protein (IBP-1). EMBO J. 7: 2417–2423, 1988.
- [44729] 18281. Brinkman, A.; Groffen, C. A. H.; Kortleve, D. J.; Drop, S. L. S.: Organization of the gene encoding the insulin-like growth factor binding protein IBP-1. Biochem. Biophys. Res. Commun. 157: 898–907, 1988.
- [44730] 18282. Ekstrand, J.; Ehrenborg, E.; Stern, I.; Stellan, B.; Zech, L.; Luthman, H.: The gene for insulin-like growth

factor binding protein-1 is localized to human chromosomal region 7p14-p12. *Genomics* 6: 413-418, 1990.

[44731] 18283. Popovici, R. M.; Lu, M.; Bhatia, S.; Faessen, G. H.; Giaccia, A. J.; Giudice, L. C.: Hypoxia regulates insulin-like growth factor-binding protein 1 in human fetal hepatocytes in primary culture: suggestive molecular mechanisms for in utero fetal growth restriction caused by uteroplacental insufficiency. *J. Clin. Endocr. Metab.* 86: 2653-2659, 2001.

[44732] 18284. Agarwal, N.; Hsieh, C. L.; Sills, D.; Swaroop, M.; Desai, B.; Francke, U.; Swaroop, A.: Sequence analysis, expression and chromosomal localization of a gene, isolated from a subtracted human retina cDNA library, that encodes an insulin-like growth factor binding protein (IGFBP2). *Exp. Eye Res.* 52: 549-561, 1991.

[44733] 18285. Allander, S. V.; Larsson, C.; Ehrenborg, E.; Suwanichkul, A.; Weber, G.; Morris, S. L.; Bajalica, S.; Kiefer, M. C.; Luthman, H.; Powell, D. R.: Characterization of the chromosomal gene and promoter for human insulin-like growth factor binding protein-5. *J. Biol. Chem.* 269:10891-10898, 1994.

[44734] 18286. Ehrenborg, E.; Vilhelmsdotter, S.; Bajalica, S.; Larsson, C.; Stern, I.; Koch, J.; Brondum-Nielsen, K.; Luthman,

H.: Structure and localization of the human insulin-like growth factor-binding protein 2 gene. *Biochem. Biophys. Res. Commun.* 176: 1250–1255, 1991.

[44735] 18287. Wood, W. I.; Cachianes, G.; Henzel, W. J.; Winslow, G. A.; Spencer, S. A.; Hellmiss, R.; Martin, J. L.; Baxter, R. C.: Cloning and expression of the growth hormone-dependent insulin-like growth factor-binding protein. *Molec. Endocr.* 2: 1176–1185, 1988.

[44736] 18288. Eklund, L.; Piuhola, J.; Komulainen, J.; Sormunen, R.; Ongvarrasopone, C.; Fassler, R.; Muona, A.; Ilves, M.; Ruskoaho, H.; Takala, T. E. S.; Pihlajaniemi, T.: Lack of type XV collagen causes a skeletal myopathy and cardiovascular defects in mice. *Proc. Nat. Acad. Sci.* 98:1194–1199, 2001.

[44737] 18289. Hagg, P. M.; Hagg, P. O.; Peltonen, S.; Autio-Harmainen, H.; Pihlajaniemi, T.: Location of type XV collagen in human tissues and its accumulation in the interstitial matrix of the fibrotic kidney. *Am. J. Path.* 150:2075–2086, 1997.

[44738] 18290. Hagg, P. M.; Horelli-Kuitunen, N.; Eklund, L.; Palotie, A.; Pihlajaniemi, T.: Cloning of mouse type XV collagen sequences and mapping of the corresponding gene to 4B1–3: comparison of mouse and human alpha–

1(XV)collagen sequences indicates divergence in the number of small collagenous domains. Genomics 45: 31–41, 1997.

[44739] 18291.Hagg, P. M.; Muona, A.; Lietard, J.; Kivirikko, S.; Pihlajaniemi, T.: Complete exon–intron organization of the human gene for the alpha–1 chain of type XV collagen (COL15A1) and comparison with the homologous Col18a1 gene. J. Biol. Chem. 273: 17824–17831, 1998.

[44740] 18292.Huebner, K.; Cannizzaro, L. A.; Jabs, E. W.; Kivirikko, S.; Manzone, H.; Pihlajaniemi, T.; Myers, J. C.: Chromosomal assignment of a gene encoding a new collagen type (COL15A1) to 9q21–q22. Genomics 14:220–224, 1992.

[44741] 18293.Kivirikko, S.; Heinamaki, P.; Rehn, M.; Honkanen, N.; Myers, J.C.; Pihlajaniemi, T.: Primary structure of the alpha–1 chain of human type XV collagen and exon–intron organization in the 3–prime region of the corresponding gene. J. Biol. Chem. 269: 4773–4779, 1994.

[44742] 18294.Muragaki, Y.; Abe, N.; Ninomiya, Y.; Olsen, B. R.; Ooshima, A.: The human alpha–1(XV) collagen chain contains a large amino–terminal non–triple helical domain with a tandem repeat structure and homology to alpha–1(XVIII) collagen. J. Biol. Chem. 269: 4042–4046, 1994.

- [44743] 18295. Myers, J. C.; Dion, A. S.; Abraham, V.; Amenta, P. S.: Type XV collagen exhibits a widespread distribution in human tissues but a distinct localization in basement membrane zones. *Cell Tissue Res.* 286:493–505, 1996.
- [44744] 18296. Myers, J. C.; Kivirikko, S.; Gordon, M. K.; Dion, A. S.; Pihlajaniemi, T.: Identification of a previously unknown human collagen chain, α -1(XV), characterized by extensive interruptions in the triple-helical region. *Proc. Nat. Acad. Sci.* 89: 10144–10148, 1992.
- [44745] 18297. Ramchandran, R.; Dhanabal, M.; Volk, R.; Waterman, M. J. F.; Segal, M.; Lu, H.; Knebelmann, B.; Sukhatme, V. P.: Antiangiogenic activity of restin, NC10 domain of human collagen XV: comparison to endostatin. *Biochem. Biophys. Res. Commun.* 255: 735–739, 1999.
- [44746] 18298. Rehn, M.; Hintikka, E.; Pihlajaniemi, T.: Primary structure of the α 1 chain of mouse type XVIII collagen, partial structure of the corresponding gene, and comparison of the α 1(XVIII) chain with its homologue, the α 1(XV) collagen chain. *J. Biol. Chem.* 269:13929–13935, 1994.
- [44747] 18299. Sasaki, T.; Larsson, H.; Tisi, D.; Claesson-Welsh, L.; Hohenester, E.; Timpl, R.: Endostatins derived from collagens XV and XVIII differ in structural and binding proper-

ties, tissue distribution and anti-angiogenic activity. *J.*

Molec. Biol. 301: 1179–1190, 2000.

[44748] 18300. Pan, T.-C.; Zhang, R.-Z.; Mattei, M.-G.; Timpl, R.; Chu, M.-L.: Cloning and chromosomal location of human alpha-1(XVI) collagen. *Proc. Nat. Acad. Sci.* 89: 6565–6569, 1992.

[44749] 18301. Yamaguchi, N.; Kimura, S.; McBride, O. W.; Hori, H.; Yamada, Y.; Kanamori, T.; Yamakoshi, H.; Nagai, Y.: Molecular cloning and partial characterization of a novel collagen chain, alpha-1(XVI), consisting of repetitive collagenous domains and cysteine-containing non-collagenous segments. *J. Biochem.* 112: 856–863, 1992.

[44750] 18302. Curtis, A. R. J.; Fey, C.; Morris, C. M.; Bindoff, L. A.; Ince, P. G.; Chinnery, P. F.; Coulthard, A.; Jackson, M. J.; Jackson, A. P.; McHale, D. P.; Hay, D.; Barker, W. A.; Markham, A. F.; Bates, D.; Curtis, A.; Burn, J.: Mutation in the gene encoding ferritin light polypeptide causes dominant adult-onset basal ganglia disease. *Nature Genet.* 28: 350–354, 2001.

[44751] 18303. Eiberg, H.; Bisgaard, M. L.; Mohr, J.: Linkage between alpha-1-B-glycoprotein (A1BG) and Lutheran (LU) red blood group system: assignment to chromosome 19: new genetic variants of A1BG. *Clin. Genet.* 36: 415–418,

1989.

- [44752] 18304.Eiberg, H.; Nielsen, L. S.; Gahne, B.; Juneja, R. K.; Mohr, J.: Exclusion data for the alpha-1-B glycoprotein (A1BG) polymorphism.(Abstract) Cytogenet. Cell Genet. 51: 994 only, 1989.
- [44753] 18305.Gahne, B.; Juneja, R. K.; Stratil, A.: Genetic polymorphism of human plasma alpha-1-B-glycoprotein: phenotyping by immunoblotting or by a simple method of 2-D electrophoresis. Hum. Genet. 76: 111-115, 1987.
- [44754] 18306.Ishioka, N.; Takahashi, N.; Putnam, F. W.: Amino acid sequence of human plasma alpha-1B-glycoprotein: homology to the immunoglobulin supergene family. Proc. Nat. Acad. Sci. 83: 2363-2367, 1986.
- [44755] 18307.Juneja, R. K.; Weitkamp, L. R.; Stratil, A.; Gahne, B.; Guttormsen, S. A.: Further studies of the plasma alpha-1-B-glycoprotein polymorphism: two new alleles and allele frequencies in Caucasians and in American blacks. Hum. Hered. 38: 267-272, 1988.
- [44756] 18308.Anderson, L.; Anderson, N. G.: High resolution two-dimensional electrophoresis of human plasma proteins. Proc. Nat. Acad. Sci. 74: 5421-5425, 1977.
- [44757] 18309.Boutin, B.; Feng, S. H.; Arnaud, P.: The genetic polymorphism of alpha(2)-HS glycoprotein: study by ultra-

thin-layer isoelectricfocusing and immunoblot. Am. J. Hum. Genet. 37: 1098–1105, 1985.

[44758] 18310.Cox, D. W.; Andrews, B. J.: Silver stain immunofixa-
tion for alpha-2-HS-glycoprotein:a new method for de-
tection of protein heterogeneity.In: Stathakos,D.: Elec-
trophoresis '82. Berlin: Walter de Gruyter (pub.) 1983.Pp.
243–247.

[44759] 18311.Cox, D. W.; Andrews, B. J.; Wills, D. E.: Genetic
polymorphismof alpha-2-HS-glycoprotein. Am. J. Hum.
Genet. 38: 699–706, 1986.

[44760] 18312.Cox, D. W.; Francke, U.; Allderdice, P. W.; McAlpine,
P. J.: Genemapping of human serum proteins using hep-
atoma hybrids and human chromosomedeletions and du-
plications. Genetics 107 (suppl.): s22–s23, 1984.

[44761] 18313.Eiberg, H.; Mohr, J.; Nielsen, L. S.: A2HS: new
methods of phenotypingand analysis of linkage relations:
assignment to chromosome 3. (Abstract) Cytogenet.Cell
Genet. 37: 461 only, 1984.

[44762] 18314.Gejyo, F.; Chang, J.–L.; Burgi, W.; Schmid, K.;
Offner, G. D.;Troxler, R. F.; Van Halbeek, H.; Dorland, L.;
Gerwig, G. J.; Vliegenthart,J. F. G.: Characterization of the
B-chain of human plasma alpha(2)HS-glycoprotein:the
complete amino acid sequence and primary structure of

its heteroglycan. J.Biol. Chem. 258: 4966–4971, 1983.

[44763] 18315.Jahnen–Dechent, W.: Personal Communication.
Meinz, Germany 7/22/1998.

[44764] 18316.Jahnen–Dechent, W.; Schinke, T.; Trindl, A.; Muller–
Esterl, W.;Sablitzky, F.; Kaiser, S.; Blessing, M.: Cloning
and targeted deletionof the mouse fetuin gene. J. Biol.
Chem. 272: 31496–31503, 1997.

[44765] 18317.Lee, C.–C.; Bowman, B. H.; Yang, F.: Human alpha–
2–HS–glycoprotein:the A and B chains with a connecting
sequence are encoded by a singlemRNA transcript. Proc.
Nat. Acad. Sci. 84: 4403–4407, 1987.

[44766] 18318.Hulsebos, T. J. M.; Jenkins, N. A.; Gilbert, D. J.;
Copeland, N.G.: The beta crystallin genes on human chro–
mosome 22 define a newregion of homology with mouse
chromosome 5. Genomics 25: 574–576,1995.

[44767] 18319.Lampi, K. J.; Ma, Z.; Shih, M.; Shearer, T. R.; Smith,
J. B.; Smith,D. L.; David, L. L.: Sequence analysis of beta–
A3, beta–B3, and beta–A4crystallins completes the identi–
fication of the major proteins inyoung human lens. J. Biol.
Chem. 272: 2268–2275, 1997.

[44768] 18320.van Rens, G.; Geurts van Kessel, A.; Bloemendal,
H.: Localizationof the beta–A4 crystallin (CRYBA4) gene on
human chromosome 22 inthe region q11.2–q13.1.

(Abstract) Cytogenet. Cell Genet. 58: 2052only, 1991.

- [44769] 18321.van Rens, G. L. M.; Geurts van Kessel, A. H. M.; Bloemendal, H.: Localization of the beta-A4-crystallin gene (CRYBA4) on human chromosome 22 in the region q11.2-q13.1. Cytogenet. Cell Genet. 61: 180-183, 1992.
- [44770] 18322.Brakenhoff, R. H.; Henskens, H. A. M.; van Rossum, M. W. P. C.; Lubsen, N. H.; Schoenmakers, J. G. G.: Activation of the gamma-E-crystallin pseudogene in the human hereditary Coppock-like cataract. Hum. Molec. Genet. 3: 279-283, 1994.
- [44771] 18323.den Dunnen, J. T.; Jongbloed, R. J. E.; Geurts van Kessel, A. H. M.; Schoenmakers, J. G. G.: Human lens gamma-crystallin sequences are located in the p12-qter region of chromosome 2. Hum. Genet. 70:217-221, 1985.
- [44772] 18324.Hejtmancik, J. F.: The genetics of cataract: our vision becomes clearer. (Editorial) Am. J. Hum. Genet. 62: 520-525, 1998.
- [44773] 18325.Heon, E.; Priston, M.; Schorderet, D. F.; Billingsley, G. D.; Girard, P. O.; Lubsen, N.; Munier, F. L.: The gamma-crystallins and human cataracts: a puzzle made clearer. Am. J. Hum. Genet. 65: 1261-1267, 1999. Note: Erratum: Am. J. Hum. Genet. 66: 753 only, 2000.
- [44774] 18326.Klopp, N.; Favor, J.; Loster, J.; Lutz, R. B.;

Neuhauser-Klaus,A.; Prescott, A.; Pretsch, W.; Quinlan, R. A.; Sandilands, A.; Vrensen,G. F. J. M.; Graw, J.: Three murine cataract mutants (Cat2) are defectivein different gamma-crystallin genes. *Genomics* 52: 152–158, 1998.

[44775] 18327.Piatigorsky, J.: Lens crystallins and their gene families. *Cell* 38:620–621, 1984.

[44776] 18328.Shiloh, Y.; Donlon, T.; Bruns, G.; Breitman, M. L.; Tsui, L.–C.: Assignment of the human gamma-crystallin gene cluster (CRYG) tothe long arm of chromosome 2, region q33–36. *Hum. Genet.* 73: 17–19,1986.

[44777] 18329.Siezen, R. J.; Thomson, J. A.; Kaplan, E. D.; Benedek, G. B.:Human lens gamma-crystallins: isolation, identification, and characterizationof the expressed gene products. *Proc. Nat. Acad. Sci.* 84: 6088–6092,1987.

[44778] 18330.Skow, L. C.; Donner, M. E.; Huang, S.–M.; Gardner, J. M.; Taylor,B. A.; Beamer, W. G.; Lalley, P. A.: Mapping of mouse gamma crystallin genes on chromosome 1. *Biochem. Genet.* 26: 557–570, 1988.

[44779] 18331.Tsui, L.–C.; Breitman, M. L.; Meakin, S. O.; Willard, H. F.; Shiloh,Y.; Donlon, T.; Bruns, G.: Localization of the human gamma-crystallin gene cluster (CRYG) to the long arm of chromosome 2, region q33–q35.(Abstract) *Cytogenet. Cell Genet.* 40: 763–764, 1985.

- [44780] 18332.Vidal, S. M.; Epstein, D. J.; Malo, D.; Weith, A.; Vekemans, M.;Gros, P.: Identification and mapping of six microdissected genomicDNA probes to the proximal region of mouse chromosome 1. *Genomics* 14:32–37, 1992.
- [44781] 18333.Fajans, S. S.; Bell, G. I.; Polonsky, K. S.: Molecular mechanismsand clinical pathophysiology of maturity–onset diabetes of the young. *NewEng. J. Med.* 345: 971–980, 2001.
- [44782] 18334.Lindner, T.; Gragnoli, C.; Furuta, H.; Cockburn, B. N.; Petzold,C.; Rietzsch, H.; Weiss, U.; Schulze, J.; Bell, G. I.: Hepatic functionin a family with a nonsense mutation (R154X) in the hepatocyte nuclearfactor–4–alpha/MODY1 gene. *J. Clin. Invest.* 100: 1400–1405, 1997.
- [44783] 18335.Moller, A. M.; Dalgaard, L. T.; Ambye, L.; Hansen, L.; Schmitz,O.; Hansen, T.; Pedersen, O.: A novel Phe75fsdelT mutation in thehepatocyte nuclear factor–4–alpha gene in a Danish pedigree with maturity–onsetdiabetes of the young. *J. Clin. Endocr. Metab.* 84: 367–369, 1999.
- [44784] 18336.Thomas, H.; Jaschkowitz, K.; Bulman, M.; Frayling, T. M.; Mitchell,S. M. S.; Roosen, S.; Lingott–Frieg, A.; Tack, C. J.; Ellard, S.;Ryffel, G. U.; Hattersley, A. T.: A distant upstream promoter ofthe HNF–4–alpha gene connects the

transcription factors involved in maturity-onset diabetes of the young. *Hum. Molec. Genet.* 10: 2089–2097, 2001.

[44785] 18337. Joseph, L. J.; Le Beau, M. M.; Jamieson, G. A., Jr.; Acharya, S.; Shows, T. B.; Rowley, J. D.; Sukhatme, V. P.: Molecular cloning, sequencing, and mapping of EGR2, a human early growth response gene encoding a protein with 'zinc-binding finger' structure. *Proc. Nat. Acad. Sci.* 85: 7164–7168, 1988.

[44786] 18338. Nagarajan, R.; Svaren, J.; Le, N.; Araki, T.; Watson, M.; Milbrandt, J.: EGR2 mutations in inherited neuropathies dominant-negatively inhibit myelin gene expression. *Neuron* 30: 355–368, 2001.

[44787] 18339. Timmerman, V.; De Jonghe, P.; Ceuterick, C.; De Vriendt, E.; Lofgren, A.; Nelis, E.; Warner, L. E.; Lupski, J. R.; Martin, J.-J.; Van Broeckhoven, C.: Novel missense mutation in the early growth response 2 gene associated with Dejerine-Sottas syndrome phenotype. *Neurology* 52: 1827–1832, 1999.

[44788] 18340. Warner, L. E.; Mancias, P.; Butler, I.; Lupski, J. R.: Mutation in the early growth response 2 (EGR2) transcription factor associated with recessive congenital hypomyelinating neuropathy (CHN). (Abstract) *Am. J. Hum. Genet.* 61 (suppl.): A350 only, 1997.

- [44789] 18341. Warner, L. E.; Mancias, P.; Butler, I. J.; McDonald, C. M.; Keppen, L.; Koob, K. G.; Lupski, J. R.: Mutations in the early growth response 2 (EGR2) gene are associated with hereditary myelinopathies. *Nature Genet.* 18: 382–384, 1998.
- [44790] 18342. Warner, L. E.; Svaren, J.; Milbrandt, J.; Lupski, J. R.: Functional consequences of mutations in the early growth response 2 gene (EGR2) correlate with severity of human myelinopathies. *Hum. Molec. Genet.* 8:1245–1251, 1999.
- [44791] 18343. Wu, J.; Joseph, L.; Sukhatme, V. P.; Kidd, K. K.: A HindIII polymorphism identified by the human early growth response gene 2 (EGR2) on chromosome
- [44792] 18344. *Nucleic Acids Res.* 16: 11855 only, 1988.
- [44793] 18345. Kaneda, Y.; Hayes, H.; Uchida, T.; Yoshida, M. C.; Okada, Y.: Regional assignment of five genes on human chromosome 19. *Chromosoma* 95:8–12, 1987.
- [44794] 18346. Hoeffler, J. P.; Meyer, T. E.; Yun, Y.; Jameson, J. L.; Habener, J. F.: Cyclic AMP–responsive DNA–binding protein: structure based on a cloned placental cDNA. *Science* 242: 1430–1433, 1988.
- [44795] 18347. Chan, E. F.; Gat, U.; McNiff, J. M.; Fuchs, E.: A common human skin tumour is caused by activating mutations in beta–catenin. *Nature Genet.* 21: 410–413, 1999.

- [44796] 18348.Horelli-Kuitunen, N.; Kvist, A.-P.; Helaakoski, T.; Kivirikko,K.; Pihlajaniemi, T.; Palotie, A.: The order and transcriptionalorientation of the human COL13A1 and P4HA genes on chromosome 10 longarm determined by high-resolution FISH. *Genomics* 46: 299–302, 1997.
- [44797] 18349.Pajunen, L.; Tamminen, M.; Solomon, E.; Pihlajaniemi, T.: Assignmentof the gene coding for the alpha 1 chain of collagen type XIII (COL13A1)to human chromosome region 10q11–qter. *Cytogenet. Cell Genet.* 52:190–193, 1989.
- [44798] 18350.Shows, T. B.; Tikka, L.; Byers, M. G.; Eddy, R. L.; Haley, L. L.;Henry, W. M.; Prockop, D. J.; Tryggvason, K.: Assignment of the humancollagen alpha–1(XIII) chain gene (COL13A1) to the q22 region of chromosome10. *Genomics* 5: 128–133, 1989.
- [44799] 18351.Tikka, L.; Pihlajaniemi, T.; Henttu, P.; Prockop, D. J.; Tryggvason,K.: Gene structure for the alpha–1 chain of a human short-chain collagen(type XIII) with alternatively spliced transcripts and translationtermination codon at the 5–prime end of the last exon. *Proc. Nat.Acad. Sci.* 85: 7491–7495, 1988.
- [44800] 18352.Cunningham, B. A.; Hemperly, J. J.; Murray, B. A.; Prediger, E.A.; Brackenbury, R.; Edelman, G. M.: Neural cell

adhesion molecule:structure, immunoglobulin-like domains, cell surface modulation, and alternative RNA splicing. *Science* 236: 799–806, 1987.

[44801] 18353.D'Eustachio, P.; Davisson, M. T.: Resolution of the staggerer(sg) mutation from the neural cell adhesion molecule locus (Ncam) on mouse chromosome 9. *Mammalian Genome* 4: 278–280, 1993.

[44802] 18354.D'Eustachio, P.; Owens, G. C.; Edelman, G. M.; Cunningham, B. A.: Chromosomal location of the gene encoding the neural cell adhesion molecule (N-CAM) in the mouse. *Proc. Nat. Acad. Sci.* 82: 7631–7635, 1985.

[44803] 18355.Lin, D. M.; Fetter, R. D.; Kopczynski, C.; Grenningloh, G.; Goodman, C. S.: Genetic analysis of fasciclin II in *Drosophila*: defasciculation, refasciculation, and altered fasciculation. *Neuron* 13: 1055–1069, 1994.

[44804] 18356.Altare, F.; Jouanguy, E.; Lamhamedi-Cherradi, S.; Fondaneche, M.-C.; Fizame, C.; Ribierre, F.; Merlin, G.; Dembic, Z.; Schreiber, R.; Lisowska-Grospierre, B.; Fischer, A.; Seboun, E.; Casanova, J.-L.: A causative relationship between mutant IFN γ R1 alleles and impaired cellular response to IFN- γ in a compound heterozygous child. (Letter) *Am. J. Hum. Genet.* 62:723–726, 1998.

[44805] 18357.Dalton, D. K.; Pitts-Meek, S.; Keshav, S.; Figari, I.

S.; Bradley,A.; Stewart, T. A.: Multiple defects of immune cell function in micewith disrupted interferon-gamma genes. Science 259: 1739-1742, 1993.

[44806] 18358.Dessein, A. J.; Hillaire, D.; Elwali, N. E. M. A.; Marquet, S.;Mohamed-Ali, Q.; Mirghani, A.; Henri, S.; Abdelhameed, A. A.; Saeed,O. K.; Magzoub, M. M. A.; Abel, L.: Severe hepatic fibrosis in Schistosomamansonii infection is controlled by a major locus that is closely linkedto the interferon-gamma receptor gene. Am. J. Hum. Genet. 65: 709-721,1999.

[44807] 18359.Fellous, M.: Personal Communication. Paris, France 10/24/1986.

[44808] 18360.Fellous, M.; Couillin, P.; Rosa, F.; Metezeau, P.; Foubert, C.;Gross, M. S.; Frezal, J.; Van Cong, N.: Receptor for human gammainterferon is specified by human chromosome 18. (Abstract) Cytogenet.Cell Genet. 40: 627-628, 1985.

[44809] 18361.Jouanguy, E.; Altare, F.; Lamhamedi, S.; Revy, P.; Emile, J.-F.;Newport, M.; Levin, M.; Blanche, S.; Seboun, E.; Fischer, A.; Casanova,J.-L.: Interferon-gamma-receptor deficiency in an infant with fatalbacille Calmette-Guerin infection. New Eng. J. Med. 335: 1956-1961,1996.

[44810] 18362.Jouanguy, E.; Dupuis, S.; Pallier, A.; Doffinger, R.;

Fondaneche, M.-C.; Fieschi, C.; Lamhamedi-Cherradi, S.; Altare, F.; Emile, J.-F.; Lutz, P.; Bordigoni, P.; Cokugras, H.; Akcakaya, N.; Landman-Parker, J.; Donnadieu, J.; Camcioglu, Y.; Casanova, J.-L.: In a novel form of IFN-gamma receptor 1 deficiency, cell surface receptors fail to bind IFN-gamma. *J. Clin. Invest.* 105: 1429–1436, 2000.

[44811] 18363. Jouanguy, E.; Lamhamedi-Cherradi, S.; Lammas, D.; Dorman, S. E.; Fondaneche, M.-C.; Dupuis, S.; Doffinger, R.; Altare, F.; Girdlestone, J.; Emile, J.-F.; Ducoulombier, H.; Edgar, D.; and 10 others: A human IFNGR1 small deletion hotspot associated with dominant susceptibility to mycobacterial infection. *Nature Genet.* 21: 370–378, 1999.

[44812] 18364. Kaplan, D. H.; Shankaran, V.; Dighe, A. S.; Stockert, E.; Aguet, M.; Old, L. J.; Schreiber, R. D.: Demonstration of an interferon gamma-dependent tumor surveillance system in immunocompetent mice. *Proc. Nat. Acad. Sci.* 95: 7556–7561, 1998.

[44813] 18365. Le Coniat, M.; Alcaide-Loridan, C.; Fellous, M.; Berger, R.: Human interferon gamma receptor 1 (IFNGR1) gene maps to chromosome region 6q23–6q24. *Hum. Genet.* 84: 92–94, 1989.

[44814] 18366. Lekstrom-Himes, J. A.; Gallin, J. I.: Immunodeficiency

ciency diseases caused by defects in phagocytes. *New Eng. J. Med.* 343: 1703–1714, 2000.

- [44815] 18367. Levin, M.; Newport, M. J.; D'Souza, S.; Kalabalikis, P.; Brown, I. N.; Lenicker, H. M.; Agius, P. V.; Davies, E. G.; Thrasher, A.; Klein, N.; Blackwell, J. M.: Familial disseminated atypical mycobacterial infection in childhood: a human mycobacterial susceptibility gene? *Lancet* 345:79–83, 1995.
- [44816] 18368. Mariano, T. M.; Kozak, C. A.; Langer, J. A.; Pestka, S.: The mouse immune interferon receptor gene is located on chromosome 10. *J. Biol. Chem.* 262: 5812–5814, 1987.
- [44817] 18369. Deloukas, P.; Schuler, G. D.; Gyapay, G.; Beasley, E. M.; Soderlund, C.; Rodriguez-Tome, P.; Hui, L.; Matisse, T. C.; McKusick, K. B.; Beckmann, J. S.; Bentolila, S.; Bihoreau, M.-T.; and 53 others: A physical map of 30,000 human genes. *Science* 282: 744–746, 1998.
- [44818] 18370. Wu, W. J.; Erickson, J. W.; Lin, R.; Cerione, R. A.: The gamma-subunit of the coatamer complex binds Cdc42 to mediate transformation. *Nature* 405:800–804, 2000.
- [44819] 18371. Davis, S. T.; Benson, B. G.; Bramson, H. N.; Chapman, D. E.; Dickerson, S. H.; Dold, K. M.; Eberwein, D. J.; Edelstein, M.; Frye, S. V.; Gampe, R. T., Jr.; Griffin, R. J.; Harris, P. A.; and 14 others: Prevention of chemotherapy-in-

duced alopecia in rats by CDK inhibitors. Science 291:134–137, 2001.

[44820] 18372. De Bondt, H. L.; Rosenblatt, J.; Jancarik, J.; Jones, H. D.; Morgan, D. O.; Kim, S.-H.: Crystal structure of cyclin-dependent kinase 2. Nature 363:595–602, 1993.

[44821] 18373. Basu, S.; Binder, R. J.; Ramalingam, T.; Srivastava, P. K.: CD91 is a common receptor for heat shock proteins gp96, hsp90, hsp70, and calreticulin. Immunity 14: 303–313, 2001.

[44822] 18374. Beisiegel, U.; Weber, W.; Ihrke, G.; Herz, J.; Stanley, K. K.: The LDL-receptor-related protein, LRP, is an apolipoprotein E-binding protein. Nature 341: 162–164, 1989.

[44823] 18375. Binder, R. J.; Han, D. K.; Srivastava, P. K.: CD91: a receptor for heat shock protein gp96. Nature Immun. 1: 151–155, 2000.

[44824] 18376. Forus, A.; Maelandsmo, G. M.; Fodstad, Y.; Myklebost, O.: The genes for the alpha-2-macroglobulin receptor/LDL receptor-related protein and GLI are located within a chromosomal segment of about 300 kilobases and are coamplified in a rhabdomyosarcoma cell line. (Abstract) Cytogenet. Cell Genet. 58: 1977 only, 1991.

[44825] 18377. Forus, A.; Myklebost, O.: A physical map of a

1.3-Mb region on the long arm of chromosome 12, spanning the GLI and LRP loci. *Genomics* 14:117–120, 1992.

- [44826] 18378. Hilliker, C.; Van Leuven, F.; Van Den Berghe, H.: Assignment of the gene coding for the α - μ 2-macroglobulin receptor to mouse chromosome 15 and to human chromosome 12q13–q14 by isotopic and nonisotopic *in situ* hybridization. *Genomics* 13: 472–474, 1992.
- [44827] 18379. Kristensen, T.; Moestrup, S. K.; Gliemann, J.; Bendtsen, L.; Sand, O.; Sottrup-Jensen, L.: Evidence that the newly cloned low-density-lipoprotein receptor related protein (LRP) is the α -2-macroglobulin receptor. *FEBS Lett.* 276: 151–155, 1990.
- [44828] 18380. Lendon, C. L.; Talbot, C. J.; Craddock, N. J.; Han, S. W.; Wragg, M.; Morris, J. C.; Goate, A. M.: Genetic association studies between dementia of the Alzheimer's type and three receptors for apolipoprotein E in a Caucasian population. *Neurosci. Lett.* 222: 187–190, 1997.
- [44829] 18381. Myklebost, O.; Arheden, K.; Rogne, S.; Geurts van Kessel, A.; Mandahl, N.; Herz, J.; Stanley, K.; Heim, S.; Mitelman, F.: The gene for the human putative apoE receptor is on chromosome 12 in the segment q13–14. *Genomics* 5: 65–69, 1989.

- [44830] 18382. Pericak-Vance, M. A.; Bass, M. P.; Yamaoka, L. H.; Gaskell, P. C.; Scott, W. K.; Terwedow, H. A.; Menold, M. M.; Conneally, P. M.; Small, G. W.; Vance, J. M.; Saunders, A. M.; Roses, A. D.; Haines, J. L.: Complete genomic screen in late-onset familial Alzheimer disease: evidence for a new locus on chromosome 12. *J.A.M.A.* 278: 1237-1241, 1997.
- [44831] 18383. Scott, W. K.; Yamaoka, L. H.; Bass, M. P.; Gaskell, P. C.; Conneally, P. M.; Small, G. W.; Farrer, L. A.; Auerbach, S. A.; Saunders, A. M.; Roses, A. D.; Haines, J. L.; Pericak-Vance, M. A.: No genetic association between the LRP receptor and sporadic or late-onset familial Alzheimer disease. *Neurogenetics* 1: 179-183, 1998.
- [44832] 18384. Strickland, D. K.; Ashcom, J. D.; Williams, S.; Burgess, W. H.; Migliorini, M.; Argraves, W. S.: Sequence identity between the alpha-2-macroglobulin receptor and low density lipoprotein receptor-related protein suggest that this molecule is a multifunctional receptor. *J. Biol. Chem.* 265:17401-17404, 1990.
- [44833] 18385. Yochum, J.; Greenwald, I.: A gene for a low density lipoprotein receptor-related protein in the nematode *Caenorhabditis elegans*. *Proc. Nat. Acad. Sci.* 90: 4572-4576, 1993.

- [44834] 18386.Ladiaz, J. A. A.; Karathanasis, S. K.: Regulation of the apolipoproteinAI gene by ARP-1, a novel member of the steroid receptor superfamily. *Science* 251:561–565, 1991.
- [44835] 18387.Modi, W. S.; Seuanez, H.; Mietus-Snyder, M.; O'Brien, S. J.; Karathanasis, S. K.: Chromosomal localization of the ARP-1 gene to 15q26. (Abstract) *Cytogenet. Cell Genet.* 58: 1995 only, 1991.
- [44836] 18388.Pereira, F. A.; Qiu, Y.; Zhou, G.; Tsai, M.-J.; Tsai, S. Y.: The orphan nuclear receptor COUP-TFII is required for angiogenesis and heart development. *Genes Dev.* 13: 1037–1049, 1999.
- [44837] 18389.Widom, R. L.; Ladiaz, J. A. A.; Kouidou, S.; Karathanasis, S. K.: Synergistic interactions between transcription factors control expression of the apolipoprotein AI gene in liver cells. *Molec. Cell. Biol.* 11:677–687, 1991.
- [44838] 18390.Ikeda, H.; Yamaguchi, M.; Sugai, S.; Aze, Y.; Narumiya, S.; Kakizuka, A.: Expanded polyglutamine in the Machado-Joseph disease protein induces cell death in vitro and in vivo. *Nature Genet.* 13: 196–202, 1996.
- [44839] 18391.Kang, H.; Sun, L. D.; Atkins, C. M.; Soderling, T. R.; Wilson, M. A.; Tonegawa, S.: An important role of neural activity-dependent CaMKIV signaling in the consolidation

of long-term memory. *Cell* 106:771–783, 2001.

[44840] 18392. Raman, V.; Blaeser, F.; Ho, N.; Engle, D. L.; Williams, C. B.; Chatila, T. A.: Requirement for Ca^{2+} /calmodulin-dependent kinase type IV/Gr in setting the thymocyte selection threshold. *J. Immun.* 167:6270–6278, 2001.

[44841] 18393. Santisteban, I.; Arredondo-Vega, F. X.; Kelly, S.; Mary, A.; Fischer, A.; Hummell, D. S.; Lawton, A.; Sorensen, R. U.; Stiehm, E. R.; Uribe, L.; Weinberg, K.; Hershfield, M. S.: Novel splicing, missense, and deletion mutations in seven adenosine deaminase-deficient patients with late/delayed onset of combined immunodeficiency disease: contribution of genotype to phenotype. *J. Clin. Invest.* 92: 2291–2302, 1993. 100. Schmalstieg, F. C.; Mills, G. C.; Tsuda, H.; Goldman, A. S.: Severe combined immunodeficiency in a child with a healthy adenosine deaminase deficient mother. *Pediatr. Res.* 17: 935–940, 1983. 101. Schrader, W. P.; Pollara, B.; Meuwissen, H. J.: Characterization of the residual adenosine deaminating activity in the spleen of a patient with combined immunodeficiency disease and adenosine deaminase deficiency. *Proc. Nat. Acad. Sci.* 75: 446–450, 1978. 102. Scott, C. R.; Chen, S.-H.; Giblett, E. R.: Deletion of the carrier state in combined im-

immunodeficiency disease associated with adenosine deaminase deficiency. *J. Clin. Invest.* 53: 1194–1196, 1974.103. Shovlin, C. L.; Hughes, J. M. B.; Simmonds, H. A.; Fairbanks, L.; Deacock, S.; Lechler, R.; Roberts, I.; Webster, A. D. B.: Adult presentation of adenosine deaminase deficiency. (Letter) *Lancet* 341:1471, 1993.104. Shovlin, C. L.; Simmonds, H. A.; Fairbanks, L. D.; Deacock, S.J.; Hughes, J. M. B.; Lechler, R. I.; Webster, A. D. B.; Sun, X.-M.; Webb, J. C.; Soutar, A. K.: Adult onset immunodeficiency caused by inherited adenosine deaminase deficiency. *J. Immun.* 153: 2331–2339, 1994.105. Spencer, N.; Hopkinson, D. A.; Harris, H.: Adenosine deaminase polymorphism in man. *Ann. Hum. Genet.* 32: 9–14, 1968.106. Stephan, V.; Wahn, V.; Le Deist, F.; Dirksen, U.; Broker, B.; Muller-Fleckenstein, I.; Horneff, G.; Schroten, H.; Fischer, A.; deSaint Basile, G.: Atypical X-linked severe combined immunodeficiency due to possible spontaneous reversion of the genetic defect in T cells. *New Eng. J. Med.* 335: 1563–1567, 1996.107. Tariverdian, G.; Ritter, H.: Adenosine deaminase polymorphism (EC 3.5.4.4): formal genetics and linkage relations. *Humangenetik* 7:176–178, 1969.108. Tischfield, J. A.; Creagan, R. P.; Nichols, E. A.; Ruddle, F.H.: Assignment of a gene for adenosine deaminase to human chromo-

some20. Hum. Hered. 24: 1–11, 1974.109. Tzall, S.; Ellenbogen, A.; Eng, F.; Hirschhorn, R.: Identification and characterization of nine RFLPs at the adenosine deaminase (ADA) locus. Am. J. Hum. Genet. 44: 864–875, 1989.110. Umetsu, D. T.; Schlossman, C. M.; Ochs, H. D.; Hershenfeld, M.S.: Heterogeneity of phenotype in two siblings with adenosine deaminase deficiency. J. Allergy Clin. Immun. 93: 543–550, 1994.111. Valerio, D.; Dekker, B. M. M.; Duyvesteyn, M. G. C.; van der Voorn, L.; Berkvens, T. M.; van Ormondt, H.; van der Eb, A. J.: One adenosine deaminase allele in a patient with severe combined immunodeficiency contains a point mutation abolishing enzyme activity. EMBO J. 5:113–119, 1986.112. Valerio, D.; Duyvesteyn, M. G. C.; Dekker, B. M. M.; Weeda, G.; Berkvens, T. M.; van der Voorn, L.; van Ormondt, H.; van der Eb, A. J.: Adenosine deaminase: characterization and expression of a gene with a remarkable promoter. EMBO J. 4: 437–443, 1985.113. Valerio, D.; Duyvesteyn, M. G. C.; Meera Khan, P.; Pearson, P.L.; Geurts van Kessel, A.; van Ormondt, H.: Direct assignment of ADA gene to chromosome 20. (Abstract) Cytogenet. Cell Genet. 37:599, 1984.114. Valerio, D.; Duyvesteyn, M. G. C.; van Ormondt, H.; Meera Khan, P.; van der Eb, A. J.: Adenosine deaminase

(ADA) deficiency in cells derived from humans with severe combined immunodeficiency is due to an aberration of the ADA protein. *Nucleic Acids Res.* 12:

1015–1024, 1984. 115. Valerio, D.; McIvor, R. S.; Williams, S. R.; Duyvesteyn, M. G.C.; van Ormondt, H.; van der Eb, A.

J.; Martin, D. W., Jr.: Cloning of human adenosine deaminase cDNA and expression in mouse cells. *Gene*

31:147–153, 1984. 116. Van der Weyden, M. B.; Kelley, W.

N.: Adenosine deaminase deficiency in severe combined immunodeficiency: evidence for a posttranslational defect.

(Abstract) *J. Clin. Invest.* 53: 81A–82A, 1974. 117.

Wakamiya, M.; Blackburn, M. R.; Jurecic, R.; McArthur, M.

J.; Geske, R. S.; Cartwright, J., Jr.; Mitani, K.; Vaishnav, S.;

Belmont, J. W.; Kellems, R. E.; Finegold, M. J.; Montgomery,

Jr., C. A.; Bradley, A.; Caskey, C. T.: Disruption of the

adenosine deaminase gene causes hepatocellular impairment and perinatal lethality in mice. *Proc. Nat. Acad. Sci.*

92: 3673–3677, 1995. 118. Weitkamp, L. R.: Further data

on the genetic linkage relationships of the adenosine deaminase locus. *Hum. Hered.* 21: 351–356, 1971. 119.

Weitkamp, L. R.: Genetic linkage relationships of the ADA and 6-PGD loci in 'Humangenetik.' (Letter) *Humangenetik*

15: 359–360, 1972. 120. Wiginton, D. A.; Adrian, G. S.;

Friedman, R. L.; Suttle, D. P.; Hutton, J. J.: Cloning of cDNA sequences of human adenosine deaminase. *Proc. Nat. Acad. Sci.* 80: 7481–7485, 1983.121. Wiginton, D. A.; Adrian, G. S.; Hutton, J. J.: Sequence of human adenosine deaminase cDNA including the coding region and a small intron. *Nucleic Acids Res.* 12: 2439–2446, 1984.122. Wiginton, D. A.; Hutton, J. J.: Immunoreactive protein in adenosine deaminase deficient human lymphoblast cell lines. *J. Biol. Chem.* 257:3211–3217, 1982.123. Wiginton, D. A.; Kaplan, D. J.; States, J. C.; Akeson, A. L.; Perme, C. M.; Bilyk, I. J.; Vaughn, A. J.; Lattier, D. L.; Hutton, J. J.: Complete sequence and structure of the gene for human adenosine deaminase. *Biochemistry* 25: 8234–8244, 1986.124. Yokoyama, S.; Hayashi, T.; Yoshimura, Y.; Irimada, K.; Saito, T.; Akiba, T.; Tsuchiya, S.: Severe combined immunodeficiency disease with adenosine deaminase deficiency. *Tohoku J. Exp. Med.* 129: 197–202, 1979.125. Yount, J.; Nichols, P.; Ochs, H. D.; Hammar, S. P.; Scott, C. R.; Chen, S.-H.; Giblett, E. R.; Wedgwood, R. J.: Absence of erythrocyte adenosine deaminase associated with severe combined immunodeficiency. *J. Pediatr.* 84: 173–177, 1974.126. Ziegler, J. B.; Lee, C. H.; Van Der Weyden, M. B.; Bagnara, A. S.; Beveridge, J.: Severe

combined immunodeficiency and adenosine deaminase deficiency: failure of enzyme replacement therapy.

Arch. Dis. Child. 55: 452–457, 1980.127. Ziegler, J. B.; Van Der Weyden, M. B.; Lee, C. H.; Daniel, A.: Prenatal diagnosis for adenosine deaminase deficiency. J. Med. Genet. 18: 154–156, 1981.

- [44842] 18394. Sikela, J. M.; Adamson, M. C.; Wilson-Shaw, D.; Kozak, C. A.: Genetic mapping of the gene for Ca(2+)/calmodulin-dependent protein kinase IV (Camk-4) to mouse chromosome 18. Genomics 8: 579–582, 1990.
- [44843] 18395. Sikela, J. M.; Law, M. L.; Kao, F.-T.; Hartz, J. A.; Wei, Q.; Hahn, W. E.: Chromosomal localization of the human gene for brain Ca(2+)/calmodulin-dependent protein kinase type IV. Genomics 4: 21–27, 1989.
- [44844] 18396. Wei, F.; Qiu, C.-S.; Liauw, J.; Robinson, D. A.; Ho, N.; Chatila, T.; Zhuo, M.: Calcium-calmodulin-dependent protein kinase IV is required for fear memory. Nature Neurosci. 5: 573–579, 2002.
- [44845] 18397. Wu, H.; Kanatous, S. B.; Thurmond, F. A.; Gallardo, T.; Isotani, E.; Bassel-Duby, R.; Williams, R. S.: Regulation of mitochondrial biogenesis in skeletal muscle by CaMK. Science 296: 349–352, 2002.
- [44846] 18398. Wu, J. Y.; Ribar, T. J.; Cummings, D. E.; Burton, K.

A.; McKnight, G. S.; Means, A. R.: Spermiogenesis and exchange of basic nuclear proteins are impaired in male germ cells lacking Camk4. *Nature Genet.* 25:448–452, 2000.

[44847] 18399. Dooley, T. P.: Personal Communication. San Antonio, Tex. 7/21/1992.

[44848] 18400. Dooley, T. P.; Weiland, K. L.; Simon, M.: cDNA sequence of human p11 calpactin I light chain. *Genomics* 13: 866–868, 1992.

[44849] 18401. Harder, T.; Kube, E.; Gerke, V.: Cloning and characterization of the human gene encoding p11: structural similarity to other members of the S-100 gene family. *Gene* 113: 269–274, 1992.

[44850] 18402. Kube, E.; Weber, K.; Gerke, V.: Primary structure of human, chicken, and *Xenopus laevis* p11, a cellular ligand of the Src-kinase substrate, annexin II. *Gene* 102: 255–259, 1991.

[44851] 18403. Okuse, K.; Malik-Hall, M.; Baker, M. D.; Poon, W.-Y. L.; Kong, H.; Chao, M. V.; Wood, J. N.: Annexin II light chain regulates sensory neuron-specific sodium channel expression. *Nature* 417: 653–656, 2002.

[44852] 18404. Saris, C. J. M.; Kristensen, T.; D'Eustachio, P.; Hicks, L. J.; Noonan, D. J.; Hunter, T.; Tack, B. F.: cDNA sequence

and tissue distribution of the mRNA for bovine and murine p11, the S100-related light chain of the protein-tyrosine kinase substrate p36 (calpactin I). *J. Biol. Chem.* 262: 10663–10671, 1987.

[44853] 18405. Inazawa, J.; Nakagawa, H.; Misawa, S.; Abe, T.; Minoshima, S.; Fukuyama, R.; Maki, M.; Murachi, T.; Hatanaka, M.; Shimizu, N.: Assignment of the human calpastatin gene (CAST) to chromosome 5 at region q14–q22. *Cytogenet. Cell Genet.* 54: 156–158, 1990.

[44854] 18406. Inazawa, J.; Nakagawa, H.; Misawa, S.; Abe, T.; Minoshima, S.; Fukuyama, R.; Maki, M.; Murachi, T.; Hatanaka, M.; Shimizu, N.: Assignment of the human calpastatin gene (CAST) to chromosome 5 at region q15–q21. (Abstract) *Cytogenet. Cell Genet.* 58: 1898 only, 1991.

[44855] 18407. Mimori, T.; Suganuma, K.; Tanami, Y.; Nojima, T.; Matsumura, M.; Fujii, T.; Yoshizawa, T.; Suzuki, K.; Akizuki, M.: Autoantibodies to calpastatin (an endogenous inhibitor for calcium-dependent neutral protease, calpain) in systemic rheumatic diseases. *Proc. Nat. Acad. Sci.* 92: 7267–7271, 1995.

[44856] 18408. Pontremoli, S.; Salamino, F.; Sparatore, B.; De Tullio, R.; Pontremoli, R.; Melloni, E.: Characterization of the

calpastatin defect in erythrocytes from patients with essential hypertension. *Biochem. Biophys. Res. Commun.* 157: 867–874, 1988.

[44857] 18409. Carritt, B.: Somatic cell genetic evidence for the presence of a gene for citrullinemia on human chromosome 9. *Cytogenet. Cell Genet.* 19:44–48, 1977.

[44858] 18410. Carritt, B.; Goldfarb, P. S. G.; Hooper, M. L.; Slack, C.: Chromosome assignment of a human gene for argininosuccinate synthetase expression in Chinese hamster-human somatic cell hybrids. *Exp. Cell Res.* 106:71–78, 1977.

[44859] 18411. Daiger, S. P.; Hoffman, N. S.; Wildin, R. S.; Su, T.-S.: Multiple, independent restriction site polymorphisms in human DNA detected with a cDNA probe to argininosuccinate synthetase (AS). *Am. J. Hum. Genet.* 36:736–749, 1984.

[44860] 18412. Daiger, S. P.; Wildin, R. S.; Su, T.-S.: Polymorphic variants of restriction fragments of human DNA detected with a probe to argininosuccinate synthetase. (Abstract) *Am. J. Hum. Genet.* 33: 136A only, 1981.

[44861] 18413. Elshourbagy, N. A.; Walker, D. W.; Boguski, M. S.; Gordon, J. I.; Taylor, J. M.: The nucleotide and derived amino acid sequence of human apolipoprotein A-IV mRNA and the close linkage of its gene to the genes of

apolipoproteins A-I and C-III. J. Biol. Chem.
261:1998-2002, 1986.

[44862] 18414. Elshourbagy, N. A.; Walker, D. W.; Paik, Y.-K.; Boguski, M. S.; Freeman, M.; Gordon, J. I.; Taylor, J. M.: Structure and expression of the human apolipoprotein A-IV gene. J. Biol. Chem. 262: 7973-7981, 1987.

- [44863] 18415.Green, P. H. R.; Glickman, R. M.; Riley, J. W.; Quinet, E.: Humanapolipoprotein A-IV: intestinal origin and distribution in plasma. J.Clin. Invest. 65: 911-919, 1980.
- [44864] 18416.Kamboh, M. I.; Ferrell, R. E.: Genetic studies of human apolipoproteins.I. Polymorphism of apolipoprotein A-IV. Am. J. Hum. Genet. 41: 119-127,1987.
- [44865] 18417.Kamboh, M. I.; Williams, E. R.; Law, J. C.; Aston, C. E.; Bunker,C. H.; Ferrell. R. E.; Pollitzer, W. S.: Molecular basis of a uniqueAfrican variant (A-IV 5) of human apolipoprotein A-IV and its significancein lipid metabolism. Genet. Epidemiol. 9: 379-388, 1992.
- [44866] 18418.Karathanasis, S. K.: Apolipoprotein multigene family: tandem organizationof human apolipoprotein AI, CIII, and AIV genes. Proc. Nat. Acad.Sci. 82: 6374-6378, 1985.
- [44867] 18419.Lohse, P.; Kindt, M. R.; Rader, D. J.; Brewer, H. B., Jr.: Geneticpolymorphism of human plasma apolipoprotein A-IV is due to nucleotidesubstitutions in the apolipoprotein A-IV gene. J. Biol. Chem. 265:10061-10064, 1990.
- [44868] 18420.Lohse, P.; Kindt, M. R.; Rader, D. J.; Brewer, H. B., Jr.: Humanplasma apolipoproteins A-IV-0 and A-IV-3: molecular basis for tworare variants of apolipoprotein A-IV-1. J. Biol. Chem. 265: 12734-12739,1990.

- [44869] 18421.Menzel, H.-J.; Boerwinkle, E.; Schrangl-Will, S.; Utermann, G.: Human apolipoprotein A-IV polymorphism: frequency and effect on lipid and lipoprotein levels. Hum. Genet. 79: 368-372, 1988.
- [44870] 18422.Menzel, H.-J.; Kovary, P. M.; Assmann, G.: Apolipoprotein A-IV polymorphism in man. Hum. Genet. 62: 349-352, 1982.
- [44871] 18423.Menzel, H.-J.; Sigurdsson, G.; Boerwinkle, E.; Schrangl-Will, S.; Dieplinger, H.; Utermann, G.: Frequency and effect of human apolipoprotein A-IV polymorphism on lipid and lipoprotein levels in an Icelandic population. Hum. Genet. 84: 344-346, 1990.
- [44872] 18424.Rogne, S.; Myklebost, O.; Olaisen, B.; Gedde-Dahl, T., Jr.; Prydz, H.: Confirmation of the close linkage between the loci for human apolipoproteins AI and AIV by the use of a cloned cDNA probe and two restriction site polymorphisms. Hum. Genet. 72: 68-71, 1986.
- [44873] 18425.Schamaun, O.; Olaisen, B.; Mevag, B.; Gedde-Dahl, T., Jr.; Ehnholm, C.; Teisberg, P.: The two apolipoprotein loci apoA-I and apoA-IV are closely linked in man. Hum. Genet. 68: 181-184, 1984.
- [44874] 18426.Tracy, R. P.; Currie, R. M.; Young, D. S.: Two-dimensional gelelectrophoresis of serum specimens from

a normal population. Clin.Chem. 28: 890–899, 1982.

- [44875] 18427.von Eckardstein, A.; Funke, H.; Schulte, M.; Erren, M.; Schulte,H.; Assmann, G.: Nonsynonymous polymorphic sites in the apolipoprotein(apo) A–IV gene are associated with changes in the concentration of apo B– and apo A–I–containing lipoproteins in a normal population. Am.J. Hum. Genet. 50: 1115–1128, 1992.
- [44876] 18428.Gautier, T.; Masson, D.; Jong, M. C.; Duverneuil, L.; Le Guern,N.; Deckert, V.; Pais de Barros, J.–P.; Dumont, L.; Bataille, A.;Zak, Z.; Jiang, X.–C.; Tall, A. R.; Havekes, L. M.; Lagrost, L.:Apolipoprotein CI deficiency markedly augments plasma lipoprotein changes mediated by human cholesteryl ester transfer protein (CETP) in CETP transgenic/ApoCI–knocked out mice. J. Biol. Chem. 277: 31354–31363,2002.
- [44877] 18429.Brown, S.; Wiebel, F. J.; Gelboin, H. V.; Minna, J. D.: Assignment of a locus required for flavoprotein–linked monooxygenase expression to human chromosome 2. Proc. Nat. Acad. Sci. 73: 4628–4632, 1976.
- [44878] 18430.Chen, Y. T.; Tukey, R. H.; Swan, D. C.; Negishi, N.; Nebert, D.W.: Characterization of the human P1–450 genomic gene. (Abstract) Clin.Res. 31: 456A, 1983.
- [44879] 18431.Corchero, J.; Pimprale, S.; Kimura, S.; Gonzalez, F.

J.: Organization of the CYP1A cluster on human chromosome 15: implications for gene regulation. *Pharmacogenetics* 11: 1–6, 2001.

[44880] 18432. Gorman, C.; Padmanabhan, R.; Howard, B. H.: High efficiency DNA-mediated transformation of primate cells. *Science* 221: 551–553, 1983.

[44881] 18433. Gorman, C. M.: CAT: an easy assay for gene expression (citation classic). *Current Contents (Life Sciences)* 36(22): 8, 1993.

[44882] 18434. Hildebrand, C. E.; Gonzalez, F. J.; Kozak, C. A.; Nebert, D. W.: Regional linkage analysis of the dioxin-inducible P-450 gene family on mouse chromosome 9. *Biochem. Biophys. Res. Commun.* 130: 396–406, 1985.

[44883] 18435. Lee, S.; Wu, X.; Reid, M. E.; Zelinski, T.; Redman, C. M.: Molecular basis of the Kell (K1) phenotype. *Blood* 85: 912–916, 1995.

[44884] 18436. Lee, S.; Zambas, E. D.; Marsh, W. L.; Redman, C. M.: The human Kell blood group gene maps to chromosome 7q33 and its expression is restricted to erythroid cells. *Blood* 81: 2804–2809, 1993.

[44885] 18437. Lee, S.; Zambas, E. D.; Marsh, W. L.; Redman, C. M.: Molecular cloning and primary structure of Kell blood group protein. *Proc. Nat. Acad. Sci.* 88: 6353–6357, 1991.

- [44886] 18438.Marsh, W. L.: Molecular biology of blood groups: cloning of theKell gene. Transfusion 32: 98–101, 1992.
- [44887] 18439.Morton, N. E.; Krieger, H.; Steinberg, A. G.; Rosenfield, R. E.: Genetic evidence confirming the localization of Sutter in the Kellblood–group system. Vox Sang. 10: 608–613, 1965.
- [44888] 18440.Murphy, M. T.; Morrison, N.; Miles, J. S.; Fraser, R. H.; Spurr,N. K.; Boyd, E.: Regional chromosomal assignment of the Kell bloodgroup locus (KEL) to chromosome 7q33–q35 by fluorescence in situ hybridization:evidence for the polypeptide nature of antigenic variation. Hum.Genet. 91: 585–588, 1993.
- [44889] 18441.Parke, J. T.; Riccardi, V. M.; Lewis, R. A.; Ferrell, R. E.:A syndrome of microcephaly and retinal pigmentary abnormalities withoutmental retardation in a family with coincidental autosomal dominanthyperreflexia. Am. J. Med. Genet. 17: 585–594, 1984.
- [44890] 18442.Purohit, K. R.; Weber, J. L.; Ward, L. J.; Keats, B. J. B.: TheKell blood group locus is close to the cystic fibrosis locus on chromosome7. Hum. Genet. 89: 457–458, 1992.
- [44891] 18443.Russo, D.; Wu, X.; Redman, C. M.; Lee, S.: Expression of Kellblood group protein in nonerythroid tissues Blood 96: 340–346, 2000.

- [44892] 18444.Stroup, M.; MacIlroy, M.; Walker, R.; Aydelotte, J. V.: Evidencethat Sutter belongs to the Kell blood group system. Transfusion 5:309–314, 1965.
- [44893] 18445.Wagner, T.; Berer, A.; Lanzer, G.; Geissler, K.: Kell is notrestricted to the erythropoietic lineage but is also expressed onmyeloid progenitor cells. Brit. J. Haemat. 110: 409–411, 2000.
- [44894] 18446.Weitkamp, L. R.; Townes, P. L.; Johnston, E.: Linkage data onurinary pepsinogen and the Kell blood group. Birth Defects Orig.Art. Ser. 11(3): 281–282, 1975. Note: Alternate: Cytogenet. CellGenet. 14: 451–452, 1975.
- [44895] 18447.Wimer, B. M.; Marsh, W. L.; Taswell, H. F.: Clinical characteristicsof the McLeod blood group phenotype. (Abstract) 19th Annual Meetingof the Am. Soc. Hemat., Boston , 1976.
- [44896] 18448.Yu, L.–C.; Twu, Y.–C.; Chang, C.–Y.; Lin, M.: Molecular basisof the Kell–null phenotype: a mutation at the splice site of humanKEL gene abolishes the expression of Kell blood group antigens. J.Biol. Chem. 276: 10247–10252, 2001.
- [44897] 18449.Zelinski, T. A.; Coghlan, G. E.; Myal, Y.; White, L. J.; Philipps,S. E.: Assignment of the Kell blood group locus to chromosome 7q.(Abstract) Cytogenet. Cell Genet. 58:

1927 only, 1991.

- [44898] 18450.Field, L. L.; Marazita, M. L.; Spence, M. A.; Crandall, B. F.;Sparkes, R. S.: Is JK linked to IGK on chromosome 2? (Abstract) Cytogenet.Cell Genet. 40: 628–629, 1985.
- [44899] 18451.Gedde–Dahl, T.: Personal Communication. Oslo, Norway 9/26/1986.
- [44900] 18452.Geitvik, G. A.; Hoyheim, B.; Gedde–Dahl, T.; Grzeschik, K. H.;Lothe, R.; Tomter, H.; Olaisen, B.: The Kidd (JK) blood group locusassigned to chromosome 18 by close linkage to a DNA–RFLP. Hum. Genet. 77:205–209, 1987.
- [44901] 18453.Hulten, M.; Lindsten, J.; Pen–Ming, L. M.; Fraccaro, M.; Mannini,A.; Trepolo, L.; Robson, E. B.; Heiken, A.; Tellingén, K. G.: Possiblelocalization of the genes for the Kidd blood group on an autosomeinvolved in a reciprocal translocation. Nature 211: 1067–1068, 1968.
- [44902] 18454.Leppert, M.; Ferrell, R.; Kamboh, M. I.; Beasley, J.; O'Connell,P.; Lathrop, M.; Lalouel, J.–M.; White, R.: Linkage of the polymorphicprotein markers F13B, C1S, C1R, and blood group antigen Kidd in CEPHreference families. (Abstract) Cytogenet. Cell Genet. 46: 647, 1987.
- [44903] 18455.Lucien, N.; Chiaroni, J.; Cartron, J.–P.; Bailly, P.: Partialdeletion in the JK locus causing a Jk(null) phenotype.

Blood 99:1079–1081, 2002.

- [44904] 18456. Lucien, N.; Sidoux–Walter, F.; Olives, B.; Moulds, J.; Le Pennec, P.–Y.; Cartron, J.–P.; Bailly, P.: Characterization of the gene encoding the human Kidd blood group/urea transporter protein: evidence for splice site mutations in Jk–null individuals. *J. Biol. Chem.* 273:12973–12980, 1998.
- [44905] 18457. Olives, B.; Mattei, M.–G.; Huet, M.; Neau, P.; Martial, S.; Cartron, J.–P.; Bailly, P.: Kidd blood group and urea transport function of human erythrocytes are carried by the same protein. *J. Biol. Chem.* 270:15607–15610, 1995.
- [44906] 18458. Womack, J. E.: Personal Communication. College Station, Texas 2/26/1990.
- [44907] 18459. Hiraoka, L. R.; Hsu, L.; Hsieh, C.–L.: Assignment of ALDH3 to human chromosome 17p11.2 and ALDH5 to human chromosome 9p13. *Genomics* 25:323–325, 1995.
- [44908] 18460. Hsu, L. C.; Chang, W.–C.; Shibuya, A.; Yoshida, A.: Human stomach aldehyde dehydrogenase cDNA and genomic cloning, primary structure, and expression in *Escherichia coli*. *J. Biol. Chem.* 267: 3030–3037, 1992.
- [44909] 18461. Kays, W. T.; Piatigorsky, J.: Aldehyde dehydrogenase class 3 expression: identification of a cornea–preferred gene promoter in transgenic mice. *Proc. Nat. Acad.*

Sci. 94: 13594–13599, 1997.

- [44910] 18462. Rogers, G. R.; Markova, N. G.; De Laurenzi, V.; Rizzo, W. B.; Compton, J. G.: Genomic organization and expression of the human fatty aldehyde dehydrogenase gene (FALDH). *Genomics* 39: 127–135, 1997.
- [44911] 18463. Santisteban, I.; Povey, S.; West, L. F.; Parrington, J. M.; Hopkinson, D. A.: Chromosome assignment, biochemical and immunological studies on a human aldehyde dehydrogenase, ALDH3. *Ann. Hum. Genet.* 49: 87–100, 1985.
- [44912] 18464. Teng, Y.-S.: Stomach aldehyde dehydrogenase: report of a new locus. *Hum. Hered.* 31: 74–77, 1981.
- [44913] 18465. Vasiliou, V.; Bairoch, A.; Tipton, K. F.; Nebert, D. W.: Eukaryotic aldehyde dehydrogenase (ALDH) genes: human polymorphisms, and recommended nomenclature based on divergent evolution and chromosomal mapping. *Pharmacogenetics* 9: 421–434, 1999.
- [44914] 18466. Wunderle, V. M.; Critcher, R.; Hastie, N.; Goodfellow, P. N.; Schedl, A.: Deletion of long-range regulatory elements upstream of SOX9 causes campomelic dysplasia. *Proc. Nat. Acad. Sci.* 95: 10649–10654, 1998.
- [44915] 18467. Young, I. D.; Zuccollo, J. M.; Maltby, E. L.; Broderick, N. J.: Campomelic dysplasia associated with a de novo 2q;17q reciprocal translocation. *J. Med. Genet.* 29:

251–252, 1992.

- [44916] 18468.von Lindern, M.; Poustka, A.; Lehrach, H.; Grosveld, G.: The (6;9)chromosome translocation, associated with a specific subtype of acutenonlymphocytic leukemia, leads to aberrant transcription of a targetgene on 9q34. *Molec. Cell. Biol.* 10: 4016–4026, 1990.
- [44917] 18469.Lynch, H. T.; Schuelke, G. S.; Kimberling, W. J.; Albano, W. A.; Lynch, J. F.; Biscione, K. A.; Lipkin, M. L.; Deschner, E. E.; Mikol, Y. B.; Sandberg, A. A.; Elston, R. C.; Bailey–Wilson, J. E.; Danes, B. S.: Hereditary nonpolyposis colorectal cancer (Lynch syndromes I and II). II. Biomarker studies. *Cancer* 56: 939–951, 1985.
- [44918] 18470.Hanauer, A.; Heilig, R.; Levin, M.; Moisan, J. P.; Grzeschik, K.H.; Mandel, J. L.: The actin gene family in man: assignment of the gene for skeletal muscle alpha-actin to chromosome 1, and presence of actin sequences on autosomes 2 and 3, and on the X and Y chromosomes. (Abstract) *Cytogenet. Cell Genet.* 37: 487–488, 1984.
- [44919] 18471.Ilkovski, B.; Cooper, S. T.; Nowak, K.; Ryan, M. M.; Yang, N.; Schnell, C.; Durling, H. J.; Roddick, L. G.; Wilkinson, I.; Kornberg, A. J.; Collins, K. J.; Wallace, G.; Gunning, P.; Hardeman, E. C.; Laing, N. G.; North, K. N.: Nemaline

myopathy caused by mutations in the muscle alpha-skeletal-actin gene. *Am. J. Hum. Genet.* 68:1333–1343, 2001.

[44920] 18472. Nowak, K. J.; Wattanasirichaigoon, D.; Goebel, H. H.; Wilce, M.; Pelin, K.; Donner, K.; Jacob, R. L.; Hubner, C.; Oexle, K.; Anderson, J. R.; Verity, C. M.; North, K. N.; and 13 others: Mutations in the skeletal muscle alpha-actin gene in patients with actin myopathy and nemaline myopathy. *Nature Genet.* 23: 208–212, 1999.

[44921] 18473. Kedes, L.; Ng, S.-Y.; Lin, C.-S.; Gunning, P.; Eddy, R.; Shows, T.; Leavitt, J.: The human beta-actin multigene family. *Trans. Assoc. Am. Phys.* 98: 42–46, 1985.

[44922] 18474. Leavitt, J.; Bushar, G.; Kakunaga, T.; Hamada, H.; Hirakawa, T.; Goldman, D.; Merrill, C.: Variations in expression of mutant beta-actin accompanying incremental increases in human fibroblast tumorigenicity. *Cell* 28:259–268, 1982.

[44923] 18475. Nakajima-Iijima, S.; Hamada, H.; Reddy, P.; Kakunaga, T.: Molecular structure of the human cytoplasmic beta-actin gene; interspecies homology of sequences in the introns. *Proc. Nat. Acad. Sci.* 82: 6133–6137, 1985.

[44924] 18476. Ng, S.-Y.; Gunning, P.; Eddy, R.; Ponte, P.; Leavitt, J.; Kedes, L.; Shows, T.: Chromosome 7 assignment of the

human beta-actin functional gene (ACTB) and the chromosomal dispersion of pseudogenes. (Abstract) Cytogenet. Cell Genet. 40: 712 only, 1985.

- [44925] 18477. Ng, S.-Y.; Gunning, P.; Eddy, R.; Ponte, P.; Leavitt, J.; Shows, T.; Kedes, L.: Evolution of the functional human beta-actin gene and its multi-pseudogene family: conservation of the noncoding regions and chromosomal dispersion of pseudogenes. Molec. Cell. Biol. 5:2720-2732, 1985.
- [44926] 18478. Toyama, S.; Toyama, S.: A variant form of beta-actin in a mutant of KB cells resistant to cytochalasin B. Cell 37: 609-614, 1984.
- [44927] 18479. Vandekerckhove, J.; Weber, K.: Mammalian cytoplasmic actins are the products of at least two genes and differ in primary structure in at least 25 identified positions from skeletal muscle actins. Proc. Nat. Acad. Sci. 75: 1106-1110, 1978.
- [44928] 18480. Allderdice, P. W.; Kaita, H.; Lewis, M.; McAlpine, P. J.; Wong, P.; Anderson, J.; Giblett, E. R.: Segregation of marker loci in families with an inherited paracentric insertion of chromosome 9. Am. J. Hum. Genet. 39: 612-617, 1986.
- [44929] 18481. Dayhoff, M. O.: Atlas of Protein Sequence and

Structure. Dehydrogenases. Washington: National Biomedical Research Foundation (pub.) 5: 1972.Pp. D141–D144.

- [44930] 18482.Osier, M. V.; Pakstis, A. J.; Soodyall, H.; Comas, D.; Goldman,D.; Odunsi, A.; Okonofua, F.; Parnas, J.; Schulz, L. O.; Bertranpetit,J.; Bonne–Tamir, B.; Lu, R.–B.; Kidd, J. R.; Kidd, K. K.: A globalperspective on genetic variation at the ADH genes reveals unusualpatterns of linkage dise–quilibrium and diversity. *Am. J. Hum. Genet.* 71:84–99, 2002.
- [44931] 18483.Smith, M.; Hopkinson, D. A.; Harris, H.: Studies on the subunitstructure and molecular size of the human de–hydrogenase isozymes determinedby the different loci, ADH(1), ADH(2), and ADH(3). *Ann. Hum. Genet.* 36:401–414, 1973.
- [44932] 18484.Comings, D. E.; Gade–Andavolu, R.; Gonzalez, N.; Blake, H.; Wu,S.; MacMurray, J. P.: Additive effect of three noradrenergic genes(ADRA2A, ADRA2C, DBH) on atten–tion–deficit hyperactivity disorderand learning disabilities in Tourette syndrome subjects. *Clin. Genet.* 55:160–172, 1999.
- [44933] 18485.Halperin, J. M.; Newcorn, J. H.; Koda, V. H.; Pick, L.; McKay,K. E.; Knott, P.: Noradrenergic mechanisms in

ADHD children with and without reading disabilities: a replication and extension. *J. Am. Acad. Child Adolesc. Psychiat.* 36: 1688–1697, 1997.

[44934] 18486. Hein, L.; Altman, J. D.; Kobilka, B. K.: Two functionally distinct α -2-adrenergic receptors regulate sympathetic neurotransmission. *Nature* 402:181–184, 1999.

[44935] 18487. Hoehe, M.; Berrettini, W.; Leppert, M.; Lalouel, J.-M.; Byerley, W.; Gershon, E.; White, R.: Genetic mapping of adrenergic receptor genes. (Abstract) *Am. J. Hum. Genet.* 45 (suppl.): A143 only, 1989.

[44936] 18488. Hoehe, M. R.; Berrettini, W. H.; Lentes, K.-U.: DNA identifies a two allele DNA polymorphism in the human α -2-adrenergic receptor gene (ADRAR), using a 5.5 kb probe (p ADRAR). *Nucleic Acids Res.* 16:9070 only, 1988.

[44937] 18489. Kobilka, B. K.; Matsui, H.; Kobilka, T. S.; Yang-Feng, T. L.; Francke, U.; Caron, M. G.; Lefkowitz, R. J.; Regan, J. W.: Cloning, sequencing, and expression of the gene coding for the human platelet α -2-adrenergic receptor. *Science* 238: 650–656, 1987.

[44938] 18490. Oakey, R. J.; Caron, M. G.; Lefkowitz, R. J.; Seldin, M. F.: Genomic organization of adrenergic and serotonin receptors in the mouse: linkage mapping of sequence-related genes provides a method for examining mammalian

chromosome evolution. *Genomics* 10: 338–344, 1991.

[44939] 18491. Philipp, M.; Brede, M. E.; Hadamek, K.; Gessler, M.; Lohse, M.J.; Hein, L.: Placental alpha-2-adrenoceptors control vascular development at the interface between mother and embryo. *Nature Genet.* 31: 311–315, 2002.

[44940] 18492. Surprenant, A.; Horstman, D. A.; Akbarali, H.; Limbird, L. E.: A point mutation of the alpha-2-adrenoceptor that blocks coupling to potassium but not calcium currents. *Science* 257: 977–980, 1992.

[44941] 18493. Yang-Feng, T. L.; Kobilka, B. K.; Caron, M. G.; Lefkowitz, R.J.; Francke, U.: Chromosomal assignment of genes for an alpha-adrenergic receptor (ADRAR) and for another member of this receptor family coupled to guanine nucleotide regulatory proteins (RG21). (Abstract) *Cytogenet. Cell Genet.* 46: 722–723, 1987.

[44942] 18494. Bruno, J. F.; Whittaker, J.; Song, J.; Berelowitz, M.: Molecular cloning and sequencing of a cDNA encoding a human alpha-1A adrenergic receptor. *Biochem. Biophys. Res. Commun.* 179: 1485–1490, 1991.

[44943] 18495. Esbenshade, T. A.; Hirasawa, A.; Tsujimoto, G.; Tanaka, T.; Yano, J.; Minneman, K. P.; Murphy, T. J.: Cloning of the human alpha-1D-adrenergic receptor and inducible expression of three subtypes in SK-N-MC cells.

Molec.Pharm. 47: 977–985, 1995.

- [44944] 18496. Forray, C.; Bard, J. A.; Wetzel, J. M.; Chiu, G.; Shapiro, E.; Tang, R.; Lepor, H.; Hartig, P. R.; Weinshank, R. L.; Branchek, T.A.; Gluchowski, C.: The alpha-1-adrenergic receptor that mediates smooth muscle contraction in human prostate has the pharmacological properties of the cloned human alpha-1C subtype. Molec. Pharm. 45:703–708, 1994.
- [44945] 18497. Loftus, S. K.; Shiang, R.; Warrington, J. A.; Bengtsson, U.; McPherson, J. D.; Wasmuth, J. J.: Genes encoding adrenergic receptors are not clustered on the long arm of human chromosome 5. Cytogenet. Cell Genet. 67: 69–74, 1994.
- [44946] 18498. Lomasney, J. W.; Cotecchia, S.; Lorenz, W.; Leung, W.-Y.; Schwinn, D. A.; Yang-Feng, T. L.; Brownstein, M.; Lefkowitz, R. J.; Caron, M. G.: Molecular cloning and expression of the cDNA for the alpha-1A-adrenergic receptor: the gene for which is located on human chromosome 5. J. Biol. Chem. 266: 6365–6369, 1991.
- [44947] 18499. Schwinn, D. A.; Johnston, G. I.; Page, S. O.; Mosley, M. J.; Wilson, K. H.; Worman, N. P.; Campbell, S.; Fidock, M. D.; Furness, L. M.; Parry-Smith, D. J.; Peter, B.; Bailey, D. S.:

Cloning and pharmacological characterization of human alpha-1 adrenergic receptors: sequence corrections and direct comparison with other species homologues. *J. Pharm. Exp. Ther.* 272: 134–142, 1995.

[44948] 18500. Schwinn, D. A.; Lomasney, J. W.: Pharmacologic characterization of cloned alpha-1-adrenoceptor subtypes: selective antagonists suggest the existence of a fourth subtype. *Europ. J. Pharm.* 227: 433–436, 1992.

[44949] 18501. Weinberg, D. H.; Trivedi, P.; Tan, C. P.; Mitra, S.; Perkins-Barrow, A.; Borkowski, D.; Strader, C. D.; Bayne, M.: Cloning, expression and characterization of human alpha adrenergic receptors alpha-1A, alpha-1B, and alpha-1C. *Biochem. Biophys. Res. Commun.* 201: 1296–1304, 1994.

[44950] 18502. Yang-Feng, T. L.; Han, H.; Lomasney, J. W.; Caron, M. G.: Localization of the cDNA for an alpha-1-adrenergic receptor subtype (ADRA1D) to chromosome band 20p13. *Cytogenet. Cell Genet.* 66: 170–171, 1994.

[44951] 18503. Allen, L. F.; Lefkowitz, R. J.; Caron, M. G.; Cotecchia, S.: G-protein-coupled receptor genes as protooncogenes: constitutively activating mutation of the alpha-1B-adrenergic receptor enhances mitogenesis and tumorigenicity. *Proc. Nat. Acad. Sci.* 88: 11354–11358,

1991.

- [44952] 18504. Markert, M. L.; Hershfield, M. S.; Wiginton, D. A.; States, J. C.; Ward, F. E.; Bigner, S. H.; Buckley, R. H.; Kaufman, R. E.; Hutton, J. J.: Identification of a deletion in the adenosine deaminase gene in a child with severe combined immunodeficiency. *J. Immun.* 138:3203–3206, 1987.
- [44953] 18505. Markert, M. L.; Hutton, J. J.; Wiginton, D. A.; States, J. C.; Kaufman, R. E.: Adenosine deaminase (ADA) deficiency due to deletion of the ADA gene promoter and first exon by homologous recombination between two Alu elements. *J. Clin. Invest.* 81: 1323–1327, 1988.
- [44954] 18506. Markert, M. L.; Norby-Slycord, C.; Ward, F. E.: A high proportion of ADA point mutations associated with a specific alanine-to-valine substitution. *Am. J. Hum. Genet.* 45: 354–361, 1989.
- [44955] 18507. Meuwissen, H. J.; Pollara, B.; Pickering, R. J.: Combined immunodeficiency disease associated with adenosine deaminase deficiency (report on a workshop held in Albany, New York, October 1, 1973). *J. Pediat.* 86:169–181, 1975.
- [44956] 18508. Migchielsen, A. A. J.; Breuer, M. L.; van Roon, M. A.; te Riele, H.; Zurcher, C.; Ossendorp, F.; Toutain, S.; Hershfield, M. S.; Berns, A.; Valerio, D.: Adenosine-deami-

nase-deficient mice die perinatally and exhibit liver-cell degeneration, atelectasis and small intestinal cell death. *Nature Genet.* 10: 279–287, 1995.

[44957] 18509. Mitchell, B. S.; Mejias, E.; Daddona, P. E.; Kelley, W. N.: Purinogenic immunodeficiency disease: selective toxicity of deoxyribonucleosides for T-cells. *Proc. Nat. Acad. Sci.* 75: 5011–5014, 1978.

[44958] 18510. Moen, R. C.; Horowitz, S. D.; Sondel, P. M.; Borchering, W. R.; Trigg, M. E.; Billing, R.; Hong, R.: Immunologic reconstitution after haploidentical bone marrow transplantation for immune deficiency disorders: treatment of bone marrow cells with monoclonal antibody CT-2 and complement. *Blood* 70:664–669, 1987.

[44959] 18511. Mohandas, T.; Sparkes, R. S.; Suh, E. J.; Hershfield, M. S.: Regional localization of the human genes for S-adenosylhomocysteine hydrolase (cen-q131) and adenosine deaminase (q131-qter) on chromosome 20. *Hum. Genet.* 66: 292–295, 1984.

[44960] 18512. Nielsen, K. B.; Tommerup, N.; Jespersen, B.; Nygaard, P.; Kleif, L.: Segregation of a t(3;20) translocation through three generations resulting in unbalanced karyotypes in six persons. *J. Med. Genet.* 23:446–451, 1986.

[44961] 18513. Orkin, S. H.; Daddona, P. E.; Shewach, D. S.;

Markham, A. F.; Bruns, G. A.; Goff, S. C.; Kelley, W. N.: Molecular cloning of human adenosine deaminase gene sequences. *J. Biol. Chem.* 258: 12753–12756, 1983.

[44962] 18514. Ozsahin, H.; Arredondo-Vega, F. X.; Santisteban, I.; Fuhrer, H.; Tuchscheid, P.; Jochum, W.; Aguzzi, A.; Lederman, H. M.; Fleischman, A.; Winkelstein, J. A.; Seger, R. A.; Herschfeld, M. S.: Adenosine deaminase deficiency in adults. *Blood* 89: 2849–2855, 1997.

[44963] 18515. Onodera, M.; Ariga, T.; Kawamura, N.; Kobayashi, I.; Ohtsu, M.; Yamada, M.; Tame, A.; Furuta, H.; Okano, M.; Matsumoto, S.; Kotani, H.; McGarrity, G. J.; Blaese, R. M.; Sakiyama, Y.: Successful peripheral T-lymphocyte-directed gene transfer for a patient with severe combined immune deficiency caused by adenosine deaminase deficiency. *Blood* 91:30–36, 1998.

[44964] 18516. Palmer, T. D.; Hock, R. A.; Osborne, W. R. A.; Miller, A. D.: Efficient retrovirus-mediated transfer and expression of a human adenosine deaminase gene in diploid skin fibroblasts from an adenosine deaminase-deficient human. *Proc. Nat. Acad. Sci.* 84: 1055–1059, 1987.

[44965] 18517. Parkman, R.; Gelfand, E. W.; Rosen, F. S.; Sanderson, A.; Hirschhorn, R.: Severe combined immunodeficiency and adenosine deaminase deficiency. *New Eng. J.*

Med. 292: 714–719, 1975.

- [44966] 18518. Petersen, M. B.; Tranebjaerg, L.; Tommerup, N.; Nygaard, P.; Edwards, H.: New assignment of the adenosine deaminase gene locus to chromosome 20q13.11 by study of a patient with interstitial deletion 20q. J. Med. Genet. 24: 93–96, 1987.
- [44967] 18519. Polmar, S. H.; Stern, R. C.; Schwartz, A. L.; Wetzler, E. M.; Chase, P. A.; Hirschhorn, R.: Enzyme replacement therapy for adenosine deaminase deficiency and severe combined immunodeficiency. New Eng. J. Med. 295: 1337–1343, 1976.
- [44968] 18520. Ratech, H.; Greco, M. A.; Gallo, G.; Rimoin, D. L.; Kamino, H.; Hirschhorn, R.: Pathologic findings in adenosine–deaminase–deficient severe combined immunodeficiency. I. Kidney, adrenal, and chondro–osseous tissue alterations. Am. J. Path. 120: 157–169, 1985.
- [44969] 18521. Ritter, H.; Wendt, G. G.; Tariverdian, G.; Zelch, J.; Rube, M.; Kirchberg, G.: Genetics and linkage analysis of adenosine deaminase. Humangenetik 14: 69–71, 1971.
- [44970] 18522. Rogers, M. H.; Lwin, R.; Fairbanks, L.; Gerritsen, B.; Gaspar, H. B.: Cognitive and behavioral abnormalities in adenosine deaminase deficient severe combined immunodeficiency. J. Pediat. 139: 44–50, 2001.

- [44971] 18523.Rothschild, C. B.; Akots, G.; Hayworth, R.; Pettenati, M. J.; Rao, P. N.; Wood, P.; Stolz, F.-M.; Hansmann, I.; Serino, K.; Keith, T. P.; Fajans, S. S.; Bowden, D. W.: A genetic map of chromosome 20q12-q13.1: multiple highly polymorphic microsatellite and RFLP markers linked to the maturity-onset diabetes of the young (MODY) locus. *Am. J. Hum. Genet.* 52: 110-123, 1993.
- [44972] 18524.Rubinstein, A.; Hirschhorn, R.; Sicklick, M.; Murphy, R. A.: In vivo and in vitro effects of thymosin and adenosine deaminase on adenosine-deaminase-deficient lymphocytes. *New Eng. J. Med.* 300:387-392, 1979.
- [44973] 18525.Rudd, N. L.; Bain, H. W.; Giblett, E.; Chen, S.-H.; Worton, R.G.: Partial trisomy 20 confirmed by gene dosage studies. *Am. J. Med. Genet.* 4: 357-364, 1979.
- [44974] 18526.Koch, G.; Shows, T. B.: Somatic cell genetics of adenosine deaminase expression and severe combined immune deficiency disease in man. *Proc. Nat. Acad. Sci.* 77: 4211-4215, 1980.
- [44975] 18527.Gautam, M.; Noakes, P. G.; Moscoso, L.; Rupp, F.; Scheller, R.H.; Merlie, J. P.; Sanes, J. R.: Defective neuromuscular synaptogenesis in agrin-deficient mutant mice. *Cell* 85: 525-535, 1996.
- [44976] 18528.Glass, D. J.; Bowen, D. C.; Stitt, T. N.; Radziejewski,

C.; Bruno, J.; Ryan, T. E.; Gies, D. R.; Shah, S.; Mattsson, K.; Burden, S. J.; DiStefano, P. S.; Valenzuela, D. M.; DeChiara, T. M.; Yancopoulos, G. D.: Agrin acts via a MuSK receptor complex. *Cell* 85: 513–523, 1996.

[44977] 18529. Khan, A. A.; Bose, C.; Yam, L. S.; Soloski, M. J.; Rupp, F.: Physiological regulation of the immunological synapse by agrin. *Science* 292: 1681–1686, 2001.

[44978] 18530. Lin, W.; Burgess, R. W.; Dominguez, B.; Pfaff, S. L.; Sanes, J. R.; Lee, K.-F.: Distinct roles of nerve and muscle in postsynaptic differentiation of the neuromuscular synapse. *Nature* 410: 1057–1064, 2001.

[44979] 18531. McMahan, U. J.: The agrin hypothesis *Cold Spring Harb. Symp. Quant. Biol.* 50: 407–418, 1990.

[44980] 18532. Rupp, F.; Ozcelik, T.; Linial, M.; Peterson, K.; Francke, U.; Scheller, R.: Structure and chromosomal localization of the mammalian agrin gene. *J. Neurosci.* 12: 3535–3544, 1992.

[44981] 18533. Rupp, F.; Payan, D. G.; Magill-Solc, C.; Cowan, D. M.; Scheller, R. H.: Structure and expression of a rat agrin. *Neuron* 6: 811–823, 1991.

[44982] 18534. Umans, L.; Serneels, L.; Overbergh, L.; Lorent, K.; Van Leuven, F.; Van den Berghe, H.: Targeted inactivation of the mouse alpha-2-macroglobulin gene. *J. Biol. Chem.*

270: 19778–19785, 1995.

- [44983] 18535.D'Alfonso, S.; Richiardi, P. M.: A polymorphic variation in a putative regulation box of the TNFA promoter region. *Immunogenetics* 39:150–154, 1994.
- [44984] 18536.Mietus–Snyder, M.; Charmley, P.; Korf, B.; Ladas, J. A. A.; Gatti, R. A. and Karathanasis, S. K.: Genetic linkage of the human apolipoprotein A1–CIII–AIV gene cluster and the neural cell adhesion molecule (NCAM) gene. *Genomics* 7: 633–637, 1990.
- [44985] 18537.Mietus–Snyder, M.; Korf, B.; Ladas, J. A.; Karathanasis, S. K.: Linkage of the human apolipoproteins A1, C3, A4 and the neural cell adhesion molecule (NCAM) genes. (Abstract) *Cytogenet. Cell Genet.* 51:1044 only, 1989.
- [44986] 18538.Nguyen, C.; Mattei, M. G.; Goridis, C.; Mattei, J. F.; Jordan, B. R.: Localization of the human NCAM gene to chromosome 11 by insitu hybridization with a murine NCAM cDNA probe. (Abstract) *Cytogenet. Cell Genet.* 40: 713 only, 1985.
- [44987] 18539.Nguyen, C.; Mattei, M. G.; Mattei, J.–F.; Santoni, M.–J.; Goridis, C.; Jordan, B. R.: Localization of the human NCAM gene to band q23 of chromosome 11: the third gene coding for a cell interaction molecule mapped to the

distal portion of the long arm of chromosome 11. J.Cell Biol. 102: 711–715, 1986.

- [44988] 18540.Rabinowitz, J. E.; Rutishauser, U.; Magnuson, T.: Targeted mutation of Ncam to produce a secreted molecule results in a dominant embryonic lethality. Proc. Nat. Acad. Sci. 93: 6421–6424, 1996.
- [44989] 18541.Rutishauser, U.; Acheson, A.; Hall, A. K.; Mann, D. M.; Sunshine, J.: The neural cell adhesion molecule (NCAM) as a regulator of cell–cell interactions. Science 240: 53–57, 1988.
- [44990] 18542.Rutishauser, U.; Goridis, C.: NCAM: the molecule and its genetics. Trends Genet. 2: 72–76, 1986.
- [44991] 18543.Telatar, M.; Lange, E.; Uhrhammer, N.; Gatti, R. A.: New localization of NCAM, proximal to DRD2 at chromosome 11q23. Mammalian Genome 6:59–60, 1995.
- [44992] 18544.Agellon, L. B.; Drover, V. A. B.; Cheema, S. K.; Gbaguidi, G. F.; Walsh, A.: Dietary cholesterol fails to stimulate the human cholesterol 7- α -hydroxylase gene (CYP7A1) in transgenic mice. J. Biol. Chem. 277:20131–20134, 2002.
- [44993] 18545.Angelin, B.; Einarsson, K.; Hellstrom, K.; Leijdt, B.: Bile acid kinetics in relation to endogenous triglyceride metabolism in various types of hyperlipoproteinemia. J.

Lipid Res. 19: 1004–1016, 1978.

- [44994] 18546. Angelin, B.; Hershon, K. S.; Brunzell, J. D.: Bile acid metabolism in hereditary forms of hypertriglyceridemia: evidence for an increased synthesis rate in monogenic familial hypertriglyceridemia. *Proc. Nat. Acad. Sci.* 84: 5434–5438, 1987.
- [44995] 18547. Cohen, J. C.; Cali, J. J.; Jelinek, D. F.; Mehrabian, M.; Sparkes, R. S.; Lusis, A. J.; Russell, D. W.; Hobbs, H. H.: Cloning of the human cholesterol 7- α -hydroxylase gene (CYP7) and localization to chromosome 8q11–q12. *Genomics* 14: 153–161, 1992.
- [44996] 18548. Drover, V. A. B.; Wong, N. C. W.; Agellon, L. B.: A distinct thyroid hormone response element mediates repression of the human cholesterol 7- α -hydroxylase (CYP7A1) gene promoter. *Molec. Endocr.* 16: 14–23, 2002.
- [44997] 18549. Goodwin, B.; Jones, S. A.; Price, R. R.; Watson, M. A.; McKee, D. D.; Moore, L. B.; Galardi, C.; Wilson, J. G.; Lewis, M. C.; Roth, M. E.; Maloney, P. R.; Willson, T. M.; Kliewer, S. A.: A regulatory cascade of the nuclear receptors FXR, SHP-1, and LRH-1 represses bile acid biosynthesis. *Molec. Cell* 6: 517–526, 2000.
- [44998] 18550. Molowa, D. T.; Chen, W. S.; Cimis, G. M.; Tan, C. P.: Transcriptional regulation of the human cholesterol 7 α -

pha-hydroxylase gene. *Biochemistry* 31:2539–2544, 1992.

- [44999] 18551.Nitta, M.; Ku, S.; Brown, C.; Okamoto, A. Y.; Shan, B.: CPF: an orphan nuclear receptor that regulates liver-specific expression of the human cholesterol 7- α -hydroxylase gene. *Proc. Nat. Acad.Sci.* 96: 6660–6665, 1999.
- [45000] 18552.Noshiro, M.; Okuda, K.: Molecular cloning and sequence analysis of cDNA encoding human cholesterol 7 α -hydroxylase. *FEBS Lett.* 268:137–140, 1990.
- [45001] 18553.Paumgartner, G.; Sauerbruch, T.: Gallstones: pathogenesis. *Lancet* 338:1117–1121, 1991.
- [45002] 18554.Wang, J.; Freeman, D. J.; Grundy, S. M.; Levine, D. M.; Guerra, R.; Cohen, J. C.: Linkage between cholesterol 7- α -hydroxylase and high plasma low-density lipoprotein cholesterol concentrations. *J.Clin. Invest.* 101: 1283–1291, 1998.
- [45003] 18555.Akita, H.; Chiba, H.; Tsuchihashi, K.; Tsuji, M.; Kumagai, M.; Matsuno, K.; Kobayashi, K.: Cholesteryl ester transfer protein gene: two common mutations and their effect on plasma high-density lipoprotein cholesterol content. *J. Clin. Endocr. Metab.* 79: 1615–1618, 1994.
- [45004] 18556.Altshuler, D.; Kruglyak, L.; Lander, E.: Genetic

polymorphisms and disease. (Letter) *New Eng. J. Med.* 338: 1626 only, 1998.

- [45005] 18557. Brown, M. L.; Inazu, A.; Hesler, C. B.; Agellon, L. B.; Mann, C.; Whitlock, M. E.; Marcel, Y. L.; Milne, R. W.; Koizumi, J.; Mabuchi, H.; Takeda, R.; Tall, A. R.: Molecular basis of lipid transfer protein deficiency in a family with increased high-density lipoproteins. *Nature* 342:448–451, 1989.
- [45006] 18558. Drayna, D.; Jarnagin, A. S.; McLean, J.; Henzel, W.; Kohr, W.; Fielding, C.; Lawn, R.: Cloning and sequencing of human cholesteryl ester transfer protein cDNA. *Nature* 327: 632–634, 1987.
- [45007] 18559. Drayna, D.; Lawn, R. M.: Multiple RFLPs at the human cholesteryl ester transfer protein (CETP) locus. *Nucleic Acids Res.* 15: 4698 only, 1987.
- [45008] 18560. Haefliger, J.-A.; Bruzzone, R.; Jenkins, N. A.; Gilbert, D. J.; Copeland, N. G.; Paul, D. L.: Four novel members of the connexin family of gap junction proteins: molecular cloning, expression, and chromosome mapping. *J. Biol. Chem.* 267: 2057–2064, 1992.
- [45009] 18561. Hsieh, C.-L.; Kumar, N. M.; Gilula, N. B.; Francke, U.: Distribution of genes for gap junction membrane channel proteins on human and mouse chromosomes. *Somat.*

Cell Molec. Genet. 17: 191–200, 1991.

- [45010] 18562.Spielman, R. S.; McGinnis, R. E.; Ewens, W. J.: Transmission test for linkage disequilibrium: the insulin gene region and insulin-dependent diabetes mellitus (IDDM). Am. J. Hum. Genet. 52: 506–516, 1993.
- [45011] 18563.Altshuler, D.; Hirschhorn, J. N.; Klannemark, M.; Lindgren, C.M.; Vohl, M.-C.; Nemesh, J.; Lane, C. R.; Schaffner, S. F.; Bolk, S.; Brewer, C.; Tuomi, T.; Gaudet, D.; Hudson, T. J.; Daly, M.; Groop, L.; Lander, E. S.: The common PPAR- γ pro12ala polymorphism is associated with decreased risk of type 2 diabetes. Nature Genet. 76–80, 2000.
- [45012] 18564.Fakhrai-Rad, H.; Nikoshkov, A.; Kamel, A.; Fernstrom, M.; Zierath, J. R.; Norgren, S.; Luthman, H.; Galli, J.: Insulin-degrading enzyme identified as a candidate diabetes susceptibility gene in GK rats. Hum. Molec. Genet. 9: 2149–2158, 2000.
- [45013] 18565.Gersuk, V. H.; Rose, T. M.; Todaro, G. J.: Molecular cloning and chromosomal localization of a pseudogene related to the human acyl-CoA binding protein/diazepam binding inhibitor. Genomics 25: 469–476, 1995.
- [45014] 18566.DeStefano, A. L.; Baldwin, C. T.; Burzstyn, M.; Gavras, I.; Handy, D. E.; Joost, O.; Martel, T.; Nicolaou, M.;

Schwartz, F.; Streeten, D. H. P.; Farrer, L. A.; Gavras, H.: Autosomal dominant orthostatic hypotensive disorder maps to chromosome 18q. *Am. J. Hum. Genet.* 63:1425–1430, 1998.

[45015] 18567. Barnett, T.; Zimmermann, W.: Workshop report: proposed nomenclature for the carcinoembryonic antigen (CEA) gene family. *Tumor Biol.* 11:59–63, 1990.

[45016] 18568. Boulton, I. C.; Gray-Owen, S. D.: Neisserial binding to CEACAM1 arrests the activation and proliferation of CD4⁺ T lymphocytes. *Nature Immun.* 3: 229–236, 2002.

[45017] 18569. Ergun, S.; Kilic, N.; Ziegeler, G.; Hansen, A.; Nollau, P.; Gotze, J.; Wurmbach, J.-H.; Horst, A.; Weil, J.; Fernando, M.; Wagener, C.: CEA-related cell adhesion molecule 1: a potent angiogenic factor and a major effector of vascular endothelial growth factor. *Molec. Cell* 5: 311–320, 2000.

[45018] 18570. Hinoda, Y.; Neumaier, M.; Hefta, S. A.; Drzeniek, Z.; Wagener, C.; Shively, L.; Hefta, L. J. F.; Shively, J. E.; Paxton, R. J.: Molecular cloning of a cDNA coding biliary glycoprotein I: primary structure of a glycoprotein immunologically crossreactive with carcinoembryonic antigen. *Proc. Nat. Acad. Sci.* 85: 6959–6963, 1988.

[45019] 18571. Neumaier, M.; Paululat, S.; Chan, A.; Matthaes, P.; Wagener, C.: Biliary glycoprotein, a potential human cell

adhesion molecule, is down-regulated in colorectal carcinomas. *Proc. Nat. Acad. Sci.* 90:10744–10748, 1993.

[45020] 18572. Poy, M. N.; Yang, Y.; Rezaei, K.; Fernstrom, M. A.; Lee, A. D.; Kido, Y.; Erickson, S. K.; Najjar, S. M.: CEACAM1 regulates insulin clearance in liver. *Nature Genet.* 30: 270–276, 2002.

[45021] 18573. Robbins, J.; Robbins, P. F.; Kozak, C. A.; Callahan, R.: The mouse biliary glycoprotein gene (Bgp): partial nucleotide sequence, expression, and chromosomal assignment. *Genomics* 10: 583–587, 1991.

[45022] 18574. Thompson, J. A.; Grunert, F.; Zimmermann, W.: Carcinoembryonic antigen gene family: molecular biology and clinical perspectives. *J. Clin. Lab. Anal.* 5: 344–366, 1991.

[45023] 18575. Willard, H. F.; Meakin, S. O.; Tsui, L.-C.; Breitman, M. L.: Assignment of human gamma crystallin multigene family to chromosome 2. *Somat. Cell Molec. Genet.* 11: 511–516, 1985.

[45024] 18576. Wistow, G.; Piatigorsky, J.: Recruitment of enzymes as lens structural proteins. *Science* 236: 1554–1556, 1987.

[45025] 18577. Zneimer, S. M.; Womack, J. E.: Regional localization of the fibronectin and gamma crystallin genes to mouse chromosome 1 by in situ hybridization. *Cytogenet. Cell*

Genet. 48: 238–241, 1988.

- [45026] 18578. Daniels, G.; King, M.-J.; Avent, N. D.; Khalid, G.; Reid, M.; Mallinson, G.; Symthe, J.; Cedergren, B.: A point mutation in the GYPC gene results in the expression of the blood group An(a) antigen on glycophorin D but not on glycophorin C: further evidence that glycophorin D is a product of the GYPC gene. *Blood* 82: 3198–3203, 1993.
- [45027] 18579. El-Maliki, B.; Blanchard, D.; Dahr, W.; Beyreuther, K.; Cartron, J.-P.: Structural homology between glycophorins C and D of human erythrocytes. *Europ. J. Biochem.* 183: 639–643, 1989.
- [45028] 18580. Jorgensen, J.; Drachmann, O.; Gavin, J.: Duch, Dh(a), a low frequency red cell antigen. *Hum. Hered.* 32: 73–75, 1982.
- [45029] 18581. King, M. J.; Avent, N. D.; Mallinson, G.; Reid, M. E.: Point mutation in the glycophorin C gene results in the expression of the blood group antigen Dh(a). *Vox Sang.* 63: 56–58, 1992.
- [45030] 18582. Le Van Kim, C.; Colin, Y.; Blanchard, D.; Dahr, W.; London, J.; Cartron, J.-P.: Gerbich blood group deficiency of the Ge:–1,–2,–3 and Ge:–1,–2,3 types: immunochemical study and genomic analysis with cDNA probes. *Europ. J. Biochem.* 165: 571–579, 1987.

- [45031] 18583.Mattei, M. G.; Colin, Y.; Le Van Kim, C.; Mattei, J. F.; Cartron,J. P.: Localization of the gene for human erythrocyte glycophorinC to chromosome 2, q14-q21. Hum. Genet. 74: 420-422, 1986.
- [45032] 18584.Pasvol, G.; Anstee, D. J.; Tanner, M. J. A.: Glycophorin C andthe invasion of red cells by Plasmodium falciparum. Lancet I: 907-908,1984.
- [45033] 18585.Race, R. R.; Sanger, R.: Blood Groups in Man. Oxford: BlackwellSci. Publ. (pub.) (6th ed.): 1975. Pp. 416-421.
- [45034] 18586.Reid, M. E.: The Gerbich blood group antigens: a review. Med.Lab. Sci. 43: 177-182, 1972.
- [45035] 18587.Reid, M. E.; Sullivan, C.; Taylor, M.; Anstee, D. J.: Inheritanceof human-erythrocyte Gerbich blood group antigens. Am. J. Hum. Genet. 41:1117-1123, 1987.
- [45036] 18588.Simmons, R. T.; Albrey, J. A.: A 'new' blood group antigen Webb(Wb) of low frequency found in two Australian families. Med. J. Aust. I:8-10, 1963.
- [45037] 18589.Sondag, D.; Alloisio, N.; Blanchard, D.; Ducluzeau, M.-T.; Colonna,P.; Bachir, D.; Bloy, C.; Cartron, J.-P.; De-launay, J.: Gerbich reactivityin 4.1(-) hereditary elliptocytosis and protein 4.1 level in bloodgroup Gerbich deficiency. Brit. J. Haemat. 65: 43-50, 1987.

- [45038] 18590.Spring, F. A.: Immunochemical characterisation of the low-incidence antigen, Dh(a). *Vox Sang.* 61: 65–68, 1991.
- [45039] 18591.Telen, M. J.; Le Van Kim, C.; Guizzo, M. L.; Cartron, J.-P.; Colin, Y.: Erythrocyte Webb-type glycophorin C variant lacks N-glycosylation due to an asparagine to serine substitution. *Am. J. Hemat.* 37: 51–52, 1991.
- [45040] 18592.Winardi, R.; Reid, M.; Conboy, J.; Mohandas, N.: Molecular analysis of glycophorin C deficiency in human erythrocytes. *Blood* 81: 2799–2803, 1993.
- [45041] 18593.Bierhuizen, M. F. A.; Mattei, M.-G.; Fukuda, M.: Expression of the developmental I antigen by a cloned human cDNA encoding a member of a beta-1,6-N-acetylglucosaminyltransferase gene family. *Genes–Dev.* 7: 468–478, 1993.
- [45042] 18594.Lin–Chu, M.; Broadberry, R. E.; Okubo, Y.; Tanaka, M.: The i phenotype and congenital cataracts among Chinese in Taiwan (Letter) *Transfusion* 31:676–677, 1991.
- [45043] 18595.Ogata, H.; Okubo, Y.; Akabane, T.: Phenotype i associated with congenital cataract in Japanese. *Transfusion* 19: 166–168, 1979.
- [45044] 18596.Yeh, J.-C.; Ong, E.; Fukuda, M.: Molecular cloning and expression of a novel beta–

1,6-N-acetylglucosaminyltransferase that forms core2, core 4, and I branches. *J. Biol. Chem.* 274: 3215–3221, 1999.

- [45045] 18597. Yu, L.-C.; Twu, Y.-C.; Chang, C.-Y.; Lin, M.: Molecular basis of the adult i phenotype and the gene responsible for the expression of the human blood group I antigen. *Blood* 98: 3840–3845, 2001.
- [45046] 18598. Allen, F. H., Jr.; Krabbe, S. M.; Corcoran, P. A.: A new phenotype (McLeod) in the Kell blood-group system. *Vox Sang.* 6: 555–560, 1961.
- [45047] 18599. Avent, N. D.; Martin, P. G.: Kell typing by allele-specific PCR (ASP). *Brit. J. Haemat.* 93: 728–730, 1996.
- [45048] 18600. Chown, B.; Lewis, M.; Kaita, K.: A 'new' Kell blood-group phenotype. *Nature* 180: 711 only, 1957.
- [45049] 18601. Conneally, P. M.; Nance, W. E.; Huntzinger, R. S.: Linkage analysis of Kell–Sutter and PTC loci. (Abstract) *Am. J. Hum. Genet.* 26: 22A only, 1974.
- [45050] 18602. Abdelhak, S.; Kalatzis, V.; Heilig, R.; Compain, S.; Samson, D.; Vincent, C.; Weil, D.; Cruaud, C.; Sahly, I.; Leibovici, M.; Bitner-Glindzicz, M.; Francis, M.; Lacombe, D.; Vigneron, J.; Charachon, R.; Boven, K.; Bedbeder, P.; Van Regemorter, N.; Weissenbach, J.; Petit, C.: A human homologue of the *Drosophila* eyes absent gene underlies bran-

chio-oto-renal(BOR) syndrome and identifies a novel gene family. *Nature Genet.* 15:157–164, 1997.

- [45051] 18603.Tommerup, N.; Schempp, W.; Meinecke, P.; Peder-
sen, S.; Bolund,L.; Brandt, C.; Goodpasture, C.; Guldberg,
P.; Held, K.; Reinwein,H.; Saugstad, O. D.; Scherer, G.;
Skjeldal, O.; Toder, R.; Westvik,J.; van der Hagen, C. B.;
Wolf, U.: Assignment of an autosomal sexreversal locus
(SRA1) and campomelic dysplasia (CMPD1) to
17q24.3–q25.1. *NatureGenet.* 4: 170–174, 1993.
- [45052] 18604.Azuma, N.; Nishina, S.; Yanagisawa, H.; Okuyama,
T.; Yamada, M.: PAX6 missense mutation in isolated foveal
hypoplasia. (Letter) *NatureGenet.* 13: 141–142, 1996.
- [45053] 18605.Curran, R. E.; Robb, R. M.: Isolated foveal hypopla-
sia. *Arch.Ophthal.* 94: 48–50, 1976.
- [45054] 18606.O'Donnell, F. E., Jr.; Pappas, H. R.: Autosomal dom-
inant fovealhypoplasia and presenile cataracts: a new syn-
drome. *Arch. Ophthal.* 100:279–281, 1982.
- [45055] 18607.Anderson, M. J.; Shelton, G. D.; Cavenee, W. K.; Ar-
den, K. C.:Embryonic expression of the tumor-associated
PAX3–FKHR fusion proteininterferes with the developmen-
tal functions of Pax3. *Proc. Nat. Acad.Sci.* 98: 1589–1594,
2001.
- [45056] 18608.Anderson, M. J.; Viars, C. S.; Czekay, S.; Cavenee,

W. K.; Arden, K. C.: Cloning and characterization of three human forkhead genes that comprise an FKHR-like gene subfamily. *Genomics* 47: 187–199, 1998.

[45057] 18609. Barr, F. G.; Galili, N.; Holick, J.; Biegel, J. A.; Rovera, G.; Emanuel, B. S.: Rearrangement of the PAX3 paired box gene in the paediatric solid tumor alveolar rhabdomyosarcoma. *Nature Genet.* 3:113–117, 1993.

[45058] 18610. Davis, R. J.; Barr, F. G.: Fusion genes resulting from alternative chromosomal translocations are overexpressed by gene-specific mechanisms in alveolar rhabdomyosarcoma. *Proc. Nat. Acad. Sci.* 94: 8047–8051, 1997.

[45059] 18611. Fredericks, W. J.; Galili, N.; Mukhopadhyay, S.; Rovera, G.; Bennicelli, J.; Barr, F. G.; Rauscher, F. J., III: The PAX3–FKHR fusion protein created by the t(2;13) translocation in alveolar rhabdomyosarcoma is a more potent transcriptional activator than PAX3. *Molec. Cell. Biol.* 15: 1522–1535, 1995.

[45060] 18612. Galili, N.; Davis, R. J.; Fredericks, W. J.; Mukhopadhyay, S.; Rauscher, F. J., III; Emanuel, B. S.; Rovera, G.; Barr, F. G.: Fusion of a fork head domain gene to PAX3 in the solid tumour alveolar rhabdomyosarcoma. *Nature Genet.* 5: 230–235, 1993.

[45061] 18613. Khan, J.; Bittner, M. L.; Saal, L. H.; Teichmann, U.;

Azorsa, D.O.; Gooden, G. C.; Pavan, W. J.; Trent, J. M.; Meltzer, P. S.: cDNAmicroarrays detect activation of a myogenic transcription program by the PAX3–FKHR fusion oncogene. *Proc. Nat. Acad. Sci.* 96: 13264–13269, 1999.

[45062] 18614. Angle, C. R.: Congenital bowing and angulation of the long bones. *Pediatrics* 13:257–268, 1954.

[45063] 18615. Bain, A. D.; Barrett, H. S.: Congenital bowing of the long bones: report of a case. *Arch. Dis. Child.* 34: 516–524, 1959.

[45064] 18616. Bell, D. M.; Leung, K. K. H.; Wheatley, S. C.; Ng, L. J.; Zhou, S.; Ling, K. W.; Sham, M. H.; Koopman, P.; Tam, P. P. L.; Cheah, K. S. E.: SOX9 directly regulates the type-II collagen gene. *Nature Genet.* 16: 174–178, 1997.

[45065] 18617. Bi, W.; Huang, W.; Whitworth, D. J.; Deng, J. M.; Zhang, Z.; Behringer, R. R.; de Crombrughe, B.: Haploinsufficiency of Sox9 results in defective cartilage primordia and premature skeletal mineralization. *Proc. Nat. Acad. Sci.* 98: 6698–6703, 2001.

[45066] 18618. Bishop, C. E.; Whitworth, D. J.; Qin, Y.; Agoulnik, A. I.; Agoulnik, I. U.; Harrison, W. R.; Behringer, R. R.; Overbeek, P. A.: A transgenic insertion upstream of Sox9 is associated with dominant XX sex reversal in the mouse. *Nature Genet.* 26: 490–494, 2000.

- [45067] 18619.Caffey, J. P.: Prenatal bowing and thickening of tubular bones,with multiple cutaneous dimples in arms and legs: a congenital syndromeof mechanical origin. Am. J. Dis. Child. 74: 543–562, 1947.
- [45068] 18620.Cameron, F. J.; Hageman, R. M.; Cooke–Yarborough, C.; Kwok, C.;Goodwin, L. L.; Sillence, D. O.; Sinclair, A. H.: A novel germ linemutation in SOX9 causes familial campomelic dysplasia and sex reversal. Hum.Molec. Genet. 5: 1625–1630, 1996.
- [45069] 18621.Cameron, F. J.; Sinclair, A. H.: Mutations in SRY and SOX9: testis–determininggenes. Hum. Mutat. 9: 388–395, 1997.
- [45070] 18622.Cooke, C. T.; Mulcahy, M. T.; Cullity, G. J.; Watson, M.; Sprague,P.: Campomelic dysplasia with sex reversal: morphological and cytogeneticstudies of a case. Pathology 17: 526–529, 1985.
- [45071] 18623.Cremin, B. J.; Orsmond, G.; Beighton, P.: Autosomal recessiveinheritance in camptomelic dwarfism.(Letter) Lancet I: 488–489,1973.
- [45072] 18624.Dagna Bricarelli, F.; Fraccaro, M.; Lindsten, J.; Muller, U.;Baggio, P.; Carbone, L. D. L.; Hjerpe, A.; Lindgren, F.; Mayerova,A.; Ringertz, H.; Ritzen, E. M.; Rovetta, D. C.; Siccherio, C.; Wolf,U.: Sex–reversed XY females with

campomelic dysplasia are H-Y negative. Hum.Genet. 57: 15-22, 1981.

[45073] 18625.Ebensperger, C.; Jager, R. J.; Lattermann, U.; Dagna Bricarelli,F.; Keutel, J.; Lindsten, J.; Rehder, H.; Muller, U.; Wolf, U.: Noevidence of mutations in four candidate genes for male sex determination/differentiationin sex-reversed XY females with campomelic dysplasia. Ann. Genet. 34:233-238, 1991.

[45074] 18626.Fontaine, G.; Walbaum, R.; Farriaux, J. P.; Tilmont, P.; Peuzin,F.; Delecour, M.: Le conseil genetique dans la dysplasie campomelique(a propos de deux observations). J. Genet. Hum. 28: 267-279, 1980.

[45075] 18627.Foster, J. W.; Dominguez-Steglich, M. A.; Guioli, S.; Kwok, C.;Weller, P. A.; Stevanovic, M.; Weissenbach, J.; Mansour, S.; Young,I. D.; Goodfellow, P. N.; Brook, J. D.; Schafer, A. J.: Campomelicdysplasia and autosomal sex reversal caused by mutations in an SRY-relatedgene. Nature 372: 525-530, 1994.

[45076] 18628.Friedrich, U.; Schaefer, E.; Meinecke, P.: Campomelic dysplasiawithout overt campomelia. Clin. Dysmorph. 1: 172-178, 1992.

[45077] 18629.Gasca, S.; Canizares, J.; de Santa Barbara, P.; Mejean, C.; Poulat,F.; Berta, P.; Boizet-Bonhoure, B.: A nu-

clear export signal within the high mobility group domain regulates the nucleocytoplasmic translocation of SOX9 during sexual determination. *Proc. Nat. Acad. Sci.* 99: 11199–11204, 2002.

- [45078] 18630. Glass, R. B. J.; Rosenbaum, K. N.: Acampomelic campomelic dysplasia: further radiographic variations. *Am. J. Med. Genet.* 69: 29–32, 1997.
- [45079] 18631. Hall, B.; Spranger, J. W.: Campomelic dysplasia: further elucidation of a distinct entity. *Am. J. Dis. Child.* 134: 285–289, 1980.
- [45080] 18632. Hoefnagel, D.; Wurster-Hill, D. H.; Dupree, W. B.; Benirschke, K.; Fuld, G. L.: Camptomelic dwarfism associated with XY-gonadal dysgenesis and chromosome anomalies. *Clin. Genet.* 13: 489–499, 1978.
- [45081] 18633. Houston, C. S.; Opitz, J. M.; Spranger, J. W.; Macpherson, R. I.; Reed, M. H.; Gilbert, E. F.; Herrmann, J.; Schinzel, A.: The campomelic syndrome: review, report of 17 cases, and follow-up on the currently 17-year-old boy first reported by Maroteaux et al in 1971. *Am. J. Med. Genet.* 15: 3–28, 1983.
- [45082] 18634. Hovmoller, M. L.; Osuna, A.; Eklof, O.; Fredga, K.; Hjerpe, A.; Lindsten, J.; Ritzen, M.; Stanescu, V.; Svenningsen, N.: Camptomelic dwarfism. A genetically deter-

mined mesenchymal disorder combined with sex reversal.
Hereditas 86: 51–62, 1977.

- [45083] 18635. Huang, W.; Chung, U.; Kronenberg, H. M.; de Crombrughe, B.: The chondrogenic transcription factor Sox9 is a target of signaling by the parathyroid hormone-related peptide in the growth plate of endochondral bones. *Proc. Nat. Acad. Sci.* 98: 160–165, 2001.
- [45084] 18636. Kanai, Y.; Koopman, P.: Structural and functional characterization of the mouse Sox9 promoter: implications for campomelic dysplasia. *Hum. Molec. Genet.* 8: 691–696, 1999.
- [45085] 18637. Medema, R. H.; Kops, G. J. P. L.; Bos, J. L.; Burgering, B. M. T.: AFX-like forkhead transcription factors mediate cell-cycle regulation by Ras and PKB through p27(kip1). *Nature* 404: 782–787, 2000.
- [45086] 18638. Eicher, E. M.; Womack, J. E.: Chromosomal location of soluble glutamic-pyruvic transaminase-1 (Gpt-1) in the mouse. *Biochem. Genet.* 15:1–8, 1977.
- [45087] 18639. Falk, C. T.; Huss, J.: Linkage data on the chromosome 16 markers HP and PGP: no additional support for or against the mapping of GPT. (Abstract) *Cytogenet. Cell Genet.* 40: 626 only, 1985.
- [45088] 18640. Ferrell, R. E.: Personal Communication. Pittsburgh,

Pa. 1/22/1988.

- [45089] 18641.Jeremiah, S. J.; Kielty, C.; Povey, S.; McLellan, T.: Immunological characterization of human GPT in hybrids supports assignment to chromosome
- [45090] 18642.(Abstract) Cytogenet. Cell Genet. 37: 498 only, 1984.8. Kalimanovska, V.; Majkic-Singh, N.; Stojanov, M.; Grozdanic, V.; Vucetic, G.; Andelic, M.; Gligorovic, V.; Tomasevic, R.: Human redcell glutamic-pyruvic transaminase polymorphism in Serbia, Yugoslavia. Hum.Hered. 33: 319-321, 1983.
- [45091] 18643.Kielty, C. M.; Povey, S.; Hopkinson, D. A.: Regulation of expression of liver-specific enzymes: II. Activation and chromosomal localization of soluble glutamate-pyruvate transaminase. Ann. Hum. Genet. 46:135-143, 1982.
- [45092] 18644.King, M.-C.; Go, R. C. P.; Elston, R. C.; Lynch, H. T.; Petrakis, N. L.: Allele increasing susceptibility to human breast cancer maybe linked to the glutamate-pyruvate transaminase locus. Science 208:406-408, 1980.
- [45093] 18645.Kompf, J.: Population genetics of soluble glutamic-pyruvic transaminase (EC:2.6.1.2): gene frequencies in southwestern Germany. Humangenetik 14:76-77, 1971.
- [45094] 18646.Lahav, M.; Szeinberg, A.: A red-cell glutamic-pyruvic transaminase polymorphism in several population

groups in Israel. Hum. Hered. 22:533–538, 1972.

[45095] 18647. Marazita, M. L.; Spence, M. A.; Sparkes, R. S.; Field, L. L.; Crandall, B. F.; Sparkes, M. C.; Crist, M.: Linkage relations of GPT (glutamic–pyruvate transaminase). (Abstract) Cytogenet. Cell Genet. 40: 690 only, 1985.

[45096] 18648. McLellan, T.: Two previously undetected variants of glutamic–pyruvic transaminase found by acidic polyacrylamide gel electrophoresis. Am. J. Hum. Genet. 34: 623–628, 1982.

[45097] 18649. McLellan, T.; Cannon, L. A.; Bishop, D. T.; Skolnick, M. H.: The cumulative lod score between a breast cancer susceptibility locus and GPT is –3.86. (Abstract) Cytogenet. Cell Genet. 37: 536–537, 1984.

[45098] 18650. Mithal, Y.; Lane, A. B.; Jenkins, T.: Absence of red cell glutamic–pyruvate transaminase: discovery of a 'silent' allele homozygote. Am. J. Hum. Genet. 32: 42–46, 1980.

[45099] 18651. O'Connell, P.; Nakamura, Y.; Lathrop, G. M.; Lepert, M.; Cartwright, P.; Lalouel, J.–M.; White, R.: Three genetic linkage groups on chromosome 8. (Abstract) Cytogenet. Cell Genet. 46: 672 only, 1987.

[45100] 18652. Olaisen, B.: Genetics of the GPT system: family, mother–child and association studies. Clin. Genet. 7: 245–254, 1975.

- [45101] 18653. Pelzer, C. F.; Norum, R. A.: Identification of human red cell glutamate-pyruvate transaminase (GPT) phenotypes by isoelectric focusing. *Am.J. Hum. Genet.* 37: 147-152, 1985.
- [45102] 18654. Rocha, J.; Amorim, A.; Almeida, V. M.; Oliveira, J. P.; Leao, M.; Tavares, M. C.; Pereira, M. S.; Vidal-Pinheiro, L.: Gene dosage evidence for the regional assignment of GPT (glutamate-pyruvate transaminase; E.C. 2.6.1.2) locus to 8q24.2-8qter. *Hum. Genet.* 80: 299-300, 1988.
- [45103] 18655. Sanders, M. F.; King, M. C.; Lattanzio, D.; Crandall, J.; Leung, R.: Absence of linkage between HP and GPT. (Abstract) *Cytogenet. Cell Genet.* 37: 536-537, 1984.
- [45104] 18656. Santachiara Benerecetti, A. S.; Beretta, M.; Pampiglione, S.: Red cell glutamic-pyruvic transaminase polymorphism in a sample of the Italian population: a new variant allele: GPT(8). *Hum. Hered.* 25: 276-278, 1975.
- [45105] 18657. Sohocki, M. M.; Sullivan, L. S.; Harrison, W. R.; Sodergren, E. J.; Elder, F. F. B.; Weinstock, G.; Tanase, S.; Daiger, S. P.: Human glutamate pyruvate transaminase (GPT): localization to 8q24.3, cDNA and genomic sequences, and polymorphic sites. *Genomics* 40: 247-252, 1997.
- [45106] 18658. Sparkes, M. C.; Crist, M.; Sparkes, R. S.: Glutamate

pyruvate transaminase null allele in seven new families.

Hum. Genet. 65:147–148, 1983.

- [45107] 18659. Wijnen, L. M. M.; Meera Khan, P.: Assignment of GPT to human chromosome 16. (Abstract) Cytogenet. Cell Genet. 32: 327 only, 1982.
- [45108] 18660. Burant, C. F.; Takeda, J.; Brot-Laroche, E.; Bell, G. I.; Davidson, N. O.: Fructose transporter in human spermatozoa and small intestine is GLUT5. J. Biol. Chem. 267: 14523–14526, 1992.
- [45109] 18661. Davidson, N. O.; Hausman, A. M. L.; Ifkovits, C. A.; Buse, J. B.; Gould, G. W.; Burant, C. F.; Bell, G. I.: Human intestinal glucose transporter expression and localization of GLUT5. Am. J. Physiol. 262:C795–C800, 1992.
- [45110] 18662. Cremonini, N.; Graziano, E.; Chiarini, V.; Sforza, A.; Zampa, G. A.: Atypical McCune–Albright syndrome associated with growth hormone–prolactin pituitary adenoma: natural history, long-term follow-up, and SMS 201–995–bromocriptine combined treatment results. J. Clin. Endocr. Metab. 75: 1166–1169, 1992.
- [45111] 18663. DeChiara, T.; Robertson, E. J.; Efstratiadis, A.: Parental imprinting of the mouse insulin-like growth factor II gene. Cell 64: 849–859, 1991.
- [45112] 18664. Eddy, M. C.; Jan de Beur, S. M.; Yandow, S. M.;

McAlister, W.H.; Shore, E. M.; Kaplan, F. S.; Whyte, M. P.; Levine, M. A.: Deficiency of the alpha-subunit of the stimulatory G protein and severe extraskeletal ossification. *J. Bone Miner. Res.* 15: 2074–2083, 2000.

[45113] 18665. Falconer, M. A.; Cope, C. L.; Robb-Smith, A. H. T.: Fibrous dysplasia of bone with endocrine disorders and cutaneous pigmentation (Albright's disease). *Quart. J. Med.* 11: 121–154, 1942.

[45114] 18666. Albertsen, H. M.; Smith, S. A.; Mazoyer, S.; Fujimoto, E.; Stevens, J.; Williams, B.; Rodriguez, P.; Cropp, C. S.; Slijepcevic, P.; Carlson, M.; Robertson, M.; Bradley, P.; Lawrence, E.; Harrington, T.; Mei Sheng, Z.; Hoopes, R.; Sternberg, N.; Brothman, A.; Callahan, R.; Ponder, B. A. J.; White, R.: A physical map and candidate genes in the BRCA1 region on chromosome 17q12–21. *Nature Genet.* 7: 472–479, 1994.

[45115] 18667. Kato, M.; Araki, S.: Paroxysmal kinesigenic choreoathetosis. *Arch. Neurol.* 20: 508–513, 1969.

[45116] 18668. Pilla, M.; Perachon, S.; Sautel, F.; Garrido, F.; Mann, A.; Wermuth, C. G.; Schwartz, J.-C.; Everitt, B. J.; Sokoloff, P.: Selective inhibition of cocaine-seeking behaviour by a partial dopamine D3 receptor agonist. *Nature* 400: 371–375, 1999.

- [45117] 18669.Demange, P.; Voges, D.; Benz, J.; Liemann, S.; Gotting, P.; Berendes,R.; Burger, A.; Huber, R.: Annexin V: the key to understanding ionselectivity and voltage regulation? Trends Biochem. Sci. 19: 272–276,1994.
- [45118] 18670.Funakoshi, T.; Heimark, R. L.; Hendrickson, L. E.; McMullen, B.A.; Fujikawa, K.: Human placental anticoagulant protein: isolationand characterization. Biochemistry 26: 5572–5578, 1987.
- [45119] 18671.Grundmann, U.; Abel, K.–J.; Bohn, H.; Lobermann, H.; Lottspeich,F.; Kupper, H.: Characterization of cDNA encoding human placentalanticoagulant protein (PP4): homology with the lipocortin family. Proc.Nat. Acad. Sci. 85: 3708–3712, 1988.
- [45120] 18672.Kaplan, R.; Jaye, M.; Burgess, W. H.; Schlaepfer, D. D.; Haigler,H. T.: Cloning and expression of cDNA for human endonexin II, a Ca(2+)and phospholipid binding protein. J. Biol. Chem. 263: 8037–8043,1988.
- [45121] 18673.Modi, W. S.; Seuanez, H.; Jaye, M.; Kaplan, R.; Haigler, H.; O'Brien,S. J.: Chromosomal mapping of the endonexin II gene. (Abstract) Cytogenet.Cell Genet. 51: 1046, 1989.
- [45122] 18674.Modi, W. S.; Seuanez, H. N.; Jaye, M.; Haigler, H. J.; Kaplan,R.; O'Brien, S. J.: The human endonexin II (ENX2)

gene is located at 4q28–q32. *Cytogenet. Cell Genet.* 52: 167–169, 1989.

[45123] 18675. Rodriguez–Garcia, M. I.; Kozak, C. A.; Morgan, R. O.; Fernandez, M. P.: Mouse annexin V chromosomal localization, cDNA sequence conservation, and molecular evolution. *Genomics* 31: 151–157, 1996.

[45124] 18676. Tait, J. F.; Frankenberry, D. A.; Shiang, R.; Murray, J. C.; Adler, D. A.; Disteché, C. M.: Chromosomal localization of the human gene for annexin V (placental anticoagulant protein I) to 4q26–q28. *Cytogenet. Cell Genet.* 57: 187–192, 1991.

[45125] 18677. Arinami, T.; Ishikawa, M.; Inoue, A.; Yanagisawa, M.; Masaki, T.; Yoshida, M. C.; Hamaguchi, H.: Chromosomal assignments of the human endothelin family genes: the endothelin–1 gene (EDN1) to 6p23–p24, the endothelin–2 gene (EDN2) to 1p34, and the endothelin–3 gene (EDN3) to 20q13.2–q13.3. *Am. J. Hum. Genet.* 48: 990–996, 1991.

[45126] 18678. Benatti, L.; Bonecchi, L.; Cozzi, L.; Sarmientos, P.: Two preproendothelin1 mRNAs transcribed by alternative promoters. *J. Clin. Invest.* 91:1149–1156, 1993.

[45127] 18679. Berge, K. E.; Berg, K.: No effect of a TaqI polymorphism in DNA at the endothelin I (EDN1) locus on normal

blood pressure level or variability. Clin. Genet. 41: 90–95, 1992.

[45128] 18680. Bloch, K. D.; Friedrich, S. P.; Lee, M.-E.; Eddy, R. L.; Shows, T. B.; Quertermous, T.: Structural organization and chromosomal assignment of the gene encoding endothelin. J. Biol. Chem. 264: 10851–10857, 1989.

[45129] 18681. Bourgeois, C.; Robert, B.; Rebourcet, R.; Mondon, F.; Mignot, T.-M.; Duc-Goiran, P.; Ferre, F.: Endothelin-1 and ET(A) receptor expression in vascular smooth muscle cells from human placenta: a new ET(A) receptor messenger ribonucleic acid is generated by alternative splicing of exon 3. J. Clin. Endocr. Metab. 82: 3116–3123, 1997.

[45130] 18682. Clouthier, D. E.; Hosoda, K.; Richardson, J. A.; Williams, S. C.; Yanagisawa, H.; Kuwaki, T.; Kumada, M.; Hammer, R. E.; Yanagisawa, M.: Cranial and cardiac neural crest defects in endothelin-A receptor-deficient mice. Development 125: 813–824, 1998.

[45131] 18683. Giaid, A.; Gibson, S. J.; Ibrahim, N. B. N.; Legon, S.; Bloom, S. R.; Yanagisawa, M.; Masaki, T.; Varndell, I. M.; Polak, J. M.: Endothelin 1, an endothelium-derived peptide, is expressed in neurons of the human spinal cord and dorsal root ganglia. Proc. Nat. Acad. Sci. 86: 7634–7638, 1989.

- [45132] 18684.Inoue, A.; Yanagisawa, M.; Kimura, S.; Kasuya, Y.; Miyauchi, T.;Goto, K.; Masaki, T.: The human endothelin family: three structurallyand pharmacologically distinct isopeptides predicted by three separategenes. Proc. Nat. Acad. Sci. 86: 2863–2867, 1989.
- [45133] 18685.Inoue, A.; Yanagisawa, M.; Takuwa, Y.; Mitsui, Y.; Kobayashi, M.;Masaki, T.: The human preproendothelin–1 gene: complete nucleotidesequence and regulation of expression. J. Biol. Chem. 264: 14954–14959,1989.
- [45134] 18686.Shaikh, S.; Ball, D.; Craddock, N.; Castle, D.; Hunt, N.; Mant,R.; Owen, M.; Collier, D.; Gill, M.: The dopamine D3 receptor gene:no association with bipolar affective disorder. J. Med. Genet. 30:308–309, 1993.
- [45135] 18687.Sokoloff, P.; Giros, B.; Martres, M.–P.; Bouthenet, M.–L.; Schwartz,J.–C.: Molecular cloning and characteriza–tion of a novel dopaminereceptor (D–3) as a target for neuroleptics. Nature 347: 146–151,1990.
- [45136] 18688.Spurlock, G.; Williams, J.; McGuffin, P.; Aschauer, H. N.; Lenzinger,E.; Fuchs, K.; Sieghart, W. C.; Meszaros, K.; Fathi, N.; Laurent,C.; Mallet, J.; Macciardi, F.; Pedrini, S.; Gill, M.; Hawi, Z.; Gibson,S.; Jazin, E. E.; Yang, H.–T.; Adolfsson, R.; Pato, C. N.; Dourado,A. M.; Owen, M. J.: Eu–ropean Multicentre Association Study of Schizophrenia:a

study of the DRD2 ser311-to-cys and DRD3 ser9-to-gly polymorphisms. *Am.J. Med. Genet.* 81: 24–28, 1998.

- [45137] 18689. Asghari, V.; Sanyal, S.; Buchwaldt, S.; Paterson, A.; Jovanovic, V.; Van Tol, H. H.: Modulation of intracellular cyclic AMP levels by different human dopamine D4 receptor variants. *J. Neurochem.* 65:1157–1165, 1995.
- [45138] 18690. Benjamin, J.; Li, L.; Patterson, C.; Greenberg, B. D.; Murphy, D. L.; Hamer, D. H.: Population and familial association between the D4 dopamine receptor gene and measures of novelty seeking. *Nature Genet.* 12: 81–84, 1996.
- [45139] 18691. Castellanos, F. X.; Lau, E.; Tayebi, N.; Lee, P.; Long, R. E.; Giedd, J. N.; Sharp, W.; Marsh, W. L.; Walter, J. M.; Hamburger, S. D.; Ginns, E. I.; Rapoport, J. L.; Sidransky, E.: Lack of an association between a dopamine-4 receptor polymorphism and attention-deficit/hyperactivity disorder: genetic and brain morphometric analyses. *Molec. Psychiat.* 3:431–434, 1998.
- [45140] 18692. Chang, F.-M.; Kidd, J. R.; Livak, K. J.; Pakstis, A. J.; Kidd, K. K.: The world-wide distribution of allele frequencies at the human dopamine D4 receptor locus. *Hum. Genet.* 98: 91–101, 1996.
- [45141] 18693. Cloninger, C. R.; Adolfsson, R.; Svrakic, N. M.: Mapping genes for human personality. *Nature Genet.* 12:

3–4, 1996.

- [45142] 18694. Cloninger, C. R.; Svrakic, D. M.; Przybeck, T. R.: A psychobiological model of temperament and character. *Arch. Gen. Psychiat.* 50: 975–990, 1993.
- [45143] 18695. Cook, E. H.; Stein, M. A.; Krasowski, M. D.; Cox, N. J.; Olkon, D. M.; Kieffer, J. E.; Leventhal, B. L.: Association of attention-deficit disorder and the dopamine transporter gene. *Am. J. Hum. Genet.* 56:993–998, 1995.
- [45144] 18696. De Luca, A.; Rizzardi, M.; Torrente, I.; Alessandroni, R.; Salvioli, G. P.; Filograsso, N.; Dallapiccola, B.; Novelli, G.: Dopamine D4 receptor (DRD4) polymorphism and adaptability trait during infancy: a longitudinal study in 1- to 5-month-old neonates. *Neurogenetics* 3:79–82, 2001.
- [45145] 18697. Ding, Y.-C.; Chi, H.-C.; Grady, D. L.; Morishima, A.; Kidd, J. R.; Kidd, K. K.; Flodman, P.; Spence, M. A.; Schuck, S.; Swanson, J. M.; Zhang, Y.-P.; Moyzis, R. K.: Evidence of positive selection acting at the human dopamine receptor D4 gene locus. *Proc. Nat. Acad. Sci.* 99: 309–314, 2002.
- [45146] 18698. Ebstein, R. P.; Novick, O.; Umansky, R.; Pirelli, B.; Osher, Y.; Blaine, D.; Bennett, E. R.; Nemanov, L.; Katz, M.; Belmaker, R. H.: Dopamine D4 receptor exon III polymorphism associated with the human personality trait of nov-

elty seeking. *Nature Genet.* 12: 78–80, 1996.

- [45147] 18699. Eisenberg, J.; Zohar, A.; Mei–Tal, G.; Steinberg, A.; Tartakovsky, E.; Gritsenko, I.; Nemanov, L.; Ebstein, R. P.: A haplotype relative risk study of the dopamine D4 receptor (DRD4) exon III repeat polymorphism and attention deficit hyperactivity disorder (ADHD). *Am. J. Med. Genet. (Neuropsychiat. Genet.)* 96: 258–261, 2000.
- [45148] 18700. Gelernter, J.; Kennedy, J. L.; Van Tol, H. H. M.; Civelli, O.; Kidd, K. K.: The D4 dopamine receptor (DRD4) maps to distal 11p close to HRAS. *Genomics* 13: 208–210, 1992.
- [45149] 18701. Gelernter, J.; Kennedy, J. L.; Van Tol, H. H. M.; Niznik, H. B.; Civelli, O.; Kidd, K. K.: The D4 dopamine receptor (DRD4) maps to distal 11p close to HRAS. (Abstract) *Cytogenet. Cell Genet.* 58:1960, 1991.
- [45150] 18702. Gelernter, J.; Kranzler, H.; Coccaro, E.; Siever, L.; New, A.; Mulgrew, C. L.: D4 dopamine–receptor (DRD4) alleles and novelty seeking in substance–dependent, personality–disorder, and control subjects. *Am. J. Hum. Genet.* 61: 1144–1152, 1997.
- [45151] 18703. Harpending, H.; Cochran, G.: In our genes. *Proc. Nat. Acad. Sci.* 99: 10–12, 2002.
- [45152] 18704. Jovanovic, V.; Guan, H.–C.; Van Tol, H. H. M.: Com–

parative pharmacological and functional analysis of the human dopamine D4.2 and D4.10 receptor variants. Pharmacogenetics 9: 561–568, 1999.

- [45153] 18705. LaHoste, G. J.; Swanson, J. M.; Wigal, S. B.; Glabe, C.; Wigal, T.; King, N.; Kennedy, J. L.: Dopamine D4 receptor gene polymorphism is associated with attention deficit hyperactivity disorder. Molec. Psychiat. 1: 121–124, 1996.
- [45154] 18706. Liu, I. S. C.; Seeman, P.; Sanyal, S.; Ulpian, C.; Rodgers-Johnson, P. E. B.; Serjeant, G. R.; Van Tol, H. H. M.: Dopamine D4 receptor variant in Africans, D4(valine194glycine), is insensitive to dopamine and clozapine: report of a homozygous individual. Am. J. Med. Genet. 61:277–282, 1996.
- [45155] 18707. Malhotra, A. K.; Virkkunen, M.; Rooney, W.; Eggert, M.; Linnoila, M.; Goldman, D.: The association between the dopamine D(4) receptor (D4DR) 16 amino acid repeat polymorphism and novelty seeking. Molec. Psychiat. 1: 388–391, 1996.
- [45156] 18708. Muramatsu, T.; Higuchi, S.; Murayama, M.; Matsushita, S.; Hayashida, M.: Association between alcoholism and the dopamine D4 receptor gene. J. Med. Genet. 33: 113–115, 1996.
- [45157] 18709. Nothen, M. M.; Cichon, S.; Hemmer, S.; Hebebrand,

J.; Remschmidt, H.; Lehmkuhl, G.; Poustka, F.; Schmidt, M.; Catalano, M.; Fimmers, R.; Korner, J.; Rietschel, M.; Propping, P.: Human dopamine D4 receptor gene: frequent occurrence of a null allele and observation of homozygosity. *Hum. Molec. Genet.* 3: 2207–2212, 1994.

[45158] 18710. Petronis, A.; Van Tol, H. H. M.; Lichter, J. B.; Livak, K. J.; Kennedy, J. L.: The D4 dopamine receptor gene maps on 11p proximal to HRAS. *Genomics* 18: 161–163, 1993.

[45159] 18711. Rowe, D. C.; Stever, C.; Giedinghagen, L. N.; Gard, J. M. C.; Cleveland, H. H.; Terris, S. T.; Mohr, J. H.; Sherman, S.; Abramowitz, A.; Waldman, I. D.: Dopamine DRD4 receptor polymorphism and attention deficit hyperactivity disorder. *Molec. Psychiat.* 3: 419–426, 1998.

[45160] 18712. Rubinstein, M.; Phillips, T. J.; Bunzow, J. R.; Falzone, T. L.; Dziewczapolski, G.; Zhang, G.; Fang, Y.; Larson, J. L.; McDougall, J. A.; Chester, J. A.; Saez, C.; Pugsley, T. A.; Gershnik, O.; Low, M. J.; Grandy, D. K.: Mice lacking dopamine D4 receptors are supersensitive to ethanol, cocaine, and methamphetamine. *Cell* 90: 991–1001, 1997.

[45161] 18713. Raskind, W. H.; Bolin, T.; Wolff, J.; Fink, J.; Matsushita, M.; Litt, M.; Lipe, H.; Bird, T. D.: Further localization of a gene for paroxysmal dystonic choreoathetosis to a 5-cM region on chromosome 2q34. *Hum. Genet.* 102:

93–97, 1998.

- [45162] 18714.Richards, R. N.; Barnett, H. J.: Paroxysmal dystonic choreoathetosis:a family study and review of the literature. *Neurology* 18: 461–469,1968.
- [45163] 18715.Stevens, H. F.: Paroxysmal choreo–athetosis: a form of reflexepilepsy. *Arch. Neurol.* 14: 415–420, 1966.
- [45164] 18716.Wagner, G. S.; McLees, B. D.; Hatcher, M. A., Jr.: Familial paroxysmalchoreo–athetosis. (Abstract) *Neurology* 16: 307, 1966.
- [45165] 18717.Williams, J.; Stevens, H.: Familial paroxysmal choreo–athetosis. *Pediatrics* 31:656–659, 1963.
- [45166] 18718.Barsh, G. S.; Seeburg, P. H.; Gelinas, R. E.: The human growthhormone gene family:structure and evolution of the chromosomal locus. *NucleicAcids Res.* 11: 3939–3958, 1983.
- [45167] 18719.Cremers, F. P. M.; Molloy, C. M.; van de Pol, D. J. R.; van denHurk, J. A. J. M.; Bach, I.; Geurts van Kessel, A. H. M.; Ropers,H.–H.: An autosomal homologue of the choroïderemia gene colocalizeswith the Usher syndrome type II locus on the distal part of chromosome1q. *Hum. Molec. Genet.* 1: 71–75, 1992.
- [45168] 18720.Halford, S.; Freedman, M. S.; Bellingham, J.; Inglis, S. L.; Poopalasundaram,S.; Soni, B. G.; Foster, R. G.; Hunt,

D. M.: Characterization of a novel human opsin gene with wide tissue expression and identification of embedded and flanking genes on chromosome 1q43. *Genomics* 72:203–208, 2001.

[45169] 18721. van Bokhoven, H.; van Genderen, C.; Molloy, C. M.; van de Pol, D. J. R.; Cremers, C. W. R. J.; van Aarem, A.; Schwartz, M.; Rosenberg, T.; Geurts van Kessel, A. H. M.; Ropers, H.-H.; Cremers, F. P. M.: Mapping of the chorioideremia-like (CHML) gene at 1q42-qter and mutation-analysis in patients with Usher syndrome type II. *Genomics* 19: 385–387, 1994.

[45170] 18722. Amato, F.; Warnes, G. M.; Kirby, C. A.; Norman, R. J.: Infertility caused by hCG autoantibody. *J. Clin. Endocr. Metab.* 87: 993–997, 2002.

[45171] 18723. Naylor, S. L.; Chin, W. W.; Goodman, H. M.; Lalley, P. A.; Grzeschik, K.-H.; Sakaguchi, A. Y.: Chromosome assignment of the genes encoding the alpha and beta subunits of the glycoprotein hormones in man and mouse. *Somat. Cell Genet.* 9: 757–770, 1983.

[45172] 18724. Hart, A. W.; Baeza, N.; Apelqvist, A.; Edlund, H.: Attenuation of FGF signalling in mouse beta-cells leads to diabetes. *Nature* 408:864–868, 2000.

[45173] 18725. Mammarella, S.; Romano, F.; Di Valerio, A.; Creati,

B.; Esposito, D. L.; Palmirotta, R.; Capani, F.; Vitullo, P.; Volpe, G.; Battista, P.; Della Loggia, F.; Mariani-Costantini, R.; Cama, A.: Interaction between the G1057D variant of IRS-2 and overweight in the pathogenesis of type 2 diabetes. *Hum. Molec. Genet.* 9: 2517–2521, 2000.

[45174] 18726. Triggs-Raine, B. L.; Kirkpatrick, R. D.; Kelly, S. L.; Norquay, L. D.; Cattini, P. A.; Yamagata, K.; Hanley, A. J. G.; Zinman, B.; Harris, S. B.; Barrett, P. H.; Hegele, R. A.: HNF1- α G319S, a transactivation-deficient mutant, is associated with altered dynamics of diabetes onset in an Oji-Cree community. *Proc. Nat. Acad. Sci.* 99:4614–4619, 2002.

[45175] 18727. Tuomi, T.; Carlsson, A.; Li, H.; Isomaa, B.; Miettinen, A.; Nilsson, A.; Nissen, M.; Ehrnstrom, B.-O.; Forsen, B.; Snickars, B.; Lahti, K.; Forsblom, C.; Saloranta, C.; Taskinen, M.-R.; Groop, L. C.: Clinical and genetic characteristics of type 2 diabetes with and without GAD antibodies. *Diabetes* 48: 150–157, 1999.

[45176] 18728. Yuan, M.; Konstantopoulos, N.; Lee, J.; Hansen, L.; Li, Z.-W.; Karin, M.; Shoelson, S. E.: Reversal of obesity- and diet-induced insulin resistance with salicylates or targeted disruption of I κ B- β . *Science* 293:1673–1677, 2001.

- [45177] 18729.Hart, T. C.; Champagne, C.; Zhou, J.; Van Dyke, T. E.: Assignment of the gene for diacylglycerol kinase (DAGK) to human chromosome 12. *Mammalian Genome* 5: 123–124, 1994.
- [45178] 18730.Hart, T. C.; Zhou, J.; Champagne, C.; Van Dyke, T. E.; Rao, P.N.; Pettenati, M. J.: Assignment of the human diacylglycerol kinase gene (DAGK) to 12q13.3 using fluorescence in situ hybridization analysis. *Genomics* 22:246–247, 1994.
- [45179] 18731.Pilz, A.; Schaap, D.; Hunt, D.; Fitzgibbon, J.: Chromosomal localization of three mouse diacylglycerol kinase (DAGK) genes: genes sharing sequence homology to the *Drosophila* retinal degeneration A (rdgA) gene. *Genomics* 26:599–601, 1995.
- [45180] 18732.Schaap, D.; de Widt, J.; van der Wal, J.; Vandekerckhove, J.; van Damme, J.; Gussow, D.; Ploegh, H. L.; van Blitterswijk, W. J.; van der Bend, R. L.: Purification, cDNA-cloning and expression of human diacylglycerol kinase. *FEBS Lett.* 275: 151–158, 1990.
- [45181] 18733.Barker, P. E.; Shipp, M. A.; D'Adamio, L.; Masteller, E. L.; Reinherz, E. L.: The common acute lymphoblastic leukemia antigen gene maps to chromosomal region 3(q21–q27). *J. Immun.* 142: 283–287, 1989.

- [45182] 18734.D'Adamio, L.; Shipp, M. A.; Masteller, E. L.; Reinherz, E. L.:Organization of the gene encoding common acute lymphoblastic leukemiaantigen (neutral endopeptidase 24.11): multiple miniexons and separate5-prime untranslated regions. Proc. Nat. Acad. Sci. 86: 7103–7107,1989.
- [45183] 18735.Debiec, H.; Guigonis, V.; Mougenot, B.; Decobert, F.; Haymann,J.–P.; Bensman, A.; Deschenes, G.; Ronco, P. M.: Antenatal membranousglomerulonephritis due to anti-neutral endopeptidase antibodies. NewEng. J. Med. 346: 2053–2060, 2002.
- [45184] 18736.Letarte, M.; Vera, S.; Tran, R.; Addis, J. B. L.; Onizuka, R. J.;Quackenbush, E. J.; Jongeneel, C. V.; McInnes, R. R.: Common acutelymphocytic leukemia antigen is identical to neutral endopeptidase. J.Exp. Med. 168: 1247–1253, 1988.
- [45185] 18737.Shipp, M. A.; Vijayaraghavan, J.; Schmidt, E. V.; Masteller, E.L.; D'Adamio, L.; Hersh, L. B.; Reinherz, E. L.: Common acute lymphoblasticleukemia antigen (CALLA) is active neutral endopeptidase 24.11 ('enkephalinase'):direct evidence by cDNA transfection analysis. Proc. Nat. Acad. Sci. 86:297–301, 1989.
- [45186] 18738.Tran–Paterson, R.; Willard, H. F.; Letarte, M.: The

common acutelymphoblastic leukemia antigen (neutral endopeptidase--3.4.24.11)gene is located on human chromosome 3. Cancer Genet. Cytogenet. 42:129-134, 1989.

[45187] 18739.Lahn, B. T.; Page, D. C.: Four evolutionary strata on the humanX chromosome. Science 286: 964-967, 1999.

[45188] 18740.Lee, F. A.; Issacs, H.; Strauss, J.: The 'campomelic' syndrome.Short life-span dwarfism with respiratory distress, hypotonia, peculiarfacies, and multiple skeletal and cartilaginous deformities. Am.J. Dis. Child. 124: 485-496, 1972.

[45189] 18741.Lynch, S. A.; Gaunt, M. L.; Minford, A. M. B.: Campomelic dysplasia:evidence of autosomal dominant inheritance. J. Med. Genet. 30: 683-686,1993.

[45190] 18742.Macpherson, R. I.; Skinner, S. A.; Donnenfeld, A. E.: Acampomeliccampomelic dysplasia. Pediat. Radiol. 20: 90-93, 1989.

[45191] 18743.Mansour, S.; Hall, C. M.; Pembrey, M. E.; Young, I. D.: A clinicaland genetic study of campomelic dysplasia. J. Med. Genet. 32: 415-420,1995.

[45192] 18744.Maraia, R.; Saal, H. M.; Wangsa, D.: A chromosome 17q de novoparacentric inversion in a patient with campomelic dysplasia; casereport and etiologic hypothesis.

Clin. Genet. 39: 401–408, 1991.

[45193] 18745.Maroteaux, P.; Spranger, J. W.; Opitz, J. M.; Kucera, J.; Lowry, R. B.; Schimke, R. N.; Kagan, S. M.: Le syndrome campomelique. PresseMed. 22: 1157–1162, 1971.

[45194] 18746.Meyer, J.; Sudbeck, P.; Held, M.; Wagner, T.; Schmitz, M. L.;Bricarelli, F. D.; Eggermont, E.; Friedrich, U.; Haas, O. A.; Kobelt, A.; Leroy, J. G.; van Maldergem, L.; Michel, E.; Mitulla, B.; Pfeiffer, R. A.; Schinzel, A.; Schmidt, H.; Scherer, G.: Mutational analysis of the SOX9 gene in campomelic dysplasia and autosomal sex reversal: lack of genotype/phenotype correlations. Hum. Molec. Genet. 6: 91–98, 1997.

[45195] 18747.Moedjono, S. J.; Crandall, B. F.; Sparkes, R. S.; Feldman, G.M.; Austin, G. E.; Perry, S.: The campomelic syndrome in a singleton and monozygotic twins. Clin. Genet. 18: 397–401, 1980.

[45196] 18748.Moog, U.; Jansen, N. J. G.; Scherer, G.; Schrandt-Stumpel, C.T. R. M.: A campomelic campomelic syndrome. Am. J. Med. Genet. 104:239–245, 2001.

[45197] 18749.Morais da Silva, S.; Hacker, A.; Harley, V.; Goodfellow, P.; Swain, A.; Lovell-Badge, R.: Sox9 expression during gonadal development implies a conserved role for the gene in testis differentiation in mammals and birds. Nature

Genet. 14: 62–68, 1996.

- [45198] 18750. Murakami, S.; Kan, M.; McKeehan, W. L.; de Crombrughe, B.: Up-regulation of the chondrogenic Sox9 gene by fibroblast growth factors is mediated by the mitogen-activated protein kinase pathway. *Proc. Nat. Acad. Sci.* 97: 1113–1118, 2000.
- [45199] 18751. Ninomiya, S.; Isomura, M.; Narahara, K.; Seino, Y.; Nakamura, Y.: Isolation of a testis-specific cDNA on chromosome 17q from a region adjacent to the breakpoint of t(12;17) observed in a patient with acampomelic campomelic dysplasia and sex reversal. *Hum. Molec. Genet.* 5: 69–72, 1996.
- [45200] 18752. Ninomiya, S.; Yokoyama, Y.; Teraoka, M.; Mori, R.; Inoue, C.; Yamashita, S.; Tamai, H.; Funato, M.; Seino, Y.: A novel mutation (296 del G) of the SOX9 gene in a patient with campomelic syndrome and sex reversal. *Clin. Genet.* 58: 224–227, 2000.
- [45201] 18753. Olney, P. N.; Kean, L. S.; Graham, D.; Elsas, L. J.; May, K. M.: Campomelic syndrome and deletion of SOX9. *Am. J. Med. Genet.* 84:20–24, 1999.
- [45202] 18754. Ozkilic, A.; Seven, M.; Yuksel, A.: A case of acampomelic campomelic dysplasia. *Genet. Counsel.* 13: 23–28, 2002.

- [45203] 18755. Patel, M.; Dorman, K. S.; Zhang, Y.-H.; Huang, B.-L.; Arnold, A. P.; Sinsheimer, J. S.; Vilain, E.; McCabe, E. R. B.: Primate DAX1, SRY, and SOX9: evolutionary stratification of sex-determination pathway. *Am. J. Hum. Genet.* 68: 275-280, 2001.
- [45204] 18756. Pfeifer, D.; Kist, R.; Dewar, K.; Devon, K.; Lander, E. S.; Birren, B.; Korniszewski, L.; Back, E.; Scherer, G.: Campomelic dysplasia translocation breakpoints are scattered over 1 Mb proximal to SOX9: evidence for an extended control region. *Am. J. Hum. Genet.* 65: 111-124, 1999.
- [45205] 18757. Puck, S. M.; Haseltine, F. P.; Francke, U.: Absence of H-Y antigen in an XY female with campomelic dysplasia. *Hum. Genet.* 57: 23-27, 1981.
- [45206] 18758. Rimoin, D. L.: Personal Communication. Torrance, Calif. 8/12/1976.
- [45207] 18759. Rodriguez, J. I.: Vascular anomalies in campomelic syndrome. *Am. J. Med. Genet.* 46: 185-192, 1993.
- [45208] 18760. Savarirayan, R.; Bankier, A.: A campomelic campomelic dysplasia with de novo 5q;17q reciprocal translocation and severe phenotype. *J. Med. Genet.* 35: 597-599, 1998.
- [45209] 18761. Schimke, R. N.: XY sex-reversed campomelia - possibly an X-linked disorder? (Letter) *Clin. Genet.* 16:

62–63, 1979.

- [45210] 18762.Shafai, T.; Schwartz, L.: Camptomelic syndrome in siblings. J.Pediat. 89: 512–513, 1976.
- [45211] 18763.Spranger, J.: Advances in bone dysplasias.
(Abstract) SixthInt. Cong. Hum. Genet. Jerusalem , 1981.
- [45212] 18764.Stuve, A.; Wiedemann, H.–R.: Congenital bowing of the long bones in two sisters. (Letter) Lancet I: 495, 1971.
- [45213] 18765.Sudbeck, P.; Schmitz, M. L.; Baeuerle, P. A.;
Scherer, G.: Sexreversal by loss of the C-terminal transactivation domain of humanSOX9. Nature Genet. 13:
230–232, 1996.
- [45214] 18766.Thong, M.–K.; Scherer, G.; Kozlowski, K.; Haan, E.;
Morris, L.: Acampomelic campomelic dysplasia with SOX9 mutation. Am. J. Med.Genet. 93: 421–425, 2000.
- [45215] 18767.Thurmon, T. F.; De Fraites, E. B.; Anderson, E. E.:
Familialcampomelic dwarfism. J. Pediat. 83: 841–843,
1973.
- [45216] 18768.Vidal, V. P. I.; Chaboissier, M.–C.; de Rooij, D. G.;
Schedl,A.: Sox9 induces testis development in XX transgenic mice. NatureGenet. 28: 216–217, 2001.
- [45217] 18769.Wagner, T.; Wirth, J.; Meyer, J.; Zabel, B.; Held, M.;
Zimmer,J.; Pasantes, J.; Dagna Bricarelli, F.; Keutel, J.; Huser, E.; Wolf,U.; Tommerup, N.; Schempp, W.; Scherer, G.:

Autosomal sex reversal and campomelic dysplasia are caused by mutations in and around the SRY-related gene SOX9. *Cell* 79: 1111–1120, 1994.

[45218] 18770. Weller, S. D. V.: Hypophosphatasia with congenital dimples. *Proc. Roy. Soc. Med.* 52: 637, 1959.

[45219] 18771. Wirth, J.; Wagner, T.; Meyer, J.; Pfeiffer, R. A.; Tietze, H.-U.; Schempp, W.; Scherer, G.: Translocation breakpoints in three patients with campomelic dysplasia and autosomal sex reversal map more than 130 kb from SOX9. *Hum. Genet.* 97: 186–193, 1996.

[45220] 18772. Wright, E.; Hargrave, M. R.; Christiansen, J.; Cooper, L.; Kun, J.; Evans, T.; Gangadharan, U.; Greenfield, A.; Koopman, P.: The Sry-related gene Sox9 is expressed during chondrogenesis in mouse embryos. *Nature Genet.* 9: 15–20, 1995.

[45221] 18773. Albritton, L. M.; Bowcock, A. M.; Eddy, R. L.; Morton, C. C.; Tseng, L.; Farrer, L. A.; Cavalli-Sforza, L. L.; Shows, T. B.; Cunningham, J. M.: The human cationic amino acid transporter (ATRC1): physical and genetic mapping to 13q12–q14. *Genomics* 12: 430–434, 1992.

[45222] 18774. Bowcock, A. M.; Gerken, S. C.; Barnes, R. I.; Shiang, R.; Jabs, E. W.; Warren, A. C.; Antonarakis, S.; Retief, A. E.; Vergnaud, G.; Leppert, M.; Lalouel, J.-M.; White, R. L.; Cav-

alli-Sforza, L. L.:The CEPH consortium linkage map of human chromosome 13. *Genomics* 16:486–496, 1993.

[45223] 18775.Kim, J. W.; Closs, E. I.; Albritton, L. M.; Cunningham, J. M.:Transport of cationic amino acids by the mouse ecotropic retrovirusreceptor. *Nature* 352: 725–728, 1991.

[45224] 18776.Kozak, C. A.; Albritton, L. M.; Cunningham, J. M.: Genetic mapping of a cloned sequence responsible for susceptibility to ecotropic murine leukemia viruses. *J. Virol.* 64: 3119–3121, 1990.

[45225] 18777.Oie, H. K.; Gazdar, A. F.; Lalley, P. A.; Russell, E. K.; Minna, J. D.; DeLarco, J.; Todaro, G. J.; Francke, U.: Mouse chromosome 5 codes for ecotropic murine leukaemia virus cell–surface receptor. *Nature* 274:60–62, 1978.

[45226] 18778.Ruddle, N. H.; Conta, B. S.; Leinwand, L.; Kozak, C.; Ruddle, F.; Besmer, P.; Baltimore, D.: Assignment of the receptor for ecotropic murine leukemia virus to mouse chromosome 5. *J. Exp. Med.* 148: 451–465, 1978.

[45227] 18779.Boldog, F.; Erlandsson, R.; Klein, G.; Sumegi, J.: Long–range restriction enzyme maps of DNF15S2, D3S2 and c–raf1 loci on the short arm of human chromosome 3. *Cancer Genet. Cytogenet.* 42: 295–306, 1989.

[45228] 18780.Cook, R. M.; Burke, B. J.; Buchhagen, D. L.; Minna, J. D.; Miller, Y. E.: Human aminoacylase–1: cloning, se–

quence, and expression analysis of a chromosome 3p21 gene inactivated in small cell lung cancer. *J. Biol. Chem.* 268: 17010–17017, 1993.

[45229] 18781. Gemmill, R. M.; Varella-Garcia, M.; Smith, D. I.; Erickson, P.; Golembieski, W.; Miller, Y.; Coyle-Morris, J.; Tommerup, N.; Drabkin, H. A.: A 2.5 Mb physical map within 3p21.1 spans the breakpoint associated with Greig cephalopolysyndactyly syndrome. *Genomics* 11: 93–102, 1991.

[45230] 18782. Ginzinger, D. G.; Shridhar, V.; Baldini, A.; Taggart, R. T.; Miller, O. J.; Smith, D. I.: The human loci DNF15S2 and D3S94 have a high degree of sequence similarity to acyl-peptide hydrolase and are located at 3p21.3. *Am. J. Hum. Genet.* 50: 826–833, 1992.

[45231] 18783. Jones, W. M.; Scaloni, A.; Bossa, F.; Popowicz, A. M.; Schneewind, O.; Manning, J. M.: Genetic relationship between acylpeptide hydrolase and acylase, two hydrolytic enzymes with similar binding but different catalytic specificities. *Proc. Nat. Acad. Sci.* 88: 2194–2198, 1991.

[45232] 18784. Miller, Y. E.; Drabkin, H.; Jones, C.; Fisher, J. H.: Aminoacylase-1: cDNA isolation, regional assignment to chromosome 3p21.1 and identification of a cross-hybridizing sequence on chromosome 18. (Abstract) *Am. J.*

Hum. Genet. 45 (suppl.): A28 only, 1989.

- [45233] 18785. Miller, Y. E.; Drabkin, H.; Jones, C.; Fisher, J. H.: Human aminoacylase-1: cloning, regional assignment to distal chromosome 3p21.1, and identification of a cross-hybridizing sequence on chromosome 18. Genomics 8: 149-154, 1990.
- [45234] 18786. Miller, Y. E.; Minna, J. D.; Gazdar, A. F.: Lack of expression of aminoacylase-1 in small cell lung cancer: evidence for inactivation of genes encoded by chromosome 3p. J. Clin. Invest. 83: 2120-2124, 1989.
- [45235] 18787. Nadeau, J. H.: A chromosomal segment conserved since divergence of lineages leading to man and mouse: the gene order of aminoacylase-1, transferrin, and beta-galactosidase on mouse chromosome 9. Genet. Res. 48: 175-178, 1986.
- [45236] 18788. Naylor, S. L.; Elliott, R. W.; Brown, J. A.; Shows, T. B.: Mapping of aminoacylase-1 and beta-galactosidase-A to homologous regions of human chromosome 3 and mouse chromosome 9 suggests location of additional genes. Am. J. Hum. Genet. 34: 235-244, 1982.
- [45237] 18789. Naylor, S. L.; Shows, T. B.; Klebe, R. J.: Bioautographic visualization of aminoacylase-1: assignment of the structural gene ACY-1 to chromosome 3 in man. Somat.

Cell Genet. 5: 11–21, 1979.

- [45238] 18790.Voss, R.; Lerer, I.; Povey, S.; Solomon, E.; Bobrow, M.: Confirmationand further regional assignment of aminoacylase 1 (ACY–1) on humanchromosome 3 using a simplified detection method. Ann. Hum. Genet. 44:1–10, 1980.
- [45239] 18791.Cook, P. W.; Piepkorn, M.; Clegg, C. H.; Plowman, G. D.; DeMay,J. M.; Brown, J. R.; Pittelkow, M. R.: Trans–genic expression of thehuman amphiregulin gene induces a psoriasis–like phenotype. J. Clin.Invest. 100: 2286–2294, 1997.
- [45240] 18792.Disteche, C. M.; Plowman, G. D.; Gronwald, R. G. K.; Kelly, J.;Bowen–Pope, D.; Adler, D. A.; Murray, J. C.: Mapping of the amphiregulinand the platelet–growth factor receptor alpha genes to the proximallong arm of chromosome 4. (Abstract) Cytogenet. Cell Genet. 51:990 only, 1989.
- [45241] 18793.Kimura, H.; Fischer, W. H.; Schubert, D.: Structure, expressionand function of a schwannoma–derived growth factor. Nature 348:257–260, 1990.
- [45242] 18794.Gillessen–Kaesbach, G.; Demuth, S.; Thiele, H.; Theile, U.; Lich,C.; Horsthemke, B.: A previously unrecognised phenotype characterisedby obesity, muscular hypo–

tonia, and ability to speak in patients with Angelman syndrome caused by an imprinting defect. *Europ. J. Hum. Genet.* 7: 638–644, 1999.

- [45243] 18795. Cockayne, D. A.; Muchamuel, T.; Grimaldi, J. C.; Muller-Steffner, H.; Randall, T. D.; Lund, F. E.; Murray, R.; Schuber, F.; Howard, M. C.: Mice deficient for the ecto-nicotinamide adenine dinucleotide glycohydrolase CD38 exhibit altered humoral immune responses. *Blood* 92:1324–1333, 1998.
- [45244] 18796. Ferrero, E.; Malavasi, F.: Human CD38, a leukocyte receptor and ectoenzyme, is a member of a novel eukaryotic gene family of nicotinamide adenine dinucleotide +/–converting enzymes: extensive structure homology with the genes for murine bone marrow stromal cell antigen 1 and aplysian ADP-ribosyl cyclase. *J. Immun.* 159: 3858–3865, 1997.
- [45245] 18797. Ferrero, E.; Saccucci, F.; Malavasi, F.: The human CD38 gene: polymorphism, CpG island, and linkage to the CD157 (BST-1) gene. *Immunogenetics* 49:597–604, 1999.
- [45246] 18798. Fukushi, Y.; Kato, I.; Takasawa, S.; Sasaki, T.; Ong, B. H.; Sato, M.; Ohsaga, A.; Sato, K.; Shirato, K.; Okamoto, H.; Maruyama, Y.: Identification of cyclic ADP-ribose-dependent mechanisms in pancreatic muscarinic

Ca(2+) signaling using CD38 knockout mice. J. Biol. Chem. 276:649–655, 2001.

- [45247] 18799.Jackson, D. G.; Bell, J. I.: Isolation of a cDNA encoding the human CD38 (T10) molecule, a cell surface glycoprotein with an unusual discontinuous pattern of expression during lymphocyte differentiation. J.Immun. 144: 2811–2815, 1990.
- [45248] 18800.Kaisho, T.; Ishikawa, J.; Oritani, K.; Inazawa, J.; Tomizawa, H.;Muraoka, O.; Ochi, T.; Hirano, T.: BST–1, a surface molecule of bonemarrow stromal cell lines that facilitates pre–B–cell growth. Proc.Nat. Acad. Sci. 91: 5325–5329, 1994.
- [45249] 18801.Nakagawara, K.; Mori, M.; Takasawa, S.; Nata, K.; Takamura, T.;Berlova, A.; Tohgo, A.; Karasawa, T.; Yonekura, H.; Takeuchi, T.;Okamoto, H.: Assignment of CD38, the gene encoding human leukocyte antigen CD38 (ADP–ribosyl cyclase/cyclic ADP–ribose hydrolase), to chromosome 4p15. Cytogenet. Cell Genet. 69: 38–39, 1995.
- [45250] 18802.Takasawa, S.; Nata, K.; Yonekura, H.; Okamoto, H.: Cyclic ADP–ribose in insulin secretion from pancreatic beta cells. Science 259: 370–373,1993.
- [45251] 18803.Takasawa, S.; Tohgo, A.; Noguchi, N.; Koguma, T.;

Nata, K.; Sugimoto, T.; Yonekura, H.; Okamoto, H.: Synthesis and hydrolysis of cyclic ADP-ribose by human leukocyte antigen CD38 and inhibition of the hydrolysis by ATP. *J. Biol. Chem.* 268: 26052–26054, 1993.

[45252] 18804. Acosta, J.; Hettinga, J.; Fluckiger, R.; Krumrei, N.; Goldfine, A.; Angarita, L.; Halperin, J.: Molecular basis for a link between complement and the vascular complications of diabetes. *Proc. Nat. Acad. Sci.* 97: 5450–5455, 2000.

[45253] 18805. Bickmore, W. A.; Longbottom, D.; Oghene, K.; Fletcher, J. M.; van Heyningen, V.: Colocalization of the human CD59 gene to 11p13 with the MIC11 cell surface antigen. *Genomics* 17: 129–135, 1993.

[45254] 18806. Davies, A.; Simmons, D. L.; Hale, G.; Harrison, R. A.; Tighe, H.; Lachmann, P. J.; Waldmann, H.: CD59, an LY-6-like protein expressed in human lymphoid cells, regulates the action of the complement membrane attack complex on homologous cells. *J. Exp. Med.* 170: 637–654, 1989.

[45255] 18807. Forsberg, U. H.; Bazil, V.; Stefanova, I.; Schroder, J.: Gene for human CD59 (likely Ly-6 homologue) is located on the short arm of chromosome 11. *Immunogenetics* 30: 188–193, 1989.

[45256] 18808. Harada, R.; Okada, N.; Fujita, T.; Okada, H.: Purifi-

cation of 1F5 antigen that prevents complement attack on homologous cell membranes. *J. Immun.* 144: 1823–1828, 1990.

- [45257] 18809. Heckl–Ostreicher, B.; Ragg, S.; Drechsler, M.; Scherthan, H.; Royer–Pokora, B.: Localization of the human CD59 gene by fluorescence in situ hybridization and pulsed–field gel electrophoresis. *Cytogenet. Cell Genet.* 63:144–146, 1993.
- [45258] 18810. Holt, D. S.; Botto, M.; Bygrave, A. E.; Hanna, S. M.; Walport, M. J.; Morgan, B. P.: Targeted deletion of the CD59 gene causes spontaneous intravascular hemolysis and hemoglobinuria. *Blood* 98: 442–449, 2001.
- [45259] 18811. Holt, D. S.; Powell, M. B.; Rushmere, N. K.; Morgan, B. P.: Genomic structure and chromosome location of the gene encoding mouse CD59. *Cytogenet. Cell Genet.* 89: 264–267, 2000.
- [45260] 18812. Huppi, K.; Duncan, R.; Potter, M.: Myc–1 is centromeric to the linkage group Ly–6–Sis–Gdc–1 on mouse chromosome 15. *Immunogenetics* 27:215–219, 1988.
- [45261] 18813. Kamiura, S.; Nolan, C. M.; Meruelo, D.: Long–range physical map of the Ly–6 complex: mapping the Ly–6 multigene family by field–inversion and two–dimensional gel electrophoresis. *Genomics* 12: 89–105, 1992.

- [45262] 18814.Low, M. G.; Saltiel, A. R.: Structural and functional roles of glycosyl-phosphatidylinositol in membranes. *Science* 239: 268–275, 1988.
- [45263] 18815.Mahoney, J. F.; Urakaze, M.; Hall, S.; DeGasperi, R.; Chang, H.-M.; Sugiyama, E.; Warren, C. D.; Borowitz, M.; Nicholson-Weller, A.; Rosse, W. F.; Yeh, E. T. H.: Defective glycosylphosphatidylinositol anchor synthesis in paroxysmal nocturnal hemoglobinuria granulocytes. *Blood* 79:1400–1403, 1992.
- [45264] 18816.Mao, M.; Yu, M.; Tong, J.-H.; Ye, J.; Zhu, J.; Huang, Q.-H.; Fu, G.; Yu, L.; Zhao, S.-Y.; Waxman, S.; Lanotte, M.; Wang, Z.-Y.; Tan, J.-Z.; Chan, S.-J.; Chen, Z.: RIG-E, a human homolog of the murine Ly-6 family, is induced by retinoic acid during the differentiation of acute promyelocytic leukemia cell. *Proc. Nat. Acad. Sci.* 93:5910–5914, 1996.
- [45265] 18817.Meri, S.; Morgan, B. P.; Davies, A.; Daniels, R. H.; Olavesen, M. G.; Waldmann, H.; Lachmann, P. J.: Human protectin (CD59), an 18,000–20,000 MW complement lysis restricting factor, inhibits C5b-8 catalysed insertion of C9 into lipid bilayers. *Immunology* 71: 1–9, 1990.
- [45266] 18818.Meri, S.; Morgan, B. P.; Wing, M.; Jones, J.; Davies, A.; Podack, E.; Lachmann, P. J.: Human protectin (CD59),

an 18–20–kD homologous complement restriction factor, does not restrict perforin–mediated lysis. *J. Exp. Med.* 172: 367–370, 1990.

[45267] 18819. Motoyama, N.; Okada, N.; Yamashina, M.; Okada, H.: Paroxysmal nocturnal hemoglobinuria due to hereditary nucleotide deletion in the HRF20 (CD59) gene. *Europ. J. Immun.* 22: 2669–2673, 1992.

[45268] 18820. Okada, N.; Harada, R.; Fujiita, T.; Okada, H.: A novel membrane glycoprotein capable of inhibiting membrane attack by homologous complement. *Int. Immun.* 1: 205–208, 1989.

[45269] 18821. Anderson, L.; Anderson, N. G.: High resolution two–dimensional electrophoresis of human plasma proteins. *Proc. Nat. Acad. Sci.* 12: 5421–5425, 1977.

[45270] 18822. Cohen, R. D.; Castellani, L. W.; Qiao, J.–H.; Van Lenten, B. J.; Lusis, A. J.; Reue, K.: Reduced aortic lesions and elevated high density lipoprotein levels in transgenic mice overexpressing mouse apolipoprotein A–IV. *J. Clin. Invest.* 99: 1906–1916, 1997.

[45271] 18823. Duverger, N.; Tresp, G.; Caillaud, J.–M.; Emmanuel, F.; Castro, G.; Fruchart, J.–C.; Steinmetz, A.; Deneffe, P.: Protection against atherogenesis in mice mediated by human apolipoprotein A–IV. *Science* 273: 966–968,

1996.

- [45272] 18824.Szpirer, C.; Riviere, M.; Cortese, R.; Nakamura, T.; Islam, M.Q.; Levan, G.; Szpirer, J.: Chromosomal localization in man and rat of the genes encoding the liver-enriched transcription factors C/EBP, DBP, and HNF1/LFB-1 (CEBP, DBP, and transcription factor 1, TCF1, respectively) and of the hepatocyte growth factor/scatter factor gene(HGF). *Genomics* 13: 293–300, 1992.
- [45273] 18825.Boorstein, W. R.; Vamvakopoulos, N. C.; Fiddes, J. C.: Human chorionic gonadotropin beta-subunit is encoded by at least eight genes arranged in tandem and inverted pairs. *Nature* 300: 419–422, 1982.
- [45274] 18826.Fiddes, J. C.; Goodman, H. M.: The cDNA for the beta-subunit of human chorionic gonadotropin suggests evolution of a gene by readthrough into the 3-prime-untranslated region. *Nature* 286: 684–687, 1980.
- [45275] 18827.Graham, M. Y.; Otani, T.; Boime, I.; Olson, M. V.; Carle, G. F.; Chaplin, D. D.: Cosmid mapping of the human chorionic gonadotropin beta subunit genes by field-inversion gel electrophoresis. *Nucleic Acids Res.* 15: 4437–4448, 1987.
- [45276] 18828.Julier, C.; Weil, D.; Couillin, P.; Cote, J. C.; Boue, A.;

Thirion, J. P.; Kaplan, J. C.; Junien, C.: Confirmation of the assignment of the genes coding for human chorionic gonadotropin beta subunit to chromosome 19. (Abstract) Cytogenet. Cell Genet. 37: 501–502, 1984.

[45277] 18829. Gersten, K. M.; Natsuka, S.; Trinchera, M.; Petryniak, B.; Kelly, R. J.; Hiraiwa, N.; Jenkins, N. A.; Gilbert, D. J.; Copeland, N. G.; Lowe, J. B.: Molecular cloning, expression, chromosomal assignment, and tissue-specific expression of a murine alpha-(1,3)-fucosyltransferase locus corresponding to the human ELAM-1 ligand fucosyl transferase. J. Biol. Chem. 270: 25047–25056, 1995.

[45278] 18830. Julier, C.; Weil, D.; Couillin, P.; Cote, J. C.; Van Cong, N.; Foubert, C.; Boue, A.; Thirion, J. P.; Kaplan, J. C.; Junien, C.: The beta chorionic gonadotropin-beta luteinizing gene cluster maps to human chromosome 19. Hum. Genet. 67: 174–177, 1984.

[45279] 18831. Lunardi-Iskander, Y.; Bryant, J. L.; Zeman, R. A.; Lam, V. H.; Samaniego, F.; Besnier, J. M.; Hermans, P.; Thierry, A. R.; Gill, P.; Gallo, R. C.: Tumorigenesis and metastasis of neoplastic Kaposi's sarcoma cell line in immunodeficient mice blocked by a human pregnancy hormone. Nature 375: 64–68, 1995.

[45280] 18832. Policastro, P.; Ovitt, C. E.; Hoshina, M.; Fukuoka,

H.; Boothby, M. R.; Bieme, I.: The beta-subunit of human chorionic gonadotropin is encoded by multiple genes. *J. Biol. Chem.* 258: 11492–11499, 1983.

[45281] 18833. Policastro, P. F.; Daniels–McQueen, S.; Carle, G.; Boime, I.: A map of the hCG-beta-LH-beta gene cluster. *J. Biol. Chem.* 261: 5907–5916, 1986.

[45282] 18834. Talmadge, K.; Vamvakopoulos, N. C.; Fiddes, J. C.: Evolution of the genes for the beta subunits of human chorionic gonadotropin and luteinizing hormone. *Nature* 307: 37–40, 1984.

[45283] 18835. Warburton, D.; Gersen, S.; Yu, M.–T.; Jackson, C.; Handelin, B.; Housman, D.: Monochromosomal rodent–human hybrids from microcell fusion of human lymphoblastoid cells containing an inserted dominant selectable marker. *Genomics* 6: 358–366, 1990.

[45284] 18836. Reseland, J. E.; Larsen, F.; Solheim, J.; Eriksen, J. A.; Hanssen, L. E.; Prydz, H.: A novel human chymotrypsin-like digestive enzyme. *J. Biol. Chem.* 272: 8099–8104, 1997.

[45285] 18837. Angeletti, R. H.: Chromogranins and neuroendocrine secretion. (Editorial) *Lab. Invest.* 55: 387–390, 1986.

[45286] 18838. Bhargava, G.; Russell, J.; Sherwood, L. M.: Phosphorylation of parathyroid secretory protein. *Proc. Nat.*

Acad. Sci. 80: 878–881,1983.

- [45287] 18839.Cetin, Y.; Aunis, D.; Bader, M.–F.; Galindo, E.; Jorns, A.; Bargsten,G.; Grube, D.: Chromostatin, a chromogranin A–derived bioactive peptide,is present in human pancre–atic insulin (beta) cells. Proc. Nat. Acad.Sci. 90: 2360–2364, 1993.
- [45288] 18840.Lu, Q.; Lemke, G.: Homeostatic regulation of the immune systemby receptor tyrosine kinases of the Tyro 3 family. Science 293:306–311, 2001.
- [45289] 18841.Abrahamson, M.: Human cysteine proteinase in–hibitors: isolation,physiological importance, inhibitory mechanism, gene structure andrelation to hereditary cere–bral hemorrhage. Scand. J. Clin. Lab.Invest. 48 (suppl. 191): 21–31, 1988.
- [45290] 18842.Abrahamson, M.; Grubb, A.: Increased body tem–perature acceleratesaggregation of the leu68–to–gln mu–tant cystatin C, the amyloid–formingprotein in hereditary cystatin C amyloid angiopathy. Proc. Nat. Acad.Sci. 91: 1416–1420, 1994.
- [45291] 18843.Abrahamson, M.; Grubb, A.; Olafsson, I.; Lundwall, A.: Molecularcloning and sequence analysis of cDNA cod–ing for the precursor ofthe human cysteine proteinase in–hibitor cystatin C. FEBS Lett. 216:229–233, 1987.

- [45292] 18844.Abrahamson, M.; Islam, M. Q.; Szpirer, J.; Szpirer, C.; Levan,G.: The human cystatin C gene (CST3), mutated in hereditary cystatinC amyloid angiopathy, is located on chromosome 20. Hum. Genet. 82:223–226, 1989.
- [45293] 18845.Abrahamson, M.; Jonsdottir, S.; Olafsson, I.; Jensson, O.; Grubb,A.: Hereditary cystatin C amyloid angiopathy: identification of the disease–causing mutation and specific diagnosis by polymerase chain reaction based analysis. Hum. Genet. 89: 377–380, 1992.
- [45294] 18846.Cohen, D. H.; Feiner, H.; Jensson, O.; Frangione, B.: Amyloidfibril in hereditary cerebral hemorrhage with amyloidosis (HCHWA)is related to the gastroentero–pancreatic neuroendocrine protein,gamma trace. J. Exp. Med. 158: 623–628, 1983.
- [45295] 18847.Ghiso, J.; Jensson, O.; Frangione, B.: Amyloid fibrils in hereditarycerebral hemorrhage with amyloidosis of Icelandic type is a variantof gamma–trace basic protein (cystatin C). Proc. Nat. Acad. Sci. 83:2974–2978, 1986.
- [45296] 18848.Ghiso, J.; Pons–Estel, B.; Frangione, B.: Hereditary cerebralamyloid angiopathy: the amyloid fibrils contain a protein which isa variant of cystatin C, an inhibitor of lysosomal cysteine proteases. Biochem.Biophys. Res. Commun. 136: 548–554, 1986.

- [45297] 18849.Grubb, A.; Jensson, O.; Gudmundsson, G.; Arnason, A.; Lofberg,H.; Malm, J.: Abnormal metabolism of gamma-trace alkaline microprotein:the basic defect in hereditary cerebral hemorrhage with amyloidosis. *NewEng. J. Med.* 311: 1547–1549, 1984.
- [45298] 18850.Galbraith, G. M. P.; Pandey, J. P.: Tumor necrosis factor alpha(TNF-alpha) gene polymorphism in alopecia areata. *Hum. Genet.* 96:433–436, 1995.
- [45299] 18851.Tazi-Ahnini, R.; di Giovine, F. S.; McDonagh, A. J. G.; Messenger,A. G.; Amadou, C.; Cox, A.; Duff, G. W.; Cork, M. J.: Structure andpolymorphism of the human gene for the interferon-induced p78 protein(MX1): evidence of association with alopecia areata in the Down syndromeregion. *Hum. Genet.* 106: 639–645, 2000.
- [45300] 18852.Webb, D. J.; Wen, J.; Lysiak, J. J.; Umans, L.; Van Leuven, F.;Gonias, S. L.: Murine alpha-macroglobulins demonstrate divergentactivities as neutralizers of transforming growth factor-beta andas inducers of nitric oxide synthesis: a possible mechanism for theendotoxin insensitivity of the alpha-2-macroglobulin gene knock-outmouse. *J. Biol. Chem.* 271: 24982–24988, 1996.
- [45301] 18853.Wilson, A. G.; di Giovine, F. S.; Blakemore, A. I. F.; Duff, G.W.: Single base polymorphism in the human tu-

mour necrosis factor alpha (TNF-alpha) gene detectable by NcoI restriction of PCR product. Hum.Molec. Genet. 1: 353 only, 1992.

[45302] 18854. Belanger, L.; Roy, S.; Allard, D.: New albumin gene 3-prime adjacent to the alpha-1-fetoprotein locus. J. Biol. Chem. 269: 5481-5484, 1994.

[45303] 18855. Lichenstein, H. S.; Lyons, D. E.; Wurfel, M. M.; Johnson, D. A.; McGinley, M. D.; Leidli, J. C.; Trollinger, D. B.; Mayer, J. P.; Wright, S. D.; Zukowski, M. M.: Afamin is a new member of the albumin, alpha-fetoprotein, and vitamin D-binding protein gene family. J. Biol. Chem. 269: 18149-18154, 1994.

[45304] 18856. Nishio, H.; Dugaiczyk, A.: Complete structure of the human alpha-albumin gene, a new member of the serum albumin multigene family. Proc. Nat. Acad. Sci. 93: 7557-7561, 1996.

[45305] 18857. Song, Y.-H.; Naumova, A. K.; Liebhaber, S. A.; Cooke, N. E.: Physical and meiotic mapping of the region of human chromosome 4q11-q13 encompassing the vitamin D binding protein DBP/Gc-globulin and albumin multigene cluster. Genome Res. 9: 581-587, 1999.

[45306] 18858. Beattie, W. G.; Dugaiczyk, A.: Structure and evolution of human alpha-fetoprotein deduced from partial se-

quence of cloned cDNA. *Gene* 20:415–422, 1982.

[45307] 18859.Belayew, A.; Tilghman, S. M.: Genetic analysis of alpha-fetoproteinsynthesis in mice. *Molec. Cell. Biol.* 2: 1427–1435, 1982.

[45308] 18860.D'Eustachio, P.; Ingram, R. S.; Tilghman, S. M.; Ruddle, F. H.: Murine alpha-fetoprotein and albumin: two evolutionarily linkedproteins encoded on the same mouse chromosome. *Somat. Cell Genet.* 7:289–294, 1981.

[45309] 18861.Eiferman, F. A.; Young, P. R.; Scott, R. W.; Tilghman, S. M.:Intragenic amplification and divergence in the mouse alpha-fetoproteingene. *Nature* 294: 713–718, 1981.

[45310] 18862.Faucett, W. A.; Greenberg, F.; Rose, E.; Alpert, E.; Bancalari,L.; Kardon, N. B.; Mizjewski, G.; Knight, G.; Haddow, J. E.: Congenitaldeficiency of alpha-fetoprotein. (Abstract) *Am. J. Hum. Genet.* 45(suppl.): A259, 1989.

[45311] 18863.Ferguson-Smith, M. A.; May, H. M.; Aitken, D. A.; O'Hare, E.; Yates,J. R. W.; Gallagher, J.; Krumlauf, R.; Tilghman, S. M.: Hereditarypersistence of alphafetoprotein (HPAFP); linkage studies with chromosome4 markers. (Abstract) *Cytogenet. Cell Genet.* 37: 469, 1984.

[45312] 18864.Ferguson-Smith, M. A.; Yates, J. R. W.; Kelly, D.; Aitken, D. A.;May, H. M.; Krumlauf, R.; Tilghman, S. M.:

Hereditary persistence of alpha-fetoprotein maps to the long arm of chromosome 4. (Abstract) *Cytogenet. Cell Genet.* 40: 628, 1985.

[45313] 18865. Gabant, P.; Forrester, L.; Nichols, J.; Van Reeth, T.; De Mees, C.; Pajack, B.; Watt, A.; Smits, J.; Alexandre, H.; Szpirer, C.; Szpirer, J.: Alpha-fetoprotein, the major fetal serum protein, is not essential for embryonic development but is required for female fertility. *Proc. Nat. Acad. Sci.* 99: 12865–12870, 2002.

[45314] 18866. Gibbs, P. E. M.; Zielinski, R.; Boyd, C.; Dugaiczyk, A.: Structure, polymorphism, and novel repeated DNA elements revealed by a complete sequence of the human alpha-fetoprotein gene. *Biochemistry* 26: 1332–1343, 1987.

[45315] 18867. Seri, M.; Celli, I.; Betsos, N.; Claudiani, F.; Camera, G.; Romeo, G.: A cys634gly substitution of the RET proto-oncogene in a family with recurrence of multiple endocrine neoplasia type 2A and cutaneous lichen amyloidosis. *Clin. Genet.* 51: 86–90, 1997.

[45316] 18868. Kamal, A.; Almenar-Queralt, A.; LeBlanc, J. F.; Roberts, E. A.; Goldstein, L. S. B.: Kinesin-mediated axonal transport of a membrane compartment containing beta-secretase and presenilin-1 requires APP. *Nature* 414: 643–648, 2001.

- [45317] 18869.Kang, D. E.; Soriano, S.; Xia, X.; Eberhart, C. G.; De Strooper,B.; Zheng, H.; Koo, E. H.: Presenilin couples the paired phosphorylationof beta-catenin independent of Axin: implications for beta-cateninactivation in tumorigenesis. *Cell* 110: 751–762, 2002.
- [45318] 18870.Hofstra, R. M. W.; Sijmons, R. H.; Stelwagen, T.; Stulp, R. P.;Kousseff, B. G.; Lips, C. J. M.; Steijlen, P. M.; Van Voorst Vader,P. C.; Buys, C. H. C. M.: RET mutation screening in familial cutaneouslichen amyloidosis and in skin amyloidosis associated with multipleendocrine neoplasia. *J. Invest. Derm.* 107: 215–218, 1996.
- [45319] 18871.Bordet, T.; Lesbordes, J.–C.; Rouhani, S.; Castelnau–Ptakhine,L.; Schmalbruch, H.; Haase, G.; Kahn, A.: Protective effects of cardiotrophin–1adenoviral gene transfer on neuromuscular degeneration in transgenicALS mice. *Hum. Molec. Genet.* 10: 1925–1933, 2001.
- [45320] 18872.Fantes, J. A.; Bickmore, W. A.; Fletcher, J. M.; Ballesta, F.;Hanson, I. M.; van Heyningen, V.: Submicroscopic deletions at theWAGR locus, revealed by nonradioactive in situ hybridization. *Am.J. Hum. Genet.* 51: 1286–1294, 1992.
- [45321] 18873.Glaser, T.; Jepeal, L.; Edwards, J. G.; Young, S. R.; Favor, J.;Maas, R. L.: PAX6 gene dosage effect in a family

with congenital cataracts, aniridia, anophthalmia and central nervous system defects. *Nature Genet.* 7: 463–471, 1994.

- [45322] 18874. Glaser, T.; Lane, J.; Housman, D.: A mouse model of the aniridia–Wilmstumor deletion syndrome. *Science* 250: 823–827, 1990.
- [45323] 18875. Gronskov, K.; Olsen, J. H.; Sand, A.; Pedersen, W.; Carlsen, N.; Jylling, A. M. B.; Lyngbye, T.; Brondum–Nielsen, K.; Rosenberg, T.: Population–based risk estimates of Wilms tumor in sporadic aniridia: a comprehensive mutation screening procedure of PAX6 identifies 80% of mutations in aniridia. *Hum. Genet.* 109: 11–18, 2001.
- [45324] 18876. Hanson, I. M.; Seawright, A.; Hardman, K.; Hodgson, S.; Zaletayev, D.; Fekete, G.; van Heyningen, V.: PAX6 mutations in aniridia. *Hum. Molec. Genet.* 2: 915–920, 1993.
- [45325] 18877. Hill, R. E.; Favor, J.; Hogan, B. L. M.; Ton, C. C. T.; Saunders, G. F.; Hanson, I. M.; Prosser, J.; Jordan, T.; Hastie, N. D.; van Heyningen, V.: Mouse small eye results from mutations in a paired–like homeobox–containing gene. *Nature* 354: 522–525, 1991.
- [45326] 18878. Jordan, T.; Hanson, I.; Zaletayev, D.; Hodgson, S.;

Prosser, J.;Seawright, A.; Hastie, N.; van Heyningen, V.:
The human PAX6 gene is mutated in two patients with
aniridia. Nature Genet. 1: 328–332,1992.

[45327] 18879.Karpen, G. H.: Position effect variegation and the
new biology of heterochromatin. Curr. Opin. Genet. Dev. 4:
281–291, 1994.

[45328] 18880.Lyon, M. F.: Personal Communication. Harwell,
England 6/9/1988.

[45329] 18881.Martha, A.; Strong, L. C.; Ferrell, R. E.; Saunders, G.
F.: Three novel aniridia mutations in the human PAX6
gene. Hum. Mutat. 6:44–49, 1995.

[45330] 18882.Matsuo, T.; Osumi–Yamashita, N.; Noji, S.; Ohuchi,
H.; Koyama,E.; Myokai, F.; Matsuo, N.; Taniguchi, S.; Doi,
H.; Iseki, S.; Ninomiya,Y.; Fujiwara, M.; Watanabe, T.; Eto,
K.: A mutation in the Pax–6 gene in rat small eye is associ-
ated with impaired migration of midbrain crest cells. Na-
ture Genet. 3: 299–304, 1993.

[45331] 18883.Oliver, M. D.; Dotan, S. A.; Chemke, J.; Abraham, F.
A.: Isolated foveal hypoplasia. Brit. J. Ophthal. 71:
926–930, 1987.

[45332] 18884.Prosser, J.; van Heyningen, V.: PAX6 mutations re-
viewed. Hum.Mutat. 11: 93–108, 1998.

[45333] 18885.Quiring, R.; Walldorf, U.; Kloter, U.; Gehring, W. J.:

Homology of the eyeless gene of *Drosophila* to the small eye gene in mice and aniridia in humans. *Science* 265: 785–789, 1994.

- [45334] 18886. Salvini-Plawen, L.; Mayr, E.: On the evolution of photoreceptors and eyes. In: Hecht, M. K.; Steere, W.; Wallace, B.: *Evolutionary Biology*. New York: Plenum Pub. (pub.) 10: 1977. Pp. 207–263.
- [45335] 18887. Schedl, A.; Ross, A.; Lee, M.; Engelkamp, D.; Rashbass, P.; van Heyningen, V.; Hastie, N. D.: Influence of PAX6 gene dosage on development: overexpression causes severe eye abnormalities. *Cell* 86: 71–82, 1996.
- [45336] 18888. Stone, D. L.; Kenyon, K. R.; Green, W. R.; Ryan, S. J.: Congenital central corneal leukoma (Peters' anomaly). *Am. J. Ophthalmol.* 81: 173–193, 1976.
- [45337] 18889. van der Meer-de Jong, R.; Dickinson, M. E.; Woychik, R. P.; Stubbs, L.; Hetherington, C.; Hogan, B. L. M.: Location of the gene involving the small eye mutation on mouse chromosome 2 suggests homology with human aniridia 2 (AN2). *Genomics* 7: 270–275, 1990.
- [45338] 18890. Newport, M. J.; Huxley, C. M.; Huston, S.; Hawrylowicz, C. M.; Oostra, B. A.; Williamson, R.; Levin, M.: A mutation in the interferon- γ -receptor gene and susceptibility to mycobacterial infection. *New Eng. J. Med.*

335:1941–1949, 1996.

- [45339] 18891.Cowan, C. A.; Yokoyama, N.; Bianchi, L. M.; Henkemeyer, M.; Fritzsche, B.: EphB2 guides axons at the midline and is necessary for normal vestibular function. *Neuron* 26: 417–430, 2000.
- [45340] 18892.Ton, C. C. T.; Hirvonen, H.; Miwa, H.; Weil, M. M.; Monaghan, P.; Jordan, T.; van Heyningen, V.; Hastie, N. D.; Meijers-Heijboer, H.; Drechsler, M.; Royer-Pokora, B.; Collins, F.; Swaroop, A.; Strong, L. C.; Saunders, G. F.: Positional cloning and characterization of a paired box- and homeobox-containing gene from the aniridia region. *Cell* 67:1059–1074, 1991.
- [45341] 18893.Novick, D.; Orchansky, P.; Revel, M.; Rubinstein, M.: The human interferon-gamma receptor: purification, characterization, and preparation of antibodies. *J. Biol. Chem.* 262: 8483–8487, 1987.
- [45342] 18894.Orchansky, P.; Rubinstein, M.; Fischer, D. G.: The interferon-gamma receptor in human monocytes is different from the one in nonhematopoietic cells. *J. Immun.* 136: 169–173, 1986.
- [45343] 18895.Papanicolaou, G. J.; Parsa, N. Z.; Meltzer, P. S.; Trent, J. M.: Assignment of interferon gamma receptor (IFNGR1) to human chromosome bands 6q24.1–q24.2 by

in situ hybridization. Cytogenet. Cell Genet. 76:181–182, 1997. Note: Erratum: Cytogenet. Cell Genet. 78: 132 only, 1997.

- [45344] 18896. Pierre–Audigier, C.; Jouanguy, E.; Lamhamedi, S.; Altare, F.; Rauzier, J.; Vincent, V.; Canioni, D.; Emile, J. F.; Fischer, A.; Blanche, S.; Gaillard, J. L.; Casanova, J. L.: Fatal disseminated *Mycobacterium smegmatis* infection in a child with inherited interferon gamma receptor deficiency. Clin. Infect. Dis. 24: 982–984, 1997.
- [45345] 18897. Rashidbaigi, A.; Langer, J. A.; Jung, V.; Jones, C.; Morse, H.G.; Tischfield, J. A.; Trill, J. J.; Kung, H.–F.; Pestka, S.: The gene for the human immune interferon receptor is located on chromosome 6. Proc. Nat. Acad. Sci. 83: 384–388, 1986.
- [45346] 18898. Rettig, W. J.; Grzeschik, K.–H.; Yenamandra, A. K.; Garcia, E.; Old, L. J.: Definition of selectable cell surface markers for human chromosomes and chromosome segments in rodent–human hybrids. Somat. Cell Molec. Genet. 14: 223–231, 1988.
- [45347] 18899. Shankaran, V.; Ikeda, H.; Bruce, A. T.; White, J. M.; Swanson, P. E.; Old, L. J.; Schreiber, R. D.: IFN–gamma and lymphocytes prevent primary tumour development and shape tumour immunogenicity. Nature 410: 1107–1111,

2001.

- [45348] 18900. Bauer, R.; Imhof, A.; Pscherer, A.; Kopp, H.; Moser, M.; Seegers, S.; Kerscher, M.; Tainsky, M. A.; Hofstaedter, F.; Buettner, R.: The genomic structure of the human AP-2 transcription factor. *Nucleic Acids Res.* 22: 1413–1420, 1994.
- [45349] 18901. Buettner, R.; Kannan, P.; Imhof, A.; Bauer, R.; Yim, S. O.; Glockshuber, R.; Van Dyke, M. W.; Tainsky, M. A.: An alternatively spliced mRNA from the AP-2 gene encodes a negative regulator of transcriptional activation by AP-2. *Molec. Cell. Biol.* 13: 4174–4185, 1993.
- [45350] 18902. Ahmad, I.: Mash-1 is expressed during ROD photoreceptor differentiation and binds an E-box, E(opsin-1), in the rat opsin gene. *Brain Res. Dev. Brain Res.* 90: 184–189, 1995.
- [45351] 18903. Ball, D. W.; Azzoli, C. G.; Baylin, S. B.; Chi, D.; Dou, S.; Donis-Keller, H.; Cumaraswamy, A.; Borges, M.; Nelkin, B. D.: Identification of a human achaete-scute homolog highly expressed in neuroendocrine tumors. *Proc. Nat. Acad. Sci.* 90: 5648–5652, 1993.
- [45352] 18904. Guillemot, F.; Lo, L.-C.; Johnson, J. E.; Auerbach, A.; Anderson, D. J.; Joyner, A. L.: Mammalian achaete-scute homolog 1 is required for the early development of olfac-

tory and autonomic neurons. *Cell* 75:463–476, 1993.

- [45353] 18905. Renault, B.; Lieman, J.; Ward, D.; Krauter, K.; Kucherlapati, R.: Localization of the human achaete–scute homolog gene (ASCL1) distal to phenylalanine hydroxylase (PAH) and proximal to tumor rejection antigen (TRA1) on chromosome 12q22–q23. *Genomics* 30: 81–83, 1995.
- [45354] 18906. Yang, B.; Ma, T.; Verkman, A. S.: Erythrocyte water permeability and renal function in double knockout mice lacking aquaporin–1 and aquaporin–3. *J. Biol. Chem.* 276: 624–628, 2001.
- [45355] 18907. Bunz, F.; Kobayashi, R.; Stillman, B.: cDNAs encoding the large subunit of human replication factor C. *Proc. Nat. Acad. Sci.* 90:11014–11018, 1993.
- [45356] 18908. Lossie, A. C.; Haugen, B. R.; Wood, W. M.; Camper, S. A.; Gordon, D. F.: Chromosomal localization of the large subunit of mouse replication factor C in the mouse and human. *Mammalian Genome* 6: 58–59, 1995.
- [45357] 18909. Luckow, B.; Bunz, F.; Stillman, B.; Lichter, P.; Schutz, G.: Cloning, expression, and chromosomal localization of the 140–kilodalton subunit of replication factor C from mice and humans. *Molec. Cell. Biol.* 14:1626–1634, 1994.
- [45358] 18910. Pennaneach, V.; Salles–Passador, I.; Munshi, A.;

Brickner, H.;Regazzoni, K.; Dick, F.; Dyson, N.; Chen, T.-T.; Wang, J. Y. J.; Fotedar,R.; Fotedar, A.: The large subunit of replication factor C promotes cell survival after DNA damage in an LxCxE motif- and Rb-dependent manner. *Molec. Cell* 7: 715–727, 2001.

[45359] 18911.Uchiumi, F.; Ohta, T.; Tanuma, S.: Replication factor C recognizes 5-prime-phosphate ends of telomeres. *Biochem. Biophys. Res. Commun.* 229:310–315, 1996.

[45360] 18912.Schmickel, R. D.: Contiguous gene syndromes: a component of recognizable syndromes. *J. Pediat.* 109: 231–241, 1986.

[45361] 18913.Olson, E.; Srivastava, D.: Molecular pathways controlling heart development. *Science* 272: 671–676, 1996.

[45362] 18914.Lamballe, F.; Klein, R.; Barbacid, M.: TRKC, a new member of the TRK family of tyrosine protein kinases, is a receptor for neurotrophin-3. *Cell* 66:967–979, 1991.

[45363] 18915.McGregor, L. M.; Baylin, S. B.; Griffin, C. A.; Hawkins, A. L.; Nelkin, B. D.: Molecular cloning of the cDNA for human TrkC (NTRK3), chromosomal assignment, and evidence for a splice variant. *Genomics* 22:267–272, 1994.

[45364] 18916.Segal, R. A.; Goumnerova, L. C.; Kwon, Y. K.; Stiles, C. D.; Pomeroy, S. L.: Expression of the neurotrophin re-

ceptor TrkC is linked to a favorable outcome in medulloblastoma. *Proc. Nat. Acad. Sci.* 91:12867–12871, 1994.

[45365] 18917.Valent, A.; Danglot, G.; Bernheim, A.: Mapping of the tyrosinekinase receptors trkA (NTRK1), trkB (NTRK2) and trkC (NTRK3) to human chromosomes 1q22, 9q22 and 15q25 by fluorescence in situ hybridization. *Europ.J. Hum. Genet.* 5: 102–104, 1997.

[45366] 18918.Kielkopf, C. L.; Rodionova, N. A.; Green, M. R.; Burley, S. K.: A novel peptide recognition mode revealed by the X-ray structure of a core U2AF35/U2AF65 heterodimer. *Cell* 106: 595–605, 2001.

[45367] 18919.Lalioti, M. D.; Gos, A.; Green, M. R.; Rossier, C.; Morris, M.A.; Antonarakis, S. E.: The gene for human U2 snRNP auxiliary factor small 35–kDa subunit (U2AF1) maps to the progressive myoclonus epilepsy (EPM1) critical region on chromosome 21q22.3. *Genomics* 33: 298–300, 1996.

[45368] 18920.Hayashizaki, Y.; Shibata, H.; Hirotsune, S.; Sugino, H.; Okazaki, Y.; Sasaki, N.; Hirose, K.; Imoto, H.; Okuizumi, H.; Muramatsu, M.; Komatsubara, H.; Shiroishi, T.; Moriwaki, K.; Katsuki, M.; Hatano, N.; Sasaki, H.; Ueda, T.; Mise, N.; Takagi, N.; Plass, C.; Chapman, V. M.: Identification of an imprinted U2af binding protein related sequence on

mouse chromosome 11 using the RLGS method. *Nature Genet.* 6:33–40, 1994.

- [45369] 18921. Zhang, M.; Zamore, P. D.; Carmo-Fonseca, M.; Lamond, A. I.; Green, M. R.: Cloning and intracellular localization of the U2 small nuclear ribonucleoprotein auxiliary factor small subunit. *Proc. Nat. Acad. Sci.* 89: 8769–8773, 1992.
- [45370] 18922. Duncan, A. M. V.; Anderson, L.; Duff, C.; Ozawa, T.; Suzuki, H.; Worton, R.; Rozen, R.: Assignment of the gene (UQCRCF1) for the Rieske iron–sulfur protein subunit of the mitochondrial cytochrome bc–1 complex to the 22q13 and 19q12–q13.1 regions of the human genome. *Genomics* 21:281–283, 1994.
- [45371] 18923. Pennacchio, L. A.; Bergmann, A.; Fukushima, A.; Okubo, K.; Salemi, A.; Lennon, G. G.: Structure, sequence and location of the UQCRCF1 gene for the human Rieske Fe–S protein. *Gene* 155: 207–211, 1995.
- [45372] 18924. Cunningham, J. M.; Vanin, E. F.; Tran, N.; Valentine, M.; Jane, S. M.: The human transcription factor CP2 (TFCP2), a component of the human gamma–globin stage selector protein, maps to chromosome region 12q13 and is within 250 kb of the NF–E2 gene. *Genomics* 30:398–399, 1995.

- [45373] 18925.Jane, S. M.; Nienhuis, A. W.; Cunningham, J. M.: Hemoglobin switching in man and chicken is mediated by a heteromeric complex between the ubiquitous transcription factor CP2 and a developmentally specific protein. EMBO J. 13: 197–105, 1995.
- [45374] 18926.Lambert, J.–C.; Goumidi, L.; Wavrant–De Vrieze, F.; Frigard, B.; Harris, J. M.; Cummings, A.; Coates, J.; Pasquier, F.; Cotel, D.; Gaillac, M.; St. Clair, D.; Mann, D. M. A.; Hardy, J.; Lendon, C.L.; Amouyel, P.; Chartier–Harlin, M.–C.: The transcriptional factor LBP–1c/CP2/LSF gene on chromosome 12 is a genetic determinant of Alzheimer's disease. Hum. Molec. Genet. 9: 2275–2280, 2000.
- [45375] 18927.Swendeman, S. L.; Spielholz, C.; Jenkins, N. A.; Gilbert, D. J.; Copeland, N. G.; Sheffery, M.: Characterization of the genomic structure, chromosomal location, promoter, and developmental expression of the alpha–globin transcription factor CP2. J. Biol. Chem. 269: 11663–11671, 1994.
- [45376] 18928.Taylor, A. E.; Yip, A.; Brayne, C.; Easton, D.; Evans, J. G.; Xuereb, J.; Cairns, N.; Esiri, M. M.; Rubinsztein, D. C.: Genetic association of an LBP–1c/CP2/LSF gene polymorphism with late onset Alzheimer's disease. J. Med. Genet.

38: 232–233, 2001.

- [45377] 18929.Chen, C.-R.; Kang, Y.; Siegel, P. M.; Massague, J.: E2F4/5 and p107 as Smad cofactors linking the TGF- β receptor to c-myc repression. *Cell* 110:19–32, 2002.
- [45378] 18930.Zhang, Y.; Venkataraj, V. S.; Fischer, S. G.; Warburton, D.; Chellappan, S. P.: Genomic cloning and chromosomal assignment of the E2F dimerization partner TFDP gene family. *Genomics* 39: 95–98, 1997.
- [45379] 18931.Moloney, D. J.; Panin, V. M.; Johnston, S. H.; Chen, J.; Shao, L.; Wilson, R.; Wang, Y.; Stanley, P.; Irvine, K. D.; Haltiwanger, R. S.; Vogt, T. F.: Fringe is a glycosyltransferase that modifies Notch. *Nature* 406: 369–375, 2000.
- [45380] 18932.Sestan, N.; Artavanis-Tsakonas, S.; Rakic, P.: Contact-dependent inhibition of cortical neurite growth mediated by Notch signaling. *Science* 286:741–746, 1999.
- [45381] 18933.Tanigaki, K.; Nogaki, F.; Takahashi, J.; Tashiro, K.; Kurooka, H.; Honjo, T.: Notch1 and Notch3 instructively restrict bFGF-responsive multipotent neural progenitor cells to an astroglial fate. *Neuron* 29:45–55, 2001.
- [45382] 18934.Barton, D. E.; Foellmer, B. E.; Du, J.; Tamm, J.; Derynck, R.; Francke, U.: Chromosomal mapping of genes for transforming growth factors β -2 and β -3 in man and mouse: dispersion of TGF- β gene family. *Oncogene*

Res. 3: 323–331, 1988.

- [45383] 18935. Dean, M.; Park, M.; Vande Woude, G. F.: Characterization of the rearranged TPR–MET oncogene breakpoint. *Molec. Cell. Biol.* 7: 921–924, 1987.
- [45384] 18936. Robinson, B. H.; Taylor, J.; Sherwood, W. G.: The genetic heterogeneity of lactic acidosis: occurrence of recognisable inborn errors of metabolism in a pediatric population with lactic acidosis. *Pediat. Res.* 14: 950–962, 1980.
- [45385] 18937. Hosokawa, Y.; Suzuki, H.; Nishikimi, M.; Matsukage, A.; Yoshida, M. C.; Ozawa, T.: Chromosomal assignment of the gene for the ubiquinone-binding protein of human mitochondrial cytochrome bc₁ complex. *Biochem. Int.* 21: 41–44, 1990.
- [45386] 18938. Suzuki, H.; Hosokawa, Y.; Toda, H.; Nishikimi, M.; Ozawa, T.: Cloning and sequencing of a cDNA for human mitochondrial ubiquinone-binding protein of complex III. *Biochem. Biophys. Res. Commun.* 156: 987–994, 1988.
- [45387] 18939. van Leeuwen, F. W.; de Kleijn, D. P. V.; van den Hurk, H. H.; Neubauer, A.; Sonnemans, M. A. F.; Sluijs, J. A.; Koycu, S.; Ramdjielal, R. D. J.; Salehi, A.; Martens, G. J. M.; Grosveld, F. G.; Burbach, J. P. H.; Hol, E. M.: Frameshift mutants of beta-amyloid precursor protein and ubiquitin-

B in Alzheimer's and Down patients. *Science* 279: 242–247, 1998.

[45388] 18940. Webb, G. C.; Baker, R. T.; Fagan, K.; Board, P. G.: Localization of the human UbB polyubiquitin gene to chromosome band 17p11.1–17p12. *Am. J. Hum. Genet.* 46: 308–315, 1990.

[45389] 18941. Vidal, R.; Frangione, B.; Rostagno, A.; Mead, S.; Revesz, T.; Plant, G.; Ghiso, J.: A stop-codon mutation in the BRI gene associated with familial British dementia. *Nature* 399: 776–781, 1999.

[45390] 18942. Adler, D. A.; Tseng, B. Y.; Wang, T. S.-F.; Distèche, C. M.: Physical mapping of the genes for three components of the mouse DNA replication complex: polymerase alpha to the X chromosome, primase p49 subunit to chromosome 10, and primase p58 subunit to chromosome 1. *Genomics* 9: 642–646, 1991.

[45391] 18943. Ogilvie, A. D.; Battersby, S.; Bubb, V. J.; Fink, G.; Harmar, A. J.; Goodwin, G. M.; Smith, C. A. D.: Polymorphism in serotonin transporter gene associated with susceptibility to major depression. *Lancet* 347: 731–733, 1996.

[45392] 18944. Doniger, J.; DiPaolo, J. A.: Coordinate N-RAS mRNA up-regulation with mutational activation in tumorigenic

guinea pig cells. *NucleicAcids Res.* 16: 969–980, 1988.

[45393] 18945.Jeffers, M.; Paciucci, R.; Pellicer, A.: Characteriza-
tion of UNR:a gene closely linked to N–RAS. *Nucleic Acids
Res.* 18: 4891–4899,1990.

[45394] 18946.Mirnics, K.; Middleton, F. A.; Marquez, A.; Lewis, D.
A.; Levitt,P.: Molecular characterization of schizophrenia
viewed by microarrayanalysis of gene expression in pre-
frontal cortex. *Neuron* 28: 53–67,2000.

[45395] 18947.Engelkamp, D.; Schafer, B. W.; Mattei, M. G.; Erne,
P.; Heizmann,C. W.: Six S100 genes are clustered on hu-
man chromosome 1q21: identificationof two genes coding
for the two previously unreported calcium–bind-
ingproteins S100D and S100E. *Proc. Nat. Acad. Sci.* 90:
6547–6551, 1993.

[45396] 18948.Herrera, G. A.; Turbat–Herrera, E. A.; Lott, R. L.: S–
100 proteinexpression by primary and metastatic adeno-
carcinomas. *Am. J. Clin.Path.* 89: 168–176, 1988.

[45397] 18949.Isobe, T.; Tsugira, A.; Okuyama, T.: Amino acid se-
quence of thesubunit structure of bovine brain S–100
protein (PAP 1–b). *J. Neurochem.* 30:921–923, 1978.

[45398] 18950.Moore, B. W.: A soluble protein characteristic of the
nervoussystem. *Biochem. Biophys. Res. Commun.* 19:
739–744, 1965.

- [45399] 18951.Morii, K.; Tanaka, R.; Takahashi, Y.; Minoshima, S.; Fukuyama,R.; Shimizu, N.; Kuwano, R.: Structure and chromosome assignmentof human S100 alpha and beta subunit genes. *Biochem. Biophys. Res.Commun.* 175: 185–191, 1991.
- [45400] 18952.Most, P.; Bernotat, J.; Ehlermann, P.; Pleger, S. T.; Reppel, M.;Borries, M.; Niroomand, F.; Pieske, B.; Janssen, P. M. L.; Eschenhagen,T.; Karczewski, P.; Smith, G. L.; Koch, W. J.; Katus, H. A.; Remppis,A.: S100A1: a regulator of myocardial contractility. *Proc. Nat.Acad. Sci.* 98: 13889–13894, 2001.
- [45401] 18953.Asada, Y.; Nadeau, J. H.: Fert is on mouse chromosome 11, notchromosome 17. *Mammalian Genome* 5: 830 only, 1994.
- [45402] 18954.Hao, Q.–L.; Heisterkamp, N.; Groffen, J.: Isolation and sequenceanalysis of a novel human tyrosine kinase gene. *Molec. Cell. Biol.* 9:1587–1593, 1989.
- [45403] 18955.Dryja, T. P.; Grondin, V. J.; Ringens, P.; Cotran, P.; Berson,E. L.; Travis, G.: Isolation of human retinal cDNA fragments homologousto the murine rds gene transcript. (Abstract) *Invest. Ophthal. Vis.Sci.* 30 (suppl.): 43 only, 1989.
- [45404] 18956.Farrar, G. J.; Jordan, S. A.; Kenna, P.; Humphries, M.

M.; Kumar–Singh,R.; McWilliam, P.; Allamand, V.; Sharp, E.; Humphries, P.: Autosomaldominant retinitis pigmentosa: localization of a disease gene (RP6)to the short arm of chromosome 6. *Genomics* 11: 870–874, 1991.

[45405] 18957.Farrar, G. J.; Kenna, P.; Jordan, S. A.; Kumar–Singh, R.; Humphries,M. M.; Sharp, E. M.; Sheils, D. M.; Humphries, P.: A three–base–pairdeletion in the peripherin–RDS gene in one form of retinitis pigmentosa. *Nature* 354:478–480, 1991.

[45406] 18958.Feist, R. M.; White, M. F., Jr.; Skalka, H.; Stone, E. M.: Choroidalneovascularization in a patient with adult foveomacular dystrophyand a mutation in the retinal degeneration slow gene (pro210–to–arg). *Am.J. Ophthal.* 118: 259–260, 1994.

[45407] 18959.Felbor, U.; Schilling, H.; Weber, B. H. F.: Adult vitelliformmacular dystrophy is frequently associated with mutations in the peripherin/RDSgene. *Hum. Mutat.* 10: 301–309, 1997.

[45408] 18960.Gass, J. D. M.: A clinicopathologic study of a peculiar foveomaculardystrophy. *Trans. Am. Ophthal. Soc.* 72: 139–155, 1974.

[45409] 18961.Jackson, K. E.; Mitchell, E. B.; Stone, E. M.; Ferrell, R. E.;Gorin, M. B.: The identification of an exon–2 periph–

erin mutation in a family with heterogeneous manifestations of a butterfly pattern macular dystrophy. (Abstract) Am. J. Hum. Genet. 53 (suppl.): 1177 only, 1993.

[45410] 18962. Jordan, S. A.; Farrar, G. J.; Kumar-Singh, R.; Kenna, P.; Humphries, M. M.; Allamand, V.; Sharp, E. M.; Humphries, P.: Autosomal dominant retinitis pigmentosa (adRP; RP6): cosegregation of RP6 and the peripherin-RD-S locus in a late-onset family of Irish origin. Am. J. Hum. Genet. 50:634-639, 1992.

[45411] 18963. Okamoto, H.; Yonemori, F.; Wakitani, K.; Minowa, T.; Maeda, K.; Shinkai, H.: A cholesteryl ester transfer protein inhibitor attenuates atherosclerosis in rabbits. Nature 406: 203-207, 2000.

[45412] 18964. Oliveira, H. C. F.; Chouinard, R. A.; Agellon, L. B.; Bruce, C.; Ma, L.; Walsh, A.; Breslow, J. L.; Tall, A. R.: Human cholesteryl ester transfer protein gene proximal promoter contains dietary cholesterol positive responsive elements and mediates expression in small intestine and periphery while predominant liver and spleen expression is controlled by 5-prime-distal sequences. J. Biol. Chem. 271: 31831-31838, 1996.

[45413] 18965. Sakai, N.; Yamashita, S.; Hirano, K.; Menju, M.; Arai, T.; Kobayashi, K.; Ishigami, M.; Yoshida, Y.; Hoshino,

T.; Nakajima, N.; Kameda-Takemura, K.; Matsuzawa, Y.: Frequency of exon 15 missense mutation (442D:G) in cholesteryl ester transfer protein gene in hyperlipoproteinemic Japanese subjects. *Atherosclerosis* 114: 139–145, 1995.

[45414] 18966. Sparkes, R. S.; Drayna, D.; Mohandas, T.; Klisak, I.; Heinzmann, C.; Lawn, R.; Lusis, A. J.: Assignment of cholesterol ester transfer protein (CETP) gene to human 16q21. (Abstract) *Cytogenet. Cell Genet.* 46:696 only, 1987.

[45415] 18967. Takahashi, K.; Jiang, X.-C.; Sakai, N.; Yamashita, S.; Hirano, K.; Bujo, H.; Yamazaki, H.; Kusunoki, J.; Miura, T.; Kussie, P.; Matsuzawa, Y.; Saito, Y.; Tall, A.: A missense mutation in the cholesteryl ester transfer protein gene with possible dominant effects on plasma high density lipoproteins. *J. Clin. Invest.* 92: 2060–2064, 1993.

[45416] 18968. Yamashita, S.; Hui, D. Y.; Sprecher, D. L.; Matsuzawa, Y.; Sakai, N.; Tarui, S.; Kaplan, D.; Wetterau, J. R.; Harmony, J. A.: Total deficiency of plasma cholesteryl ester transfer protein in subjects homozygous and heterozygous for the intron 14 splicing defect. *Biochem. Biophys. Res. Commun.* 170: 1346–1351, 1990.

[45417] 18969. Zhong, S.; Sharp, D. S.; Grove, J. S.; Bruce, C.; Yano,

K.; Curb, J. D.; Tall, A. R.: Increased coronary heart disease in Japanese-American men with mutation in the cholesterol ester transfer protein gene despite increased HDL levels. *J. Clin. Invest.* 97: 2917–2923, 1996.

[45418] 18970. Chung, B.-C.; Matteson, K. J.; Voutilainen, R.; Mohandas, T. K.; Miller, W. L.: Human cholesterol side-chain cleavage enzyme, P450scc: cDNA cloning, assignment of the gene to chromosome 15, and expression in the placenta. *Proc. Nat. Acad. Sci.* 83: 8962–8966, 1986.

[45419] 18971. Durocher, F.; Morissette, J.; Simard, J.: Genetic linkage mapping of the CYP11A1 gene encoding the cholesterol side-chain cleavage P450(scc) close to the CYP1A1 gene and D15S204 in the chromosome 15q22.33–q23 region. *Pharmacogenetics* 8: 49–53, 1998.

[45420] 18972. Billingsley, G. D.; Walter, M. A.; Hammond, G. L.; Cox, D. W.: Physical mapping of four serpin genes: alpha-1-antitrypsin, alpha-1-antichymotrypsin, corticosteroid-binding globulin, and protein C inhibitor, within a 280-kb region on chromosome 14q32.1. *Am. J. Hum. Genet.* 52: 343–353, 1993.

[45421] 18973. Wang, J.; Hannon, G. J.; Beach, D. H.: Risky immortalization by telomerase. (Letter) *Nature* 405: 755–756, 2000.

- [45422] 18974.Wu, K.-J.; Grandori, C.; Amacker, M.; Simon-Ver-
mot, N.; Polack,A.; Lingner, J.; Dalla-Favera, R.: Direct ac-
tivation of TERT transcriptionby c-MYC. *Nature Genet.* 21:
220-224, 1999.
- [45423] 18975.Greco, A.; Ittmann, M.; Barletta, C.; Basilico, C.;
Croce, C. M.;Cannizzaro, L. A.; Huebner, K.: Chromosomal
localization of humangen es required for G(1) progression
in mammalian cells. *Genomics* 4:240-245, 1989.
- [45424] 18976.Ittmann, M.; Greco, A.; Basilico, C.: Isolation of the
human genethat complements a temperature-sensitive
cell cycle mutation in BHKcells. *Molec. Cell. Biol.* 7:
3386-3393, 1987.
- [45425] 18977.Ip, N. Y.; Stitt, T. N.; Tapley, P.; Klein, R.; Glass, D.
J.; Fandl,J.; Greene, L. A.; Barbacid, M.; Yancopoulos, G. D.:
Similaritiesand differences in the way neurotrophins inter-
act with the Trk receptorsin neuronal and nonneuronal
cells. *Neuron* 10: 137-149, 1993.
- [45426] 18978.Knezevich, S. R.; McFadden, D. E.; Tao, W.; Lim, J.
F.; Sorensen,P. H. B.: A novel ETV6-NTRK3 gene fusion in
congenital fibrosarcoma. *NatureGenet.* 18: 184-187,
1998.
- [45427] 18979.Bonnefont, J.-P.; Chretien, D.; Rustin, P.; Robinson,
B.; Vassault,A.; Aupetit, J.; Charpentier, C.; Rabier, D.;

Saudubray, J.-M.; Munnich, A.: Alpha-ketoglutarate dehydrogenase deficiency presenting as congenital lactic acidosis. *J. Pediatr.* 121: 255–258, 1992.

[45428] 18980. Guffon, N.; Lopez-Mediavilla, C.; Dumoulin, R.; Mousson, B.; Godinot, C.; Carrier, H.; Collombet, J. M.; Divry, P.; Mathieu, M.; Guibaud, P.: 2-Ketoglutarate dehydrogenase deficiency, a rare cause of primary hyperlactataemia: report of a new case. *J. Inherit. Metab. Dis.* 16: 821–830, 1993.

[45429] 18981. Kohlschütter, A.; Behbehani, A.; Langenbeck, U.; Albani, M.; Heidemann, P.; Hoffmann, G.; Kleineke, J.; Lehnert, W.; Wendel, U.: A familial progressive neurodegenerative disease with 2-oxoglutaric aciduria. *Europ. J. Pediatr.* 138: 32–37, 1982.

[45430] 18982. Koike, K.: The gene encoding human 2-oxoglutarate dehydrogenase: structural organization and mapping to chromosome 7p13–p14. *Gene* 159: 261–266, 1995.

[45431] 18983. Koike, K.; Urata, Y.; Goto, S.: Cloning and nucleotide sequence of the cDNA encoding human 2-oxoglutarate dehydrogenase (lipoamide). *Proc. Nat. Acad. Sci.* 89: 1963–1967, 1992.

[45432] 18984. Szabo, P.; Cai, X.; Ali, G.; Blass, J. P.: Localization of

the gene (OGDH) coding for the E1 α component of the α -ketoglutarate dehydrogenase complex to chromosome 7p13-p11.2. *Genomics* 20: 324-326, 1994.

[45433] 18985. Kury, S.; Dreno, B.; Bezieau, S.; Giraudet, S.; Kharfi, M.; Kamoun, R.; Moisan, J.-P.: Identification of SLC39A4, a gene involved in acrodermatitis enteropathica. *Nature Genet.* 31: 239-240, 2002.

[45434] 18986. Rogers, E. E.; Eide, D. J.; Guerinot, M. L.: Altered selectivity in an *Arabidopsis* metal transporter. *Proc. Nat. Acad. Sci.* 97: 12356-12360, 2000.

[45435] 18987. Wang, K.; Pugh, E. W.; Griffen, S.; Doheny, K. F.; Mostafa, W. Z.; al-Aboosi, M. M.; el-Shanti, H.; Gitschier, J.: Homozygosity mapping places the acrodermatitis enteropathica gene on chromosomal region 8q24.3. *Am. J. Hum. Genet.* 68: 1055-1060, 2001.

[45436] 18988. Haberle, J.; Pauli, S.; Linnebank, M.; Kleijer, W. J.; Bakker, H. D.; Wanders, R. J. A.; Harms, E.; Koch, H. G.: Structure of the human argininosuccinate synthetase gene and an improved system for molecular diagnostics in patients with classical and mild citrullinemia. *Hum. Genet.* 110: 327-333, 2002.

[45437] 18989. Barbosa, J.; Rich, S.; Dunsworth, T.; Swanson, J.: Linkage disequilibrium between insulin-dependent dia-

betes and the Kidd blood group Jk(b)allele. J. Clin. Endocr. Metab. 55: 193–195, 1982.

[45438] 18990.Harper, P. A.; Healy, P. J.; Dennis, J. A.; O'Brien, J. J.; Rayward,D. H.: Citrullinaemia as a cause of neurological disease in neonatalFriesian calves. Aust. Vet. J. 63: 378–379, 1986.

[45439] 18991.Harper, P. A. W.; Healy, P. J.; Dennis, J. A.: Animal model ofhuman disease: citrullinemia (argininosuccinate synthetase deficiency). Am.J. Path. 135: 1213–1215, 1989.

[45440] 18992.Jackson, M. J.; Surh, L. C.; O'Brien, W. E.; Beaudet, A. L.:Assignment of the structural gene for argininosuccinate synthetaseto proximal mouse chromosome 2. Genomics 6: 545–547, 1990.

[45441] 18993.Kobayashi, K.; Ichiki, H.; Saheki, T.; Tatsuno, M.; Uchiyama,C.; Nukada, O.; Yoda, T.: Structure of an abnormal messenger RNAfor argininosuccinate synthetase in citrullinemia. Hum. Genet. 76:27–32, 1987.

[45442] 18994.Kobayashi, K.; Jackson, M. J.; Tick, D. B.; O'Brien, W. E.; Beaudet,A. L.: Characterization of nine mutant alleles causing citrullinemia.(Abstract) Am. J. Hum. Genet. 45 (suppl.): A201 only, 1989.

[45443] 18995.Kobayashi, K.; Jackson, M. J.; Tick, D. B.; O'Brien,

W. E.; Beaudet, A. L.: Heterogeneity of mutations in argininosuccinate synthetase causing human citrullinemia. *J. Biol. Chem.* 265: 11361–11367, 1990.

[45444] 18996. Kobayashi, K.; Kakinoki, H.; Fukushige, T.; Shaheen, N.; Terazono, H.; Saheki, T.: Nature and frequency of mutations in the argininosuccinate synthetase gene that cause classical citrullinemia. *Hum. Genet.* 96:454–463, 1995.

[45445] 18997. Kobayashi, K.; Rosenbloom, C.; Beaudet, A. L.; O'Brien, W. E.: Additional mutations in argininosuccinate synthetase causing citrullinemia. *Molec. Biol. Med.* 8: 95–100, 1991.

[45446] 18998. Kobayashi, K.; Saheki, T.; Imamura, Y.; Noda, T.; Inoue, I.; Matuo, S.; Hagihara, S.; Nomiya, H.; Jinno, Y.; Shimada, K.: Messenger RNA coding for argininosuccinate synthetase in citrullinemia. *Am. J. Hum. Genet.* 38: 667–680, 1986.

[45447] 18999. Kobayashi, K.; Shaheen, N.; Terazono, H.; Saheki, T.: Mutations in argininosuccinate synthetase mRNA of Japanese patients, causing classical citrullinemia. *Am. J. Hum. Genet.* 55: 1103–1112, 1994.

[45448] 19000. Li, C.-M.; Chao, H.-K.; Liu, Y.-F.; Su, T.-S.: A non-sense mutation is responsible for the RNA-negative phe-

notype in human citrullinaemia. *Europ.J. Hum. Genet.* 9: 685–689, 2001.

[45449] 19001.Northrup, H.; Lathrop, M.; Lu, S.–Y.; Daiger, S. P.; Beaudet,A. L.; O'Brien, W. E.: Multilocus linkage analysis with the humanargininosuccinate synthetase gene. *Genomics* 5: 442–444, 1989.

[45450] 19002.Sase, M.; Kobayashi, K.; Imamura, Y.; Saheki, T.; Nakano, K.;Miura, S.; Mori, M.: Level of translatable messenger RNA coding forargininosuccinate synthetase in the liver of the patients with quantitative–typecitrullinemia. *Hum. Genet.* 69: 130–134, 1985.

[45451] 19003.Todd, S.; Naylor, S. L.: New chromosomal mapping assignmentsfor argininosuccinate synthetase pseudogene 1, interferon–beta–3 gene,and the diazepam binding inhibitor gene. *Somat. Cell Molec. Genet.* 18:381–385, 1992.

[45452] 19004.Rousseau–Merck, M. F.; Pizon, V.; Tavitian, A.; Berger, R.: Chromosomemapping of the human RAS related RAP1A, RAP1B and RAP2 genes to chromosomes1p13–12, 12q14 and 13q34, respectively. (Abstract) *Cytogenet. CellGenet.* 51: 1070 only, 1989.

[45453] 19005.Rousseau–Merck, M. F.; Pizon, V.; Tavitian, A.; Berger, R.: Chromosomemapping of the human RAS–

related RAP1A, RAP1B, and RAP2 genes to chromosomes 1p12–p13, 12q14, and 13q34, respectively. Cytogenet. Cell Genet. 53:2–4, 1990.

- [45454] 19006. Sebzda, E.; Bracke, M.; Tugal, T.; Hogg, N.; Cantrell, D. A.: Rap1a positively regulates T cells via integrin activation rather than inhibiting lymphocyte signaling. Nature Immun. 3: 251–258, 2002.
- [45455] 19007. Takai, S.; Nishino, N.; Kitayama, H.; Ikawa, Y.; Noda, M.: Mapping of the KREV1 transformation suppressor gene and its pseudogene (KREV1P) to human chromosome 1p13.3 and 14q24.3, respectively, by fluorescence in situ hybridization. Cytogenet. Cell Genet. 63: 59–61, 1993.
- [45456] 19008. Zhu, J. J.; Qin, Y.; Zhao, M.; Van Aelst, L.; Malinow, R.: Ras and Rap control AMPA receptor trafficking during synaptic plasticity. Cell 110:443–455, 2002.
- [45457] 19009. Ahmad, F.; Goldstein, B. J.: Functional association between the insulin receptor and the transmembrane protein-tyrosine phosphatase LAR in intact cells. J. Biol. Chem. 272: 448–457, 1997.
- [45458] 19010. Disteche, C. M.; Adler, D. A.; Tedder, T. F.; Saito, H.: Mapping of the genes for LYAM1, a new lymphocyte adhesion molecule, and for LAR, a new receptor-linked

protein tyrosine phosphatase, to human chromosome 1
(Abstract) Cytogenet. Cell Genet. 51: 990 only, 1989.

- [45459] 19011. Harder, K. W.; Saw, J.; Miki, N.; Jirik, F.: Coexisting amplifications of the chromosome 1p32 genes (PTPRF and MYCL1) encoding protein tyrosine phosphatase LAR and L-myc in a small cell lung cancer line. Genomics 27:552–553, 1995.
- [45460] 19012. Jirik, F. R.; Harder, K. W.; Melhado, I. G.; Anderson, L. L.; Duncan, A. M. V.: The gene for leukocyte antigen-related tyrosine phosphatase (LAR) is localized to human chromosome 1p32, a region frequently deleted in tumors of neuroectodermal origin. Cytogenet. Cell Genet. 61:266–268, 1992.
- [45461] 19013. Nam, H.-J.; Poy, F.; Krueger, N. X.; Saito, H.; Frederick, C. A.: Crystal structure of the tandem phosphatase domains of RPTP LAR. Cell 97:449–457, 1999.
- [45462] 19014. O'Grady, P.; Krueger, N. X.; Streuli, M.; Saito, H.: Genomic organization of the human LAR protein tyrosine phosphatase gene and alternative splicing in the extracellular fibronectin type-III domains. J. Biol. Chem. 269: 25193–25199, 1994.
- [45463] 19015. Schaapveld, R. Q.; Schepens, J. T.; Robinson, G. W.; Attema, J.; Oerlemans, F. T.; Fransen, J. A.; Streuli, M.;

Wieringa, B.; Hennighausen, L.; Hendriks, W. J.: Impaired mammary gland development and function in mice lacking LAR receptor-like tyrosine phosphatase activity. *Dev. Biol.* 188: 134–146, 1997.

[45464] 19016. Schaapveld, R. Q. J.; van den Maagdenberg, A. M. J. M.; Schepens, J. T. G.; Olde Weghuis, D.; Geurts van Kessel, A.; Wieringa, B.; Hendriks, W. J. A. J.: The mouse gene *Ptprf* encoding the leukocyte common antigen-related molecule LAR: cloning, characterization, and chromosomal localization. *Genomics* 27:124–130, 1995.

[45465] 19017. Tsujikawa, K.; Kawakami, N.; Uchino, Y.; Ichijo, T.; Furukawa, T.; Saito, H.; Yamamoto, H.: Distinct functions of the two protein tyrosine phosphatase domains of LAR (leukocyte common antigen-related) on tyrosine dephosphorylation of insulin receptor. *Molec. Endocr.* 15:271–280, 2001.

[45466] 19018. Ali, R. R.; Sarra, G.-M.; Stephens, C.; de Alwis, M.; Bainbridge, J. W. B.; Munro, P. M.; Fauser, S.; Reichell, M. B.; Kinnon, C.; Hunt, D. M.; Bhattacharya, S. S.; Thrasher, A. J.: Restoration of photoreceptor ultrastructure and function in retinal degeneration slow mice by gene therapy. *Nature Genet.* 25: 306–310, 2000.

[45467] 19019. Bascom, R. A.; Connell, G.; Garcia-Heras, J.;

Collins, L.; Ledbetter, D.; Molday, R. S.; Kalnins, V.;
McInnes, R. R.: Molecular and ultrastructural characterization of the products of the human retinopathy candidate-genes ROM1 and RDS. (Abstract) Am. J. Hum. Genet. 47 (suppl.): A101 only, 1990.

[45468] 19020. Connell, G.; Bascom, R.; Molday, L.; Reid, D.;
McInnes, R. R.; Molday, R. S.: Photoreceptor peripherin is the normal product of the gene responsible for retinal degeneration in the rds mouse. Proc. Nat. Acad. Sci. 88: 723–726, 1991.

[45469] 19021. Demant, P.; Ivanyi, D.; van Nie, R.: The map position of the rds gene on the 17th chromosome of the mouse. Tissue Antigens 13: 53–55, 1979.

[45470] 19022. Makela, T. P.; Kere, J.; Winqvist, R.; Alitalo, K.: Intrachromosomal rearrangements fusing L-myc and rlf in small-cell lung cancer. Molec. Cell. Biol. 11: 4015–4021, 1991.

[45471] 19023. Makela, T. P.; Saksela, K.; Evan, G.; Alitalo, K.: A fusion protein formed by L-myc and a novel gene in SCLC. EMBO J. 10: 1331–1335, 1991.

[45472] 19024. Brody, L. C.; Abel, K. J.; Castilla, L. H.; Couch, F. J.; McKinley, D. R.; Yin, G.-Y.; Ho, P. P.; Merajver, S.; Chandrasekharappa, S. C.; Xu, J.; Cole, J. L.; Struwing, J. P.;

Valdes, J. M.; Collins, F. S.; Weber, B. L.: Construction of a transcription map surrounding the BRCA1 locus of human chromosome 17. *Genomics* 25: 238–247, 1995.

[45473] 19025. Hloch, P.; Schiedner, G.; Stahl, H.: Complete cDNA sequence of the human p68 protein. *Nucleic Acids Res.* 18: 3045, 1990.

[45474] 19026. Iggo, R.; Gough, A.; Xu, W.; Lane, D. P.; Spurr, N. K.: Chromosome mapping of the human gene encoding the 68-kDa nuclear antigen (p68) by using the polymerase chain reaction. *Proc. Nat. Acad. Sci.* 86: 6211–6214, 1989.

[45475] 19027. Iggo, R. D.; Xu, W.; Lane, D. P.; Gough, A.; Spurr, N. K.: Assignment of P68 to chromosome 17 using PCR. (Abstract) *Cytogenet. Cell Genet.* 51: 1017, 1989.

[45476] 19028. Acker, J.; Mattei, M.-G.; Wintzerith, M.; Roedel, N.; Depetris, D.; Vigneron, M.; Keding, C.: Chromosomal localization of human RNA polymerase II subunit genes. *Genomics* 20: 496–499, 1994.

[45477] 19029. Bourquin, J.-P.; Stagljar, I.; Meier, P.; Moosmann, P.; Silke, J.; Baechi, T.; Georgiev, O.; Schaffner, W.: A serine/arginine-rich nuclear matrix cyclophilin interacts with the C-terminal domain of RNA polymerase II. *Nucleic Acids Res.* 25: 2055–2061, 1997.

[45478] 19030. Schulze, A.; Hansen, C.; Skakkebaek, N. E.; Bron-

dum-Nielsen, K.;Ledbetter, D. H.; Tommerup, N.: Exclusion of SNRPN as a major determinant of Prader-Willi syndrome by a translocation breakpoint. *Nature Genet.* 12:452-454, 1996.

[45479] 19031.Schweizer, J.; Zynger, D.; Francke, U.: In vivo nuclease hypersensitivity studies reveal multiple sites of parental origin-dependent differential chromatin conformation in the 150 kb SNRPN transcription unit. *Hum.Molec. Genet.* 8: 555-566, 1999.

[45480] 19032.Shemer, R.; Hershko, A. Y.; Perk, J.; Mostoslavsky, R.; Tsuberi, B.; Cedar, H.; Buiting, K.; Razin, A.: The imprinting box of the Prader-Willi/Angelman syndrome domain. *Nature Genet.* 26: 440-443, 2000.

[45481] 19033.Sun, Y.; Nicholls, R. D.; Butler, M. G.; Saitoh, S.; Hainline, B. E.; Palmer, C. G.: Breakage in the SNRPN locus in a balanced 46,XY,t(15;19) Prader-Willi syndrome patient. *Hum. Molec. Genet.* 5: 517-524, 1996.

[45482] 19034.Sutcliffe, J. S.; Nakao, M.; Christian, S.; Orstavik, K. H.; Tommerup, N.; Ledbetter, D. H.; Beaudet, A. L.: Deletions of a differentially methylated CpG island at the SNRPN gene define a putative imprinting control region. *Nature Genet.* 8: 52-58, 1994.

[45483] 19035.Wirth, J.; Back, E.; Huttenhofer, A.; Nothwang,

H.-G.; Lich, C.; Gross, S.; Menzel, C.; Schinzel, A.; Kioschis, P.; Tommerup, N.; Ropers, H.-H.; Horsthemke, B.; Buiting, K.: A translocation breakpoint cluster disrupts the newly defined 3-prime end of the SNURF-SNRPN transcription unit on chromosome 15. *Hum. Molec. Genet.* 10: 201-210, 2001.

[45484] 19036. Stevanovic, M.; Zuffardi, O.; Collignon, J.; Lovell-Badge, R.; Goodfellow, P.: The cDNA sequence and chromosomal location of the human SOX2 gene. *Mammalian Genome* 5: 640-642, 1994.

[45485] 19037. Critcher, R.; Stitson, R. N. M.; Wade-Martins, R.; Easty, D. J.; Farr, C. J.: Assignment of Sox4 to mouse chromosome 13 bands A3-A5 by fluorescence in situ hybridization; refinement of the human SOX4 location to 6p22.3 and of SOX20 to chromosome 17p12.3. *Cytogenet. Cell Genet.* 81: 294-295, 1998.

[45486] 19038. Denny, P.; Swift, S.; Connor, F.; Ashworth, A.: An SRY-related gene expressed during spermatogenesis in the mouse encodes a sequence-specific DNA-binding protein. *EMBO J.* 11: 3705-3712, 1992.

[45487] 19039. Farr, C. J.; Easty, D. J.; Ragoussis, J.; Collignon, J.; Lovell-Badge, R.; Goodfellow, P. N.: Characterization and mapping of the human SOX4 gene. *Mammalian Genome* 4:

577–584, 1993.

- [45488] 19040.Hansen, G. M.; Skapura, D.; Justice, M. J.: Genetic profile of insertion mutations in mouse leukemias and lymphomas. *Genome Res.* 10:237–243, 2000.
- [45489] 19041.Li, J.; et al; et al: Leukaemia disease genes: large-scale cloning and pathway predictions. *Nature Genet.* 23: 348–353, 1999.
- [45490] 19042.Lund, A. H.; Turner, G.; Trubetskoy, A.; Verhoeven, E.; Wientjens, E.; Hulsman, D.; Russell, R.; DePinho, R. A.; Lenz, J.; van Lohuizen, M.: Genome-wide retroviral insertional tagging of genes involved in cancer in *Cdkn2a*-deficient mice. *Nature Genet.* 32: 160–165, 2002.
- [45491] 19043.Suzuki, T.; Shen, H.; Akagi, K.; Morse, H. C., III; Malley, J.D.; Naiman, D. Q.; Jenkins, N. A.; Copeland, N. G.: New genes involved in cancer identified by retroviral tagging. *Nature Genet.* 32: 166–174, 2002. Note: Erratum: *Nature Genet.* 32: 331 only, 2002.
- [45492] 19044.van de Wetering, M.; Oosterwegel, M.; van Norren, K.; Clevers, H.: Sox-4, an Sry-like HMG box protein, is a transcriptional activator in lymphocytes. *EMBO J.* 12: 3847–3854, 1993.
- [45493] 19045.Shoyab, M.; McDonald, V. L.; Bradley, J. G.; Todaro, G. J.: Amphiregulin: a bifunctional growth-modulating gly-

coprotein produced by the phorbol12–myristate
13–acetate–treated human breast adenocarcinoma cell
lineMCF–7. Proc. Nat. Acad. Sci. 85: 6528–6532, 1988.

- [45494] 19046.Shoyab, M.; Plowman, G. D.; McDonald, V. L.;
Bradley, J. G.; Todaro,G. J.: Structure and function of hu-
man amphiregulin: a member ofthe epidermal growth fac-
tor family. Science 243: 1074–1076, 1989.
- [45495] 19047.Donahue, R. P.; Bias, W. B.; Renwick, J. H.; McKu-
sick, V. A.:Probable assignment of the Duffy blood group
locus to chromosome 1in man. Proc. Nat. Acad. Sci. 61:
949–955, 1968.
- [45496] 19048.Dracopoli, N. C.; Meisler, M. H.: Mapping the hu-
man amylase genecluster on the proximal short arm of
chromosome 1 using a highly informative(CA)_n repeat.
Genomics 7: 97–102, 1990.
- [45497] 19049.Knoll, J. H. M.; Nicholls, R. D.; Magenis, R. E.; Glatt,
K.; Graham,J. M., Jr.; Kaplan, L.; Lalande, M.: Angelman
syndrome: three molecularclasses identified with chromo-
some 15q11q13–specific DNA markers. Am.J. Hum.
Genet. 47: 149–155, 1990.
- [45498] 19050.Ono, H.; Kuno, Y.; Tanaka, H.; Yamashina, M.;
Tsuyoshi, T.; Kondo,N.; Orii, T.: A case of paroxysmal
nocturnal hemoglobinuria withoutdeficiency of decay–

accelerating factor on erythrocytes. *Blood* 75:1746–1747, 1990.

[45499] 19051. Petranka, J. G.; Fleenor, D. E.; Sykes, K.; Kaufman, R. E.; Rosse, W. F.: Structure of the CD59–encoding gene: further evidence of a relationship to murine lymphocyte antigen Ly-6 protein. *Proc. Nat. Acad. Sci.* 89: 7876–7879, 1992.

[45500] 19052. Rosse, W. F.: Personal Communication. Durham, N. C. 6/3/1993.

[45501] 19053. Rosse, W. F.; Parker, C. J.: Paroxysmal nocturnal hemoglobinuria. *Clin. Haemat.* 14: 105–125, 1985.

[45502] 19054. Rother, R. P.; Rollins, S. A.; Mennone, J.; Chodera, A.; Fidel, S. A.; Bessler, M.; Hillmen, P.; Squinto, S. P.: Expression of recombinant transmembrane CD59 in paroxysmal nocturnal hemoglobinuria B cells confers resistance to human complement. *Blood* 84: 2604–2611, 1994.

[45503] 19055. Tone, M.; Walsh, L. A.; Waldmann, H.: Gene structure of human CD59 and demonstration that discrete mRNAs are generated by alternative polyadenylation. *J. Molec. Biol.* 227: 971–976, 1992.

[45504] 19056. Walsh, L. A.; Tone, M.; Thiru, S.; Waldmann, H.: The CD59 antigen--a multifunctional molecule. *Tissue Antigens* 40: 213–220, 1992.

- [45505] 19057.Yamashina, M.; Ueda, E.; Kinoshita, T.; Takami, T.; Ojima, A.;Ono, H.; Tanaka, H.; Kondo, N.; Orii, T.; Okada, N.; Okada, H.; Inoue,K.; Kitani, T.: Inherited complete deficiency of 20–kilodalton homologousrestriction factor (CD59) as a cause of paroxysmal nocturnal hemoglobinuria. *NewEng. J. Med.* 323: 1184–1189, 1990.
- [45506] 19058.Kumanogoh, A.; Watanabe, C.; Lee, I.; Wang, X.; Shi, W.; Araki,H.; Hirata, H.; Iwahori, K.; Uchida, J.; Yasui, T.; Matsumoto, M.;Yoshida, K.; Yakura, H.; Pan, C.; Parnes, J. R.; Kikutani, H.: Identificationof CD72 as a lymphocyte receptor for the class IV semaphorin CD100:a novel mechanism for regulating B cell signaling. *Immunity* 13:621–631, 2000.
- [45507] 19059.Von Hoegen, I.; Hsieh, C.–L.; Scharting, R.; Francke, U.; Parnes,J. R.: Identity of human Lyb–2 and CD72 and localization of the geneto chromosome 9. *Europ. J. Immun.* 21: 1425–1431, 1991.
- [45508] 19060.Cambiaggi, C.; Scupoli, M. T.; Cestari, T.; Gerosa, F.; Carra,G.; Tridente, G.; Accolla, R. S.: Constitutive expression of CD69in interspecies T–cell hybrids and locus assignment to human chromosome12. *Immunogenetics* 36: 117–120, 1992.
- [45509] 19061.Lopez–Cabrera, M.; Santis, A. G.; Fernandez–Ruiz,

E.; Blacher, R.; Esch, F.; Sanchez-Mateos, P.; Sanchez-Madrid, F.: Molecular cloning, expression, and chromosomal localization of the human earliest lymphocyte activation antigen AIM/CD69, a new member of the C-type animal lectin superfamily of signal-transmitting receptors. *J. Exp. Med.* 178:537–547, 1993.

[45510] 19062. Chandra, T.; Stackhouse, R.; Kidd, V. J.; Robson, K. J. H.; Woo, S. L. C.: Sequence homology between human alpha-1-antichymotrypsin, alpha-1-antitrypsin, and antithrombin III. *Biochemistry* 22: 5055–5061, 1983.

[45511] 19063. Eriksson, S.; Lindmark, B.; Lilia, H.: Familial alpha-1-antichymotrypsin deficiency. *Acta Med. Scand.* 220: 447–453, 1986.

[45512] 19064. Gilfix, B. M.; Briones, L.: Absence of the A1252G mutation in alpha 1-antichymotrypsin in a North American population suffering from dementia. *J. Cereb. Blood Flow Metab.* 17: 233–235, 1997.

[45513] 19065. Haines, J. L.; Pritchard, M. L.; Saunders, A. M.; Schildkraut, J. M.; Growdon, J. H.; Gaskell, P. C.; Farrer, L. A.; Auerbach, S. A.; Gusella, J. F.; Locke, P. A.; Rosi, B. L.; Yamaoka, L.; Small, G. W.; Conneally, P. M.; Roses, A. D.; Pericak-Vance, M. A.: No genetic effect of alpha-1-antichymotrypsin in Alzheimer disease. *Genomics*

33:53–56, 1996.

- [45514] 19066.Haines, J. L.; Scott, W. K.; Pericak–Vance, M. A.: Reply to 'Geneticeffect of alpha–1–antichymotrypsin on the risk of Alzheimer disease.'(Letter) Genomics 40: 384–385, 1997.
- [45515] 19067.Kamboh, M. I.; Aston, C. E.; Ferrell, R. E.; Dekosky, S. T.: Geneticeffect of alpha–1–antichymotrypsin on the risk of Alzheimer disease.(Letter) Genomics 41: 382–385, 1997.
- [45516] 19068.Kamboh, M. I.; Sanghera, D. K.; Ferrell, R. E.; DeKosky, S. T.: APOE*4–associated Alzheimer's disease risk is modified by alpha–1–antichymotrypsinpolymorphism. Nature Genet. 10: 486–488, 1995.
- [45517] 19069.Kelsey, G. D.; Abeliovich, D.; McMahon, C. J.; Whitehouse, D.;Corney, G.; Povey, S.; Hopkinson, D. A.; Wolfe, J.; Mieli–Vergani,G.; Mowat, A. P.: Cloning of the human alpha–1 antichymotrypsin geneand genetic analysis of the gene in relation to alpha–1 antitrypsindeficiency. J. Med. Genet. 25: 361–368, 1988.
- [45518] 19070.Morgan, K.; Licastro, F.; Tilley, L.; Ritchie, A.; Morgan, L.;Pedrini, S.; Kalsheker, N.: Polymorphism in the alpha–1–antichymotrypsin(ACT) gene promoter: effect on

expression in transfected glial and liver cell lines and plasma ACT concentrations. Hum. Genet. 109:303–310, 2001.

- [45519] 19071. Morgan, K.; Morgan, L.; Carpenter, K.; Lowe, J.; Lam, L.; Cave, S.; Xuereb, J.; Wischik, C.; Harrington, C.; Kalsheker, N. A.: Microsatellite polymorphism of the alpha-1-antichymotrypsin gene locus associated with sporadic Alzheimer's disease. Hum. Genet. 99: 27–31, 1997.
- [45520] 19072. Munoz, E.; Obach, V.; Oliva, R.; Marti, M. J.; Ezquerro, M.; Pastor, P.; Ballesta, F.; Tolosa, E.: Alpha-1-antichymotrypsin gene polymorphism and susceptibility to Parkinson's disease. Neurology 52: 297–301, 1999.
- [45521] 19073. Poller, W.; Faber, J.-P.; Scholz, S.; Weidinger, S.; Bartholome, K.; Olek, K.; Eriksson, S.: Mis-sense mutation of alpha-1-antichymotrypsin gene associated with chronic lung disease. (Letter) Lancet 339:1538, 1992.
- [45522] 19074. Poller, W.; Faber, J.-P.; Weidinger, S.; Tief, K.; Scholz, S.; Fischer, M.; Olek, K.; Kirchgesser, M.; Heidtmann, H.-H.: A leucine-to-proline substitution causes a defective alpha-1-antichymotrypsin allele associated with familial obstructive lung disease. Genomics 17: 740–743, 1993.
- [45523] 19075. Rabin, M.; Watson, M.; Breg, W. R.; Kidd, V.; Woo, S.

L. C.; Ruddle, F. H.: Human alpha-1-antichymotrypsin and alpha-1-antitrypsin (PI) genes map to the same region on chromosome 14. (Abstract) Cytogenet. Cell Genet. 40: 728, 1985.

[45524] 19076. Rabin, M.; Watson, M.; Kidd, V.; Woo, S. L. C.; Breg, W. R.; Ruddle, F. H.: Regional location of alpha-1-antichymotrypsin and alpha-1-antitrypsin genes on human chromosome 14. Somat. Cell Molec. Genet. 12: 209-214, 1986.

[45525] 19077. Han, P.; Fletcher, C. F.; Copeland, N. G.; Jenkins, N. A.; Yaremko, L. M.; Michaeli, T.: Assignment of the mouse Pde7A gene to the proximal region of chromosome 3 and of the human PDE7A gene to chromosome 8q13. Genomics 48:275-276, 1998.

[45526] 19078. Michaeli, T.; Bloom, T. J.; Martins, T.; Loughney, K.; Ferguson, K.; Riggs, M.; Rodgers, L.; Beavo, J. A.; Wigler, M.: Isolation and characterization of a previously undetected human cAMP phosphodiesterase by complementation of cAMP phosphodiesterase-deficient *Saccharomyces cerevisiae*. J. Biol. Chem. 268: 12925-12932, 1993.

[45527] 19079. Milatovich, A.; Bolger, G.; Michaeli, T.; Francke, U.: Chromosomal localizations of genes for five cAMP-specific

phosphodiesterases in man and mouse. *Somat. Cell Molec. Genet.* 20: 75–86, 1994.

[45528] 19080. Loughney, K.; Martins, T. J.; Harris, E. A. S.; Sadhu, K.; Hicks, J. B.; Sonnenburg, W. K.; Beavo, J. A.; Ferguson, K.: Isolation and characterization of cDNAs corresponding to two human calcium, calmodulin-regulated, 3'-prime, 5'-prime-cyclic nucleotide phosphodiesterases. *J. Biol. Chem.* 271:796–806, 1996.

[45529] 19081. Wilson, D. E.; McKenna, L.: Assignment of the human gene for phosphodiesterase 1A to chromosome 4. (Abstract) *Am. J. Hum. Genet.* 43: A162 only, 1988.

[45530] 19082. Ritter, J. K.; Chen, F.; Sheen, Y. Y.; Tran, H. M.; Kimura, S.; Yeatman, M. T.; Owens, I. S.: A novel complex locus UGT1 encodes human bilirubin, phenol, and other UDP-glucuronosyltransferase isozymes with identical carboxyl termini. *J. Biol. Chem.* 267: 3257–3261, 1992.

[45531] 19083. Tukey, R. H.; Strassburg, C. P.: Human UDP-glucuronosyltransferases: metabolism, expression, and disease. *Annu. Rev. Pharm. Toxicol.* 40:581–616, 2000.

[45532] 19084. James, P. F.; Grupp, I. L.; Grupp, G.; Woo, A. L.; Askew, G. R.; Croyle, M. L.; Walsh, R. A.; Lingrel, J. B.: Identification of a specific role for the Na,K-ATPase alpha-2 isoform as a regulator of calcium in the heart. *Molec. Cell*

3: 555–563, 1999.

- [45533] 19085.Kawakami, K.; Ohta, T.; Nojima, H.; Nagano, K.: Primary structure of the alpha-subunit of human Na,K-ATPase deduced from cDNA sequence. *J.Biochem.* 100: 389–397, 1986.
- [45534] 19086.Kent, R. B.; Fallows, D. A.; Geissler, E.; Glaser, T.; Emanuel, J. R.; Lalley, P. A.; Levenson, R.; Housman, D. E.: Genes encoding alpha and beta subunits of Na,K-ATPase are located on three different chromosomes in the mouse. *Proc. Nat. Acad. Sci.* 84: 5369–5373, 1987.
- [45535] 19087.Ruiz-Opazo, N.; Barany, F.; Hirayama, K.; Herrera, V. L. M.: Confirmation of mutant alpha-1 Na,K-ATPase gene and transcript in Dahl salt-sensitive/JR rats. *Hypertension* 24: 260–270, 1994.
- [45536] 19088.Ovchinnikov, Y. A.; Monastyrskaya, G. S.; Broude, N. E.; Allikmets, R. L.; Ushkaryov, Y. A.; Melkov, A. M.; Smirnov, Y. V.; Malyshev, I. V.; Dulubova, I. E.; Petrukhin, K. E.; Gryshin, A. V.; Sverdlov, V. E.; Kiyatkin, N. I.; Kostina, M. B.; Modyanov, N. N.; Sverdlov, E. D.: The family of human Na⁺,K⁺-ATPase genes: a partial nucleotide sequence related to the alpha-subunit. *FEBS Lett.* 213: 73–80, 1987.
- [45537] 19089.Shull, M. M.; Lingrel, J. B.: Multiple genes encode the human Na⁺,K⁺-ATPase catalytic subunit. *Proc. Nat.*

Acad. Sci. 84: 4039–4043, 1987.

- [45538] 19090. Sverdlov, E. D.; Broude, N. E.; Sverdlov, V. E.; Monastyrskaya, G. S.; Grishin, A. V.; Petrukhin, K. E.; Akopyanz, N. S.; Modyanov, N. N.; Ovchinnikov, Y. A.: Family of Na⁺, K⁺–ATPase genes: intra-individual tissue-specific restriction fragment length polymorphism. FEBS Lett. 221:129–133, 1987.
- [45539] 19091. Yang–Feng, T. L.; Schneider, J. W.; Lindgren, V.; Shull, M. M.; Benz, E. J., Jr.; Lingrel, J. B.; Francke, U.: Chromosomal localization of human Na⁺, K⁺–ATPase alpha- and beta-subunit genes. Genomics 2:128–138, 1988.
- [45540] 19092. Gloor, S.; Antonicek, H.; Sweadner, K. J.; Pagliusi, S.; Frank, R.; Moos, M.; Schachner, M.: The adhesion molecule on glia (AMOG) is a homologue of the beta subunit of the Na,K–ATPase. J. Cell Biol. 110:165–174, 1990.
- [45541] 19093. Hsieh, C.–L.; Cheng–Deutsch, A.; Gloor, S.; Schachner, M.; Francke, U.: Assignment of Amog (adhesion molecule on glia) gene to mouse chromosome 11 near Zfp–3 and Asgr–1,2 and to human chromosome 17. Somat. Cell Molec. Genet. 16: 401–405, 1990.
- [45542] 19094. Malo, D.; Schurr, E.; Levenson, R.; Gros, P.: Assignment of Na,K–ATPase beta–(2)–subunit gene (Atpb–2) to mouse chromosome 11. Genomics 6:697–699, 1990.

- [45543] 19095.Martin–Vasallo, P.; Dackowski, P.; Emanuel, J. R.; Levenson, R.: Identification of a putative isoform of the Na,K–ATPase beta subunit:primary structure and tissue-specific expression. *J. Biol. Chem.* 164:4613–4618, 1989.
- [45544] 19096.Pagliusi, S.; Antonicek, H.; Gloor, S.; Frank, R.; Moos, M.; Schachner,M.: Identification of a cDNA clone specific for the neural adhesionmolecule AMOG. *J. Neu–rosce. Res.* 22: 113–119, 1989.
- [45545] 19097.Gallango, M. L.; Muller, A.; Suinaga, R.: Biochemical characterizationof a red cell UMP kinase variant found in the Warao Indians of Venezuela. *Biochem.Genet.* 16: 1085–1093, 1978.
- [45546] 19098.Gallango, M. L.; Suinaga, R.: Uridine monophos–phate kinase polymorphism in two Venezuelan popula–tions. *Am. J. Hum. Genet.* 30: 215–218, 1978.
- [45547] 19099.Giblett, E. R.; Anderson, J. E.; Chen, S.–H.; Teng, Y.–S.; Cohen,F.: Uridine monophosphate kinase: a new genetic polymorphism withpossible clinical implications. *Am. J. Hum. Genet.* 26: 627–635,1974.
- [45548] 19100.Giblett, E. R.; Anderson, J. E.; Lewis, M.; Kaita, H.: A new polymorphicezyme, uridine monophosphate kinase: gene frequencies and a linkageanalysis. *Birth Defects Orig. Art. Ser.* 11(3): 159–161, 1975. Note:Alternate: Cy–

togenet. Cell Genet. 14: 329–331, 1975.

- [45549] 19101. Medrano, L.; Green, H.: A uridine kinase deficient mutant of 3T3 and a selective method for cells containing the enzyme. Cell 1:23–26, 1974.
- [45550] 19102. Petersen, G. M.; Silimperi, D. R.; Scott, E. M.; Hall, D. B.; Rotter, J. I.; Ward, J. I.: Uridine monophosphate kinase 3: a genetic marker for susceptibility to Haemophilus influenzae type B disease. Lancet II:417–418, 1985.
- [45551] 19103. Ranzani, G.; Bertolotti, E.; Santachiara-Benerecetti, A. S.: The polymorphism of the red cell uridine monophosphate kinase in two samples of the Italian population. Hum. Hered. 27: 332–335, 1977.
- [45552] 19104. Ruddle, F. H.; Creagan, R. P.: Parasexual approaches to the genetics of man. Ann. Rev. Genet. 9: 407–486, 1975.
- [45553] 19105. Satlin, A.; Kucherlapati, R. S.; Ruddle, F. H.: Assignment of the gene for human uridine monophosphate kinase to chromosome 1 using somatic cell hybrid clone panels. Cytogenet. Cell Genet. 15: 146–152, 1975.
- [45554] 19106. Anderson, J. A.; Teng, Y.-S.; Giblett, E. R.: Stains for six enzymes potentially applicable to chromosomal assignment by cell hybridization. Cytogenet. Cell Genet. 14: 295–299, 1975.

- [45555] 19107.Hoglund, L.; Reichard, P. :J. Biol. Chem. 265: 6589–6595, 1990.
- [45556] 19108.Paglia, D. E.; Valentine, W. N.; Brockway, R. A.: Identification of thymidine nucleotidase and deoxyribonucleotidase activities among normal isozymes of 5–prime–nucleotidase in human erythrocytes. Proc.Nat. Acad. Sci. 81: 588–592, 1984.
- [45557] 19109.Rampazzo, C.; Gallinaro, L.; Milanesi, E.; Frigimelica, E.; Reichard,P.; Bianchi, V.: A deoxyribonucleotidase in mitochondria: involvement in regulation of dNTP pools and possible link to genetic disease. Proc.Nat. Acad. Sci. 97: 8239–8244, 2000.
- [45558] 19110.Rampazzo, C.; Johansson, M.; Gallinaro, L.; Ferraro, P.; Hellman,U.; Karlsson, A.; Reichard, P.; Bianchi, V.: Mammalian 5–prime(3–prime)–deoxyribonucleotidase, cDNA cloning, and overexpression of the enzyme in Escherichia coli and mammalian cells. J. Biol. Chem. 275: 5409–5415,2000.
- [45559] 19111.Swallow, D. M.; Turner, V. S.; Hopkinson, D. A.: Isozymes of rodent 5–prime–nucleotidase: evidence for two independent structural loci UMPH–1 and UMPH–2. Ann. Hum. Genet. 47: 9–17, 1983.
- [45560] 19112.Tjernshaugen, H.; Fritzson, P. :Biochem. J. 154:

77–80, 1976.

- [45561] 19113. Wilson, D. E.; Swallow, D. M.; Povey, S.: Assignment of the humangene for uridine 5–prime–monophosphate phosphohydrolase (UMPH2) to the long arm of chromosome 17. *Ann. Hum. Genet.* 50: 223–227, 1986.
- [45562] 19114. Wilson, D. E.; Woodard, D.; Sandler, A.; Erickson, J.; Gurney, A.: Provisional assignment of the gene for uridine monophosphatase–2 (Umph–2) to mouse chromosome 11. *Biochem. Genet.* 25: 1–6, 1987.
- [45563] 19115. Xiong, J.–P.; Stehle, T.; Diefenbach, B.; Zhang, R.; Dunker, R.; Scott, D. L.; Joachimiak, A.; Goodman, S. L.; Arnaout, M. A.: Crystal structure of the extracellular segment of integrin alpha–V–beta–3. *Science* 294:339–345, 2001.
- [45564] 19116. Xiong, J.–P.; Stehle, T.; Zhang, R.; Joachimiak, A.; Frech, M.; Goodman, S. L.; Arnaout, M. A.: Crystal structure of the extracellular segment of integrin alpha–V–beta–3 in complex with an Arg–Gly–Asp ligand. *Science* 296: 151–155, 2002.
- [45565] 19117. Disteche, C. M.; Plowman, G. D.; Gronwald, R. G. K.; Kelly, J.; Bowen–Pope, D.; Adler, D. A.; Murray, J. C.: Mapping of the amphiregulin and the platelet–growth factor receptor alpha genes to the proximal long arm of chromosome 4. (Abstract) *Cytogenet. Cell Genet.* 51:990,

1989.

- [45566] 19118.Gronwald, R. G. K.; Adler, D. A.; Kelly, J. D.; Distèche, C. M.; Bowen-Pope, D. F.: The human PDGF receptor alpha-subunit gene maps to chromosome 4 in close proximity to c-kit. *Hum. Genet.* 85: 383–385, 1990.
- [45567] 19119.Hol, F. A.; Geurds, M. P. A.; Chatkupt, S.; Shugart, Y. Y.; Balling, R.; Schrandt-Stumpel, C. T. R. M.; Johnson, W. G.; Hamel, B. C. J.; Mariman, E. C. M.: PAX genes and human neural tube defects: an amino acid substitution in PAX1 in a patient with spina bifida. *J. Med. Genet.* 8: 655–660, 1996.
- [45568] 19120.Hsieh, C.-L.; Navankasattusas, S.; Escobedo, J. A.; Williams, L.T.; Francke, U.: Chromosomal localization of the gene for AA-type platelet-derived growth factor receptor (PDGFRA) in humans and mice. *Cytogenet. Cell Genet.* 56: 160–163, 1991.
- [45569] 19121.Ikuno, Y.; Kazlauskas, A.: TGF-beta-1-dependent contraction of fibroblasts is mediated by the PDGF-alpha receptor. *Invest. Ophthalmol. Vis. Sci.* 43: 41–46, 2002.
- [45570] 19122.Joosten, P. H. L. J.; Hol, F. A.; van Beersum, S. E. C.; Peters, H.; Hamel, B. C. J.; Afink, G. B.; van Zoelen, E. J. J.; Mariman, E. C. M.: Altered regulation of platelet-derived growth factor receptor-alpha gene transcription in vitro by

spina bifida-associated mutant Pax1 proteins. Proc. Nat. Acad. Sci. 95: 14459–14463, 1998.

[45571] 19123.Sanke, T.; Bell, G. I.; Sample, C.; Rubenstein, A. H.; Steiner, D. F.: An islet amyloid peptide is derived from an 89-amino acid precursor by proteolytic processing. J. Biol. Chem. 263: 17243–17246, 1988.

[45572] 19124.Verchere, C. B.; D'Alessio, D. A.; Palmiter, R. D.; Weir, G. C.; Bonner-Weir, S.; Baskin, D. G.; Kahn, S. E.: Islet amyloid formation associated with hyperglycemia in transgenic mice with pancreatic beta cell expression of human islet amyloid polypeptide. Proc. Nat. Acad. Sci. 93: 3492–3496, 1996.

[45573] 19125.Westermarck, P.; Engstrom, U.; Johnson, K. H.; Westermarck, G. T.; Betsholtz, C.: Islet amyloid polypeptide: pinpointing amino acid residues linked to amyloid fibril formation. Proc. Nat. Acad. Sci. 87:5036–5040, 1990.

[45574] 19126.Lin, F.; Worman, H. J.: Structural organization of the human gene (LMNB1) encoding nuclear lamin B1. Genomics 27: 230–236, 1995.

[45575] 19127.Maeno, H.; Sugimoto, K.; Nakajima, N.: Genomic structure of the mouse gene (Lmnbl) encoding nuclear lamin B1. Genomics 30: 342–346, 1995.

[45576] 19128.Oklu, R.; Hesketh, R.: The latent transforming

growth factor betabinding protein (LTBP) family. *Biochem. J.* 352: 601–610, 2000.

[45577] 19129. Brodsky, G. L.; Muntoni, F.; Miodini, S.; Sinagra, G.; Sewry, C.; Mestroni, L.: Lamin A/C gene mutation associated with dilated cardiomyopathy with variable skeletal muscle involvement. *Circulation* 101: 473–476, 2000.

[45578] 19130. Cao, H.; Hegele, R. A.: Nuclear lamin A/C R482Q mutation in Canadian kindreds with Dunnigan-type familial partial lipodystrophy. *Hum. Molec. Genet.* 9: 109–112, 2000.

[45579] 19131. De Sandre-Giovannoli, A.; Chaouch, M.; Kozlov, S.; Vallat, J.-M.; Tazir, M.; Kassouri, N.; Szepietowski, P.; Hammadouch, T.; Vandenberghe, A.; Stewart, C. L.; Grid, D.; Levy, N.: Homozygous defects in LMNA, encoding lamin A/C nuclear-envelope proteins, cause autosomal recessive axonal neuropathy in human (Charcot-Marie-Tooth disorder type 2) and mouse. *Am. J. Hum. Genet.* 70: 726–736, 2002. Note: Erratum: *Am. J. Hum. Genet.* 70: 1075 only, 2002.

[45580] 19132. Fatkin, D.; MacRae, C.; Sasaki, T.; Wolff, M. R.; Porcu, M.; Frenneaux, M.; Atherton, J.; Vidaillet, H. J., Jr.; Spudich, S.; De Girolami, U.; Seidman, J. G.; Seidman, C. E.: Missense mutations in the rod domain of the lamin A/C

gene as causes of dilated cardiomyopathy and conduction-system disease. *New Eng. J. Med.* 341: 1715–1724, 1999.

- [45581] 19133. Fisher, D. Z.; Chaudhary, N.; Blobel, G.: cDNA sequencing of nuclear lamins A and C reveals primary and secondary structural homology to intermediate filament proteins. *Proc. Nat. Acad. Sci.* 83: 6450–6454, 1986.
- [45582] 19134. Flier, J. S.: Pushing the envelope on lipodystrophy. *Nature Genet.* 24:103–104, 2000.
- [45583] 19135. Garg, A.; Vainathar, M.; Weatherall, P. T.; Bowcock, A. M.: Phenotypic heterogeneity in patients with familial partial lipodystrophy (Dunnigan variety) related to the site of missense mutations in lamin A/C gene. *J. Clin. Endocr. Metab.* 86: 59–65, 2001.
- [45584] 19136. Genschel, J.; Schmidt, H. H.-J.: Mutations in the LMNA gene encoding lamin A/C. *Hum. Mutat.* 16: 451–459, 2000.
- [45585] 19137. Guilly, M. N.; Bensussan, A.; Bourge, J. F.; Bornens, M.; Courvalin, J. C.: A human T lymphoblastic cell line lacks lamins A and C. *EMBO J.* 6: 3795–3799, 1987.
- [45586] 19138. Hegele, R. A.; Cao, H.; Harris, S. B.; Zinman, B.; Hanley, A. J.; Anderson, C. M.: Genetic variation in LMNA modulates plasma leptin and indices of obesity in aboriginal Canadians. *Physiol. Genomics* 3:39–44, 2000.

- [45587] 19139.Hegele, R. A.; Cao, H.; Huff, M. W.; Anderson, C. M.: LMNA R482Qmutation in partial lipodystrophy associated with reduced plasma leptinconcentration. J. Clin. Endocr. Metab. 85: 3089–3093, 2000.
- [45588] 19140.Hegele, R. A.; Huff, M. W.; Young, T. K.: Common genomic variationin LMNA modulates indexes of obesity in Inuit. J. Clin. Endocr. Metab. 86:2747–2751, 2001.
- [45589] 19141.Krohne, G.; Benavente, R.: The nuclear lamins: a multigene familyof proteins in evolution and differentiation. Exp. Cell Res. 162:1–10, 1986.
- [45590] 19142.Lebel, S.; Raymond, Y.: Lamin A is not synthesized as a largerprecursor polypeptide. Biochem. Biophys. Res. Commun. 149: 417–423,1987.
- [45591] 19143.Lin, F.; Worman, H. J.: Structural organization of the humangene encoding nuclear lamin A and nuclear lamin C. J. Biol. Chem. 268:16321–16326, 1993.
- [45592] 19144.Lloyd, D. J.; Trembath, R. C.; Shackleton, S.: A novel interactionbetween lamin A and SREBP1: implications for partial lipodystrophyand other laminopathies. Hum. Molec. Genet. 11: 769–777, 2002.
- [45593] 19145.McKeon, F. D.; Kirschner, M. W.; Caput, D.: Homologies in bothprimary and secondary structure between nuclear envelope and intermediatefilament proteins. Na–

ture 319: 463–468, 1986.

- [45594] 19146. Muchir, A.; Bonne, G.; van der Kooi, A. J.; van Mee-
gen, M.; Baas, F.; Bolhuis, P. A.; de Visser, M.; Schwartz, K.:
Identification of mutations in the gene encoding lamins A/
C in autosomal dominant limb girdle muscular dystrophy
with atrioventricular conduction disturbances (LGMD1B).
Hum. Molec. Genet. 9: 1453–1459, 2000.
- [45595] 19147. Novelli, G.; Muchir, A.; Sangiulio, F.; Helbling-
Leclerc, A.; D'Apice, M. R.; Massart, C.; Capon, F.; Sbraccia,
P.; Federici, M.; Lauro, R.; Tudisco, C.; Pallotta, R.; Scarano,
G.; Dallapiccola, B.; Merlini, L.; Bonne, G.: Mandibuloacral
dysplasia is caused by a mutation in LMNA—encoding lamin
A/C. Am. J. Hum. Genet. 71: 426–431, 2002.
- [45596] 19148. Raffaele di Barletta, M.; Ricci, E.; Galluzzi, G.;
Tonali, P.; Mora, M.; Morandi, L.; Romorini, A.; Voit, T.;
Orstavik, K. H.; Merlini, L.; Trevisan, C.; Biancalana, V.;
Housmanowa-Petrusewicz, I.; Bione, S.; Ricotti, R.;
Schwartz, K.; Bonne, G.; Toniolo, D.: Different mutations in
the LMNA gene cause autosomal dominant and autosomal
recessive Emery–Dreifuss muscular dystrophy. Am. J.
Hum. Genet. 66: 1407–1412, 2000.
- [45597] 19149. Schmidt, H. H.-J.; Genschel, J.; Baier, P.; Schmidt,
M.; Ockenga, J.; Tietge, U. J. F.; Propsting, M.; Buttner, C.;

Manns, M. P.; Lochs, H.; Brabant, G.: Dyslipemia in familial partial lipodystrophy caused by and R482W mutation in the LMNA gene. *J. Clin. Endocr. Metab.* 86:2289–2295, 2001.

[45598] 19150. Shackleton, S.; Lloyd, D. J.; Jackson, S. N. J.; Evans, R.; Niermeijer, M. F.; Singh, B. M.; Schmidt, H.; Brabant, G.; Kumar, S.; Durrington, P. N.; Gregory, S.; O'Rahilly, S.; Trembath, R. C.: LMNA, encoding lamin A/C, is mutated in partial lipodystrophy. *Nature Genet.* 24:153–156, 2000.

[45599] 19151. Wydner, K. L.; McNeil, J. A.; Lin, F.; Worman, H. J.; Lawrence, J. B.: Chromosomal assignment of human nuclear envelope protein genes LMNA, LMNB1, and LBR by fluorescence in situ hybridization. *Genomics* 32:474–478, 1996.

[45600] 19152. Furukawa, K.; Hotta, Y.: cDNA cloning of a germ cell specific lamin B3 from mouse spermatocytes and analysis of its function by ectopic expression in somatic cells. *EMBO J.* 12: 97–106, 1993.

[45601] 19153. Furukawa, K.; Inagaki, H.; Hotta, Y.: Identification and cloning of an mRNA coding for a germ cell-specific A-type lamin in mice. *Exp. Cell Res.* 212: 426–430, 1994.

[45602] 19154. Garbarz, M.; Devaux, I.; Bournier, O.; Grandchamp, B.; Dhermy, D.: Protein 4.1 Lille, a novel mutation in the downstream initiation codon of protein 4.1 gene associ-

ated with heterozygous 4,1(-) hereditary elliptocytosis.

Hum. Mutat. 5: 339–340, 1995.

- [45603] 19155. Garbarz, M.; Dhermy, D.; Lecomte, M. C.; Feo, C.; Chaveroche, I.; Galand, C.; Bournier, O.; Bertrand, O.; Boivin, P.: A variant of erythrocyte membrane skeletal protein band 4.1 associated with hereditary elliptocytosis. Blood 64: 1006–1015, 1984.
- [45604] 19156. Geerdink, R. A.; Nijenhuis, L. E.; Huizinga, J.: Hereditary elliptocytosis: linkage data in man. Ann. Hum. Genet. 30: 363–378, 1967.
- [45605] 19157. Jensson, O.; Jonasson, T.; Olafsson, O.: Hereditary elliptocytosis in Iceland. Brit. J. Haemat. 13: 844–854, 1967.
- [45606] 19158. Kan, Y.-W.: Personal Communication. San Francisco, Calif. 2/28/1986.
- [45607] 19159. Kuroda, S.; Takeuchi, T.; Nagamori, H.: Data on the linkage between elliptocytosis and Rh blood type. Jpn. J. Hum. Genet. 5: 112–118, 1960.
- [45608] 19160. Lambert, S.; Conboy, J.; Zail, S.: A molecular study of heterozygous protein 4.1 deficiency in hereditary elliptocytosis. Blood 72: 1926–1929, 1988.
- [45609] 19161. Lambert, S.; Zail, S.: Partial deficiency of protein 4.1 in hereditary elliptocytosis. Am. J. Hemat. 26:

263–272, 1987.

[45610] 19162.Lipton, E. L.: Elliptocytosis with hemolytic anemia: the effectsof splenectomy. *Pediatrics* 15: 67–82, 1955.

[45611] 19163.Lux, S. E.; Wolfe, L. C.: Inherited disorders of the red cellmembrane skeleton. *Pediat. Clin. N. Am.* 27: 463–486, 1980.

[45612] 19164.Marchesi, S. L.; Conboy, J.; Agre, P.; Letsinger, J. T.; Marchesi,V. T.; Speicher, D. W.; Mohandas, N.: Molecular analysis of insertion/deletionmutations in protein 4.1 in elliptocytosis. I. Biochemical identificationof rearrange-ments in the spectrin/actin binding domain and function-alcharacterizations. *J. Clin. Invest.* 86: 516–523, 1990.

[45613] 19165.McGuire, M.; Agre, P.: Three distinct variants of protein 4.1in Caucasian hereditary elliptocytosis. (Abstract) *Clin. Res.* 35:428A, 1987.

[45614] 19166.McGuire, M.; Smith, B. L.; Agre, P.: Distinct variants of erythrocyteprotein 4.1 inherited in linkage with ellipto-cytosis and Rh type inthree white families. *Blood* 72: 287–293, 1988.

[45615] 19167.Morle, L.; Garbarz, M.; Alloisio, N.; Girot, R.; Chaveroche, I.;Boivin, P.; Delaunay, J.: The characterization of protein 4.1 Presles,a shortened variant of RBC mem-brane protein 4.1. *Blood* 65: 1511–1517,1985.

- [45616] 19168.Morle, L.; Pothier, B.; Alloisio, N.; Ducluzeau, M.-T.; Marques,S.; Olim, G.; Martins e Silva, J.; Feo, C.; Garbarz, M.; Chaver Roche,I.; Boivin, P.; Delaunay, J.: Red cell membrane alteration involving protein 4.1 and protein 3 in a case of recessively inherited haemolytic anemia. *Europ. J. Haemat.* 38: 447–455, 1987.
- [45617] 19169.Morton, N. E.: The detection and estimation of linkage between the genes for elliptocytosis and the Rh blood type. *Am. J. Hum. Genet.* 8:80–96, 1956.
- [45618] 19170.Nielsen, J. A.; Strunk, K. W.: Homozygous hereditary elliptocytosis as the cause of haemolytic anemia in infancy. *Scand. J. Haemat.* 5:486–496, 1968.
- [45619] 19171.Parra, M.; Gascard, P.; Walensky, L. D.; Snyder, S. H.; Mohandas,N.; Conboy, J. G.: Cloning and characterization of 4.1G (EPB41L2), a new member of the skeletal protein 4.1 (EPB41) gene family. *Genomics* 49:298–306, 1998.
- [45620] 19172.Peters, J. C.; Rowland, M.; Israels, L. G.; Zipursky, A.: Erythrocyte sodium transport in hereditary elliptocytosis. *Canad. J. Physiol. Pharm.* 44: 817–827, 1966.
- [45621] 19173.Roberts, J. A. F.: Genetic linkage in man, with particular reference to the usefulness of very small bodies of data. *Quart. J. Med.* 14:27–33, 1945.

- [45622] 19174. Shi, Z.-T.; Afzal, V.; Coller, B.; Patel, D.; Chasis, J. A.; Parra, M.; Lee, G.; Paszty, C.; Stevens, M.; Walensky, L.; Peters, L. L.; Mohandas, N.; Rubin, E.; Conboy, J. G.: Protein 4.1R-deficient mice are viable but have erythroid membrane skeleton abnormalities. *J. Clin. Invest.* 103: 331–340, 1999.
- [45623] 19175. Takakuwa, Y.; Tchernia, G.; Rossi, M.; Benabadji, M.; Mohandas, N.: Restoration of normal membrane stability to unstable protein 4.1-deficient erythrocyte membranes by incorporation of purified protein 4.1. *J. Clin. Invest.* 78: 80–85, 1986.
- [45624] 19176. Tang, C.-J. C.; Tang, T. K.: Rapid localization of membrane skeletal protein 4.1 (EL1) to human chromosome 1p33–p34.2 by nonradioactive in situ hybridization. *Cytogenet. Cell Genet.* 57: 119, 1991.
- [45625] 19177. Tang, T. K.; Leto, T. L.; Correia, I.; Alonso, M. A.; Marchesi, V. T.; Benz, E. J., Jr.: Selective expression of an erythroid-specific isoform of protein 4.1. *Proc. Nat. Acad. Sci.* 85: 3713–3717, 1988.
- [45626] 19178. Tchernia, G.; Mohandas, N.; Shohet, S. B.: Deficiency of skeletal membrane protein band 4.1 in homozygous hereditary elliptocytosis: implications for erythrocyte membrane stability. *J. Clin. Invest.* 68: 454–460, 1981.

- [45627] 19179.Tomaselli, M. B.; John, K. M.; Lux, S. E.: Elliptical erythrocytemembrane skeletons and heat-sensitive spectrin in hereditary elliptocytosis. *Proc.Nat. Acad. Sci.* 78: 1911–1915, 1981.
- [45628] 19180.Brands, J. H. G. M.; Maassen, J. A.; Van Hemert, F. J.; Amons,R.; Moller, W.: The primary structure of the alpha subunit of humanelongation factor 1: structural aspects of guanine–nucleotide–bindingsites. *Europ. J. Biochem.* 155: 167–171, 1986.
- [45629] 19181.Ditzel, H. J.; Masaki, Y.; Nielsen, H.; Farnaes, L.; Burton, D.R.: Cloning and expression of a novel human antibody--antigen pairassociated with Felty's syndrome. *Proc. Nat. Acad. Sci.* 97: 9234–9239,2000.
- [45630] 19182.Lund, A.; Knudsen, S. M.; Vissing, H.; Clark, B.; Tommerup, N.: Assignment of human elongation factor 1-alpha genes: EEF1A mapsto chromosome 6q14 and EEF1A2 to 20q13.3. *Genomics* 36: 359–361,1996.
- [45631] 19183.Opdenakker, G.; Cabeza-Arvelaiz, Y.; Fiten, P.; Dijkmans, R.; VanDamme, J.; Volckaert, G.; Billiau, A.; Van Elsen, A.; Van der Schueren,B.; Van den Berghe, H.; Cassiman, J.-J.: Human elongation factor1-alpha: a polymorphic and conserved multigene family with multiplechromosomal localizations. *Hum. Genet.* 75: 339–344, 1987.

- [45632] 19184.Cohen, A. J.; Li, F. P.; Berg, S.; Marchetto, D. J.; Tsai, S.; Jacobs, S. C.; Brown, R. S.: Hereditary renal-cell carcinoma associated with chromosomal translocation. *New Eng. J. Med.* 301: 592–595, 1979.
- [45633] 19185.Gemmill, R. M.; West, J. D.; Boldog, F.; Tanaka, N.; Robinson, L. J.; Smith, D. I.; Li, F.; Drabkin, H. A.: The hereditary renal cell carcinoma 3;8 translocation fuses FHIT to a patched-related gene, TRC8. *Proc. Nat. Acad. Sci.* 95: 9572–9577, 1998.
- [45634] 19186.Nakano, A.; Pulkkinen, L.; Murrell, D.; Rico, J.; Lucky, A. W.; Garzon, M.; Stevens, C. A.; Robertson, S.; Pfendner, E.; Uitto, J.: Epidermolysis bullosa with congenital pyloric atresia: novel mutations in the beta-4 integrin gene (ITGB4) and genotype/phenotype correlations. *Pediatr. Res.* 49: 618–626, 2001.
- [45635] 19187.Pulkkinen, L.; Bruckner-Tuderman, L.; August, C.; Uitto, J.: Compound heterozygosity for missense mutation (L156P) and nonsense (R554X) mutations in the beta-4 integrin gene (ITGB4) underlies mild, nonlethal phenotype of epidermolysis bullosa with pyloric atresia. *Am. J. Path.* 152:935–941, 1998.
- [45636] 19188.Pulkkinen, L.; Rouan, F.; Bruckner-Tuderman, L.; Wallerstein, R.; Garzon, M.; Brown, T.; Smith, L.; Carter, W.;

Uitto, J.: Novel ITGB4 mutations in lethal and nonlethal variants of epidermolysis bullosa with pyloric atresia: missense versus nonsense. *Am. J. Hum. Genet.* 63:1376–1387, 1998.

- [45637] 19189. Shaw, L. M.; Rabinovitz, I.; Wang, H. H.-F.; Toker, A.; Mercurio, A. M.: Activation of phosphoinositide 3-OH kinase by the alpha-6/beta-4 integrin promotes carcinoma invasion. *Cell* 91: 949–960, 1997.
- [45638] 19190. Suzuki, S.; Naitoh, Y.: Amino acid sequence of a novel integrin beta-4 subunit and primary expression of the mRNA in epithelial cells. *EMBO J.* 9: 757–763, 1990.
- [45639] 19191. Vidal, F.; Aberdam, D.; Miquel, C.; Christiano, A. M.; Pulkkinen, L.; Uitto, J.; Ortonne, J.-P.; Meneguzzi, G.: Integrin beta-4 mutations associated with junctional epidermolysis bullosa with pyloric atresia. *Nature Genet.* 10: 229–234, 1995.
- [45640] 19192. Krissansen, G. W.; Yuan, Q.; Jenkins, D.; Jiang, W.-M.; Rooke, L.; Spurr, N. K.; Eccles, M.; Leung, E.; Watson, J. D.: Chromosomal locations of the genes coding for the integrin beta-6 and beta-7 subunits. *Immunogenetics* 35: 58–61, 1992.
- [45641] 19193. Baker, E.; Sutherland, G. R.; Jiang, W.-M.; Yuan, Q.; Leung, E.; Watson, J. D.; Krissansen, G. W.: Mapping of the

human integrin beta-7 gene (ITG-beta-7) to 12q13.13 by non-isotopic in situ hybridization. *Mammalian Genome* 2: 272-273, 1992.

- [45642] 19194. Erle, D. J.; Ruegg, C.; Sheppard, D.; Pytela, R.: Complete amino acid sequence of an integrin beta subunit (beta-7) identified in leukocytes. *J. Biol. Chem.* 266: 11009-11016, 1991.
- [45643] 19195. Yuan, Q.; Kozak, C. A.; Jiang, W.; Hollander, D.; Watson, J. D.; Krissansen, G. W.: Genetic mapping of the gene coding for the integrin beta-7 subunit to the distal part of mouse chromosome 15. *Immunogenetics* 35:403-407, 1992.
- [45644] 19196. Chany, C.; Vignal, M.; Couillin, P.; Van Cong, N.; Boue, J.; Boue, A.: Chromosomal localization of human genes governing the interferon-induced antiviral state. *Proc. Nat. Acad. Sci.* 72: 3129-3133, 1975.
- [45645] 19197. Dithmar, S.; Rusciano, D.; Lynn, M. J.; Lawson, D. H.; Armstrong, C. A.; Grossniklaus, H. E.: Neoadjuvant interferon alpha-2b treatment in a murine model for metastatic ocular melanoma: a preliminary study. *Arch. Ophthalmol.* 118: 1085-1089, 2000.
- [45646] 19198. Ezekowitz, R. A. B.; Mulliken, J. B.; Folkman, J.: Interferon alpha-2a therapy for life-threatening hemangiomas

of infancy. *NewEng. J. Med.* 326: 1456–1463, 1992.

[45647] 19199.Hejny, C.; Sternberg, P., Jr.; Lawson, D. H.; Greiner, K.; Aaberg,T. M., Jr.: Retinopathy associated with high-dose interferon alfa-2btherapy. *Am. J. Ophthal.* 131: 782–787, 2001.

[45648] 19200.Aplin, H. M.; Hirst, K. L.; Crosby, A. H.; Dixon, M. J.: Mappingof the human dentin matrix acidic phosphoprotein gene (DMP1) to thedentinogenesis imperfecta type II critical region at chromosome 4q21. *Genomics* 30:347–349, 1995.

[45649] 19201.Ho, A. S. Y.; Liu, Y.; Khan, T. A.; Hsu, D.-H.; Bazan, J. F.; Moore,K. W.: A receptor for interleukin 10 is related to interferon receptors. *Proc.Nat. Acad. Sci.* 90: 11267–11271, 1993.

[45650] 19202.Liu, Y.; Wei, S. H.-Y.; Ho, A. S.-Y.; de Waal Malefyt, R.; Moore,K. W.: Expression cloning and characterization of a human Il-10 receptor. *J.Immun.* 152: 1821–1829, 1994.

[45651] 19203.Blixt, A.; Mahlapuu, M.; Aitola, M.; Pelto-Huikko, M.; Enerback,S.; Carlsson, P.: A forkhead gene, FoxE3, is essential for lens epithelialproliferation and closure of the lens vesicle. *Genes Dev.* 14: 245–254,2000.

[45652] 19204.Semina, E. V.; Brownell, I.; Mintz-Hittner, H. A.;

Murray, J. C.; Jamrich, M.: Mutations in the human fork-head transcription factor FOXE3 associated with anterior segment ocular dysgenesis and cataracts. Hum.Molec. Genet. 10: 231–236, 2001.

[45653] 19205. Siegelmann–Danieli, N.; Buetow, K. H.: Constitutional genetic variation at the human aromatase gene (Cyp19) and breast cancer risk. Brit.J. Cancer 79: 456–463, 1999.

[45654] 19206. Simpson, E. R.; Michael, M. D.; Agarwal, V. R.; Hinchelwood, M.M.; Bulun, S. E.; Zhao, Y.: Expression of the CYP19 (aromatase) gene: an unusual case of alternative promoter usage. FASEB J. 11: 29–36, 1997.

[45655] 19207. Sparkes, R. S.; Mohandas, T.; Chen, S.; Besman, M. J.; Zollman, S.; Shively, J. E.: Assignment of the aromatase gene to human chromosome 15q21. (Abstract) Cytogenet. Cell Genet. 46: 696–697, 1987.

[45656] 19208. Toda, K.; Merashima, M.; Kawamoto, T.; Sumimoto, H.; Yokoyama, Y.; Kuribayashi, I.; Mitsuuchi, Y.; Maeda, T.; Yamamoto, Y.; Sagara, Y.; Ikeda, H.; Shizuta, Y.: Structural and functional characterization of human aromatase P-450 gene. Europ. J. Biochem. 193: 559–565, 1990.

[45657] 19209. Wang, Z. J.; Jeffs, B.; Ito, M.; Achermann, J. C.; Yu, R. N.; Hales, D. B.; Jameson, J. L.: Aromatase (Cyp19) ex-

pression is up-regulated by targeted disruption of Dax1.

Proc. Nat. Acad. Sci. 98: 7988–7993, 2001.

[45658] 19210. Whitlock, J. P., Jr.: The regulation of cytochrome P-450 gene expression. Annu. Rev. Pharm. Toxicol. 26: 333–369, 1986.

[45659] 19211. Zhou, D.; Pompon, D.; Chen, S.: Structure–function studies of human aromatase by site-directed mutagenesis: kinetic properties of mutants pro308-to-phe, tyr361-to-phe, tyr361-to-leu, and phe406-to-arg. Proc. Nat. Acad. Sci. 88: 410–414, 1991.

[45660] 19212. Bruneau, G.; Gross, M.-S.; Krieger, M.; Bernheim, A.; Thibault, J.; Nguyen, V. C.: Preparation of a human DOPA decarboxylase cDNA probe by PCR and its assignment to chromosome 7. Ann. Genet. 33: 208–213, 1990.

[45661] 19213. Christenson, J. G.; Dairman, W.; Udenfriend, S.: On the identity of DOPA decarboxylase and 5-hydroxytryptophan decarboxylase (immunological titration–aromatic L-amino acid decarboxylase–serotonin–dopamine–norepinephrine). Proc. Nat. Acad. Sci. 69: 343–347, 1972.

[45662] 19214. Craig, S. P.; Le Van Thai, A.; Weber, M.; Craig, I. W.: Localisation of the gene for human aromatic L-amino acid decarboxylase (DDC) to chromosome 7p13–p11 by in situ

hybridisation. Cytogenet. Cell Genet. 61:114–116, 1992.

- [45663] 19215. Ichinose, H.; Kurosawa, Y.; Titani, K.; Fujita, K.; Nagatsu, T.: Isolation and characterization of a cDNA clone encoding human aromatic L-amino acid decarboxylase. Biochem. Biophys. Res. Commun. 164:1024–1030, 1989.
- [45664] 19216. Scherer, L. J.; McPherson, J. D.; Wasmuth, J. J.; Marsh, J. L.: Human dopa decarboxylase: localization to human chromosome 7p11 and characterization of hepatic cDNAs. Genomics 13: 469–471, 1992.
- [45665] 19217. Sumi-Ichinose, C.; Ichinose, H.; Takahashi, E.; Hori, T.; Nagatsu, T.: Molecular cloning of genomic DNA and chromosomal assignment of the gene for human aromatic L-amino acid decarboxylase, the enzyme for catecholamine and serotonin biosynthesis. Biochemistry 31:2229–2238, 1992.
- [45666] 19218. Luttrell, L. M.; Ferguson, S. S. G.; Daaka, Y.; Miller, W. E.; Maudsley, S.; Della Rocca, G. J.; Lin, F.-T.; Kawakatsu, H.; Owada, K.; Luttrell, D. K.; Caron, M. G.; Lefkowitz, R. J.: Beta-arrestin-dependent formation of beta-2 adrenergic receptor–Src protein kinase complexes. Science 283:655–661, 1999.
- [45667] 19219. Attramadal, H.; Arriza, J. L.; Aoki, C.; Dawson, T. M.; Codina, J.; Kwatra, M. M.; Snyder, S. H.; Caron, M. G.;

Lefkowitz, R. J.:Beta-arrestin-2, a novel member of the arrestin/beta-arrestin gene family. *J. Biol. Chem.* 267: 17882–17890, 1992.

- [45668] 19220.Bohn, L. M.; Gainetdinov, R. R.; Lin, F.-T.; Lefkowitz, R. J.;Caron, M. G.: Mu-opioid receptor desensitization by beta-arrestin-2determines morphine tolerance but not dependence. *Nature* 408: 720–723,2000.
- [45669] 19221.Bohn, L. M.; Lefkowitz, R. J.; Gainetdinov, R. R.; Peppel, K.;Caron, M. G.; Lin, F.-T.: Enhanced morphine analgesia in mice lackingbeta-arrestin 2. *Science* 286: 2495–2498, 1999.
- [45670] 19222.Fong, F. M.; Premont, R. T.; Richardson, R. M.; Yu, Y.-R. A.; Lefkowitz,R. J.; Patel, D. D.: Defective lymphocyte chemotaxis in beta-arrestin2-and GRK6-deficient mice. *Proc. Nat. Acad. Sci.* 99: 7478–7483, 2002.
- [45671] 19223.Kiselev, A.; Socolich, M.; Vinos, J.; Hardy, R. W.; Zuker, C. S.;Ranganathan, R.: A molecular pathway for light-dependent photoreceptorapoptosis in *Drosophila*. *Neuron* 28: 139–152, 2000.
- [45672] 19224.McDonald, P. H.; Chow, C.-W.; Miller, W. E.; Laporte, S. A.; Field,M. E.; Lin, F.-T.; Davis, R. J.; Lefkowitz, R. J.: Beta-arrestin2: a receptor-regulated MAPK scaffold for the activation of JNK3. *Science* 290:1574–1577, 2000.

- [45673] 19225.Karathanasis, S. K.; Oettgen, P.; Haddad, I. A.; Antonarakis, S. E.: Structure, evolution, and polymorphisms of the human apolipoproteinA4 gene (APOA4). *Proc. Nat. Acad. Sci.* 83: 8457–8461, 1986.
- [45674] 19226.Carrington, M.; Kissner, T.; Gerrard, B.; Ivanov, S.; O'Brien, S. J.; Dean, M.: Novel alleles of the chemokine–receptor gene CCR5. *Am.J. Hum. Genet.* 61: 1261–1267, 1997.
- [45675] 19227.Chaganti, R. S. K.: Personal Communication. New York, N. Y. 10/22/1993.
- [45676] 19228.Chaudhuri, A.; Polyakova, J.; Zbrzezna, V .; Pogo, A. O.: The coding sequence of Duffy blood group gene in humans and simians: restriction fragment length polymorphism, antibody and malarial parasite specificities, and expression in nonerythroid tissues in Duffy–negative individuals. *Blood* 85:615–621, 1995.
- [45677] 19229.Chaudhuri, A.; Polyakova, J.; Zbrzezna, V.; Williams, K.; Gulati, S.; Pogo, A. O.: Cloning of glycoprotein D cDNA, which encodes the major subunit of the Duffy blood group system and the receptor for the *Plasmodium vivax* malaria parasite. *Proc. Nat. Acad. Sci.* 90:10793–10797, 1993.
- [45678] 19230.Cook, P. J. L.; Page, B. M.; Johnston, A. W.; Stan–

ford, W. K.;Gavin, J.: Four further families informative for 1q and the Duffyblood group. Cytogenet. Cell Genet. 22: 378–380, 1978.

[45679] 19231.Cook, P. J. L.; Robson, E. B.; Buckton, K. E.; Jacobs, P. A.; Polani,P. E.: Segregation of genetic markers in families with chromosomepolymorphisms and structural rearrangements involving chromosome no.1. Ann. Hum. Genet. 37: 261–274, 1974.

[45680] 19232.Crawford, M. N.; Punnett, H. H.; Carpenter, G. G.: Deletion ofthe long arm of chromosome 16 and an unexpected Duffy blood groupphenotype reveal a possible autosomal linkage. Nature 215: 1075–1076,1967.

[45681] 19233.Gelpi, A. P.; King, M. C.: Association of Duffy blood groupswith the sickle cell trait. Hum. Genet. 32: 65–68, 1976.

[45682] 19234.Hadley, T. J.; David, P. H.; McGinniss, M. H.; Miller, L. H.:Identification of an erythrocyte component carrying the Duffy bloodgroup Fy–a antigen. Science 223: 597–599, 1984.

[45683] 19235.Hadley, T. J.; Peiper, S. C.: From malaria to chemokine receptor:the emerging physiologic role of the Duffy blood group antigen. Blood 89:3077–3091, 1997.

[45684] 19236.Hamblin, M. T.; Di Rienzo, A.: Detection of the sig–

nature of natural selection in humans: evidence from the Duffy blood group locus. *Am.J. Hum. Genet.* 66: 1669–1679, 2000.

- [45685] 19237. Hamblin, M. T.; Thompson, E. E.; Di Rienzo, A.: Complex signatures of natural selection at the Duffy blood group locus. *Am. J. Hum. Genet.* 70: 369–383, 2002.
- [45686] 19238. Hessner, M. J.; Pircon, R. A.; Johnson, S. T.; Luhm, R. A.: Prenatal genotyping of the Duffy blood group system by allele-specific polymerase chain reaction. *Prenatal Diag.* 19: 41–45, 1999.
- [45687] 19239. Horuk, R.; Chitnis, C. E.; Darbonne, W. C.; Colby, T. J.; Rybicki, A.; Hadley, T. J.; Miller, L. H.: A receptor for the malarial parasite *Plasmodium vivax*: the erythrocyte chemokine receptor. *Science* 261: 1182–1184, 1993.
- [45688] 19240. Howard, P. N.; Stoddard, G. R.; Goddard, M. W.; Seely, J. R.: Giemsa banding of chromosome 1qh+ and linkage analysis. *J. Med. Genet.* 12: 44–48, 1975.
- [45689] 19241. Iwamoto, S.; Li, J.; Sugimoto, N.; Okuda, H.; Kajii, E.: Characterization of the Duffy gene promoter: evidence for tissue-specific abolishment of expression in Fy(a–b–) of black individuals. *Biochem. Biophys. Res. Commun.* 222: 852–859, 1996.
- [45690] 19242. Lautenberger, J. A.; Stephens, J. C.; O'Brien, S. J.;

Smith, M.W.: Significant admixture linkage disequilibrium across 30 cM around the FY locus in African Americans. *Am. J. Hum. Genet.* 66: 969–978, 2000.

[45691] 19243. Lee, C. S. N.; Ying, K. L.; Bowen, P.: Position of the Duffy locus on chromosome 1 in relation to breakpoints for structural rearrangements. *Am. J. Hum. Genet.* 26: 93–102, 1974.

[45692] 19244. Li, J.; Iwamoto, S.; Sugimoto, N.; Okuda, H.; Kajii, E.: Dinucleotide repeat in the 3-prime flanking region provides a clue to the molecular evolution of the Duffy gene. *Hum. Genet.* 99: 573–577, 1997.

[45693] 19245. Braun, A.; Kammerer, S.; Bohme, E.; Muller, B.; Roscher, A. A.: Identification of polymorphic sites of the human bradykinin B(2) receptor gene. *Biochem. Biophys. Res. Commun.* 211: 234–240, 1995.

[45694] 19246. Erdmann, J.; Hegemann, N.; Weidemann, A.; Kallisch, H.; Hummel, M.; Hetzer, R.; Fleck, E.; Regitz-Zagrosek, V.: Screening the human bradykinin B2 receptor gene in patients with cardiovascular diseases: identification of a functional mutation in the promoter and a new coding variant (T21M). *Am. J. Med. Genet.* 80: 521–525, 1998.

[45695] 19247. Hess, J. F.; Borkowski, J. A.; Young, G. S.; Strader,

C. D.; Ransom, R. W.: Cloning and pharmacological characterization of a human bradykinin(BK-2) receptor. *Biochem. Biophys. Res. Commun.* 184: 260–268, 1992.

[45696] 19248. Kammerer, S.; Braun, A.; Arnold, N.; Roscher, A. A.: The human bradykinin B(2) receptor gene: full length cDNA, genomic organization and identification of the regulatory region. *Biochem. Biophys. Res. Commun.* 211: 226–233, 1995.

[45697] 19249. Ma, J.; Wang, D.; Ward, D. C.; Chen, L.; Dessai, T.; Chao, J.; Chao, L.: Structure and chromosomal localization of the gene (BDKRB2) encoding human bradykinin B2 receptor. *Genomics* 23: 362–369, 1994.

[45698] 19250. Powell, S. J.; Slynn, G.; Thomas, C.; Hopkins, B.; Briggs, I.; Graham, A.: Human bradykinin B2 receptor: nucleotide sequence analysis and assignment to chromosome 14. *Genomics* 15: 435–438, 1993.

[45699] 19251. Taketo, M.; Yokoyama, S.; Rochelle, J.; Kimura, S.; Higashida, H.; Taketo, M.; Seldin, M. F.: Mouse B2 bradykinin receptor gene maps to distal chromosome 12. *Genomics* 27: 222–223, 1995.

[45700] 19252. Chen, H.; Weber, A. J.: BDNF enhances retinal ganglion cell survival in cats with optic nerve damage. *Invest. Ophthalm. Vis. Sci.* 42:966–974, 2001.

- [45701] 19253. Conover, J. C.; Erickson, J. T.; Katz, D. M.; Bianchi, L. M.; Poueymirou, W. T.; McClain, J.; Pan, L.; Helgren, M.; Ip, N. Y.; Boland, P.; Friedman, B.; Wiegand, S.; Vejsada, R.; Kato, A. C.; DeChiara, T. M.; Yancopoulos, G. D.: Neuronal deficits, not involving motor neurons, in mice lacking BDNF and/or NT4. *Nature* 375: 235–238, 1995.
- [45702] 19254. Hanson, I. M.; Seawright, A.; van Heyningen, V.: The human BDNF gene maps between FSHB and HVBS1 at the boundary of 11p13–p14. *Genomics* 13:1331–1333, 1992.
- [45703] 19255. Hofer, M. M.; Barde, Y.-A.: Brain-derived neurotrophic factor prevents neuronal death in vivo. *Nature* 331: 261–262, 1988.
- [45704] 19256. Huang, Z. J.; Kirkwood, A.; Pizzorusso, T.; Porciatti, V.; Morales, B.; Bear, M. F.; Maffei, L.; Tonegawa, S.: BDNF regulates the maturation of inhibition and the critical period of plasticity in mouse visual cortex. *Cell* 98: 739–755, 1999.
- [45705] 19257. Kovalchuk, Y.; Hanse, E.; Kafitz, K. W.; Konnerth, A.: Postsynaptic induction of BDNF-mediated long-term potentiation. *Science* 295:1729–1734, 2002.
- [45706] 19258. Liu, X.; Ernfors, P.; Wu, H.; Jaenisch, R.: Sensory but not motor neuron deficits in mice lacking NT4 and BDNF.

Nature 375: 238–241,1995.

- [45707] 19259.Lu, Q.; Sun, E. E.; Klein, R. S.; Flanagan, J. G.: Ephrin–B reversesignaling is mediated by a novel PDZ–RGS protein and selectively inhibitsG protein–coupled chemoattraction. Cell 105: 69–79, 2001.
- [45708] 19260.Masi, L.; Becherini, L.; Colli, E.; Gennari, L.; Mansani, R.; Falchetti,A.; Becorpi, A. M.; Cepollaro, C.; Gonnelli, S.; Tanini, A.; Brandi,M. L.: Polymorphisms of the calcitonin receptor gene are associatedwith bone mineral density in postmenopausal Italian women. Biochem.Biophys. Res. Commun. 248: 190–195, 1998.
- [45709] 19261.Masi, L.; Becherini, L.; Gennari, L.; Colli, E.; Mansani, R.; Falchetti,A.; Cepollaro, C.; Gonnelli, S.; Tanini, A.; Brandi, M. L.: Allelicvariants of human calci–tonin receptor: distribution and associationwith bone mass in postmenopausal Italian women. Biochem. Bio–phys.Res. Commun. 245: 622–626, 1998.
- [45710] 19262.Nakamura, M.; Zhang, Z. Q.; Shan, L.; Hisa, T.; Sasaki, M.; Tsukino,R.; Yokoi, T.; Kaname, A.; Kakudo, K.: Allelic variants of humancalcitonin receptor in the Japanese population. Hum. Genet. 99:38–41, 1997.
- [45711] 19263.Perez Jurado, L. A.; Li, X.; Francke, U.: The human calcitoninreceptor gene (CALCR) at 7q21.3 is outside the

deletion associated with the Williams syndrome. Cytogenet. Cell Genet. 70: 246–249, 1995.

- [45712] 19264. Taboulet, J.; Frenco, J. L.; Delage-Murroux, R.; Pichaud, F.; de Vernejoul, M. C.; Jullienne, A.: Evidence for 2 allelic forms of calcitonin receptor gene: distribution in normal and osteoporotic women. (Abstract) J. Bone Miner. Res. 11 (suppl. 1): S204, 1996.
- [45713] 19265. Taboulet, J.; Frenkian, M.; Frenco, J. L.; Feingold, N.; Jullienne, A.; de Vernejoul, M. C.: Calcitonin receptor polymorphism is associated with a decreased fracture risk in post-menopausal women. Hum. Molec. Genet. 7: 2129–2133, 1998.
- [45714] 19266. Amara, S. G.; Arriza, J. L.; Leff, S. E.; Swanson, L. W.; Evans, R. M.; Rosenfeld, M. G.: Expression in brain of a messenger RNA encoding a novel neuropeptide homologous to calcitonin gene-related peptide. Science 229: 1094–1097, 1985.
- [45715] 19267. Hoppener, J. W. M.; Steenbergh, P. H.; Zandberg, J.; Geurts van Kessel, A. H. M.; Baylin, S. B.; Nelkin, B. D.; Jansz, H. S.; Lips, C. J. M.: The second human calcitonin/CGRP gene is located on chromosome 11. Hum. Genet. 70: 259–263, 1985.
- [45716] 19268. Steenbergh, P. H.; Hoppener, J. W. M.; Zandberg, J.;

Lips, C. J.M.; Jansz, H. S.: A second human calcitonin/CGRP gene. *FEBS Lett.* 183:403–407, 1985.

[45717] 19269.Arthur, J. S. C.; Elce, J. S.; Hegadorn, C.; Williams, K.; Greer, P. A.: Disruption of the murine calpain small subunit gene, *Capn4*:calpain is essential for embryonic development but not for cell growth and division. *Molec. Cell. Biol.* 20: 4474–4481, 2000.

[45718] 19270.Ohno, S.; Emori, Y.; Suzuki, K.: Nucleotide sequence of a cDNA coding for the small subunit of human calcium-dependent protease. *Nucleic Acids Res.* 14: 5559 only, 1986.

[45719] 19271.Ohno, S.; Minoshima, S.; Kudoh, J.; Fukuyama, R.; Ohmi-Imajoh, S.; Suzuki, K.; Shimizu, Y.; Shimizu, N.: Four genes for the calpain family locate on four distinct human chromosomes. (Abstract) *Cytogenet. Cell Genet.* 51: 1054–1055, 1989.

[45720] 19272.Ohno, S.; Minoshima, S.; Kudoh, J.; Fukuyama, R.; Shimizu, Y.; Ohmi-Imajoh, S.; Shimizu, N.; Suzuki, K.: Four genes for the calpain family locate on four distinct human chromosomes. *Cytogenet. Cell Genet.* 53: 225–229, 1990.

[45721] 19273.Sakihama, T.; Kakidani, H.; Zenita, K.; Yumoto, N.; Kikuchi, T.; Sasaki, T.; Kannagi, R.; Nakanishi, S.; Ohmori, M.; Takio, K.; Titani, K.; Murachi, T.: A putative

Ca(2+)-binding protein: structure of the light subunit of porcine calpain elucidated by molecular cloning and protein sequence analysis. *Proc. Nat. Acad. Sci.* 82: 6075-6079, 1985.

- [45722] 19274. Berchtold, M. W.; Egli, R.; Rhyner, J. A.; Hameister, H.; Strehler, E. E.: Localization of the human bona fide calmodulin genes CALM1, CALM2, and CALM3 to chromosomes 14q24-q31, 2p21.1-p21.3, and 19q13.2-q13.3. *Genomics* 16:461-465, 1993.
- [45723] 19275. Chin, D.; Winkler, K. E.; Means, A. R.: Characterization of substrate phosphorylation and use of calmodulin mutants to address implications from the enzyme crystal structure of calmodulin-dependent protein kinase I. *J. Biol. Chem.* 272: 31235-31240, 1997.
- [45724] 19276. Drum, C. L.; Yan, S.-Z.; Bard, J.; Shen, Y.-Q.; Lu, D.; Soelaiman, S.; Grabarek, Z.; Bohm, A.; Tang, W.-J.: Structural basis for the activation of anthrax adenyl cyclase exotoxin by calmodulin. *Nature* 415:396-402, 2002.
- [45725] 19277. Koller, M.; Schnyder, B.; Strehler, E. E.: Structural organization of the human CaMIII calmodulin gene. *Biochim. Biophys. Acta* 1087:180-189, 1990.
- [45726] 19278. Kretsinger, R. H.; Rudnick, S. E.; Weissman, L. J.: Crystal structure of calmodulin. *J. Inorganic Biochem.* 28:

289–302, 1986.

- [45727] 19279. McPherson, J. D.; Hickie, R. A.; Wasmuth, J. J.; Meyskens, F. L.; Perham, R. N.; Strehler, E. E.; Graham, M. T.: Chromosomal localization of multiple genes encoding calmodulin. (Abstract) *Cytogenet. Cell Genet.* 58: 1951 only, 1991.
- [45728] 19280. Pegues, J. C.; Friedberg, F.: Multiple mRNAs encoding human calmodulin. *Biochem. Biophys. Res. Commun.* 172: 1145–1149, 1990.
- [45729] 19281. Rhyner, J. A.; Ottiger, M.; Wicki, R.; Greenwood, T. M.; Strehler, E. E.: Structure of the human CALM1 calmodulin gene and identification of two CALM1-related pseudogenes CALM1P1 and CALM1P2. *Europ. J. Biochem.* 225: 71–82, 1994.
- [45730] 19282. Scambler, P. J.; McPherson, M. A.; Bates, G.; Bradbury, N. A.; Dormer, R. L.; Williamson, R.: Biochemical and genetic exclusion of calmodulin as the site of the basic defect in cystic fibrosis. *Hum. Genet.* 76: 278–282, 1987.
- [45731] 19283. Schumacher, M. A.; Rivard, A. F.; Bachinger, H. P.; Adelman, J. P.: Structure of the gating domain of a $\text{Ca}(2+)$ -activated K^+ channel complexed with $\text{Ca}(2+)/$ calmodulin. *Nature* 410: 1120–1124, 2001.
- [45732] 19284. Sen Gupta, B.; Detera-Wadleigh, S. D.; McBride, O.

W.; Friedberg, F.: A calmodulin pseudogene on human chromosome 17. *Nucleic Acids Res.* 17: 2868 only, 1989.

[45733] 19285. Powers, P. A.; Liu, S.; Hogan, K.; Gregg, R. G.: Molecular characterization of the gene encoding the gamma subunit of the human skeletal muscle 1,4-dihydropyridine-sensitive Ca^{2+} channel (CACNLG), cDNA sequence, gene structure, and chromosomal location. *J. Biol. Chem.* 268: 9275–9279, 1993.

[45734] 19286. Ambartsumian, N.; Tarabykina, S.; Grigorian, M.; Tulchinsky, E.; Hulgaard, E.; Georgiev, G.; Lukanidin, E.: Characterization of two splice variants of metastasis-associated human mts1 gene. *Gene* 159:125–130, 1995.

[45735] 19287. Jackson-Grusby, L. L.; Swiergiel, J.; Linzer, D. I.: A growth-related mRNA in cultured mouse cells encodes a placental calcium binding protein. *Nucleic Acids Res.* 15: 6677–6690, 1987.

[45736] 19288. Iles, D. E.; Segers, B.; Weghuis, D. O.; Suikerbuijk, R.; Wieringa, B.: Localization of the gamma-subunit of the skeletal muscle L-type voltage-dependent calcium channel gene (CACNLG) to human chromosome band 17q24 by in situ hybridization and identification of a polymorphic repetitive DNA sequence at the gene locus. *Cytogenet. Cell Genet.* 64:227–230, 1993.

- [45737] 19289.Saris, C. J.; Kristensen, T.; D'Eustachio, P.; Hicks, L. J.; Noonan,D. J.; Hunter, T.; Tack, B. F.: cDNA sequence and tissue distributionof the mRNA for bovine and murine p11, the S100-related light chainof the protein-tyrosine kinase substrate p36 (calpactin I). *J. Biol.Chem.* 262: 10663–10671, 1987.
- [45738] 19290.Lefort, A.; Lecocq, R.; Libert, F.; Lamy, F.; Swillens, S.; Vassart,G.; Dumont, J. E.: Cloning and sequencing of a calcium-binding proteinregulated by cyclic AMP in the thyroid. *EMBO J.* 8: 111–116, 1989.
- [45739] 19291.Loughlin, J.; Irven, C.; Sykes, B.: Exclusion of the cartilagelink protein and the cartilage matrix protein genes as the mutantloci in several heritable chondrodysplasias. *Hum. Genet.* 94: 698–700,1994.
- [45740] 19292.Osborne-Lawrence, S. L.; Sinclair, A. K.; Hicks, R. C.; Lacey,S. W.; Eddy, R. L., Jr.; Byers, M. G.; Shows, T. B.; Duby, A. D.:Complete amino acid sequence of human cartilage link protein (CRTL1)deduced from cDNA clones and chromosomal assignment of the gene. *Genomics* 8:562–567, 1990.
- [45741] 19293.Watanabe, H.; Yamada, Y.: Mice lacking link protein develop dwarfismand craniofacial abnormalities. *Nature Genet.* 21: 225–229, 1999.

- [45742] 19294.Deak, F.; Piecha, D.; Bachrati, C.; Paulsson, M.; Kiss, I.: Primary structure and expression of matrilin-2, the closest relative of cartilage matrix protein within the von Willebrand factor type A-like module superfamily. *J. Biol. Chem.* 272: 9268–9274, 1997.
- [45743] 19295.Deak, F.; Wagener, R.; Kiss, I.; Paulsson, M.: The matrilins: a novel family of oligomeric extracellular matrix proteins. *Matrix Biol.* 18: 55–64, 1999.
- [45744] 19296.Hansson, A.-S.; Heinegard, D.; Holmdahl, R.: A new animal model for relapsing polychondritis, induced by cartilage matrix protein (matrilin-1). *J. Clin. Invest.* 104: 589–598, 1999.
- [45745] 19297.Jenkins, R. N.; Osborne-Lawrence, S. L.; Sinclair, A. K.; Eddy, R. L., Jr.; Byers, M. G.; Shows, T. B.; Duby, A. D.: Structure and chromosomal location of the human gene encoding cartilage matrix protein. *J. Biol. Chem.* 265: 19624–19631, 1990.
- [45746] 19298.Lang, B.; Rothenfusser, A.; Lanchbury, J. S.; Rauh, G.; Breedveld, F. C.; Urlacher, A.; Albert, E. D.; Peter, H.-H.; Melchers, I.: Susceptibility to relapsing polychondritis is associated with HLA-DR4. *Arthritis Rheum.* 36: 660–664, 1993.
- [45747] 19299.Muratoglu, S.; Krysan, K.; Balazs, M.; Sheng, H.;

Zakany, R.; Modis, L.; Kiss, I.; Deak, F.: Primary structure of human matrilin-2, chromosomal location of the MATN2 gene and conservation of an AT-AC intron in matrilin genes. *Cytogenet. Cell Genet.* 90: 323-327, 2000.

[45748] 19300. Wagener, R.; Kobbe, B.; Paulsson, M.: Primary structure of matrilin-3, a new member of a family of extracellular matrix proteins related to cartilage matrix protein (matrilin-1) and von Willebrand factor. *FEBS Lett.* 413: 129-134, 1997.

[45749] 19301. Zeuner, M.; Straub, R. H.; Rauh, G.; Albert, E. D.; Scholmerich, J.; Lang, B.: Relapsing polychondritis: clinical and immunogenetic analysis of 62 patients. *J. Rheum.* 24: 96-101, 1997.

[45750] 19302. Boldyreff, B.; Klett, C.; Gottert, E.; Geurts van Kessel, A.; Hameister, H.; Issinger, O.-G.: Assignment of casein kinase 2 alpha sequence to two different human chromosomes. *Hum. Genet.* 89: 79-82, 1992.

[45751] 19303. Doray, B.; Ghosh, P.; Griffith, J.; Geuze, H. J.; Kornfeld, S.: Cooperation of GGAs and AP-1 in packaging MPRs at the trans-Golgi network. *Science* 297: 1700-1703, 2002.

[45752] 19304. Meisner, H.; Heller-Harrison, R.; Buxton, J.; Czech, M. P.: Molecular cloning of the human casein kinase II al-

pha subunit. *Biochemistry* 28:4072–4076, 1989.

[45753] 19305. Wirkner, U.; Voss, H.; Lichter, P.; Ansorge, W.; Pyerin, W.: The human gene (CSNK2A1) coding for the casein kinase II subunit alpha is located on chromosome 20 and contains tandemly arranged Alu repeats. *Genomics* 19:257–265, 1994.

[45754] 19306. Wirkner, U.; Voss, H.; Lichter, P.; Weitz, S.; Ansorge, W.; Pyerin, W.: Human casein kinase II subunit alpha: sequence of a processed (pseudo) gene and its localization on chromosome 11. *Biochim. Biophys. Acta* 1131: 220–222, 1992.

[45755] 19307. Yang-Feng, T. L.; Zheng, K.; Kopatz, I.; Naiman, T.; Canaani, D.: Mapping of the human casein kinase II catalytic subunit genes: two loci carrying the homologous sequences for the alpha subunit. *Nucleic Acids Res.* 19: 7125–7129, 1991.

[45756] 19308. Lozeman, F. J.; Litchfield, D. W.; Piening, C.; Takio, K.; Walsh, K. A.; Krebs, E. G.: Isolation and characterization of human cDNA clones encoding the alpha and the alpha-prime subunits of casein kinase II. *Biochemistry* 29: 8436–8447, 1990.

[45757] 19309. Xu, X.; Toselli, P. A.; Russell, L. D.; Seldin, D. C.: Globozoospermia in mice lacking the casein kinase II al-

pha-prime catalytic subunit. *Nature Genet.* 23: 118–121, 1999.

[45758] 19310. Abrieu, A.; Kahana, J. A.; Wood, K. W.; Cleveland, D. W.: CENP-E is an essential component of the mitotic checkpoint in vitro. *Cell* 102:817–826, 2000.

[45759] 19311. Wood, K. W.; Sakowicz, R.; Goldstein, L. S. B.; Cleveland, D. W.: CENP-E is a plus end-directed kinetochore motor required for metaphase chromosome alignment. *Cell* 97: 357–366, 1997.

[45760] 19312. Yao, X.; Abrieu, A.; Zheng, Y.; Sullivan, K. F.; Cleveland, D. W.: CENP-E forms a link between attachment of spindle microtubules to kinetochores and the mitotic checkpoint. *Nature Cell Biol.* 2:484–491, 2000.

[45761] 19313. Yen, T. J.; Compton, D. A.; Wise, D.; Zinkowski, R. P.; Brinkley, B. R.; Earnshaw, W. C.; Cleveland, D. W.: CENP-E, a novel human centromere-associated protein required for progression from metaphase to anaphase. *EMBO J.* 10: 1245–1254, 1991.

[45762] 19314. Yen, T. J.; Li, G.; Schaar, B. T.; Szilak, I.; Cleveland, D. W.: CENP-E is a putative kinetochore motor that accumulates just before mitosis. *Nature* 359: 536–539, 1992.

[45763] 19315. Geissler, E. N.; Liao, M.; Brook, J. D.; Martin, F. H.; Zsebo, K. M.; Housman, D. E.; Galli, S. J.: Stem cell factor

(SCF), a novel hematopoietic growth factor and ligand for c-kit tyrosine kinase receptor, maps on human chromosome 12 between 12q14.3 and 12qter. *Somat. Cell Molec. Genet.* 17: 207–214, 1991.

[45764] 19316. Heissig, B.; Hattori, K.; Dias, S.; Friedrich, M.; Ferris, B.; Hackett, N. R.; Crystal, R. G.; Besmer, P.; Lyden, D.; Moore, M. A. S.; Werb, Z.; Rafii, S.: Recruitment of stem and progenitor cells from the bone marrow niche requires MMP-9 mediated release of Kit-ligand. *Cell* 109:625–637, 2002.

[45765] 19317. Martin, F. H.; Suggs, S. V.; Langley, K. E.; Lu, H. S.; Ting, J.; Okino, K. H.; Morris, C. F.; McNiece, I. K.; Jacobsen, F. W.; Mendiaz, E. A.; Birkett, N. C.; Smith, K. A.; and 15 others: Primary structure and functional expression of rat and human stem cell factor DNAs. *Cell* 63:203–211, 1990.

[45766] 19318. Mathew, S.; Murty, V. V. V. S.; Hunziker, W.; Chaganti, R. S. K.: Subregional mapping of 13 single-copy genes on the long arm of chromosome 12 by fluorescence in situ hybridization. *Genomics* 14: 775–779, 1992.

[45767] 19319. Li, X.-Y.; Mattei, M. G.; Zaleska-Rutczynska, Z.; Hooft van Huijsduijnen, R.; Figueroa, F.; Nadeau, J.; Benoist, C.; Mathis, D.: One subunit of the transcription

factor NF- κ B maps close to the major histocompatibility-complex in murine and human chromosomes. *Genomics* 11: 630–634, 1991.

[45768] 19320. Johnston, J.; Bollekens, J.; Allen, R. H.; Berliner, N.: Structure of the cDNA encoding transcobalamin I, a neutrophil granule protein. *J. Biol. Chem.* 264: 15754–15757, 1989.

[45769] 19321. Johnston, J.; Yang-Feng, T.; Berliner, N.: Genomic structure and mapping of the chromosomal gene for transcobalamin I (TCN1): comparison to human intrinsic factor. *Genomics* 12: 459–464, 1992.

[45770] 19322. Yang-Feng, T. L.; Berliner, N.; Deverajan, P.; Johnston, J.: Assignment of two human neutrophil secondary granule protein genes, transcobalamin I and neutrophil collagenase to chromosome 11. (Abstract) *Cytogenet. Cell Genet.* 58: 1974 only, 1991.

[45771] 19323. Courey, A. J.; Holtzman, D. A.; Jackson, S. P.; Tjian, R.: Synergistic activation by the glutamine-rich domains of human transcription factor Sp1. *Cell* 59: 827–836, 1989.

[45772] 19324. Gaynor, R. B.; Shieh, B.-H.; Klisak, I.; Sparkes, R. S.; Lusk, A. J.: Localization of the transcription factor SP1 gene to human chromosome 12q12–q13.2. *Cytogenet. Cell Genet.* 64: 210–212, 1993.

- [45773] 19325.Kadonaga, J. T.; Carner, K. R.; Masiarz, F. R.; Tjian, R.: Isolation of a cDNA encoding transcription factor Sp1 and functional analysis of the DNA binding domain. *Cell* 51: 1079–1090, 1987.
- [45774] 19326.Matera, A. G.; Ward, D. C.: Localization of the human Sp1 transcription factor gene to 12q13 by fluorescence in situ hybridization. *Genomics* 17:793–794, 1993.
- [45775] 19327.Szpirer, J.; Szpirer, C.; Riviere, M.; Levan, G.; Marynen, P.; Cassiman, J.-J.; Wiese, R.; DeLuca, H. F.: The Sp1 transcription factor gene (SP1) and the 1,25-dihydroxyvitamin D(3) receptor gene (VDR) are colocalized on human chromosome arm 12q and rat chromosome
- [45776] 19328.*Genomics* 11: 168–173, 1991.7. Wimmer, E. A.; Jackle, H.; Pfeifle, C.; Cohen, S. M.: A *Drosophila* homologue of human Sp1 is a head-specific segmentation gene. *Nature* 366:690–694, 1993.
- [45777] 19329.Abbott, C.; Piaggio, G.; Ammendola, R.; Solomon, E.; Povey, S.; Gounari, F.; De Simone, V.; Cortese, R.: Mapping of the gene TCF2 for the transcription factor LFB3 to human chromosome 17 by polymerase chain reaction. *Genomics* 8: 165–167, 1990.
- [45778] 19330.Bach, I.; Mattei, M.-G.; Cereghini, S.; Yaniv, M.: Two

members of an HNF1 homeoprotein family are expressed in human liver. *Nucleic Acids Res.* 19: 3553–3559, 1991.

[45779] 19331. Bingham, C.; Ellard, S.; Allen, L.; Bulman, M.; Shepherd, M.; Frayling, T.; Berry, P. J.; Clark, P. M.; Lindner, T.; Bell, G. I.; Ryffel, G. U.; Nicholls, A. J.; Hattersley, A. T.: Abnormal nephron development associated with a frameshift mutation in the transcription factor hepatocyte nuclear factor-1-beta. *Kidney Int.* 57: 898–907, 2000.

[45780] 19332. Horikawa, Y.; Iwasaki, N.; Hara, M.; Furuta, H.; Hinokio, Y.; Cockburn, B. N.; Lindner, T.; Yamagata, K.; Ogata, M.; Tomonaga, O.; Kuroki, H.; Kasahara, T.; Iwamoto, Y.; Bell, G. I.: Mutation in hepatocyte nuclear factor-1-beta gene (TCF2) associated with MODY. (Letter) *Nature Genet.* 17: 384–385, 1997.

[45781] 19333. Menzel, R.; Kaisaki, P. J.; Rjasanowski, I.; Heinke, P.; Kerner, W.; Menzel, S.: A low renal threshold for glucose in diabetic patients with a mutation in the hepatocyte nuclear factor-1-alpha (HNF-1-alpha) gene. *Diabet. Med.* 15: 816–820, 1998.

[45782] 19334. Nishigori, H.; Yamada, S.; Kohama, T.; Tomura, H.; Sho, K.; Horikawa, Y.; Bell, G. I.; Takeuchi, T.; Takeda, J.: Frameshift mutation, A263fsinsGG, in the hepatocyte nuclear factor-1-beta gene associated with diabetes and re-

nal dysfunction. Diabetes 47: 1354–1355, 1998.

[45783] 19335. Wild, W.; Pogge von Strandmann, E.; Nastos, A.; Senkel, S.; Lingott-Frieg, A.; Bulman, M.; Bingham, C.; El-lard, S.; Hattersley, A. T.; Ryffel, G. U.: The mutated human gene encoding hepatocyte nuclear factor 1-beta inhibits kidney formation in developing *Xenopus* embryos.

Proc. Nat. Acad. Sci. 97: 4695–4700, 2000.

[45784] 19336. Aplan, P. D.; Jones, C. A.; Chervinsky, D. S.; Zhao, X.; Ellsworth, M.; Wu, C.; McGuire, E. A.; Gross, K. W.: An *scl* gene product lacking the transactivation domain induces bony abnormalities and cooperates with LMO1 to generate T-cell malignancies in transgenic mice. EMBO J. 16: 2408–2419, 1997.

[45785] 19337. Aplan, P. D.; Lombardi, D. P.; Ginsberg, A. M.; Cossman, J.; Bertness, V. L.; Kirsch, I. L.: Disruption of the human SCL locus by 'illegitimate' V-(D)-J recombinase activity. Science 250: 1426–1429, 1990.

[45786] 19338. Aplan, P. D.; Lombardi, D. P.; Reaman, G. H.; Sather, H. N.; Hammond, G. D.; Kirsch, I. R.: Involvement of the putative hematopoietic transcription factor SCL in T-cell acute lymphoblastic leukemia. Blood 79: 1327–1333, 1992.

[45787] 19339. Brown, L.; Cheng, J.-T.; Chen, Q.; Siciliano, M. J.;

Crist, W.;Buchanan, G.; Baer, R.: Site-specific recombination of the tal-1gene is a common occurrence in human T-cell leukemia. EMBO J. 9:3343–3351, 1990.

- [45788] 19340.Collazo-Garcia, N.; Scherer, P.; Aplan, P. D.: Cloning and characterization of a murine SIL gene. Genomics 30: 506–513, 1995.
- [45789] 19341.Izraeli, S.; Lowe, L. A.; Bertness, V. L.; Good, D. J.; Dorward, D. W.; Kirsch, I. R.; Kuehn, M. R.: The SIL gene is required for mouse embryonic axial development and left-right specification. Nature 399:691–694, 1999.
- [45790] 19342.Roy, A. L.; Du, H.; Gregor, P. D.; Novina, C. D.; Martinez, E.;Roeder, R. G.: Cloning of an Inr- and E-box binding protein, TFIID-I, that interacts physically and functionally with USF1. EMBO J. 16:7091–7104, 1997.
- [45791] 19343.Steingrimsson, E.; Sawadogo, M.; Gilbert, D. J.; Zervos, A. S.;Brent, R.; Blonar, M. A.; Fisher, D. E.; Copeland, N. G.; Jenkins, N. A.: Murine chromosomal location of five bHLH-Zip transcription factor genes. Genomics 28: 179–183, 1995.
- [45792] 19344.Zhong, G.; Fan, P.; Ji, H.; Dong, F.; Huang, Y.: Identification of a chlamydial protease-like activity factor responsible for the degradation of host transcription factors. J. Exp. Med. 193: 935–942, 2001.

- [45793] 19345.Aasland, R.; Olsen, L. C.; Spurr, N. K.; Krokan, H. E.; Helland, D. E.: Chromosomal assignment of human uracil–DNA glycosylase to chromosome 12. *Genomics* 7: 139–141, 1990.
- [45794] 19346.Dinner, A. R.; Blackburn, G. M.; Karplus, M.: Uracil–DNA glycosylase acts by substrate autocatalysis. *Nature* 413: 752–755, 2001.
- [45795] 19347.Haug, T.; Skorpen, F.; Kvaloy, K.; Eftedal, I.; Lund, H.; Krokan, H. E.: Human uracil–DNA glycosylase gene: sequence organization, methylation pattern, and mapping to chromosome 12q23–q24.1. *Genomics* 36:408–416, 1996.
- [45796] 19348.Haug, T.; Skorpen, F.; Lund, H.; Krokan, H. E.: Structure of the gene for human uracil–DNA glycosylase and analysis of the promoter function. *FEBS Lett.* 353: 180–184, 1994.
- [45797] 19349.Meyer–Sieglar, K.; Mauro, D. J.; Seal, G.; Wurzer, J.; deRiel, J. K.; Sirover, M. A.: A human nuclear uracil DNA glycosylase is the 37–kDa subunit of glyceraldehyde–3–phosphate dehydrogenase. *Proc. Nat. Acad. Sci.* 88: 8460–8464, 1991.
- [45798] 19350.Nilsen, H.; Rosewell, I.; Robins, P.; Skjelbred, C. F.; Andersen, S.; Slupphaug, G.; Daly, G.; Krokan, H. E.; Lindahl, T.; Barnes, D.E.: Uracil–DNA glycosylase

(UNG)-deficient mice reveal a primary role of the enzyme during DNA replication. *Molec. Cell* 5: 1059–1065, 2000.

[45799] 19351. Olsen, L. C.; Aasland, R.; Wittwer, C. U.; Krokan, H. E.; Helland, D. E.: Molecular cloning of human uracil-DNA glycosylase, a highly conserved DNA repair enzyme. *EMBO J.* 8: 3121–3125, 1989.

[45800] 19352. Vollberg, T. M.; Siegler, K. M.; Cool, B. L.; Sirover, M. A.: Isolation and characterization of the human uracil DNA glycosylase gene. *Proc. Nat. Acad. Sci.* 86: 8693–8697, 1989.

[45801] 19353. Momeni, P.; Glockner, G.; Schmidt, O.; von Holtum, D.; Albrecht, B.; Gillessen-Kaesbach, G.; Hennekam, R.; Meinecke, P.; Zabel, B.; Rosenthal, A.; Horsthemke, B.; Ludecke, H.-J.: Mutations in a new gene, encoding a zinc-finger protein, cause tricho-rhino-phalangeal syndrome type I. *Nature Genet.* 24: 71–74, 2000.

[45802] 19354. Komuro, I.; Wenninger, K. E.; Philipson, K. D.; Izumo, S.: Molecular cloning and characterization of the human cardiac Na(+)/Ca(2+) exchanger cDNA. *Proc. Nat. Acad. Sci.* 89: 4769–4773, 1992.

[45803] 19355. Kraev, A.; Chumakov, I.; Carafoli, E.: The organization of the human gene NCX1 encoding the sodium-calcium exchanger. *Genomics* 37: 105–112, 1996.

- [45804] 19356.McDaniel, L. D.; Lederer, W. J.; Kofuji, P.; Schulze, D. H.; Kieval,R.; Schultz, R. A.: Mapping of the human cardiac $\text{Na}^+/\text{Ca}^{2+}$ exchanger gene (NCX1) by fluorescent in situ hybridization to chromosome region 2p22–p23. *Cytogenet. Cell Genet.* 63: 192–193, 1993.
- [45805] 19357.Shieh, B.–H.; Xia, Y.; Sparkes, R. S.; Klisak, I.; Lysis, A. J.; Nicoll, D. A.; Philipson, K. D.: Mapping of the gene for the cardiac sarcolemmal $\text{Na}^+-\text{Ca}^{2+}$ exchanger to human chromosome 2p21–p23. *Genomics* 12:616–617, 1992.
- [45806] 19358.Wakimoto, K.; Kobayashi, K.; Kuro-o, M.; Yao, A.; Iwamoto, T.; Yanaka, N.; Kita, S.; Nishida, A.; Azuma, S.; Toyoda, Y.; Omori, K.; Imahie, H.; Oka, T.; Kudoh, S.; Kohmoto, O.; Yazaki, Y.; Shigekawa, M.; Imai, Y.; Nabeshima, Y.; Komura, I.: Targeted disruption of $\text{Na}^+/\text{Ca}^{2+}$ exchanger gene leads to cardiomyocyte apoptosis and defects in heartbeat. *J. Biol. Chem.* 275: 36991–36998, 2000.
- [45807] 19359.Chong, S. S.; Kristjansson, K.; Zoghbi, H. Y.; Hughes, M. R.: Molecular cloning of the cDNA encoding a human renal sodium phosphate transport protein and its assignment to chromosome 6p21.3–p23. *Genomics* 18:355–359, 1993.

- [45808] 19360.Kos, C. H.; Tihiy, F.; Murer, H.; Lemieux, N.; Tenenhouse, H. S.: Comparative mapping of Na(+)-phosphate cotransporter genes, NPT1 and NPT2, in human and rabbit. *Cytogenet. Cell Genet.* 75: 22–24, 1996.
- [45809] 19361.Zhang, X.-X.; Tenenhouse, H. S.; Hewson, A. S.; Eydoux, P.: Assignment of renal specific Na(+)-phosphate cotransporter gene Slc17a1 to mouse chromosome bands 13A3–A4 by in situ hybridization. *Cytogenet. Cell Genet.* 76: 180 only, 1997.
- [45810] 19362.Ghishan, F. K.; Knobel, S.; Dasuki, M.; Butler, M.; Phillips, J.: Chromosomal localization of the human renal sodium phosphate transporter to chromosome 5: implications for X-linked hypophosphatemia. *Pediatr. Res.* 35: 510–513, 1994.
- [45811] 19363.Hartmann, C. M.; Hewson, A. S.; Kos, C. H.; Hilfiker, H.; Soumounou, Y.; Murer, H.; Tenenhouse, H. S.: Structure of murine and human renal type II Na(+)-phosphate cotransporter genes (Npt2 and NPT2). *Proc. Nat. Acad. Sci.* 93: 7409–7414, 1996.
- [45812] 19364.Kos, C. H.; Tihiy, F.; Econs, M. J.; Murer, H.; Lemieux, N.; Tenenhouse, H. S.: Localization of a renal sodium-phosphate cotransporter gene to human chromosome 5q35. *Genomics* 19: 176–177, 1994.

- [45813] 19365.Magagnin, S.; Werner, A.; Markovich, D.; Sorribas, V.; Stange,G.; Biber, J.; Murer, H.: Expression cloning of human and rat renal cortex Na/Pi cotransport. *Proc. Nat. Acad. Sci.* 90: 5979–5983, 1993.
- [45814] 19366.McPherson, J. D.; Krane, M. C.; Wagner–McPherson, C. B.; Kos, C.H.; Tenenhouse, H. S.: High resolution mapping of the renal sodium–phosphate cotransporter gene (NPT2) confirms its localization to human chromosome 5q35. *Pediat. Res.* 41: 632–634, 1997.
- [45815] 19367.Tenenhouse, H. S.; Werner, A.; Biber, J.; Ma, S.; Martel, J.; Roy,S.; Murer, H.: Renal Na(+)-phosphate cotransport in murine X-linked hypophosphatemic rickets: molecular characterization. *J. Clin. Invest.* 93:671–676, 1994.
- [45816] 19368.Zhang, X.-X.; Tenenhouse, H. S.; Hewson, A. S.; Murer, H.; Eydoux,P.: Assignment of renal-specific Na(+)-phosphate cotransporter gene Slc17a2 to mouse chromosome band 13B by in situ hybridization. *Cytogenet.Cell Genet.* 77: 304–305, 1997.
- [45817] 19369.Barnard, R.; Kelly, G.; Manzetti, S. O.; Harris, E. L.: Neither the New Zealand genetically hypertensive strain nor Dahl salt-sensitive strain has an A1079T transversion in the alpha-1 isoform of the Na(+),K(+)-ATPase gene. *Hy-*

pertension 38: 786–792, 2001.

- [45818] 19370. Chehab, F. F.; Kan, Y. W.; Law, M. L.; Hartz, J.; Kao, F.-T.; Blostein, R.: Human placental Na⁺,K⁺-ATPase alpha subunit: cDNA cloning, tissue expression, DNA polymorphism, and chromosomal localization. *Proc. Nat. Acad. Sci.* 84: 7901–7905, 1987.
- [45819] 19371. Dahl, L. K.; Heine, M.; Tassinari, L.: Role of genetic factors in susceptibility to experimental hypertension due to chronic excess salt ingestion. *Nature* 194: 480–482, 1972.
- [45820] 19372. Dahl, L. K.; Heine, M.; Thompson, K.: Genetic influence of the kidneys on blood pressure: evidence from chronic renal homografts in rats with opposite predispositions to hypertension. *Circ. Res.* 40: 94–101, 1974.
- [45821] 19373. Glorioso, N.; Filigheddu, F.; Troffa, C.; Soro, A.; Parpaglia, P. P.; Tsikoudakis, A.; Myers, R. H.; Herrera, V. L. M.; Ruiz-Opazo, N.: Interaction of alpha-1-Na,K-ATPase and Na,K,2Cl-cotransporter genes in human essential hypertension. *Hypertension* 38: 204–209, 2001.
- [45822] 19374. Herrera, V. L. M.; Ruiz-Opazo, N.: Alteration of alpha-1 Na⁺,K⁽⁺⁾-ATPase 86R-beta(+) influx by a single amino acid substitution. *Science* 249: 1023–1026, 1990.
- [45823] 19375. Andersson, S.; Russell, D. W.: Structural and bio-

chemical properties of cloned and expressed human and rat steroid 5- α -reductases. *Proc. Nat. Acad. Sci.* 87: 3640-3644, 1990.

[45824] 19376. Ellis, J. A.; Stebbing, M.; Harrap, S. B.: Genetic analysis of male pattern baldness and the 5- α -reductase genes. *J. Invest. Derm.* 110: 849-853, 1998.

[45825] 19377. Harris, G.; Azzolina, B.; Baginsky, W.; Cimis, G.; Rasmusson, G.H.; Tolman, R. L.; Raetz, C. R. H.; Ellsworth, K.: Identification and selective inhibition of an isozyme of steroid 5- α -reductase in human scalp. *Proc. Nat. Acad. Sci.* 89: 10787-10791, 1992.

[45826] 19378. Hsieh, C.-L.; Milatovich, A.; Russell, D.; Francke, U.: Chromosomal mapping of human steroid 5 α -reductase gene (SRD5A1) and pseudogene (SRD5AP1) in human and mouse. (Abstract) *Cytogenet. Cell Genet.* 58:1897 only, 1991.

[45827] 19379. Jenkins, E. P.; Andersson, S.; Imperato-McGinley, J.; Wilson, J.D.; Russell, D. W.: Genetic and pharmacological evidence for more than one human steroid 5- α -reductase. *J. Clin. Invest.* 89: 293-300, 1992.

[45828] 19380. Jenkins, E. P.; Hsieh, C.-L.; Milatovich, A.; Normington, K.; Berman, D. M.; Francke, U.; Russell, D. W.: Characterization and chromosomal mapping of a human

steroid 5- α -reductase gene and pseudogene
and mapping of the mouse homologue. *Genomics* 11:
1102–1112, 1991.

[45829] 19381. Thigpen, A. E.; Silver, R. I.; Guileyardo, J. M.; Casey, M. L.; McConnell, J. D.; Russell, D. W.: Tissue distribution and ontogeny of steroid 5- α -reductase isozyme expression. *J. Clin. Invest.* 92:903–910, 1993.

[45830] 19382. He, Z.; Yamamoto, R.; Furth, E. E.; Schantz, L. J.; Naylor, S. L.; George, H.; Billheimer, J. T.; Strauss, J. F., III: cDNAs encoding members of a family of proteins related to human sterol carrier protein 2 and assignment of the gene to human chromosome 1p21-pter. *DNA Cell Biol.* 10: 559–569, 1991.

[45831] 19383. Vesa, J.; Hellsten, E.; Barnoski, B. L.; Emanuel, B. S.; Billheimer, J. T.; Mead, S.; Cowell, J. K.; Strauss, J. F., III; Peltonen, L.: Assignment of sterol carrier protein X/sterol carrier protein 2 to 1p32 and its exclusion as the causative gene for infantile neuronal ceroid lipofuscinosis. *Hum. Molec. Genet.* 3: 341–346, 1994.

[45832] 19384. Welch, C. L.; Xia, Y.-R.; Billheimer, J. T.; Strauss, J. F., III; Lusi, A. J.: Assignment of the mouse sterol carrier protein gene (Scp2) to chromosome 4. *Mammalian Genome* 7: 624–625, 1996.

- [45833] 19385.Yamamoto, R.; Kallen, C. B.; Babalola, G. O.; Ren-
nert, H.; Billheimer, J. T.; Strauss, J. F., III: Cloning and ex-
pression of a cDNA encoding human sterol carrier protein
2. *Proc. Nat. Acad. Sci.* 88: 463–467, 1991.
- [45834] 19386.Yamamoto, R.; Naylor, S. L.; George, H.; Billheimer,
J. T.; Strauss, J. F., III: Assignment of the gene encoding
sterol carrier protein 2 to human chromosome 1pter–p21.
(Abstract) *Cytogenet. Cell Genet.* 58:1866–1867, 1991.
- [45835] 19387.DeBose-Boyd, R. A.; Brown, M. S.; Li, W.-P.; No-
hturfft, A.; Goldstein, J. L.; Espenshade, P. J.: Transport-
dependent proteolysis of SREBP: relocation of Site-1 pro-
tease from Golgi to ER obviates the need for SREBP trans-
port to Golgi. *Cell* 99: 703–712, 1999.
- [45836] 19388.Hua, X.; Wu, J.; Goldstein, J. L.; Brown, M. S.;
Hobbs, H. H.: Structure of the human gene encoding sterol
regulatory element binding protein-1 (SREBF1) and local-
ization of SREBF1 and SREBF2 to chromosomes 17p11.2
and 22q13. *Genomics* 25: 667–673, 1995.
- [45837] 19389.Osborne, T. F.: CREating a SCAP-less liver keeps
SREBPs pinned in the ER membrane and prevents increased
lipid synthesis in response to low cholesterol and high in-
sulin. *Genes Dev.* 15: 1873–1878, 2001.
- [45838] 19390.Shimano, H.; Shimomura, I.; Hammer, R. E.; Herz,

J.; Goldstein, J. L.; Brown, M. S.; Horton, J. D.: Elevated levels of SREBP-2 and cholesterol synthesis in livers of mice homozygous for a targeted disruption of the SREBP-1 gene. *J. Clin. Invest.* 100: 2115–2124, 1997.

[45839] 19391. Shimomura, I.; Hammer, R. E.; Richardson, J. A.; Ikemoto, S.; Bashmakov, Y.; Goldstein, J. L.; Brown, M. S.: Insulin resistance and diabetes mellitus in transgenic mice expressing nuclear SREBP-1c in adipose tissue: model for congenital generalized lipodystrophy. *Genes Dev.* 12: 3182–3194, 1998.

[45840] 19392. Tobe, K.; Suzuki, R.; Aoyama, M.; Yamauchi, T.; Kamon, J.; Kubota, N.; Terauchi, Y.; Matsui, J.; Akanuma, Y.; Kimura, S.; Tanaka, J.; Abe, M.; Ohsumi, J.; Nagai, R.; Kadowaki, T.: Increased expression of the sterol regulatory element-binding protein-1 gene in insulin receptor substrate-2 $-/-$ mouse liver. *J. Biol. Chem.* 276: 38337–38340, 2001.

[45841] 19393. Wang, X.; Sato, R.; Brown, M. S.; Hua, X.; Goldstein, J. L.: SREBP-1, a membrane-bound transcription factor released by sterol-regulated proteolysis. *Cell* 77: 53–62, 1994.

[45842] 19394. Yokoyama, C.; Wang, X.; Briggs, M. R.; Admon, A.; Wu, J.; Hua, X.; Goldstein, J. L.; Brown, M. S.: SREBP-1, a

basic-helix-loop-helix-leucinezipper protein that controls transcription of the low density lipoproteinreceptor gene. Cell 75: 187–197, 1993.

- [45843] 19395.Joosten, P. H. L. J.; Toepoel, M.; Mariman, E. C. M.; Van Zoelen,E. J. J.: Promoter haplotype combinations of the platelet-derivedgrowth factor alpha-receptor gene predispose to human neural tubedefects. Nature Genet. 27: 215–217, 2001.
- [45844] 19396.Travis, G. H.; Sutcliffe, J. G.; Bok, D.: The retinal degenerationslow (rds) gene product is a photoreceptor disc membrane-associatedglycoprotein. Neuron 6: 61–70, 1991.
- [45845] 19397.Morris, C.; Heisterkamp, N.; Hao, Q. L.; Testa, J. R.; Groffen,J.: The human tyrosine kinase gene (FER) maps to chromosome 5 andis deleted in myeloid leukemias with a del(5q). Cytogenet. Cell Genet. 53:196–200, 1990.
- [45846] 19398.Herrera, V. L. M.; Xie, H. X.; Lopez, L. V.; Schork, N. J.; Ruiz-Opazo,N.: The alpha-1 Na,K-ATPase gene is a susceptibility hypertensiongene in the Dahl salt-sen-sitive-HSD rat. J. Clin. Invest. 102: 1102–1111,1998.
- [45847] 19399.Williams, T. M.; Montoya, G.; Wu, Y.; Eddy, R. L.; Byers, M. G.;Shows, T. B.: The TCF8 gene encoding a zinc finger protein (Nil-2-a)resides on human chromosome

10p11.2. Genomics 14: 194–196, 1992.

- [45848] 19400.van Nie, R.; Ivanyi, D.; Demant, P.: A new H-2-linked mutation, rds, causing retinal degeneration in the mouse. *Tissue Antigens* 12:106–108, 1978.
- [45849] 19401.Vine, A. K.; Schatz, H.: Adult-onset foveomacular pigment epithelial dystrophy. *Am. J. Ophthal.* 89: 680–691, 1980.
- [45850] 19402.Weleber, R. G.; Carr, R. E.; Murphey, W. H.; Sheffield, V. C.; Stone, E. M.: Phenotypic variation including retinitis pigmentosa, pattern dystrophy, and fundus flavimaculatus in a single family with a deletion of codon 153 or 154 of the peripherin/RDS gene. *Arch. Ophthal.* 111: 1531–1542, 1993.
- [45851] 19403.Wells, J.; Wroblewski, J.; Keen, J.; Inglehearn, C.; Jubb, C.; Eckstein, A.; Jay, M.; Arden, G.; Bhattacharya, S.; Fitzke, F.; Bird, A.: Mutations in the human retinal degeneration slow (RDS) gene can cause either retinitis pigmentosa or macular dystrophy. *Nature Genet.* 3:213–218, 1993.
- [45852] 19404.Wroblewski, J. J.; Wells, J. A., III; Eckstein, A.; Fitzke, F.; Jubb, C.; Keen, J.; Inglehearn, C.; Bhattacharya, S.; Arden, G. B.; Jay, M.; Bird, A. C.: Macular dystrophy associated with mutations at codon 172 in the human retinal de-

generation slow gene. *Ophthalmology* 101:12–22, 1994.

[45853] 19405.Eph Nomenclature Committee: Unified nomenclature for the Ephfamily receptors and their ligands, the ephrins. *Cell* 90: 403–404,1997.

[45854] 19406.Hattori, M.; Osterfield, M.; Flanagan, J. G.: Regulated cleavageof a contact-mediated axon repellent. *Science* 289: 1360–1365, 2000.

[45855] 19407.Maru, Y.; Hirai, H.; Yoshida, M. C.; Takaku, F.: Evolution, expression,and chromosomal location of a novel receptor tyrosine kinase gene,eph. *Molec. Cell. Biol.* 8: 3770–3776, 1988.

[45856] 19408.Yoshida, M. C.; Maru, H.; Hirai, H.; Takau, F.: Chromosomal locationof a novel receptor tyrosine kinase gene, EPH, on chromosome 7. (Abstract) *Cytogenet.Cell Genet.* 51: 1113 only, 1989.

[45857] 19409.Boyd, A. W.; Ward, L. D.; Wicks, I. P.; Simpson, R. J.; Salvaris,E.; Wilks, A.; Welch, K.; Loudovaris, M.; Rockman, S.; Busmanis, I.: Isolation and characterization of a novel receptor-type proteintyrosine kinase (hek) from a human pre-B cell line. *J. Biol. Chem.* 267:3262–3267, 1992.

[45858] 19410.Fox, G. M.; Holst, P. L.; Chute, H. T.; Lindberg, R. A.; Janssen,A. M.; Basu, R.; Welcher, A. A.: cDNA cloning and tissue distributionof five human EPH-like receptor

protein-tyrosine kinases. *Oncogene* 10:897-905, 1995.

[45859] 19411.Wicks, I. P.; Lapsys, N. M.; Baker, E.; Campbell, L. J.; Boyd,A. W.; Sutherland, G. R.: Localization of a human receptor tyrosinekinase (ETK1) to chromosome region 3p11.2. *Genomics* 19: 38-41, 1994.

[45860] 19412.Wicks, I. P.; Wilkinson, D.; Salvaris, E.; Boyd, A. W.: Molecularcloning of HEK, the gene encoding a receptor tyrosine kinase expressedby human lymphoid tumor cell lines. *Proc. Nat. Acad. Sci.* 89: 1611-1615,1992.

[45861] 19413.Elsea, S. H.; Mykytyn, K.; Ferrell, K.; Coulter, K. L.; Das, P.;Dubiel, W.; Patel, P. I.; Metherall, J. E.: Hemizygosity for theCOP9 signalosome subunit gene, SGN3, in the Smith-Magenis syndrome. *Am.J. Med. Genet.* 87: 342-348, 1999.

[45862] 19414.Koyama, K.; Fukushima, Y.; Inazawa, J.; Tomot-sune, D.; Takahashi,N.; Nakamura, Y.: The human homologue of the murine Llglh gene (LLGL)maps within the Smith-Magenis syndrome region in 17p11.2. *Cytogenet.Cell Genet.* 72: 78-82, 1996.

[45863] 19415.Forsberg, E.; Hirsch, E.; Frohlich, L.; Meyer, M.; Ekblom, P.;Aszodi, A.; Werner, S.; Fassler, R.: Skin wounds and severed nervesheal normally in mice lacking tenascin-C. *Proc. Nat. Acad. Sci.* 93:6594-6599, 1996.

- [45864] 19416. Gulcher, J. R.; Alexakos, M. J.; Le Beau, M. M.; Lemons, R. S.; Stefansson, K.: Chromosomal localization of the human hexabrachion (tenascin) gene and evidence for recent reduplication within the gene. *Genomics* 6:616–622, 1990.
- [45865] 19417. Gulcher, J. R.; Nies, D. E.; Alexakos, M. J.; Ravikant, N. A.; Sturgill, M. E.; Marton, L. S.; Stefansson, K.: Structure of the human hexabrachion (tenascin) gene. *Proc. Nat. Acad. Sci.* 88: 9438–9442, 1991.
- [45866] 19418. Mitrovic, N.; Schachner, M.: Detection of tenascin–C in the nervous system of the tenascin–C mutant mouse. *J. Neurosci. Res.* 42: 710–717, 1995.
- [45867] 19419. Nies, D. E.; Hemesath, T. J.; Kim, J.–H.; Gulcher, J. R.; Stefansson, K.: The complete cDNA sequence of human hexabrachion (tenascin): a multidomain protein containing unique epidermal growth factor repeats. *J. Biol. Chem.* 266: 2818–2823, 1991.
- [45868] 19420. Williams, T. M.; Moolten, D.; Burlein, J.; Romano, J.; Bhaerman, R.; Godillot, A.; Mellon, M.; Rauscher, F. J., III; Kant, J. A.: Identification of a zinc finger protein that inhibits IL–2 gene expression. *Science* 254: 1791–1794, 1991.
- [45869] 19421. Ludecke, H.–J.; Schaper, J.; Meinecke, P.; Momeni,

P.; Gross, S.; von Holtum, D.; Hirche, H.; Abramowicz, M. J.; Albrecht, B.; Apacik, C.; Christen, H.-J.; Claussen, U.; and 28 others: Genotypic and phenotypic spectrum in trichorhino-phalangeal syndrome types I and III. *Am. J. Hum. Genet.* 68: 81–91, 2001.

[45870] 19422. Chiang, L.; Contreras, L.; Chiang, J.; Ward, P. H.: Human prostatic gastricsinogen: the precursor of seminal fluid acid proteinase. *Arch. Biochem. Biophys.* 210: 14–20, 1981.

[45871] 19423. Hayano, T.; Sogawa, K.; Ichihara, Y.; Fujii-Kuriyama, Y.; Takahashi, K.: Primary structure of human pepsinogen C gene. *J. Biol. Chem.* 263:1382–1385, 1988.

[45872] 19424. Pals, G.; Azuma, T.; Mohandas, T. K.; Bell, G. I.; Bacon, J.; Samloff, I. M.; Walz, D. A.; Barr, P. J.; Taggart, R. T.: Human pepsinogen C (progastricsin) polymorphism: evidence for a single locus located at 6p21.1-pter. *Genomics* 4: 137–145, 1989.

[45873] 19425. Randolph, L. M.; Azuma, T.; Petersen, G. M.; Sparkes, R. S.; Toyoda, M.; Wang, S. J.; Taggart, R. T.: Assignment of pepsinogen C (PGC) to 6p21.3-p21.1. (Abstract) *Cytogenet. Cell Genet.* 51: 1063–1064, 1989.

[45874] 19426. Szymura, J. M.; Klein, J.: Linkage of a gene controlling urinary pepsinogen with the major histocompatibility

complex of the mouse. *Immunogenetics* 13: 267–271, 1981.

- [45875] 19427. Taggart, R. T.; Cass, L. G.; Mohandas, T. K.; Derby, P.; Barr, P. J.; Pals, G.; Bell, G. I.: Human pepsinogen C (progastricsin): isolation of cDNA clones, localization to chromosome 6, and sequence homology with pepsinogen A. *J. Biol. Chem.* 264: 375–379, 1989.
- [45876] 19428. Taggart, R. T.; Mohandas, T. K.; Bell, G. I.: Assignment of human preprogastricsin (PGC) to chromosome 6 and regional localization of PGC (6pter–p21.1), prolactin PRL (6pter–p21.1). (Abstract) *Cytogenet. Cell Genet.* 46: 701–702, 1987.
- [45877] 19429. Takahara, K.; Fukushige, S.; Murotsu, T.; Ichihara, Y.; Hayano, T.; Ishihara, T.; Takahashi, K.: Assignment of human pepsinogen C (PGC) gene to chromosome 6. *Cytogenet. Cell Genet.* 52: 100–101, 1989.
- [45878] 19430. Miwa, S.; Sato, T.; Murao, H.; Kozuru, M.; Ibayashi, H.: A new type of phosphofructokinase deficiency: hereditary nonspherocytic hemolytic anemia. *Acta Haemat. Jpn.* 35: 113–118, 1972.
- [45879] 19431. Bernardi, G.: Personal Communication. Paris, France 5/13/1992.
- [45880] 19432. Boulard, M. R.; Bois, M.; Reviron, M.; Najean, Y.:

Red-cell phosphofructokinase deficiency. New Eng. J. Med. 291: 978–979, 1974.

[45881] 19433. Chadeaux, B.; Rethore, M. O.; Allard, D.: Regional mapping of liver type 6-phosphofructokinase isoenzyme on chromosome 21. Hum. Genet. 68: 136–137, 1984.

- [45882] 19434.Cox, D. R.; Kawashima, H.; Vora, S.; Epstein, C. J.: Regional mapping of SOD-1, PRGS, and PFK-L on human chromosome 21. (Abstract) Cytogenet.Cell Genet. 37: 441-442, 1984.
- [45883] 19435.Elson, A.; Levanon, D.; Brandeis, M.; Dafni, N.; Bernstein, Y.;Danciger, E.; Groner, Y.: The structure of the human liver-type phosphofructokinase gene. Genomics 7: 47-56, 1990.
- [45884] 19436.Etiemble, J.; Kahn, A.; Boivin, P.; Bernard, J. F.; Goudemand,M.: Hereditary hemolytic anemia with erythrocyte phosphofructokinase deficiency. Hum. Genet. 31: 83-91, 1976.
- [45885] 19437.Etiemble, J.; Picat, C.; Simeon, J.; Blatrix, C.; Boivin, P.:Inherited erythrocyte phosphofructokinase deficiency: molecular mechanism. Hum.Genet. 55: 383-390, 1980.
- [45886] 19438.Gehnrich, S. C.; Gekakis, N.; Sul, H. S.: Liver (B-type) phosphofructokinase mRNA: cloning, structure, and expression. J. Biol. Chem. 263: 11755-11759,1988.
- [45887] 19439.Kahn, A.; Etiemble, J.; Meienhofer, M. C.; Boivin, P.: Erythrocyte phosphofructokinase deficiency associated with an unstable variant of muscle phosphofructokinase. Clin. Chim. Acta 61: 415-419, 1975.
- [45888] 19440.Levanon, D.; Danciger, E.; Dafni, N.; Groner, Y.:

Genomic clones of the human liver-type phosphofructokinase. *Biochem. Biophys. Res. Commun.* 141: 374–380, 1986.

- [45889] 19441. Pantelakis, S. N.; Karaklis, A. G.; Alexiou, D.; Vardas, E.; Valaes, T.: Red cell enzymes in trisomy 21. *Am. J. Hum. Genet.* 22: 184–193, 1970.
- [45890] 19442. Van Keuren, M.; Drabkin, H.; Hart, I.; Harker, D.; Patterson, D.; Vora, S.: Regional assignment of human liver-type 6-phosphofructokinase to chromosome 21q22.3 by using somatic cell hybrids and a monoclonal anti-L antibody. *Hum. Genet.* 74: 34–40, 1986.
- [45891] 19443. Vora, S.; Davidson, M.; Seaman, C.; Miranda, A. F.; Noble, N. A.; Tanaka, K. R.; Frenkel, E. P.; DiMauro, S.: Heterogeneity of the molecular lesions in inherited phosphofructokinase deficiency. *J. Clin. Invest.* 72: 1995–2006, 1983.
- [45892] 19444. Vora, S.; Durham, S.; de Martinville, B.; Francke, U.: Assignment of the genes for liver type phosphofructokinase (PFKL) to chromosome 21 and for muscle type (PFKM) to region p32–q32 of chromosome 1. (Abstract) *Cytogenet. Cell Genet.* 32: 324 only, 1982.
- [45893] 19445. Vora, S.; Francke, U.: Assignment of the human gene for liver type phosphofructokinase isozyme to chro-

mosome 21 using somatic cellhybrids. (Abstract) *Pediat. Res.* 15: 570 only, 1981.

[45894] 19446.Vora, S.; Francke, U.: Assignment of the human gene for liver-type6-phosphofructokinase isozyme (PFKL) to chromosome 21 by using somaticcell hybrids and monoclonal anti-L antibody. *Proc. Nat. Acad. Sci.* 78:3738–3742, 1981.

[45895] 19447.Wang, D.; Fang, H.; Cantor, C. R.; Smith, C. L.: A contiguousNotI restriction map of band q22.3 of human chromosome 21. *Proc.Nat. Acad. Sci.* 89: 3222–3226, 1992.

[45896] 19448.Vidal, R.; Revesz, T.; Rostagno, A.; Kim, E.; Holton, J. L.; Bek,T.; Bojsen-Moller, M.; Braendgaard, H.; Plant, G.; Ghiso, J.; Frangione,B.: A decamer duplication in the 3-prime region of the BRI gene originatesan amyloid peptide that is associated with dementia in a Danish kindred. *Proc.Nat. Acad. Sci.* 97: 4920–4925, 2000.

[45897] 19449.Fathallah-Shaykh, H.; Wolf, S.; Wong, E.; Posner, J. B.: Cloningof a leucine-zipper protein recognized by the sera of patients withantibody-associated paraneoplastic cerebellar degeneration. *Proc.Nat. Acad. Sci.* 88: 3451–3454, 1991.

[45898] 19450.Gress, T.; Baldini, A.; Rocchi, M.; Furneaux, H.;

Posner, J. B.;Siniscalco, M.: In situ mapping of the gene coding for a leucinezipper DNA binding protein (CDR 2) to the region between two rarefragile sites of autosome 16 (16p12–p13.1). (Abstract) Cytogenet.Cell Genet. 58: 1999–2000, 1991.

[45899] 19451.Gress, T.; Baldini, A.; Rocchi, M.; Furneaux, H.;

Posner, J. B.;Siniscalco, M.: In situ mapping of the gene coding for a leucinezipper DNA binding protein (CDR62) to 16p12–16p13.1. Genomics 13:1340–1342, 1992.

[45900] 19452.Raimondi, E.; Rubboli, F.; Moralli, D.; Chini, B.; For-

nasari, D.;Tarroni, P.; De Carli, L.; Clementi, F.: Chromosomal localizationand physical linkage of the genes encoding the human alpha–3, alpha–5,and beta–4 neuronal nicotinic receptor subunits. Genomics 12: 849–850,1992.

[45901] 19453.Anand, R.; Lindstrom, J.: Nucleotide sequence of the human nicotinicacetylcholine receptor beta–2 subunit gene. Nucleic Acids Res. 18:4272, 1990.

[45902] 19454.De Fusco, M.; Becchetti, A.; Patrignani, A.; Annesi,

G.; Gambardella,A.; Quattrone, A.; Ballabio, A.; Wanke, E.; Casari, G.: The nicotinicreceptor beta–2 subunit is mutant in nocturnal frontal lobe epilepsy. NatureGenet. 26: 275–276, 2000.

[45903] 19455.Lueders, K. K.; Elliott, R. W.; Marenholz, I.; Mischke,

D.; DuPree, M.; Hamer, D.: Genomic organization and mapping of the human and mouse neuronal beta-1-nicotinic acetylcholine receptor genes. *Mammalian Genome* 10: 900–905, 1999.

[45904] 19456. Phillips, H. A.; Favre, I.; Kirkpatrick, M.; Zuberi, S. M.; Goudie, D.; Heron, S. E.; Scheffer, I. E.; Sutherland, G. R.; Berkovic, S. F.; Bertrand, D.; Mulley, J. C.: CHRNA2 is the second acetylcholine receptor subunit associated with autosomal dominant nocturnal frontal lobe epilepsy. *Am. J. Hum. Genet.* 68: 225–231, 2001.

[45905] 19457. Picciotto, M. R.; Zoli, M.; Lena, C.; Bessis, A.; Lallemand, Y.; LeNovere, N.; Vincent, P.; Pich, E. M.; Brulet, P.; Changeux, J.-P.: Abnormal avoidance learning in mice lacking functional high-affinity nicotine receptor in the brain. *Nature* 374: 65–67, 1995.

[45906] 19458. Zoli, M.; Picciotto, M. R.; Ferrari, R.; Cocchi, D.; Changeux, J.-P.: Increased neurodegeneration during ageing in mice lacking high-affinity nicotine receptors. *EMBO J.* 18: 1235–1244, 1999.

[45907] 19459. Tarroni, P.; Rubboli, F.; Chini, B.; Zwart, R.; Oortgiesen, M.; Sher, E.; Clementi, F.: Neuronal-type nicotinic receptors in human neuroblastoma and small-cell lung carcinoma cell lines. *FEBS Lett.* 312:66–70, 1992.

- [45908] 19460.Bonner, T. I.; Buckley, N. J.; Young, A. C.; Brann, M. R.: Identification of a family of muscarinic acetylcholine receptor genes. *Science* 237:527–532, 1987.
- [45909] 19461.Schwenk, W. F.; Rizza, R. A.; Mandarino, L. J.; Gerich, J. E.; Hayles, A. B.; Haymond, M. W.: Familial insulin resistance and acanthosis nigricans: presence of a post-binding defect. *Diabetes* 35: 33–37, 1986.
- [45910] 19462.Speckman, R. A.; Garg, A.; Du, F.; Bennett, L.; Veile, R.; Arioglu, E.; Taylor, S. I.; Lovett, M.; Bowcock, A. M.: Mutational and haplotype analyses of families with familial partial lipodystrophy (Dunnigan variety) reveal recurrent missense mutations in the globular C-terminal domain of lamin A/C. *Am. J. Hum. Genet.* 66: 1192–1198, 2000.
Note: Erratum: *Am. J. Hum. Genet.* 67: 775 only, 2000.
- [45911] 19463.Chan, J.; Watt, V. M.: Eek and erk, new members of the eph subclass of receptor protein-tyrosine kinases. *Oncogene* 6: 1057–1061, 1991.
- [45912] 19464.Park, S.; Frisen, J.; Barbacid, M.: Aberrant axonal projections in mice lacking EphA8 (Eek) tyrosine protein kinase receptors. *EMBO J.* 16: 3106–3114, 1997.
- [45913] 19465.Ganju, P.; Shigemoto, K.; Brennan, J.; Entwistle, A.; Reith, A.D.: The Eck receptor tyrosine kinase is implicated in pattern formation during gastrulation, hindbrain seg-

mentation and limb development. *Oncogene* 9:1613–1624, 1994.

- [45914] 19466.Lindberg, R. A.; Hunter, T.: cDNA cloning and characterization of eck, an epithelial cell receptor–tyrosine kinase in the eph/elk family of protein kinases. *Molec. Cell. Biol.* 10: 6316–6324, 1990.
- [45915] 19467.Sulman, E. P.; Tang, X. X.; Allen, C.; Biegel, J. A.; Pleasure, D. E.; Brodeur, G. M.; Ikegaki, N.: ECK, a human EPH–related gene, maps to 1p36.1, a common region of alteration in human cancers. *Genomics* 40:371–374, 1997.
- [45916] 19468.Colucci, F.; Schweighoffer, E.; Tomasello, E.; Turner, M.; Ortaldo, J. R.; Vivier, E.; Tybulewicz, V. L. J.; Di Santo, J. P.: Natural cytotoxicity uncoupled from the Syk and ZAP–70 intracellular kinases. *Nature Immun.* 3: 288–294, 2002.
- [45917] 19469.Ku, G.; Malissen, B.; Mattei, M.–G.: Chromosomal location of the Syk and ZAP–70 tyrosine kinase genes in mice and humans. *Immunogenetics* 40:300–302, 1994.
- [45918] 19470.Toyabe, S.–I.; Watanabe, A.; Harada, W.; Karasawa, T.; Uchiyama, M.: Specific immunoglobulin E responses in ZAP–70–deficient patients are mediated by Syk–dependent T–cell receptor signalling. *Immunology* 103:164–171,

2001.

- [45919] 19471.Sudbrak, R.; Golla, A.; Hogan, K.; Powers, P.; Gregg, R.; Du Chesne,I.; Lehmann–Horn, F.; Deufel, T.: Exclusion of malignant hyperthermiasusceptibility (MHS) from a putative MHS2 locus on chromosome 17qand of the alpha–1, beta–1, and gamma subunits of the dihydropyridinereceptor calcium channel as candidates for the molecular defect. *Hum.Molec. Genet.* 2: 857–862, 1993.
- [45920] 19472.Naim, H. Y.; Sterchi, E. E.; Lentze, M. J.: Structure, biosynthesis,and glycosylation of human small intestinal maltase–glucoamylase. *J.Biol. Chem.* 263: 19709–19717, 1988.
- [45921] 19473.Nichols, B. L.; Eldering, J.; Avery, S.; Hahn, D.; Quaroni, A.;Sterchi, E.: Human small intestinal maltase–glucoamylase cDNA cloning:homology to sucrase–isomaltase. *J. Biol. Chem.* 273: 3076–3081, 1998.
- [45922] 19474.Schuler, G. D.; Boguski, M. S.; Stewart, E. A.; Stein, L. D.; Gyapay,G.; Rice, K.; White, R. E.; Rodriguez–Tome, P.; Aggarwal, A.; Bajorek,E.; Bentolila, S.; Birren, B. B.; Butler, A.; Castle, A. B.; Chiannilkulchai,N.; Chu, A.; Clee, C.; Cowles, S.; Day, P. J. R.; Dibling, T.; and84 others: A gene map of the human genome. *Science* 274: 540–546,1996.
- [45923] 19475.Tanahashi, N.; Suzuki, M.; Fujiwara, T.; Takahashi,

E.; Shimbara,N.; Chung, C. H.; Tanaka, K.: Chromosomal localization and immunological analysis of a family of human 26S proteasomal ATPases. *Biochem. Biophys. Res. Commun.* 243: 229–232, 1998.

[45924] 19476. Levine, A.; Cantoni, G. L.; Razin, A.: Inhibition of promoter activity by methylation: possible involvement of protein mediators. *Proc. Nat. Acad. Sci.* 88: 6515–6518, 1991.

[45925] 19477. Nan, X.; Meehan, R. R.; Bird, A.: Dissection of the methyl–CpG binding domain from the chromosomal protein MeCP2. *Nucleic Acids Res.* 21: 4886–4892, 1993.

[45926] 19478. Ohki, I.; Shimotake, N.; Fujita, N.; Jee, J.–G.; Ikegami, T.; Nakao, M.; Shirakawa, M.: Solution structure of the methyl–CpG binding domain of human MBD1 in complex with methylated DNA. *Cell* 105: 487–497, 2001.

[45927] 19479. Carrera, C. J.; Eddy, R. L.; Shows, T. B.; Carson, D. A.: Assignment of the gene for methylthioadenosine phosphorylase to human chromosome 9 by mouse–human somatic cell hybridization. *Proc. Nat. Acad. Sci.* 81: 2665–2668, 1984.

[45928] 19480. Chilcote, R. R.; Brown, E.; Rowley, J. D.: Lymphoblastic leukemia with lymphomatous features associated with abnormalities of the short arm of chromosome 9.

New Eng. J. Med. 313: 286–291, 1985.

- [45929] 19481.Nobori, T.; Takabayashi, K.; Tran, P.; Orvis, L.; Batova, A.; Yu, A. L.; Carson, D. A.: Genomic cloning of methylthioadenosine phosphorylase: a purine metabolic enzyme deficient in multiple different cancers. Proc. Nat. Acad. Sci. 93: 6203–6208, 1996.
- [45930] 19482.Olopade, O. I.; Jenkins, R. B.; Ransom, D. T.; Malik, K.; Pomykala, H.; Nobori, T.; Cowan, J. M.; Rowley, J. D.; Diaz, M. O.: Molecular analysis of deletions of the short arm of chromosome 9 in human gliomas. Cancer Res. 52: 2523–2529, 1992.
- [45931] 19483.Olopade, O. I.; Pomykala, H. M.; Hagos, F.; Sveen, L. W.; Espinosa, R., III; Dreyling, M. H.; Gursky, S.; Stadler, W. M.; Le Beau, M. M.; Bohlander, S. K.: Construction of a 2.8-megabase yeast artificial chromosome contig and cloning of the human methylthioadenosine phosphorylase gene from the tumor suppressor region on 9p21. Proc. Nat. Acad. Sci. 92:6489–6493, 1995.
- [45932] 19484.Ragione, F. D.; Takabayashi, K.; Mastropietro, S.; Mercurio, C.; Oliva, A.; Russo, G. L.; Pietra, V. D.; Borriello, A.; Nobori, T.; Carson, D. A.; Zappia, V.: Purification and characterization of recombinant human 5-prime-methylthioadenosine phosphorylase: definite

identification of coding cDNA. *Biochem. Biophys. Res. Commun.* 223: 514–519, 1996.

- [45933] 19485. Williams–Ashman, H. G.; Seidenfeld, J.; Galletti, P.: Trends in the biochemical pharmacology of 5–prime–deoxy–5–prime–methylthioadenosine. *Biochem. Pharm.* 31: 277–288, 1982.
- [45934] 19486. Cirullo, R. E.; Wasmuth, J. J.: Assignment of the human gene encoding methionyl–tRNA synthetase to chromosome 12. (Abstract) *Cytogenet. Cell Genet.* 37: 437 only, 1984.
- [45935] 19487. Cirullo, R. E.; Wasmuth, J. J.: Assignment of the human MARS gene, encoding methioninyl–tRNA synthetase, to chromosome 12 using human x Chinese hamster cell hybrids. *Somat. Cell Molec. Genet.* 10: 225–234, 1984.
- [45936] 19488. Grewal, P. K.; Holzfeind, P. J.; Bittner, R. E.; Hewitt, J. E.: Mutant glycosyltransferase and altered glycosylation of alpha–dystroglycan in the myodystrophy mouse. *Nature Genet.* 28: 151–154, 2001.
- [45937] 19489. Zabel, B. U.; Eddy, R. L.; Scott, J.; Shows, T. B.: The human nerve growth factor gene (NGF) is located on the short arm of chromosome 1. (Abstract) *Cytogenet. Cell Genet.* 37: 614, 1984.
- [45938] 19490. Adams, D. S.; Kiyokawa, M.; Getman, M. E.;

Shashoua, V. E.: Genes encoding giant danio and golden shiner ependymin. *Neurochem. Res.* 21:377–384, 1996.

[45939] 19491. de Groen, P. C.; Eggen, B. J. L.; Gispen, W. H.; Schotman, P.; Schrama, L. H.: Cloning and promoter analysis of the human B-50/GAP-43 gene. *J. Molec. Neurosci.* 6: 109–119, 1995.

[45940] 19492. Kosik, K. S.; Orecchio, L. D.; Bruns, G. A. P.; Benowitz, L. I.; MacDonald, G. P.; Cox, D. R.; Neve, R. L.: Human GAP-43: its deduced amino acid sequence and chromosomal localization in mouse and human. *Neuron* 1:127–132, 1988.

[45941] 19493. Reeves, R. H.; O'Hara, B. F.; Gearhart, J. D.: Localization on mouse chromosome 16 of Smst and Gap43: a conserved synteny with human chromosome 3. (Abstract) *Cytogenet. Cell Genet.* 51: 1064, 1989.

[45942] 19494. Strittmatter, S. M.; Fankhauser, C.; Huang, P. L.; Mashimo, H.; Fishman, M. C.: Neuronal pathfinding is abnormal in mice lacking the neuronal growth cone protein GAP-43. *Cell* 80: 445–452, 1995.

[45943] 19495. Bespalova, I. N.; Farjo, Q.; Mortlock, D. P.; Jackson, A. U.; Meisler, M. H.; Swaroop, A.; Burmeister, M.: Mapping of the neural retinal leucine zipper gene, *Nrl*, to mouse chromosome 14. *Mammalian Genome* 4:618–620, 1993.

- [45944] 19496. Bessant, D. A. R.; Payne, A. M.; Mitton, K. P.; Wang, Q.-L.; Swain, P. K.; Plant, C.; Bird, A. C.; Zack, D. J.; Swaroop, A.; Bhattacharya, S. S.: A mutation in NRL is associated with autosomal dominant retinitis pigmentosa. *Nature Genet.* 21: 355–356, 1999.
- [45945] 19497. Bessant, D. A. R.; Payne, A. M.; Plant, C.; Bird, A. C.; Swaroop, A.; Bhattacharya, S. S.: NRL S50T mutation and the importance of 'founder effects' in inherited retinal dystrophies. *Europ. J. Hum. Genet.* 8: 783–787, 2000.
- [45946] 19498. Dahl, S. P.; Jackson, A.; Kimberling, W. J.; Blackwood, D.; Swaroop, A.: Genetic mapping of NRL, a human retina-specific gene located on chromosome 14. (Abstract) *Am. J. Hum. Genet.* 51 (suppl.): A185 only, 1992.
- [45947] 19499. Farjo, Q.; Jackson, A.; Pieke-Dahl, S.; Scott, K.; Kimberling, W. J.; Sieving, P. A.; Richards, J. E.; Swaroop, A.: Human bZIP transcription factor gene NRL: structure, genomic sequence, and fine linkage mapping at 14q11.2 and negative mutation analysis in patients with retinal degeneration. *Genomics* 45: 395–401, 1997.
- [45948] 19500. Farjo, Q.; Jackson, A. U.; Xu, J.; Gryzenia, M.; Skolnick, C.; Agarwal, N.; Swaroop, A.: Molecular characterization of the murine neural retina leucine zipper gene, *Nrl*. *Genomics* 18: 216–222, 1993.

- [45949] 19501.Mears, A. J.; Kondo, M.; Swain, P. K.; Takada, Y.; Bush, R. A.;Saunders, T. L.; Sieving, P. A.; Swaroop, A.: Nrl is required forrod photoreceptor development. *Nature Genet.* 29: 447–452, 2001.
- [45950] 19502.Swaroop, A.; Xu, J.; Agarwal, N.; Weissman, S. M.: A simple andefficient cDNA library subtraction procedure: isolation of human retina–specificcDNA clones. *Nucleic Acids Res.* 19: 1954 only, 1991.
- [45951] 19503.Swaroop, A.; Xu, J.; Pawar, H.; Jackson, A.; Skolnick, C.; Agarwal,N.: A conserved retina–specific gene encodes a basic motif/leucinezipper domain. *Proc. Nat. Acad. Sci.* 89: 266–270, 1992.
- [45952] 19504.Wright, A. F.; Mansfield, D. C.; Bruford, E. A. Teague, P. W.;Thomson, K. L.; Riise, R.; Jay, M.; Patton, M. A.; Jeffery, S.; Schinzel,A.; Tommerup, N.: Fossarello, M.: Genetic studies in autosomal recessiveforms of retinitis pigmentosa.In: Anderson, R. E.; LaVail, M. M.;Hollyfield, J. G. (eds.): *Degenerative diseases of the retina*. New York: Plenum Press (pub.) 1995. Pp. 293–302.
- [45953] 19505.Yang–Feng, T. L.; Swaroop, A.: Neural retina–specific leucinezipper gene NRL (D14S46E) maps to human chromosome 14q11.1–q11.2. *Genomics* 14:491–492, 1992.

- [45954] 19506.Eddy, R. L.; Kretschmer, P. J.; Fairhurst, J. L.; Shows, T. B.; Bohlen, P.; O'Hara, B.; Kovesdi, I.: A human gene family of neuriteoutgrowth-promoting proteins: the gene for a heparin binding neuriteoutgrowth-promoting factor maps to 7q22-qter. (Abstract) Cytogenet.Cell Genet. 58: 1920 only, 1991.
- [45955] 19507.Lai, S.; Czubayko, F.; Riegel, A. T.; Wellstein, A.: Structureof the human heparin-binding growth factor gene pleiotrophin. Biochem.Biophys. Res. Commun. 187: 1113-1122, 1992.
- [45956] 19508.Li, Y.-S.; Hoffman, R. M.; Le Beau, M. M.; Espinosa, R., III; Jenkins,N. A.; Gilbert, D. J.; Copeland, N. G.; Deuel, T. F.: Characterizationof the human pleiotrophin gene: promoter region and chromosomal localization. J.Biol. Chem. 267: 26011-26016, 1992.
- [45957] 19509.Li, Y.-S.; Milner, P. G.; Chauhan, A. K.; Watson, M. A.; Hoffman,R. M.; Kodner, C. M.; Milbrandt, J.; Deuel, T. F.: Cloning and expressionof a developmentally regulated protein that induces mitogenic andneurite outgrowth activity. Science 250: 1690-1694, 1990.
- [45958] 19510.Milner, P. G.; Shah, D.; Veile, R.; Donis-Keller, H.; Kumar, B.V.: Cloning, nucleotide sequence, and chromosome localization ofthe human pleiotrophin gene. Bio-

chemistry 31: 12023–12028, 1992.

- [45959] 19511. Brightbill, H. D.; Libraty, D. H.; Krutzik, S. R.; Yang, R.-B.; Bellsie, J. T.; Bieharski, J. R.; Maitland, M.; Norgard, M. V.; Plevy, S. E.; Smale, S. T.; Brennan, P. J.; Bloom, B. R.; Godowski, P. J.; Modlin, R. L.: Host defense mechanisms triggered by microbial lipoproteins through toll-like receptors. *Science* 285: 732–736, 1999.
- [45960] 19512. Jankovic, D.; Kullberg, M. C.; Hieny, S.; Caspar, P.; Collazo, C. M.; Sher, A.: In the absence of IL-12, CD4⁺ T cell responses to intracellular pathogens fail to default to a Th2 pattern and are host protective in an IL-10^{-/-} setting. *Immunity* 16: 429–439, 2002.
- [45961] 19513. Jouanguy, E.; Lamhamedi-Cherradi, S.; Altare, F.; Fondaneche, M.-C.; Tuerlinckx, D.; Blanche, S.; Emile, J.-F.; Gaillard, J.-L.; Schreiber, R.; Levin, M.; Fischer, A.; Hivroz, C.; Casanova, J.-L.: Partial interferon-gamma receptor 1 deficiency in a child with tuberculoid bacillus Calmette-Guerin infection and a sibling with clinical tuberculosis. *J. Clin. Invest.* 100: 2658–2664, 1997.
- [45962] 19514. Warrington, J. A.; Bengtsson, U.: High-resolution physical mapping of human 5q31–q33 using three methods: radiation hybrid mapping, interphase fluorescence in situ hybridization, and pulsed-field gel electrophoresis.

Genomics 24:395–398, 1994.

- [45963] 19515. Anderson, S. K.; Gallinger, S.; Roder, J.; Frey, J.; Young, H.A.; Ortaldo, J. R.: A cyclophilin-related protein involved in the function of natural killer cells. Proc. Nat. Acad. Sci. 90: 542–546, 1993.
- [45964] 19516. Frey, J. L.; Bino, T.; Kantor, R. R. S.; Segal, D. M.; Giardina, S. L.; Roder, J.; Anderson, S.; Ortaldo, J. R.: Mechanism of target cell recognition by natural killer cells: characterization of a novel triggering molecule restricted to CD3– large granular lymphocytes. J. Exp. Med. 174: 1527–1536, 1991.
- [45965] 19517. Rinfret, A.; Anderson, S. K.: IL-2 regulates the expression of the NK-TR gene via an alternate RNA splicing mechanism. Molec. Immun. 30:1307–1313, 1993.
- [45966] 19518. Simons-Evelyn, M.; Young, H. A.; Anderson, S. K.: Characterization of the mouse Nktr gene and promoter. Genomics 40: 94–100, 1997.
- [45967] 19519. Young, H. A.; Jenkins, N. A.; Copeland, N. G.; Simek, S.; Lerman, M. I.; Zbar, B.; Glenn, G.; Ortaldo, J. R.; Anderson, S. K.: Localization of a novel natural killer triggering receptor locus to human chromosome 3p23–p21 and mouse chromosome 9. Genomics 16: 548–549, 1993.
- [45968] 19520. Scott, I. C.; Halila, R.; Jenkins, J. M.; Mehan, S.;

Apostolou,S.; Winqvist, R.; Callen, D. F.; Prockop, D. J.; Peltonen, L.; Kadler,K. E.: Molecular cloning, expression and chromosomal localization of a human gene encoding a 33 kDa putative metallopeptidase (PRSM1). *Gene* 174:135–143, 1996.

[45969] 19521.Aliprantis, A. O.; Yang, R.-B.; Mark, M. R.; Suggett, S.; Devaux,B.; Radolf, J. D.; Klimpel, G. R.; Godowski, P.; Zychlinsky, A.:Cell activation and apoptosis by bacterial lipoproteins through toll-like receptor-2. *Science* 285: 736–739, 1999.

[45970] 19522.Baldwin, A. S., Jr.: The NF-kappa-B and I-kappa-B proteins: new discoveries and insights. *Annu. Rev. Immun.* 14: 649–683, 1996.

[45971] 19523.Barnes, P. J.; Karin, M.: Nuclear factor-kappa-B -- A pivotal transcription factor in chronic inflammatory diseases. *New Eng. J.Med.* 336: 1066–1071, 1997.

[45972] 19524.Chen, F.; Castranova, V.; Shi, X.; Demers, L. M.: New insights into the role of nuclear factor-kappa-B, a ubiquitous transcription factor in the initiation of diseases. *Clin. Chem.* 45: 7–17, 1999.

[45973] 19525.Heron, E.; Deloukas, P.; van Loon, A. P. G. M.: The complete exon-intron structure of the 156-kb human gene NFKB1, which encodes the p105 and p50 proteins of

transcription factors NF-kappa-B and I-kappa-B-gamma: implications for NF-kappa-B-mediated signal transduction. *Genomics* 30:493-505, 1995.

- [45974] 19526. Huxford, T.; Huang, D.-B.; Malek, S.; Ghosh, G.: The crystal structure of the I-kappa-B-alpha/NF-kappa-B complex reveals mechanisms of NF-kappa-B inactivation. *Cell* 95: 759-770, 1998.
- [45975] 19527. Lin, L.; DeMartino, G. N.; Greene, W. C.: Cotranslational biogenesis of NF-kappa-B p50 by the 26S proteasome. *Cell* 92: 819-828, 1998.
- [45976] 19528. Liptay, S.; Schmid, R. M.; Perkins, N. D.; Meltzer, P.; Altherr, M. R.; McPherson, J. D.; Wasmuth, J. J.; Nabel, G. J.: Related subunits of NF-kappa-B map to two distinct loci associated with translocations in leukemia, NFKB1 and NFKB2. *Genomics* 13: 287-292, 1992.
- [45977] 19529. Mathew, S.; Murty, V. V. V. S.; Dalla-Favera, R.; Chaganti, R. S. K.: Chromosomal localization of genes encoding the transcription factors, c-rel, NF-kappa-Bp50, NF-kappa-Bp65, and Iy-10 by fluorescence in situ hybridization. *Oncogene* 8: 191-193, 1993.
- [45978] 19530. Meyer, R.; Hatada, E. N.; Hohmann, H. P.; Haiker, M.; Bartsch, C.; Rothlisberger, U.; Lahm, H. W.; Schlaeger, E. J.; van Loon, A. P.; Scheidereit, C.: Cloning of the DNA-

binding subunit of human nuclear factor kappa B: the level of its mRNA is strongly regulated by phorbol ester or tumor necrosis factor alpha. *Proc. Nat. Acad. Sci.* 88: 966–970, 1991.

[45979] 19531. Ozes, O. N.; Mayo, L. D.; Gustin, J. A.; Pfeffer, S. R.; Pfeffer, L. M.; Donner, D. B.: NF-kappa-B activation by tumor necrosis factor requires the Akt serine-threonine kinase. *Nature* 401: 82–85, 1999.

[45980] 19532. Romashkova, J. A.; Makarov, S. S.: NF-kappa-B is a target of AKT in anti-apoptotic PDGF signalling. *Nature* 401: 86–90, 1999.

[45981] 19533. Ryan, K. M.; Ernst, M. K.; Rice, N. R.; Voudsen, K. H.: Role of NF-kappa-B in p53-mediated programmed cell death. *Nature* 404: 892–897, 2000.

[45982] 19534. Sen, R.; Baltimore, D.: Multiple nuclear factors interact with the immunoglobulin enhancer sequences. *Cell* 46: 705–716, 1986.

[45983] 19535. Sha, W. C.; Liou, H.-C.; Tuomanen, E. I.; Baltimore, D.: Targeted disruption of the p50 subunit of NF-kappa-B leads to multifocal defects in immune responses. *Cell* 80: 321–330, 1995.

[45984] 19536. Zhong, H.; May, M. J.; Jimi, E.; Ghosh, S.: The phosphorylation status of nuclear NF-kappa-B determines its

association with CBP/p300 or HDAC-1. *Molec. Cell* 9: 625–636, 2002.

- [45985] 19537. Claudio, E.; Brown, K.; Park, S.; Wang, H.; Siebenlist, U.: BAFF-induced NEMO-independent processing of NF-kappa-B2 in maturing B cells. *Nature Immun.* 3: 958–965, 2002.
- [45986] 19538. Neri, A.; Chang, C.-C.; Lombardi, L.; Salina, M.; Corradini, P.; Maiolo, A. T.; Chaganti, R. S. K.; Dalla-Favera, R.: B cell lymphoma-associated chromosomal translocation involves candidate oncogene *lyt-10*, homologous to NF-kappa-B p50. *Cell* 67: 1075–1087, 1991.
- [45987] 19539. Taparowsky, E.; Shimizu, K.; Goldfarb, M.; Wigler, M.: Structure and activation of the human N-ras gene. *Cell* 34: 581–586, 1983.
- [45988] 19540. Yuasa, Y.; Gol, R. A.; Chang, A.; Chiu, I.-M.; Reddy, E. P.; Tronick, S. R.; Aaronson, S. A.: Mechanism of activation of an N-ras oncogene of SW-1271 human lung carcinoma cells. *Proc. Nat. Acad. Sci.* 81: 3670–3674, 1984.
- [45989] 19541. Bakin, A. V.; Curran, T.: Role of DNA 5-methylcytosine transferase in cell transformation by fos. *Science* 283: 387–390, 1999.
- [45990] 19542. Barker, P. E.; Rabin, M.; Watson, M.; Breg, W. R.; Ruddle, F. H.; Verma, I. M.: Human c-fos oncogene

mapped within chromosomal region 14q21–q31. Proc. Nat. Acad. Sci. 81: 5826–5830, 1984.

[45991] 19543. Dony, C.; Gruss, P.: Proto-oncogene c-fos expression in growth regions of fetal bone and mesodermal web tissue. Nature 328: 711–714, 1987.

[45992] 19544. Ekstrand, A. J.; Zech, L.: Human c-fos proto-oncogene mapped to chromosome 14, band q24.3–q31: possibilities for oncogene activation by chromosomal rearrangements in human neoplasms. Exp. Cell Res. 169:262–266, 1987.

[45993] 19545. Glover, J. N. M.; Harrison, S. C.: Crystal structure of the heterodimeric bZIP transcription factor c-Fos–c-Jun bound to DNA. Nature 373:257–261, 1995.

[45994] 19546. Grigoriadis, A. E.; Wang, Z.-Q.; Cecchini, M. G.; Hofstetter, W.; Felix, R.; Fleisch, H. A.; Wagner, E. F.: c-Fos: a key regulator of osteoclast–macrophage lineage determination and bone remodeling. Science 266:443–448, 1994.

[45995] 19547. Grigoriadis, A. E.; Wang, Z.-Q.; Wagner, E. F.: Fos and bone cell development: lessons from a nuclear oncogene. Trends Genet. 11:436–441, 1995.

[45996] 19548. Hayflick, L.: The limited in vitro lifetime of human diploid cell strains. Exp. Cell Res. 37: 614–636, 1965.

[45997] 19549. Johnson, R. S.; Spiegelman, B. M.; Papaioannou, V.:

Pleiotropic effects of a null mutation in the c-fos proto-oncogene. *Cell* 71:577–586, 1992.

[45998] 19550. Muller, R.; Tremblay, J. M.; Adamson, E. D.; Verma, I. M.: Tissue and cell type-specific expression of two human c-onc genes. *Nature* 304:454–456, 1983.

[45999] 19551. Rogaev, E. I.; Lukiw, W. J.; Vaula, G.; Haines, J. L.; Rogaeva, E. A.; Tsuda, T.; Alexandrova, N.; Liang, Y.; Mortilla, M.; Amaducci, L.; Bergamini, L.; Bruni, A. C.; Foncin, J.-F.; Macciardi, F.; Montesi, M. P.; Sorbi, S.; Rainero, I.; Pinessi, L.; Polinsky, R. J.; Frommelt, P.; Duara, R.; Lopez, R.; Pollen, D.; Gusella, J. F.; Tanzi, R.; Crapper MacLachlan, D.; St. George-Hyslop, P. H.: Analysis of the c-FOS gene on chromosome 14 and the promoter of the amyloid precursor protein gene in familial Alzheimer's disease. *Neurology* 43: 2275–2279, 1993.

[46000] 19552. Ruther, U.; Garber, C.; Komitowski, D.; Muller, R.; Wagner, E. F.: Deregulated c-fos expression interferes with normal bone development in transgenic mice. *Nature* 325: 412–416, 1987.

[46001] 19553. Saez, E.; Rutberg, S. E.; Mueller, E.; Oppenheim, H.; Smoluk, J.; Yuspa, S. H.; Spiegelman, B. M.: c-fos is required for malignant progression of skin tumors. *Cell* 82: 721–732, 1995.

- [46002] 19554.Sanyal, S.; Sandstrom, D. J.; Hoeffler, C. A.; Ramaswami, M.:AP-1 function upstream of CREB to control synaptic plasticity in *Drosophila*. *Nature* 416:870–874, 2002.
- [46003] 19555.Seshadri, T.; Campisi, J.: Repression of c-fos transcription and an altered genetic program in senescent human fibroblasts. *Science* 247:205–209, 1990.
- [46004] 19556.van Straaten, F.; Muller, R.; Curran, T.; Van Beveren, C.; Verma, I. M.: Complete nucleotide sequence of a human c-onc gene: deduced amino acid sequence of the human c-fos protein. *Proc. Nat. Acad.Sci.* 80: 3183–3187, 1983.
- [46005] 19557.Visvader, J.; Sassone-Corsi, P.; Verma, I. M.: Two adjacent promoter elements mediate nerve growth factor activation of the c-fos gene and bind distinct nuclear complexes. *Proc. Nat. Acad. Sci.* 85: 9474–9478, 1988.
- [46006] 19558.Wang, Z.-Q.; Grigoriadis, A. E.; Mohle-Steinlein, U.; Wagner, E. F.: A novel target cell for c-fos-induced oncogenesis: development of chondrogenic tumours in embryonic stem cell chimeras. *EMBO J.* 10:2437–2450, 1991.
- [46007] 19559.Wang, Z.-Q.; Ovitt, C.; Grigoriadis, A. E.; Mohle-Steinlein, U.; Ruther, U.; Wagner, E. F.: Bone and haematopoietic defects in mice lacking c-fos. *Nature* 360:

741–745, 1992.

- [46008] 19560.Zhang, J.; Zhang, D.; McQuade, J. S.; Behbehani, M.; Tsien, J.Z.; Xu, M.: c-fos regulates neuronal excitability and survival. *NatureGenet.* 30: 416–420, 2002.
- [46009] 19561.Arheden, K.; Mandahl, N.; Strombeck, B.; Isaksson, M.; Mitelman, F.: Chromosome localization of the human oncogene INT1 to 12q13 by in situ hybridization. *Cytogenet. Cell Genet.* 47: 86–87, 1988.
- [46010] 19562.Gavin, B. J.; McMahon, J. A.; McMahon, A. P.: Expression of multiple novel Wnt-1/int-1-related genes during fetal and adult mouse development. *GenesDev.* 4: 2319–2332, 1990.
- [46011] 19563.Kirikoshi, H.; Sekihara, H.; Katoh, M.: WNT10A and WNT6, clustered in human chromosome 2q35 region with head-to-tail manner, are strongly coexpressed in SW480 cells. *Biochem. Biophys. Res. Commun.* 283:798–805, 2001.
- [46012] 19564.McMahon, A. P.: The Wnt family of developmental regulators. *TrendsGenet.* 8: 236–242, 1992.
- [46013] 19565.Nusse, R.; Brown, A.; Papkoff, J.; Scambler, P.; Shackleford, G.; McMahon, A.; Moon, R.; Varmus, H.: A new nomenclature for int-1 and related genes: the Wnt gene family. (Letter) *Cell* 64: 231–232, 1991.

- [46014] 19566.Nusse, R.; van't Veer, L.; Geurts van Kessel, A.; van Agthoven,A.; Bootsma, D.; Varmus, H.: Chromosomal localization of a humanhomologue of a putative mammary tumor oncogene. (Abstract) Cytogenet.Cell Genet. 37: 556–557, 1984.
- [46015] 19567.Pellegrino, J. E.; Lensch, M. W.; Muenke, M.; Chance, P. F.: Clinicaland molecular analysis in Joubert syndrome. Am. J. Med. Genet. 72:59–62, 1997.
- [46016] 19568.Thomas, K. R.; Capecchi, M. R.: Targeted disruption of the murineint–1 proto–oncogene resulting in severe abnormalities in midbrainand cerebellar development. Nature 346: 847–850, 1990.
- [46017] 19569.Braaten, D.; Wellington, S.; Warburton, D.; Luban, J.: Assignmentof cyclophilin A (PPIA) to human chromosome band 7p13 by in situ hybridization. Cytogenet.Cell Genet. 74: 262 only, 1996.
- [46018] 19570.Liu, J.; Albers, M. W.; Chen, C.–M.; Schreiber, S. L.; Walsh, C.T.: Cloning, expression, and purification of human cyclophilin inEscherichia coli and assessment of the catalytic role of cysteinesby site–directed mutagenesis. Proc. Nat. Acad. Sci. 87: 2304–2308,1990.
- [46019] 19571.Luban, J.; Bossolt, K. L.; Franke, E. K.; Kalpana, G. V.; Goff,S. P.: Human immunodeficiency virus type 1 gag

protein binds to cyclophilins A and B. *Cell* 73: 1067–1078, 1993.

[46020] 19572. Takahashi, N.; Hayano, T.; Suzuki, M.: Peptidyl-prolyl cis-trans isomerase is the cyclosporin A-binding protein cyclophilin. *Nature* 337:473–475, 1989.

[46021] 19573. Willenbrink, W.; Halaschek, J.; Schuffenhauer, S.; Kunz, J.; Steinkasserer, A.: Cyclophilin A, the major intracellular receptor for the immunosuppressant cyclosporin A, maps to chromosome 7p11.2–p13: four pseudogenes map to chromosomes 3, 10, 14, and 18. *Genomics* 28: 101–104, 1995.

[46022] 19574. Dickinson, D. P.; Zhao, Y.; Thiesse, M.; Siciliano, M. J.: Direct mapping of seven genes encoding human type 2 cystatins to a single site located at 20p11.2. *Genomics* 24: 172–175, 1994.

[46023] 19575. Saitoh, E.; Isemura, S.; Sanada, K.; Ohnishi, K.: The human cysteine gene family: cloning of three members and evolutionary relationship between cystatins and Bowman-Birk type proteinase inhibitors. *Biomed. Biochim. Acta* 50: 599–605, 1991.

[46024] 19576. Takahashi, E.; Hori, T.; O'Connell, P.; Leppert, M.; White, R.: R-banding and nonisotopic in situ hybridization: precise localization of the human type II collagen

gene (COL2A1). Hum. Genet. 86: 14–16,1990.

[46025] 19577.Takahashi, E.; Yamauchi, M.; Tsuji, H.; Hitomi, A.; Meuth, M.;Hori, T.: Chromosome mapping of the human cytidine–5–prime–triphosphatesynthetase (CTPS) gene to band 1p34.1–p34.3 by fluorescence in situhybridization. Hum. Genet. 88: 119–121, 1991.

[46026] 19578.Takahashi, E.–I.; Yamauchi, M.; Ayusawa, D.; Kaneda, S.; Seno,T.; Meuth, M.; Hori, T.–A.: Chromosome mappings of the human cytidine–5–prime–triphosphatesynthetase (CTPS) gene and the human ubiquitin–activating enzyme UBE1gene by fluorescence in situ hybridization. (Abstract) Cytogenet.Cell Genet. 58: 1864 only, 1991.

[46027] 19579.Thomas, P. E.; Sen, S.; Lamb, B. J.; Chu, E. H. Y.: Cloning andexpression of mammalian CTP synthetase genes. (Abstract) Am. J. Hum.Genet. 45 (suppl.): A11 only, 1989.

[46028] 19580.Whelan, J.; Phear, G.; Yamauchi, M.; Meuth, M.: Clustered basesubstitutions in CTP synthetase conferring drug resistance in Chinesehamster ovary cells. Nature Genet. 3: 317–322, 1993.

[46029] 19581.Yamauchi, M.; Takahashi, E.; Whelan, J.; Phear, G.; Meuth, M.:Mapping and functional analysis of the cytidine

triphosphate synthetase(CTPS) gene. (Abstract) Human Genome Mapping Workshop 93 1 only,1993.

- [46030] 19582.Nebert, D. W.; Gonzalez, F. J.: P450 genes: structure, evolution,and regulation. Annu. Rev. Biochem. 56: 945–993, 1987.
- [46031] 19583.Paolini, M.; Cantelli–Forti, G.; Perocco, P.; Pedulli, G. F.;Abdel–Rahman, S. Z.; Legator, M. S.: Co–carcinogenic effect of beta–carotene.(Letter) Nature 398: 760–761, 1999.
- [46032] 19584.Espinoza, H.; Cox, C. J.; Semina, E. V.; Amendt, B. A.: A molecularbasis for differential developmental anomalies in Axenfeld–Riegersyndrome. Hum. Molec. Genet. 11: 743–753, 2002.
- [46033] 19585.Leticic, K.; Zoncu, R.; Rakic, P.: Origin of GABAergic neuronsin the human neocortex. Nature 417: 645–649, 2002.
- [46034] 19586.McGuinness, T.; Porteus, M. H.; Smiga, S.; Bulfone, A.; Kingsley,C.; Qiu, M.; Liu, J. K.; Long, J. E.; Xu, D.; Rubenstein, J. L. R.: Sequence, organization, and transcription of the Dlx–1 and the Dlx–2locus. Genomics 35: 473–485, 1996.
- [46035] 19587.Ozcelik, T.; Porteus, M. H.; Rubenstein, J. L. R.; Francke, U.: DLX2 (TES1), a homeobox gene of the Distal–

less family, assigned to conserved regions on human and mouse chromosomes 2. *Genomics* 13:1157–1161, 1992.

[46036] 19588. Porteus, M. H.; Brice, A. E. J.; Bulfone, A.; Usdin, T. B.; Ciaranello, R. D.; Rubenstein, J. L. R.: Isolation and characterization of a library of cDNA clones that are preferentially expressed in the embryonic telencephalon.

Molec. Brain Res. 12: 7–22, 1992.

[46037] 19589. Qiu, M.; Bulfone, A.; Martinez, S.; Meneses, J. J.; Shimamura, K.; Pedersen, R. A.; Rubenstein, J. L.: Null mutation of *Dlx-2* results in abnormal morphogenesis of proximal first and second branchial arch derivatives and abnormal differentiation in the forebrain. *Genes Dev.* 9: 2523–2538, 1995.

[46038] 19590. Salvador, J. M.; Hollander, M. C.; Nguyen, A. T.; Kopp, J. B.; Barisoni, L.; Moore, J. K.; Ashwell, J. D.; Fornace, A. J., Jr.: Mice lacking the p53-effector gene *Gadd45a* develop a lupus-like syndrome. *Immunity* 16:499–508, 2002.

[46039] 19591. Takekawa, M.; Saito, H.: A family of stress-inducible GADD45-like proteins mediate activation of the stress-responsive MTK1/MEKK4 MAPKKK. *Cell* 95:521–530, 1998.

[46040] 19592. Dhermy, D.; Garbarz, M.; Lecomte, M. C.; Feo, C.;

Bournier, O.;Chaveroche, I.; Gautero, H.; Galand, C.;
Boivin, P.: Hereditary elliptocytosis:clinical, morphological
and biochemical studies of 38 cases. *Nouv.Rev. Franc.
Hemat.* 28: 129–140, 1986.

[46041] 19593.Fairbanks, G.; Steck, T. L.; Wallach, D. F. H.: Elec-
trophoreticanalysis of the major polypeptides of the hu-
man erythrocyte membrane. *Biochemistry* 10:2606–2617,
1971.

[46042] 19594.Feo, C. J.; Fischer, S.; Piau, J. P.; Grange, M. J.; Tch-
ernia,G.: Premiere observation de l'absence d'une proteine
de la membraneerythrocytaire (bande 4–1) dans un cas
d'anemie elliptocytaire familiale. *Nouv.Rev. Franc. Hemat.*
22: 315–325, 1980.

[46043] 19595.Eddy, R. L.; Mahnke–Zizelman, D. K.; Bausch–Ju-
rken, M. T.; Sabina,R. L.; Shows, T. B.: Distribution of the
AMP deaminase multigenefamily within the human
genome: assignment of the AMPD2 to chromo-
some1p21–p34 and AMPD3 to chromosome 11p13–pter.
(Abstract) *Human GenomeMapping Workshop* 93 24 only,
1993.

[46044] 19596.Ogasawara, N.; Goto, H.; Yamada, Y.; Nishigaki, I.;
Itoh, T.; Hasegawa,I.; Park, K. S.: Deficiency of AMP deam-
inase in erythrocytes. *Hum.Genet.* 75: 15–18, 1987.

- [46045] 19597.Sermsuvitayawong, K.; Wang, X.; Nagabukuro, A.; Matsuda, Y.; Morisaki,H.; Toyama, K.; Mukai, T.; Morisaki, T.: Genomic organization ofAmpd3, heart-type AMPD gene, located in mouse chromosome 7. Mam-malianGenome 8: 767-769, 1997.
- [46046] 19598.Yamada, Y.; Goto, H.; Ogasawara, N.: Cloning and nucleotide sequenceof the cDNA encoding human erythrocyte-specific AMP deaminase. Biochim.Biophys. Acta 1171: 125-128, 1992.
- [46047] 19599.Yamada, Y.; Goto, H.; Ogasawara, N.: A point mutation responsiblefor human erythrocyte AMP deaminase deficiency. Hum. Molec. Genet. 3:331-334, 1994.
- [46048] 19600.Allegrucci, C.; Liguori, L.; Mezzasoma, I.; Minelli, A.: A1 adenosinereceptor in human spermatozoa: its role in the fertilization process. Molec.Genet. Metab. 71: 381-386, 2000.
- [46049] 19601.Boman, H.; Hermodson, M.; Hammond, C. A.; Motulsky, A. G.: Analbuminemiain an American Indian girl. Clin. Genet. 9: 513-526, 1976.